Craniofacial Malformations

Basic distinction used in the BCSC Peds book

?   ?
Craniofacial Malformations

Basic distinction used in the BCSC Peds book

Craniosynostoses  Not Craniosynostoses
To what does the term craniosynostosis refer?
To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)
Cranial sutures of the newborn

Craniofacial Malformations
To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s) a thing?

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

Is failure of closing of cranial suture(s) a thing?
The opposite of craniosynostosis—ie, the failure of sutures to completely close—is that a thing?

What clinically significant finding might result?
The outpouching of CNS contents through the residual opening.

What would such an outpouching be called?
That depends upon its contents… --If the outpouching contains meninges, it’s called a meningocele. --If it contains brain tissue, it’s called an encephalocele. --If it contains both meninges and brain tissue, it’s a meningoencephalocele.
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s) a thing? Yes

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face.
Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

Is failure of craniosynostosis a thing? Yes

The opposite of craniosynostosis--ie, the failure of sutures to completely close--is that a thing?
Indeed it is

What clinically significant finding might result?
The outpouching of CNS contents through the residual opening
What would such an outpouching be called?
That depends upon its contents…
--If the outpouching contains meninges, it's called a meningocele
--If it contains brain tissue, it's called an encephalocele
--If it contains both meninges and brain tissue, it's a meningoencephalocele
To what does the term craniosynostosis refer? Yes

What results from premature suture closing? Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

The opposite of craniosynostosis—ie, the failure of sutures to completely close—is that a thing? Indeed it is.

What clinically significant finding might result? The outpouching of CNS contents through the residual opening...
Craniofacial Malformations

Craniosynostoses

To what does the term craniosynostosis refer?

To the premature closing of cranial suture(s) a thing? Yes

What results from premature suture closing?

Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

Is failure of closing of cranial suture(s) a thing? Yes

The opposite of craniosynostosis—ie, the failure of sutures to completely close—is that a thing? Indeed it is

What clinically significant finding might result?

The outpouching of CNS contents through the residual opening
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

To what does the term craniosynostosis refer? To the premature closing of cranial suture(s) a thing? Yes

What results from premature suture closing? Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

Is failure of closing of cranial suture(s) a thing? Yes

The opposite of craniosynostosis--ie, the failure of sutures to completely close--is that a thing? Indeed it is

What clinically significant finding might result? The outpouching of CNS contents through the residual opening

What would such an outpouching be called?
To what does the term craniosynostosis refer?

Is failure of closing of cranial suture(s) a thing? Yes

The opposite of craniosynostosis--ie, the failure of sutures to completely close--is that a thing?

Indeed it is

What clinically significant finding might result?

The outpouching of CNS contents through the residual opening

What would such an outpouching be called?

That depends upon its contents…

--If the outpouching contains meninges, it's called a meningocele.

--If it contains brain tissue, it's called an encephalocele.

--If it contains both meninges and brain tissue, it's a meningoencephalocele.
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s) a thing? Yes

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face.
Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

Is failure of closing of cranial suture(s) a thing? Yes

The opposite of craniosynostosis--ie, the failure of sutures to completely close--is that a thing?
Indeed it is

What clinically significant finding might result?
The outpouching of CNS contents through the residual opening

What would such an outpouching be called?
That depends upon its contents…
--If the outpouching contains meninges, it's called a meningocele
--If it contains brain tissue, it's called an encephalocele
--If it contains both meninges and brain tissue, it's a meningoencephalocele
Craniofacial Malformations

Encephalocele
If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?

- If the outpouching contains meninges, it's called a **meningocele**.
- If it contains brain tissue, it's called an **encephalocele**.
- If it contains both meninges and brain tissue, it's a **meningoencephalocele**.
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
At the medial canthus

---If the outpouching contains meninges, it's called a meningocoele
---If it contains brain tissue, it's called an encephalocele
---If it contains both meninges and brain tissue, it's a meningoencephalocele
If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located? At the medial canthus

Given this location, what entity is it likely to be mistaken for?

--If the outpouching contains meninges, it’s called a meningoccele
--If it contains brain tissue, it’s called an encephalocele
--If it contains both meninges and brain tissue, it’s a meningoencephalocele
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
At the medial canthus

Given this location, what entity is it likely to be mistaken for?
A dacryocele

--If the outpouching contains meninges, it's called a meningoecele
--If it contains brain tissue, it's called an encephalocele
--If it contains both meninges and brain tissue, it's a meningoencephalocele
Craniofacial Malformations

Nasal encephalocele

Dacryocele
If an -oceles comes to the attention of an ophthalmologist, where is it most likely to be located? At the medial canthus

Given this location, what entity is it likely to be mistaken for? A dacryocele

What is a dacryocele?

--If the outpouching contains meninges, it’s called a meningocoele
--If it contains brain tissue, it’s called an encephalocele
--If it contains both meninges and brain tissue, it’s a meningoencephalocele
Craniofacial Malformations

Craniosynostoses  Not Craniosynostoses

To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s) a thing? Yes

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

Is failure of the opposite of craniosynostosis--ie, the failure of sutures to completely close--a thing? Indeed it is

What clinically significant finding might result?
The outpouching of CNS contents through the residual opening

What would such an outpouching be called?
That depends upon its contents…
--If the outpouching contains meninges, it's called a meningoceles
--If it contains brain tissue, it's called an encephalocele
--If it contains both meninges and brain tissue, it's a meningoencephalocele

If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
At the medial canthus

Given this location, what entity is it likely to be mistaken for?
A dacryocele

What is a dacryocele?
A congenital swelling of the lacrimal sac
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s) a thing? Yes

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

Is failure of the opposite of craniosynostosis--ie, the failure of sutures to completely close--a thing? Indeed it is

What clinically significant finding might result?
The outpouching of CNS contents through the residual opening

What would such an outpouching be called?
That depends upon its contents…
--If the outpouching contains meninges, it's called a **meningocele**
--If it contains brain tissue, it's called an **encephalocele**
--If it contains both meninges and brain tissue, it's a **meningoencephalocele**

If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
At the medial canthus

Given this location, what entity is it likely to be mistaken for?
A dacryocele

To determine the nature of a nasal canthal cystic mass, what confirmatory test should be performed?
Fine needle biopsy, perhaps?

Um, no. Just no. Please don't stick a needle (or anything else) into what may be the brain.

OK then smart guy, what should be done to differentiate between the two?
Neuroimaging
Craniofacial Malformations

Craniosynostoses

If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
At the medial canthus

Given this location, what entity is it likely to be mistaken for?
A dacryocele

To determine the nature of a nasal canthal cystic mass, what confirmatory test should be performed?
Fine needle biopsy, perhaps?
Um, no. Just no. Please don’t stick a needle (or anything else) into what may be the brain.

Not Craniosynostoses

- If the outpouching contains meninges, it’s called a **meningocele**
- If it contains brain tissue, it’s called an **encephalocele**
- If it contains both meninges and brain tissue, it’s a **meningoencephalocele**
If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
At the medial canthus

Given this location, what entity is it likely to be mistaken for?
A dacryocoele

To determine the nature of a nasal canthal cystic mass, what confirmatory test should be performed?
Fine needle biopsy, perhaps?
Um, no. Just no. Please don’t stick a needle (or anything else) into what may be the brain.

OK then smart guy, what should be done to differentiate between the two?

--If the outpouching contains meninges, it’s called a **meningocele**
--If it contains brain tissue, it’s called an **encephalocele**
--If it contains both meninges and brain tissue, it’s a **meningoencephalocele**
If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
At the medial canthus

Given this location, what entity is it likely to be mistaken for?
A dacryocele

To determine the nature of a nasal canthal cystic mass, what confirmatory test should be performed?
Fine needle biopsy, perhaps?
Um, no. Just no. Please don’t stick a needle (or anything else) into what may be the brain.

OK then smart guy, what should be done to differentiate between the two?
Neuroimaging

--If the outpouching contains meninges, it’s called a **meningocele**
--If it contains brain tissue, it’s called an **encephalocele**
--If it contains both meninges and brain tissue, it’s a **meningoencephalocele**
If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located? At the medial canthus

Given this location, what entity is it likely to be mistaken for? A dacryocele

To determine the nature of a nasal canthal cystic mass, what confirmatory test should be performed? Fine needle biopsy, perhaps?

Um, no. Just no. Please don't stick a needle (or anything else) into what may be the brain.

Are there clinical signs that can differentiate between the two?

OK then smart guy, what should be done to differentiate between the two?

Neuroimaging

--If the outpouching contains meninges, it's called a meningocele
--If it contains brain tissue, it's called an encephalocele
--If it contains both meninges and brain tissue, it's a meningoencephalocele
### Craniofacial Malformations

#### Craniosynostoses

To what does the term **craniosynostosis** refer?

To the premature closing of cranial suture(s) a thing? Yes

What results from premature suture closing?

Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

#### Not Craniosynostoses

Is failure of the opposite of craniosynostosis--ie, the failure of sutures to completely close--a thing? Indeed it is

What clinically significant finding might result?

The outpouching of CNS contents through the residual opening

What would such an outpouching be called?

That depends upon its contents…

--If the outpouching contains meninges, it's called a **meningocele**

--If it contains brain tissue, it's called an **encephalocele**

--If it contains both meninges and brain tissue, it's a **meningoencephalocele**

If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?

At the medial canthus

Given this location, what entity is it likely to be mistaken for?

A dacryocle

To determine the nature of a nasal canthal cystic mass, what confirmatory test should be performed?

*Fine needle biopsy, perhaps?*

Um, no. Just no. Please don’t stick a needle (or anything else) into what may be the brain.

OK then smart guy, what **should** be done to *differentiate between the two?*

*Neuroimaging*

Are there clinical signs that can

--- If the -ocele is pulsatile?

--- If it contains meninges?

--- If it contains brain tissue?

--- If it contains both meninges and brain tissue?

<table>
<thead>
<tr>
<th>Relation to medial canthal tendon</th>
<th>Dacryocle</th>
<th>Meningoencephalocele</th>
</tr>
</thead>
<tbody>
<tr>
<td>?</td>
<td></td>
<td>?</td>
</tr>
</tbody>
</table>

---

---
If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
At the medial canthus

Given this location, what entity is it likely to be mistaken for?
A dacryocoele

To determine the nature of a nasal canthal cystic mass, what confirmatory test should be performed?
Fine needle biopsy, perhaps?
Um, no. Just no. Please don’t stick a needle (or anything else) into what may be the brain.

Are there clinical signs that can differentiate between the two? Yes

<table>
<thead>
<tr>
<th>Relation to medial canthal tendon</th>
<th>Dacryocoele</th>
<th>Mening/enceph-ocele</th>
</tr>
</thead>
<tbody>
<tr>
<td>Below it</td>
<td>Above it</td>
<td></td>
</tr>
</tbody>
</table>
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
At the medial canthus

Given this location, what entity is it likely to be mistaken for?
A dacrocele

To determine the nature of a nasal canthal cystic mass, what confirmatory test should be performed?
Fine needle biopsy, perhaps?
Um, no. Just no. Please don’t stick a needle (or anything else) into what may be the brain.

OK then smart guy, what should be done to differentiate between the two?
Neuroimaging

Are there clinical signs that can differentiate between the two? Yes

<table>
<thead>
<tr>
<th>Relation to medial canthal tendon</th>
<th>Dacrocele</th>
<th>Mening/enceph-ocele</th>
</tr>
</thead>
<tbody>
<tr>
<td>Below it</td>
<td>Above it</td>
<td></td>
</tr>
</tbody>
</table>

Pulsatile?

? ?
Craniofacial Malformations

Craniosynostoses
Not Craniosynostoses

If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
At the medial canthus

Given this location, what entity is it likely to be mistaken for?
A dacryocele

To determine the nature of a nasal canthal cystic mass, what confirmatory test should be performed?
Fine needle biopsy, perhaps?
Um, no. Just no. Please don’t stick a needle (or anything else) into what may be the brain.

Are there clinical signs that can differentiate between the two? Yes

<table>
<thead>
<tr>
<th>Relation to medial canthal tendon</th>
<th>Dacryocele</th>
<th>Mening/enceph-ocele</th>
</tr>
</thead>
<tbody>
<tr>
<td>Below it</td>
<td>Above it</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Pulsatile?</th>
<th>No</th>
<th>Yes</th>
</tr>
</thead>
</table>
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
At the medial canthus

Given this location, what entity is it likely to be mistaken for?
A dacryocele

To determine the nature of a nasal canthal cystic mass, what confirmatory test should be performed?
Fine needle biopsy, perhaps?
Um, no. Just no. Please don’t stick a needle (or anything else) into what may be the brain.

Are there clinical signs that can help determine the nature of the mass?
OK then smart guy, what should be done to differentiate between the two? Yes

Neuroimaging

<table>
<thead>
<tr>
<th>Relation to medial canthal tendon</th>
<th>Dacryocele</th>
<th>Mening/enceph-ocele</th>
</tr>
</thead>
<tbody>
<tr>
<td>Below</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pulsatile?</td>
<td>No</td>
<td>Yes</td>
</tr>
</tbody>
</table>

What causes a mening/enceph-ocele to pulsate?

Direct transmission of the intracranial pulse pressure
If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located? At the medial canthus

Given this location, what entity is it likely to be mistaken for? A dacryocele

To determine the nature of a nasal canthal cystic mass, what confirmatory test should be performed? Fine needle biopsy, perhaps? Um, no. Just no. Please don’t stick a needle (or anything else) into what may be the brain.

Are there clinical signs that can differentiate between the two? Yes Neuroimaging

<table>
<thead>
<tr>
<th>Relation to medial canthal tendon</th>
<th>Dacryocele</th>
<th>Mening/enceph-ocele</th>
</tr>
</thead>
<tbody>
<tr>
<td>If the mass is above the tendon</td>
<td>Bettei</td>
<td>What causes a mening/encephocele to pulsate? Direct transmission of the intracranial pulse pressure</td>
</tr>
<tr>
<td>If the mass is below the tendon</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pulsatile?</td>
<td>No</td>
<td>Yes</td>
</tr>
</tbody>
</table>

What causes a mening/enceph-ocele to pulsate? Direct transmission of the intracranial pulse pressure
Craniofacial Malformations

Craniosynostoses

To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
It can be an isolated/sporadic finding, but is more often syndromic
To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
It can be an isolated/sporadic finding, but is more often syndromic

In addition to the skull/face, what other bodypart(s) is/are often involved in the syndromic craniosynostoses?
Craniofacial Malformations

Craniosynostoses

To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
It can be an isolated/sporadic finding, but is more often syndromic

In addition to the skull/face, what other bodypart(s) is/are often involved in the syndromic craniosynostoses?
The hands and feet
To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
It can be an isolated/sporadic finding, but is more often syndromic

What two specific abnormalities of the hands/feet are characteristic of the craniosynostosis syndromes?
Hands and feet
To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
It can be an isolated/sporadic finding, but is more often syndromic

In addition to the skull/face, what other bodypart(s) is/are often involved in the syndromic craniosynostoses?
The hands and feet

What two specific abnormalities of the hands/feet are characteristic of the craniosynostosis syndromes?
Syndactyly and brachydactyly
**Craniofacial Malformations**

**Craniosynostoses**

*To what does the term craniosynostosis refer?*
To the premature closing of cranial suture(s)

*What results from premature suture closing?*
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

*How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?*
It can be an isolated/sporadic finding, but is more often syndromic

In addition to the skull/face, what other body part(s) is/are often involved in the syndromic craniosynostoses? The hands and feet

*What two specific abnormalities of the hands/feet are characteristic of the craniosynostosis syndromes?*
Syndactyly and brachydactyly

*What is syndactyly?*
To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
It can be an isolated/sporadic finding, but is more often syndromic

In addition to the skull/face, what other bodypart(s) is/are often involved in the syndromic craniosynostoses?
The hands and feet

What two specific abnormalities of the hands/feet are characteristic of the craniosynostosis syndromes?
Syndactyly and brachydactyly

What is syndactyly?
Partial fusion of the digits
Craniofacial Malformations

Craniosynostoses

To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
It can be an isolated/sporadic finding, but is more often syndromic

In addition to the skull/face, what other bodypart(s) is/are often involved in the syndromic craniosynostoses?
The hands and feet

What two specific abnormalities of the hands/feet are characteristic of the craniosynostosis syndromes?
Syndactyly and brachydactyly

What is syndactyly?
Partial fusion of the digits

What is brachydactyly?
To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

How does craniosynostosis present (i.e., isolated/sporadic; syndromic; etc)?
It can be an isolated/sporadic finding, but is more often syndromic

In addition to the skull/face, what other body part(s) is/are often involved in the syndromic craniosynostoses?
The hands and feet

What two specific abnormalities of the hands/feet are characteristic of the craniosynostosis syndromes?
Syndactyly and brachydactyly

What is syndactyly?
Partial fusion of the digits

What is brachydactyly?
Abnormally short digits
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
It can be an isolated/sporadic finding, but is more often syndromic

In addition to the skull/face, what other bodypart(s) is/are often involved in the syndromic craniosynostoses?
The hands and feet

There are four classic craniofacial synostosis syndromes. What are they?
Craniofacial Malformations

Craniosynostoses

To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)

What results from premature suture closing?
Premature closure produces abnormal growth patterns of the skull and face. Depending upon which suture(s) closes prematurely, specific and well-recognized patterns of craniofacial malformation result.

How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
It can be an isolated/sporadic finding, but is more often syndromic

In addition to the skull/face, what other bodypart(s) is/are often involved in the syndromic craniosynostoses?
The hands and feet

There are four classic craniofacial synostosis syndromes. What are they?
Coming in hot…
Craniofacial Malformations

Craniosynostoses

- ?
- ?
- ?
- ?

Not Craniosynostoses

Most ophtho residents can name two of these, if not all three

You a **baller** if you can name this one
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Most ophthalmic residents can name two of these, if not all three

You a **baller** if you can name this one
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- **Saethre-Chotzen syndrome**

Not Craniosynostoses

*How is this pronounced?*
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

How is this pronounced? SAY-three CHOT-zen
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

All four have the same inheritance pattern--what is it?
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

All four have the same inheritance pattern--what is it?
AD
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies--which ones?
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies--which ones?
Craniofacial Malformations

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies - which ones?

What orbital features are common to all three conditions?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies— which ones?

What orbital features are common to all three conditions?
Proptosis and hypertelorism
Craniofacial Malformations

Crouzon syndrome: Proptosis
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies--which ones?

What orbital features are common to all three conditions? Proptosis and hypertelorism

Why do they have proptosis, ie what is the cause?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies—which ones?

What orbital features are common to all three conditions? Proptosis and hypertelorism

Why do they have proptosis, i.e., what is the cause?
The orbits are abnormally shallow
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Three have similar facies—three of which?

What orbital features are common to all three conditions?

- Proptosis
- Hypertelorism

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies — which ones?

What orbital features are common to all three conditions? Proptosis and hypertelorism

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

What orbital features are common to all three conditions?

- Proptosis
- Hypertelorism

Why do they have proptosis, ie what is the cause?

- The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?

- Exposure keratopathy

Is the proptosis accompanied by lid retraction?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies— which ones?

What orbital features are common to all three conditions? Proptosis and hypertelorism

Why do they have proptosis, i.e., what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No— if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)
Craniofacial Malformations

Crouzon syndrome: Ptosis + inferior scleral show
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies—-which ones?

What orbital features are common to all three conditions?
- Proptosis
- Hypertelorism

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Three have similar facies

Not Craniosynostoses

What orbital features are common to all three conditions?

- Proptosis
- Hypertelorism

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies—which ones?

What orbital features are common to all three conditions?

- Proptosis
- Hypertelorism

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What is hypertelorism?
An abnormally large distance between the medial orbital walls

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies

What orbital features are common to all three conditions?

Proptosis and hypertelorism

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What is hypertelorism?
An abnormally large distance between the medial orbital walls

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance, ie, the PD
Craniofacial Malformations

In addition to an increased interpupillary distance, hypertelorism will result in an increased distance between what other orbital structures?

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What is hypertelorism?
An abnormally large distance between the medial orbital walls

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance

Hypertelorism always leads to telecanthus, but is telecanthus always associated with hypertelorism?
No, telecanthus can present as a soft-tissue abnormality absent hypertelorism (it is called primary telecanthus when it occurs in the absence of hypertelorism, and secondary telecanthus when occurring with it)

Yes
Craniofacial Malformations

In addition to an increased interpupillary distance, hypertelorism will result in an increased distance between what other orbital structures?
The medial canthi

What orbital features are common to all three conditions?
Proptosis and hypertelorism

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What is hypertelorism?
An abnormally large distance between the medial orbital walls

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance

Hypertelorism always leads to telecanthus, but is telecanthus always associated with hypertelorism?
No, telecanthus can present as a soft-tissue abnormality absent hypertelorism (it is called primary telecanthus when it occurs in the absence of hypertelorism, and secondary telecanthus when occurring with it)

Is telecanthus a common occurrence in the syndromic craniosynostoses?
Yes
In addition to an increased interpupillary distance, hypertelorism will result in an increased distance between what other orbital structures?
The medial canthi

What is the name for an abnormally increased distance between the medial canthi?

What orbital features are common to all three conditions? Proptosis and hypertelorism

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What is hypertelorism?
An abnormally large distance between the medial orbital walls

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance
Craniofacial Malformations

In addition to an increased interpupillary distance, hypertelorism will result in an increased distance between what other orbital structures?
The medial canthi

What is the name for an abnormally increased distance between the medial canthi?
Telecanthus

What orbital features are common to all three conditions? 
Proptosis and hypertelorism

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What is hypertelorism?
An abnormally large distance between the medial orbital walls

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance

Hypertelorism always leads to telecanthus, but is telecanthus always associated with hypertelorism?
No, telecanthus can present as a soft-tissue abnormality absent hypertelorism (it is called primary telecanthus when it occurs in the absence of hypertelorism, and secondary telecanthus when occurring with it)

Yes
Craniofacial Malformations

**Craniosynostoses Not Craniosynostoses**

Crouzon syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

Three have similar facies—which ones?

What orbital features are common to all three conditions?

- **Proptosis**
- Hypertelorism

What is **hypertelorism**?

An abnormally large distance between the medial orbital walls

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?

**The interpupillary distance**

*Why do they have proptosis, ie what is the cause?*

The orbits are abnormally shallow

*What serious sequelae can result from the proptosis/shallow orbits?*

Exposure keratopathy

*Is the proptosis accompanied by lid retraction?*

No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

*What is the name for an abnormally increased distance between the medial canthi?*

**Telecanthus**

**Hypertelorism always leads to telecanthus, but is telecanthus always associated with hypertelorism?**

No, telecanthus can present as a soft-tissue abnormality absent hypertelorism (it is called **primary telecanthus** when it occurs in the absence of hypertelorism, and **secondary telecanthus** when occurring with it)

*Is telecanthus a common occurrence in the syndromic craniosynostoses?*

Yes
In addition to an increased interpupillary distance, hypertelorism will result in an increased distance between what other orbital structures?
The medial canthi

What is the name for an abnormally increased distance between the medial canthi?
**Telecanthus**

Hyptelorism always leads to telecanthus, but is telecanthus always associated with hypertelorism?
No, telecanthus can present as a soft-tissue abnormality absent hypertelorism (it is called primary telecanthus when it occurs in the absence of hypertelorism, and secondary telecanthus when occurring with it)

What orbital features are common to all three conditions?
**Proptosis and hypertelorism**

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What is hypertelorism?
An abnormally large distance between the medial orbital walls

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance

The interpupillary distance
Craniofacial Malformations

In addition to an increased interpupillary distance, hypertelorism will result in an increased distance between what other orbital structures?
The medial canthi

What is the name for an abnormally increased distance between the medial canthi?
Telecanthus

Hypertelorism always leads to telecanthus, but is telecanthus always associated with hypertelorism?
No, telecanthus can present as a soft-tissue abnormality absent hypertelorism (it is called primary telecanthus when it occurs in the absence of hypertelorism, and secondary telecanthus when occurring with it)

Is telecanthus a characteristic feature of Crouzon, Apert, and Pfeiffer syndromes?

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What is hypertelorism?
An abnormally large distance between the medial orbital walls

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance

What orbital features are common to all three conditions?
Proptosis and hypertelorism and telecanthus?
Craniofacial Malformations

In addition to an increased interpupillary distance, hypertelorism will result in an increased distance between what other orbital structures?
The medial canthi

What is the name for an abnormally increased distance between the medial canthi?
Telecanthus

Hypertelorism always leads to telecanthus, but is telecanthus always associated with hypertelorism?
No, telecanthus can present as a soft-tissue abnormality absent hypertelorism (it is called primary telecanthus when it occurs in the absence of hypertelorism, and secondary telecanthus when occurring with it)

Is telecanthus a characteristic feature of Crouzon, Apert, and Pfeiffer syndromes?
Yes

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What is hypertelorism?
An abnormally large distance between the medial orbital walls

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)
Craniofacial Malformations

Craniosynostoses
- **Crouzon syndrome**
- **Apert syndrome**
- **Pfeiffer syndrome**
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies— which ones? Proptosis and hypertelorism

What orbital features are common to all three conditions?

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

Why is hypertelorism? An abnormally large distance between the medial orbital walls. What easily obtainable measurement is used as a proxy? The interpupillary distance, ie, the PD

Is the proptosis accompanied by lid retraction? No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these?

What serious sequelae can result from the proptosis/shallow orbits? Exposure keratopathy

What is hypertelorism? An abnormally large distance between the medial orbital walls. What easily obtainable measurement is used as a proxy? The interpupillary distance, ie, the PD
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome

Not Craniosynostoses

Three have similar facies— which ones?

What orbital features are common to all three conditions?

- Proptosis
- Hypertelorism

Why do they have proptosis, i.e., what is the cause?
The orbits are abnormally shallow.

What is hypertelorism? An abnormally large distance between the medial orbital walls.

What serious sequelae can result from the proptosis/shallow orbits?

Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No— if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon).

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these?

They are intorted and diverge excessively from one another.

The interpupillary distance, i.e., the PD is used as a proxy.
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies—which ones?

What orbital features are common to all three conditions?

- Proptosis
- Hypertelorism

Why do they have proptosis, i.e., what is the cause?
The orbits are abnormally shallow

What is hypertelorism?
An abnormally large distance between the medial orbital walls

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No—if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these?
They are extorted, and diverge excessively from one another

The interpupillary distance, i.e., the PD
Craniofacial Malformations

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence?

Craniosynostoses Not Craniosynostoses

Crouzon syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

What orbital features are common to all three conditions?

- Proptosis
- Hypertelorism
- Extreme divergence

Why do they have proptosis, ie what is the cause?

The orbits are abnormally shallow

What is hypertelorism?

An abnormally large distance between the medial orbital walls

What serious sequelae can result from the proptosis/shallow orbits?

- Exposure keratopathy

Is the proptosis accompanied by lid retraction?

No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What easily obtainable measurement is used as a proxy?

The interpupillary distance, ie, the PD

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence?

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these?

- They are extorted and diverge excessively from one another

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence?

Because the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

Elevation during adduction…What relatively common strabismus-related problem does that sound like?

- Inferior oblique overaction

It sounds like pseudo-IO overaction.

An abnormally large distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?

The interpupillary distance, ie, the PD
Craniofacial Malformations

**OK, the orbits are extorted. Does this lead to clinical sequelae of consequence?**

Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

---

**Proptosis and hypertelorism**

What orbital features are common to all three conditions?

Why do they have proptosis, ie what is the cause?

The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?

Exposure keratopathy

Is the proptosis accompanied by lid retraction?

No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What is hypertelorism?

An abnormally large distance between the medial orbital walls.

What easily obtainable measurement is used as a proxy?

The interpupillary distance, ie, the PD

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these?

**Extorted** and telecanthus and...extorted, excessively divergent

---

**The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these?**

They are extorted and diverge excessively from one another.
Craniofacial Malformations

**OK, the orbits are extorted. Does this lead to clinical sequelae of consequence?**
Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

Elevation during adduction…What relatively common strabismus-related problem does that sound like?

It sounds like inferior oblique overaction—but it's not. Rather, it is pseudo-I0 overaction.

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these?
They are **extorted**, and diverge excessively from one another.

What orbital features are common to all three conditions?
**Proptosis** and **hypertelorism** and telecanthus and... **extorted**, excessively divergent.

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow.

What is hypertelorism?
An abnormally large distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance, ie, the PD.
Craniofacial Malformations

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

Elevation during adduction…What relatively common strabismus-related problem does that sound like?
It sounds like inferior oblique overaction--but it's not. Rather, it is pseudo-IO overaction.

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these?
They are extorted, and diverge excessively from one another.

What is proptosis? The orbits are abnormally shallow.
Why do they have proptosis, ie what is the cause? The orbits are abnormally shallow.
What is hypertelorism? An abnormally large distance between the medial orbital walls. What easily obtainable measurement is used as a proxy? The interpupillary distance, ie, the PD.

Is the proptosis accompanied by lid retraction? No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon).
Craniofacial Malformations

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

Elevation during adduction…What relatively common strabismus-related problem does that sound like?
It sounds like **inferior oblique overaction**--but it’s not. Rather, it is *pseudo*-IO overaction.

What determines the amount of divergence between the orbits?

What orbital features are common to all three conditions?
Three have similar facies--which ones?

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

The interpupillary distance, ie, the PD

What is hypertelorism?
An abnormally large distance between the medial orbital walls. Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance, ie, the PD
Craniofacial Malformations

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

What orbital features are common to all three conditions? Proptosis and hypertelorism. What is hypertelorism? An abnormally large distance between the medial orbital walls. Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy? The interpupillary distance, ie, the PD.

What determines the amount of divergence between the orbits? The angle formed by the lateral orbital walls with respect to one another.

What serious sequelae can result from the proptosis/shallow orbits? Exposure keratopathy. Is the proptosis accompanied by lid retraction? No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon).

What is hypertelorism? An abnormally large distance between the medial orbital walls.

What does elevation during adduction sound like? Inferior oblique overaction. It sounds like inferior oblique overaction--but it's not. Rather, it is pseudo-IO overaction.

What determines the amount of divergence between the orbits? The angle formed by the lateral orbital walls with respect to one another.

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these? They are extorted, and diverge excessively from one another.

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

What orbital features are common to all three conditions? Proptosis and hypertelorism. What is hypertelorism? An abnormally large distance between the medial orbital walls. Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy? The interpupillary distance, ie, the PD.

What determines the amount of divergence between the orbits? The angle formed by the lateral orbital walls with respect to one another.

What serious sequelae can result from the proptosis/shallow orbits? Exposure keratopathy. Is the proptosis accompanied by lid retraction? No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon).

What is hypertelorism? An abnormally large distance between the medial orbital walls. What does elevation during adduction sound like? Inferior oblique overaction. It sounds like inferior oblique overaction--but it's not. Rather, it is pseudo-IO overaction.

What determines the amount of divergence between the orbits? The angle formed by the lateral orbital walls with respect to one another.

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these? They are extorted, and diverge excessively from one another.
Craniofacial Malformations

Craniosynostoses Not Craniosynostoses

Crouzon syndrome
Apert syndrome
Pfeiffer syndrome
Saethre-Chotzen syndrome

Three have similar facies—which ones?

What orbital features are common to all three conditions?

Proptosis and hypertelorism

What is hypertelorism?

An abnormally large distance between the medial orbital walls.

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?

The interpupillary distance, ie, the PD

Why do they have proptosis, ie what is the cause?

The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?

Exposure keratopathy

Is the proptosis accompanied by lid retraction?

No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these?

They are extorted, and diverge excessively

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence?

Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

Elevation during adduction…What relatively common strabismus-related problem does that sound like?

It sounds like inferior oblique overaction--but it’s not. Rather, it is pseudo-IO overaction.

What determines the amount of divergence between the orbits?

The angle formed by the lateral orbital walls with respect to one another

What is the normal angle formed by these walls?

Ninety degrees

What is it in the Crouzon-facies craniosynostoses?

It varies, but is often well above 90°
Craniofacial Malformations

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

Elevation during adduction...What relatively common strabismus-related problem does that sound like? It sounds like inferior oblique overaction—but it’s not. Rather, it is pseudo-IO overaction.

What determines the amount of divergence between the orbits? The angle formed by the lateral orbital walls with respect to one another.

What is the normal angle formed by these walls? Ninety degrees.

What serious sequelae can result from the proptosis/shallow orbits? Exposure keratopathy.

Is the proptosis accompanied by lid retraction? No—if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon).

What orbital features are common to all three conditions? Proptosis and hypertelorism.

What is hypertelorism? An abnormally large distance between the medial orbital walls. Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy? The interpupillary distance, ie, the PD.

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these? They are extorted, and diverge excessively from one another.

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.
Craniofacial Malformations

Normal orbital divergence
Craniofacial Malformations

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

Elevation during adduction…What relatively common strabismus-related problem does that sound like?

It sounds like inferior oblique overaction--but it’s not. Rather, it is pseudo-IO overaction.

What determines the amount of divergence between the orbits?
The angle formed by the lateral orbital walls with respect to one another

What is the normal angle formed by these walls?
Ninety degrees

What is it in the Crouzon-facies craniosynostoses?

They are extorted, and diverge excessively

What serious sequelae can result from the proptosis/shallow orbits?

Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What orbital features are common to all three conditions?

Proptosis and hypertelorism

What is hypertelorism?

An abnormally large distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?

The interpupillary distance, ie, the PD

What is the angle formed by the lateral orbital walls with respect to one another?

Ninety degrees

What is it in the Crouzon-facies craniosynostoses?

It varies, but is often well above 90
Craniofacial Malformations

Craniosynostoses Not Craniosynostoses

Crouzon syndrome
Apert syndrome
Pfeiffer syndrome

Three have similar facies--which ones?

What orbital features are common to all three conditions?
Proptosis and hypertelorism

What is hypertelorism?
An abnormally large distance between the medial orbital walls.

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance, ie, the PD

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?
Exposure keratopathy

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these?
They are extorted, and diverge excessively from one another

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbitalis are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

Elevation during adduction…What relatively common strabismus-related problem does that sound like?
It sounds like inferior oblique overaction--but it’s not. Rather, it is pseudo-IO overaction.

What determines the amount of divergence between the orbits?
The angle formed by the lateral orbital walls with respect to one another

What is the normal angle formed by these walls?
Ninety degrees

What is it in the Crouzon-facies craniosynostoses?
It varies, but is often well above 90
Craniofacial Malformations

Normal orbital divergence

Excessive orbital divergence in Crouzon
Craniofacial Malformations

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

Elevation during adduction…What relatively common strabismus-related problem does that sound like?

It sounds like inferior oblique overaction--but it’s not. Rather, it is pseudo-IO overaction.

What determines the amount of divergence between the orbits? The angle formed by the lateral orbital walls with respect to one another.

As expected, the myriad orbital problems lead to disordered movement of the globes. In this regard, what sort of strabismus pattern is typical of the synostoses that present with the Crouzon-type facies? They are extorted, and diverge excessively from one another.

Is the proptosis accompanied by lid retraction? No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon).

What orbital features are common to all three conditions? Proptosis and...extorted, excessively divergent.

What is hypertelorism?

An abnormally large distance between the medial orbital walls. Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?

The interpupillary distance, ie, the PD.
**Craniofacial Malformations**

**Craniosynostoses and Not Craniosynostoses**

Crouzon syndrome

Apert syndrome

Pfeiffer syndrome

Three have similar facies — which ones?

What orbital features are common to all three conditions?

Proptosis and hypertelorism

What is hypertelorism?

An abnormally large distance between the medial orbital walls.

Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?

The interpupillary distance, ie, the PD

Why do they have proptosis, ie, what is the cause?

The orbits are abnormally shallow

What serious sequelae can result from the proptosis/shallow orbits?

Exposure keratopathy

Is the proptosis accompanied by lid retraction?

No — if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What?

They are extorted, and diverge excessively from one another

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence?

Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

Elevation during adduction...What relatively common strabismus-related problem does that sound like?

It sounds like inferior oblique overaction -- but it’s not. Rather, it is pseudo-IO overaction.

What determines the amount of divergence between the orbits?

The angle formed by the lateral orbital walls with respect to one another

As expected, the myriad orbital problems lead to disordered movement of the globes. In this regard, what sort of strabismus pattern is typical of the synostoses that present with the Crouzon-type facies?

Well, the excessive divergence produces... word, and the extortion causes the... word to be worse in... direction of gaze

Putting it all together, we can see that these pts have a V-pattern exotropia.

Is the proptosis accompanied by lid retraction?

No -- if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What easily obtainable measurement is used as a proxy?

The interpupillary distance, ie, the PD
Craniofacial Malformations

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbital walls are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

What are the orbital features common to all three conditions? Proptosis and hypertelorism.

What is hypertelorism? An abnormally large distance between the medial orbital walls. Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy? The interpupillary distance, ie, the PD.

What determines the amount of divergence between the orbits? The angle formed by the lateral orbital walls with respect to one another.

As expected, the myriad orbital problems lead to disordered movement of the globes. In this regard, what sort of strabismus pattern is typical of the synostoses that present with the Crouzon-type facies? Well, the excessive divergence produces exotropia, and the extortion causes the exotropia to be worse in upgaze.

Is the proptosis accompanied by lid retraction? No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon).

What is it in the Crouzon-facies craniosynostoses? It varies, but is often well above 90°.

What is it in the other conditions? Ninety degrees.

Putting it all together, we can see that these pts have a V-pattern exotropia.
Craniofacial Malformations

Crouzon syndrome: Exotropia
Craniofacial Malformations

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.

Elevation during adduction…What relatively common strabismus-related problem does that sound like?

It sounds like inferior oblique overaction—but it’s not. Rather, it is pseudo-IO overaction.

What determines the amount of divergence between the orbits?
The angle formed by the lateral orbital walls with respect to one another.

As expected, the myriad orbital problems lead to disordered movement of the globes. In this regard, what sort of strabismus pattern is typical of the synostoses that present with the Crouzon-type facies? Well, the excessive divergence produces exotropia, and the extorsion causes the exotropia to be worse in upgaze. Putting it all together, we can see that these pts have a specific type of exotropia.

They are extorted, and diverge excessively from one another.

Is the proptosis accompanied by lid retraction?
No—if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

What orbital features are common to all three conditions?
Proptosis and...extorted, excessively divergent

What is hypertelorism?
An abnormally large distance between the medial orbital walls. Absent imaging, it is impossible to directly measure the distance between the medial orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance, ie, the PD
**Craniofacial Malformations**

*OK, the orbits are extorted. Does this lead to clinical sequelae of consequence? Indeed it does. Consider: Because the orbitals are extorted, the positions of the rectus muscles are extorted as well. As an example, take the medial recti. Because of the orbital extorsion, these are located superonasally rather than medially. Thus, when the MR contract, the eyes will elevate in addition to adducting.*

*Elevation during adduction…What relatively common strabismus-related problem does that sound like? It sounds like **in inferior oblique overaction**--but it’s not. Rather, it is **pseudo-IO overaction**.*

*What determines the amount of divergence between the orbits? The angle formed by the lateral orbital walls with respect to one another.*

*As expected, the myriad orbital problems lead to disordered movement of the globes. In this regard, what sort of strabismus pattern is typical of the synostoses that present with the Crouzon-type facies? Well, the excessive divergence produces **exotropia**, and the extortion causes the **exotropia** to be worse in **upgaze**. Putting it all together, we can see that these pts have a **V-pattern exotropia**.*

*They are extorted, and diverge excessively from one another.*
Craniofacial Malformations

Crouzon syndrome: V-pattern exotropia
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies--which ones?

What orbital features are common to all three conditions?
- Proptosis
- Hypertelorism
- Telecanthus

Why do they have proptosis, ie what is the cause?
The orbits are abnormally shallow

What is hypertelorism?
An abnormally large distance between the medial orbital walls

Is the proptosis accompanied by lid retraction?
No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

Speaking of putting it all together…There is a word--admittedly, ill-defined--that serves as an umbrella term for the orbital abnormalities found in the craniosynostoses with Crouzon-type facies. What is it?

The interpupillary distance, ie, the PD
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

Three have similar facies--which ones?

What orbital features are common to all three conditions?

Proptosis and hypertelorism

Why do they have proptosis, i.e., what is the cause?

The orbits are abnormally shallow

What is hypertelorism?

An abnormally large distance between the medial orbital walls

Is the proptosis accompanied by lid retraction?

No--if anything, ptosis may be present (although inferior scleral show is characteristic of Crouzon)

Speaking of putting it all together... There is a word—admittedly, ill-defined—that serves as an umbrella term for the orbital abnormalities found in the craniosynostoses with Crouzon-type facies. What is it?

‘Exorbitism’

The interpupillary distance, i.e., the PD
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- **Saethre-Chotzen syndrome**

Not Craniosynostoses

*What facial features characterize Saethre-Chotzen syndrome (SCS)?*
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- **Saethre-Chotzen syndrome**

*What facial features characterize Saethre-Chotzen syndrome (SCS)?*  
Firstly, it’s worth reiterating what features **don’t** characterize it, ie, it does **not** present with the exorbitism which is the hallmark of the Crouzon-type craniosynostoses. Rather, SCS facies are more subtle--facial asymmetry, flat forehead, ptosis and ear abnormalities are the rule.
Craniofacial Malformations

Saethre-Chotzen syndrome
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings:
- Syndactyly
- Brachydactyly

two terms mentioned previously
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings: Syndactyly and/or brachydactyly
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings: Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in… Apert syndrome?
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings: Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in… Apert syndrome? Apert is known for severe
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- **Apert syndrome**
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings:
Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests.

In that regard, how do they tend to manifest in…

Apert syndrome? Apert is known for severe **syndactyly**
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings: Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…

Apert syndrome? Apert is known for severe syndactyly, which typically involves complete fusion of the index through pinky fingers--only the thumb is free. Likewise, most or all of the toes will be fused as well.
Craniofacial Malformations

Apert syndrome
Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…

Apert syndrome? **Apert is known for severe syndactyly**, which typically involves complete fusion of the index through pinky fingers, only the thumb is free. Likewise, most or all of the toes will be syndactylous.

*Got a mnemonic for this important factoid?*
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings:
Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…

Apert syndrome? **Apert is known for severe syndactyly**, which typically involves complete fusion of the index through pinky fingers; only the thumb is free. Likewise, most or all of the toes will be syndactylized.

Got a mnemonic for this important factoid? Pts with Apert syndrome can't pull their fingers ‘apert’ (ie, ‘apart’).
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- **Apert syndrome**
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings: Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in...

**Apert syndrome**? Apert is known for severe **syndactyly**, which typically involves complete fusion of the index through pinky fingers--only the thumb is free. Likewise, most or all of the toes will be fused as well.

_Fingers all together, the thumb separate--this is the basis for the garment-based name given to this condition. What is that name?_
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- **Apert syndrome**
- Pfeiffer syndrome
- **Saethre-Chotzen syndrome**

Not Craniosynostoses

*These three have common hand/feet findings: Syndactyly and/or brachydactyly*

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in… Apert syndrome? Apert is known for severe **syndactyly**, which typically involves complete fusion of the index through pinky fingers--only the thumb is free. Likewise, most or all of the toes will be fused as well.

Fingers all together, the thumb separate--this is the basis for the garment-based name given to this condition. What is that name? **Mitten deformity** (‘A’ for effort if you said ‘boxing glove deformity’)

---

114
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings: Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…

Apert syndrome? Apert is known for severe syndactyly, which typically involves complete fusion of the index through pinky fingers--only the thumb is free. Likewise, most or all of the toes will be fused as well.

Pfeiffer syndrome?
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- **Apert syndrome**
- **Pfeiffer syndrome**
- **Saethre-Chotzen syndrome**

Not Craniosynostoses

These three have common hand/feet findings: **Syndactyly and/or brachydactyly**

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…

**Apert syndrome**? Apert is known for severe **syndactyly**, which typically involves complete fusion of the index through pinky fingers--only the thumb is free. Likewise, most or all of the toes will be fused as well.

**Pfeiffer syndrome**? The syndactyly is much **less severe** than in Apert.
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- **Apert syndrome**
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

*These three have common hand/feet findings: Syndactyly and/or brachydactyly*

*Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…*

*Apert syndrome?* Apert is known for severe **syndactyly**, which typically involves complete fusion of the index through pinky fingers--only the thumb is free. Likewise, most or all of the toes will be fused as well.

*Pfeiffer syndrome?* The syndactyly is much **less** severe than in Apert.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings:
Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…

Apert syndrome? Apert is known for severe syndactyly, which typically involves complete fusion of the index through pinky fingers--only the thumb is free. Likewise, most or all of the toes will be fused as well.

Pfeiffer syndrome? The syndactyly is much less severe than in Apert. The defining feature in Pfeiffer syndrome is that the thumbs and great toes are very short and hand feature
foot feature
short vs long
narrow vs broad
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings:

Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…

Apert syndrome? Apert is known for severe syndactyly, which typically involves complete fusion of the index through pinky fingers--only the thumb is free. Likewise, most or all of the toes will be fused as well.

Pfeiffer syndrome? The syndactyly is much less severe than in Apert. The defining feature in Pfeiffer syndrome is that the thumbs and great toes are very short and broad.
Craniofacial Malformations

Pfeiffer syndrome
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- **Apert syndrome**
- **Pfeiffer syndrome**
- **Saethre-Chotzen syndrome**

Not Craniosynostoses

*These three have common hand/feet findings: Syndactyly and/or brachydactyly*

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…

**Apert syndrome?** Apert is known for severe syndactyly, which typically involves complete fusion of the index through pinky fingers—only the thumb is free. Likewise, most or all of the toes will be fused as well.

**Pfeiffer syndrome?** The syndactyly is much less severe than in Apert. The defining feature in Pfeiffer syndrome is that the thumbs and great toes are very short and broad.

*Got a mnemonic for remembering this?*

*Pfeiffer syndrome is that the thumbs and great toes are very short and broad*
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings: Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…

Apert syndrome? Apert is known for severe syndactyly, which typically involves complete fusion of the index through pinky fingers—only the thumb is free. Likewise, most or all of the toes will be fused as well.

Pfeiffer syndrome? The syndactyly is much less severe than in Apert. The defining feature in Pfeiffer syndrome is that the thumbs and great toes are very short and broad

Got a mnemonic for remembering this?
I remember it because the actress Michelle Pfeiffer is well-known for having short/fat thumbs and great toes—that’s why her hands and feet are never seen on camera*

*None of this is actually true, but wouldn’t it make for a great mnemonic if it was?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings:
Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests.
In that regard, how do they tend to manifest in…

Apert syndrome? Apert is known for severe syndactyly, which typically involves complete fusion of the index through pinky fingers—only the thumb is free. Likewise, most or all of the toes will be fused as well.

Pfeiffer syndrome? The syndactyly is much less severe than in Apert. The defining feature in Pfeiffer syndrome is that the thumbs and great toes are very short and broad.

Saethre-Chotzen syndrome? These pts have mild syndactyly.
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have common hand/feet findings: Syndactyly and/or brachydactyly

Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…

Apert syndrome? Apert is known for severe syndactyly, which typically involves complete fusion of the index through pinky fingers—only the thumb is free. Likewise, most or all of the toes will be fused as well.

Pfeiffer syndrome? The syndactyly is much less severe than in Apert. The defining feature in Pfeiffer syndrome is that the thumbs and great toes are very short and broad.

Saethre-Chotzen syndrome? These pts have mild syndactyly
Craniofacial Malformations

Saethre-Chotzen syndrome: Mild syndactyly
Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests. In that regard, how do they tend to manifest in…

**Apert syndrome?** Apert is known for severe *syndactyly*, which typically involves complete fusion of the index through pinky fingers—only the thumb is free. Likewise, most or all of the toes will be fused as well.

**Pfeiffer syndrome?** The syndactyly is much *less* severe than in Apert. The defining feature in Pfeiffer syndrome is that the *thumbs* and *great toes* are very *short* and *broad*

**Saethre-Chotzen syndrome?** These pts have *mild* syndactyly, and their toes tend to be *short* and deviated *laterally* vs *medially*
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- **Apert syndrome**
- **Pfeiffer syndrome**
- **Saethre-Chotzen syndrome**

Not Craniosynostoses

*These three have common hand/feet findings:*

Syndactyly and/or brachydactyly

*Each of the three has a characteristic manner in which the syndactyly/brachydactyly manifests.*

**In that regard, how do they tend to manifest in…**

**Apert syndrome?** Apert is known for severe *syndactyly*, which typically involves complete fusion of the index through pinky fingers--only the thumb is free. Likewise, most or all of the toes will be fused as well.

**Pfeiffer syndrome?** The syndactyly is much *less* severe than in Apert. The defining feature in Pfeiffer syndrome is that the *thumbs* and *great toes* are very *short* and *broad*

**Saethre-Chotzen syndrome?** These pts have *mild* syndactyly, and their toes tend to be *short* and deviated *laterally*
Craniofacial Malformations

Saethre-Chotzen syndrome: Short, laterally deviated toes
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

*These three present with exorbitism, ie, ‘bug eyed’*

**TLDR**
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three present with exorbitism, ie, ‘bug eyed’

This one does not

TLDR
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

These three have hand/feet involvement

TLDR
Craniofacial Malformations

- Craniosynostoses
  - Crouzon syndrome
  - Apert syndrome
  - Pfeiffer syndrome
  - Saethre-Chotzen syndrome

- Not Craniosynostoses
  - This one does not

TLDR

These three have hand/feet involvement
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
  - This one does not
- Apert syndrome
  - These three present with exorbitism, ie, ‘bug eyed’
- Pfeiffer syndrome
  - These three have hand/feet involvement
- Saethre-Chotzen syndrome
  - This one does not

Not Craniosynostoses

TLDR

The status of the eyes and hands/feet provide strong clues for identifying the craniosynostosis syndrome!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- ?
- ?
- ?
- ?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
Craniofacial Malformations

- Craniosynostoses
  - Crouzon syndrome
  - Saethre-Chotzen syndrome

- Not Craniosynostoses
  - Goldenhar syndrome?
  - Treacher Collins syndrome?
  - Pierre Robin sequence?
  - Fetal alcohol syndrome?

*Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?*
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-syntotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

Before we go any further: What is a branchial arch?
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Goldenhar syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins

Before we go any further: What is a branchial arch? Arch-like features of the early embryo; several give rise to the head and neck.
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called **branchial arch** syndromes. Which two? Goldenhar and Treacher Collins

Before we go any further: What is a branchial arch? Arch-like features of the early embryo; several give rise to the head and neck

By what other name are branchial arches called?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins

Before we go any further: What is a branchial arch? Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too)
Craniofacial Malformations
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called **branchial arch** syndromes. Which two? Goldenhar and Treacher Collins

Before we go any further: What is a branchial arch?
Arch-like features of the early embryo; several give rise to the head and neck

By what other name are branchial arches called?
**Pharyngeal** arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too)

How many arches are there, and which are we concerned with?
Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins

Before we go any further: What is a branchial arch? Arch-like features of the early embryo; several give rise to the head and neck

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too)

How many arches are there, and which are we concerned with? There are five or six (depends on who you ask), but we’re only concerned with the first and the second
Before we go any further: What is a branchial arch?
Arch-like features of the early embryo; several give rise to the head and neck

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the Peds book uses 'branchial,' so we will too)

How many arches are there, and which are we concerned with? There are five or six (depends on who you ask), but we're only concerned with the first and the second

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins

What is the non-numeric name of the first arch? The mandibular arch

What nerve innervates the musculature of the mandibular arch? The mandibular division of the trigeminal (V3)

What are the muscles of the mandibular arch? The muscles of mastication, and MATT (that's a mnemonic, BTW)

The mandibular arch contributes to the development of another facial structure of note—what is it? The ear (both internal and external)
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with? There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Crouzon syndrome
- Saethre-Chotzen syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

What is the non-numeric name of the first arch? The mandibular arch.

What nerve innervates the musculature of the mandibular arch? The mandibular division of the trigeminal (V3).

What are the muscles of the mandibular arch? The muscles of mastication, and MATT (that’s a mnemonic, BTW).

The mandibular arch contributes to the development of another facial structure of note--what is it? The ear (both internal and external).
Craniofacial Malformations
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the Peds book uses 'branchial,' so we will too).

How many arches are there, and which are we concerned with? There are five or six (depends on who you ask), but we're only concerned with the first and the second.

148 Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

What is the non-numeric name of the first arch? Mandibular arch

Can I infer that the mandibular arch gives rise to the mandible? You can indeed. Less intuitively, it also gives rise to other bones of the midface including the zygoma and maxilla, as well as the temporal bones.
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?

Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with?

There are five or six (depends on who you ask), but we're only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

- Saethre-Chotzen syndrome

Not Craniosynostoses

- Goldenhar syndrome

- Treacher Collins syndrome

- Pierre Robin sequence

- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

What is the non-numeric name of the first arch?

The mandibular arch.

What nerve innervates the musculature of the mandibular arch?

The mandibular division of the trigeminal (V3).

What are the muscles of the mandibular arch?

The muscles of mastication, and MATT (that’s a mnemonic, BTW).

The mandibular arch contributes to the development of another facial structure of note—what is it?

The ear (both internal and external).

Can I infer that the mandibular arch gives rise to the mandible?

You can indeed. Less intuitively, it also gives rise to other bones of the midface including the zygoma and maxilla, as well as the temporal bones.

the first
Before we go any further: What is a branchial arch?
Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?
Pharyngeal arches (in fact, this name is preferred, but the Peds book uses 'branchial,' so we will too).

How many arches are there, and which are we concerned with?
There are five or six (depends on who you ask), but we're only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldnhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Goldenhar and Treacher Collins are examples of what are called branchial arch syndromes. Which two?

What is the non-numeric name of the first arch?
The mandibular arch

What nerve innervates the musculature of the mandibular arch?
The mandibular division of the trigeminal (V3)

The mandibular arch contributes to the development of another facial structure of note—what is it?
The ear (both internal and external)
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?

Pharyngeal arches (in fact, this name is preferred, but the peds book uses 'branchial,' so we will too).

How many arches are there, and which are we concerned with?

There are five or six (depends on who you ask), but we're only concerned with the first and the second.

- Craniofacial Malformations
- Craniosynostoses
  - Crouzon syndrome
  - Saethre-Chotzen syndrome
- Not Craniosynostoses
  - Goldenhar syndrome
  - Treacher Collins syndrome
  - Goldenhar and Treacher Collins
  - Pfeiffer syndrome
  - Saethre-Chotzen syndrome
  - Goldenhar syndrome
  - Treacher Collins syndrome
  - Pierre Robin sequence
  - Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is the non-numeric name of the first arch?

The mandibular arch

What nerve innervates the musculature of the mandibular arch?

The mandibular division of the trigeminal (V3)
Before we go any further: What is a branchial arch?
Arch-like features of the early embryo; several give rise to the head and neck

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too)

How many arches are there, and which are we concerned with? There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Craniosynostoses
- Goldenhar and Treacher Collins syndromes

Crouzon syndrome

Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too)

What is the non-numeric name of the first arch?
The mandibular arch

What nerve innervates the musculature of the mandibular arch? The mandibular division of the trigeminal (V3)

What are the muscles of the mandibular arch?

The ear (both internal and external)
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?

Pharyngeal arches (in fact, this name is preferred, but the *Peds* book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with?

There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

---

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is the non-numeric name of the first arch?

The mandibular arch

What nerve innervates the musculature of the mandibular arch?

The mandibular division of the trigeminal (V3)

What are the muscles of the mandibular arch?

The muscles of mastication, and MATT (that’s a mnemonic, BTW)
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?

Pharyngeal arches (in fact, this name is preferred, but the Peds book uses 'branchial,' so we will too)

How many arches are there, and which are we concerned with?

There are five or six (depends on who you ask), but we're only concerned with the first and the second.

What is the non-numeric name of the first arch?

The mandibular arch

What nerve innervates the musculature of the mandibular arch?

The mandibular division of the trigeminal (V3)

What are the muscles of the mandibular arch?

The muscles of mastication, and MATT (that's a mnemonic, BTW)

The mandibular arch contributes to the development of another facial structure of note—what is it?

The ear (both internal and external)

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Craniofacial Malformations

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What are the muscles of mastication?

- --
- --
- --

muscles of mastication

the first
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?

Pharyngeal arches (in fact, this name is preferred, but the Peds book uses 'branchial,' so we will too).

How many arches are there, and which are we concerned with?

There are five or six (depends on who you ask), but we're only concerned with the first and the second.

What is the non-numeric name of the first arch?

The mandibular arch.

What nerve innervates the musculature of the mandibular arch?

The mandibular division of the trigeminal (V3).

What are the muscles of the mandibular arch?

The muscles of mastication, and MATT (that's a mnemonic, BTW).

The mandibular arch contributes to the development of another facial structure of note—what is it?

The ear (both internal and external).

Two of the non-syndromic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins.

The muscles of mastication are:

--Medial (aka internal) pterygoid
--Lateral (aka external) pterygoid
--Masseter
--Temporalis

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Goldenhar and Treacher Collins
- Pierre Robin sequence
- Fetal alcohol syndrome
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Goldenhar and Treacher Collins
- Pierre Robin sequence
- Fetal alcohol syndrome
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

What are the muscles of mastication?

--Medial (aka internal) pterygoid
--Lateral (aka external) pterygoid
--Masseter
--Temporalis

the first
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck

By what other name are branchial arches called?
Pharyngeal arches (in fact, this name is preferred, but the *Peds* book uses ‘branchial,’ so we will too)

How many arches are there, and which are we concerned with?
There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

What is the non-numeric name of the first arch?
The *mandibular* arch

What nerve innervates the musculature of the mandibular arch?
The mandibular division of the trigeminal (V3)

What are the muscles of the mandibular arch?
The muscles of mastication, and

What are the muscles of the mandibular arch?
MATT (that’s a mnemonic, BTW)

The mandibular arch contributes to the development of another facial structure of note—what is it?
The ear (both internal and external)

**Craniofacial Malformations**

**Craniosynostoses**
- Crouzon syndrome
- Saethre-Chotzen syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins

**Not Craniosynostoses**
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome

What are these MATT muscles of which you speak?
--M
--A
--T
--T

What are the muscles of the mandibular arch?
MATT

The first
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?

Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with?

There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

What is the non-numeric name of the first arch?

The mandibular arch.

What nerve innervates the musculature of the mandibular arch?

The mandibular division of the trigeminal (V3).

What are the muscles of the mandibular arch?

The muscles of mastication, and MATT (that’s a mnemonic, BTW).

The mandibular arch contributes to the development of another facial structure of note—what is it?

The ear (both internal and external).

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-syostotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins.

What are these MATT muscles of which you speak?

- Mylohyoid
- Anterior belly of the digastric
- Tensor tympani
- Tensor veli palatini

two Latin words

What are the muscles of the mandibular arch?

The muscles of mastication, and MATT (that’s a mnemonic, BTW).

the first
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the Peds book uses 'branchial,' so we will too).

How many arches are there, and which are we concerned with? There are five or six (depends on who you ask), but we're only concerned with the first and the second.

What is the non-numeric name of the first arch? The mandibular arch.

What nerve innervates the musculature of the mandibular arch? The mandibular division of the trigeminal (V3).

What are the muscles of the mandibular arch? The muscles of mastication, and **MATT** (that's a mnemonic, BTW). The mandibular arch contributes to the development of another facial structure of note--what is it? The ear (both internal and external).

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

What are these MATT muscles of which you speak? 

-- **Mylohyoid** 
-- **A**nterior belly of the digastric 
-- **T**ensor tympani 
-- **T**ensor veli palatini

What are the muscles of the mandibular arch? The muscles of mastication, and **MATT** (that's a mnemonic, BTW). 

the first
Before we go any further: What is a branchial arch?
Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with? There are five or six (depends on who you ask), but we're only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Apert syndrome
- Pfeiffer syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Crouzon syndrome
- Saethre-Chotzen syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

What is the non-numeric name of the first arch? The mandibular arch.

What nerve innervates the musculature of the mandibular arch? The mandibular division of the trigeminal (V3).

What are the muscles of the mandibular arch? The muscles of mastication, and MATT (that’s a mnemonic, BTW).

The mandibular arch contributes to the development of another facial structure of note--what is it? The ear (both internal and external).
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the *Peds* book uses ‘branchial,’ so we will too)

How many arches are there, and which are we concerned with? There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

**Craniofacial Malformations**

**Craniosynostoses**
- Crouzon syndrome
- Saethre-Chotzen syndrome

**Not Craniosynostoses**
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Goldenhar and Treacher Collins
-金has the non-numeric name of the first arch? The **mandibular** arch

What nerve innervates the musculature of the mandibular arch? The mandibular division of the trigeminal (V3)

What are the muscles of the mandibular arch? The muscles of mastication, and MATT (that’s a mnemonic, BTW)

The mandibular arch contributes to the development of another facial structure of note—what is it? The ear (both internal and external)
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?
Pharyngeal arches (in fact, this name is preferred, but the "Peds" book uses 'branchial,' so we will too.

How many arches are there, and which are we concerned with?
There are five or six (depends on who you ask), but we're only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pfeiffer syndrome
- Crouzon syndrome
- Apert syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome

Not Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pfeiffer syndrome
- Crouzon syndrome
- Apert syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins.

What is the non-numeric name of the second arch?
The hyoid arch.

What nerve innervates the musculature of the hyoid arch?
The facial nerve.

What are the muscles of the hyoid arch?
The muscles of facial expression, and PASS (yep, another mnemonic).

Does the hyoid arch contribute to the ear like the first arch?
It does indeed.
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?

Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too)

How many arches are there, and which are we concerned with?

There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
- Other head and neck

Goldenhar syndrome and Treacher Collins syndrome are examples of what are called branchial arch syndromes.

What is the non-numeric name of the second arch?

The hyoid arch

Does the hyoid arch contribute to the ear like the first arch?

It does indeed.
Craniofacial Malformations

First (mandibular) arch
Second (hyoid) arch
Third
Fourth
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the *Peds* book uses 'branchial,' so we will too).

How many arches are there, and which are we concerned with? There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

---

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
- Goldenhar and Treacher Collins
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
- Goldenhar and Treacher Collins
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is the non-numeric name of the second arch?

The hyoid arch

Likewise, can I infer that the hyoid arch gives rise to the bone of the same name?

It gives rise to part of the hyoid (and let's leave it at that).

---

There are five or six (depends on who you ask), but we’re only concerned with the first and the second.
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?

Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with?

There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is the non-numeric name of the second arch?

The hyoid arch

Likewise, can I infer that the hyoid arch gives rise to the bone of the same name?

It gives rise to part of the hyoid (and let’s leave it at that).

What nerve innervates the musculature of the hyoid arch?

The facial nerve

What are the muscles of the hyoid arch?

The muscles of facial expression, and PASS (yep, another mnemonic)

Does the hyoid arch contribute to the ear al la the first arch?

It does indeed.

Likewise, can I infer that the hyoid arch gives rise to the bone of the same name?

It gives rise to part of the hyoid (and let’s leave it at that).
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with? There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pfeiffer syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

What is the non-numeric name of the second arch? The hyoid arch.

What nerve innervates the musculature of the hyoid arch? The facial nerve.

What are the muscles of the hyoid arch? The muscles of facial expression, and PASS (yep, another mnemonic).

Does the hyoid arch contribute to the ear like the first arch? It does indeed.
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called? Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with? There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Goldenhar and Treacher Collins
- Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is the non-numeric name of the second arch? The hyoid arch.

What nerve innervates the musculature of the hyoid arch? The facial nerve.

There are five or six (depends on who you ask), but we’re only concerned with the first and the second.
What is a branchial arch?
Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?
Pharyngeal arches (in fact, this name is preferred, but the *Peds* book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with?
There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

---

**Craniofacial Malformations**

**Craniosynostoses**
- Crouzon syndrome

**Not Craniosynostoses**
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins.

What is the non-numeric name of the second arch?
The hyoid arch.

What nerve innervates the musculature of the hyoid arch?
The facial nerve.

What are the muscles of the hyoid arch?
The muscles of facial expression, and PASS (yep, another mnemonic).

Does the hyoid arch contribute to the ear like the first arch?
It does indeed.
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?
Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with?
There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Syndactyly
- Facial alcohol syndrome
- VACTERL

Craniosynostoses
- Craniosynostoses

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins.

What is the non-numeric name of the second arch?
The hyoid arch.

What nerve innervates the musculature of the hyoid arch?
The facial nerve.

What are the muscles of the hyoid arch?
The muscles of facial expression, and PASS (yep, another mnemonic).

Does the hyoid arch contribute to the ear like the first arch?
It does indeed.
Before we go any further: What is a branchial arch?
Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?
Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with?
There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Craniosynostoses
- Goldenhar and Treacher Collins

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins

What is the non-numeric name of the second arch?
The hyoid arch.

What nerve innervates the musculature of the hyoid arch?
The facial nerve.

What are the muscles of the hyoid arch?
The muscles of facial expression, and PASS (yep, another mnemonic).

Does the hyoid arch contribute to the ear like the first arch?
It does indeed.

What are these PASS muscles?
- P
- A
- S
- S

There are five or six (depends on who you ask), but we’re only concerned with the first and the second.
Before we go any further: What is a branchial arch?
Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?
Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with?
There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins.

What is the non-numeric name of the second arch?
The hyoid arch.

What nerve innervates the musculature of the hyoid arch?
The facial nerve.

What are the muscles of the hyoid arch?
The muscles of facial expression, and PASS (yep, another mnemonic).

Does the hyoid arch contribute to the ear like the first arch?
It does indeed.

What are these PASS muscles?
- Posterior belly of the digastric
- Auricular muscles
- Stylohyoid
- Stapedius

Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses

Craniosynostoses
- Crouzon syndrome

Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins.

What is the non-numeric name of the second arch?
The hyoid arch.

What nerve innervates the musculature of the hyoid arch?
The facial nerve.

What are the muscles of the hyoid arch?
The muscles of facial expression, and PASS (yep, another mnemonic).

Does the hyoid arch contribute to the ear like the first arch?
It does indeed.

What are these PASS muscles?
- Posterior belly of the digastric
- Auricular muscles
- Stylohyoid
- Stapedius

The second

There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

Craniofacial Malformations
What is a branchial arch?
Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?
Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with?
There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins.

What is the non-numeric name of the second arch?
The hyoid arch.

What nerve innervates the musculature of the hyoid arch?
The facial nerve.

What are the muscles of the hyoid arch?
The muscles of facial expression, and PASS (yep, another mnemonic).

Does the hyoid arch contribute to the ear al la the first arch?
Yes, the second.

There are five or six (depends on who you ask), but we’re only concerned with the first and the second.
Before we go any further: What is a branchial arch?

Arch-like features of the early embryo; several give rise to the head and neck.

By what other name are branchial arches called?

Pharyngeal arches (in fact, this name is preferred, but the Peds book uses ‘branchial,’ so we will too).

How many arches are there, and which are we concerned with?

There are five or six (depends on who you ask), but we’re only concerned with the first and the second.

---

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Apert syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins.

What is the non-numeric name of the second arch?

The hyoid arch.

What nerve innervates the musculature of the hyoid arch?

The facial nerve.

What are the muscles of the hyoid arch?

The muscles of facial expression, and PASS (yep, another mnemonic).

Does the hyoid arch contribute to the ear al la the first arch?

It does indeed.

The second...
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-sysyototic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

At long last: What is a branchial arch syndrome?
At long last: What is a branchial arch syndrome?
As one might expect, it is a constellation of craniofacial abnormalities secondary to maldevelopment or dysgenesis of structures arising from the mandibular and/or hyoid branchial arches.

Two of the non-systotic conditions are examples of what are called _branchial arch syndromes_. Which two? Goldenhar and Treacher Collins.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-synostotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

At long last: What is a branchial arch syndrome? As one might expect, it is a constellation of craniofacial abnormalities secondary to maldevelopment or dysgenesis of structures arising from the mandibular and/or hyoid branchial arches.

Which structures are those?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

At long last: What is a branchial arch syndrome?
As one might expect, it is a constellation of craniofacial abnormalities secondary to maldevelopment or dysgenesis of structures arising from the mandibular and/or hyoid branchial arches.

Which structures are those?
For starters: The mandible, the maxilla, the muscles of facial expression and mastication, and the ears.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-cystotic conditions are examples of what are called **branchial arch syndromes**. Which two? Goldenhar and Treacher Collins.

What is the incidence of Goldenhar?

About 1/4000 live births

What is its inheritance pattern?

It is sporadic

Is there a sex predilection?

Yes, males are twice as likely to be affected.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins

What is the incidence of Goldenhar?
About 1/4000 live births

What is its inheritance pattern?
It is sporadic

Is there a sex predilection?
Yes, males are twice as likely to be affected
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

What is the incidence of Goldenhar? About 1/4000 live births

What is its inheritance pattern?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins

What is the incidence of Goldenhar? About 1/4000 live births

What is its inheritance pattern? It is sporadic in the majority of cases
Craniofacial Malformations

Craniosynostostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called **branchial arch syndromes**. Which two? Goldenhar and Treacher Collins.

What is the incidence of Goldenhar? About 1/4000 live births

What is its inheritance pattern? It is sporadic in the majority of cases

Is there a sex predilection?
Craniofacial Malformations

- Craniosynostoses
  - Crouzon syndrome
  - Saethre-Chotzen syndrome

- Not Craniosynostoses
  - Goldenhar syndrome
  - Treacher Collins syndrome
  - Pierre Robin sequence
  - Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

What is the incidence of Goldenhar?
About 1/4000 live births

What is its inheritance pattern?
It is sporadic in the majority of cases

Is there a sex predilection?
Yes, M vs F are #x as likely to be affected
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

What is the incidence of Goldenhar?
About 1/4000 live births

What is its inheritance pattern?
It is sporadic in the majority of cases

Is there a sex predilection?
Yes, males are twice as likely to be affected
Craniofacial Malformations

Craniosynostoses

Crouzon syndrome

Not Craniosynostoses

Goldenhar syndrome

Treacher Collins syndrome

Pierre Robin sequence

Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name? (the answer will start with an ‘O’)

Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?

Lid coloboma

Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?

Ear abnormalities (pre-auricular appendages; aural fistulae)

Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?

At the limbus

Are Goldenhar individuals cognitively impaired?

A minority (~10%) have mental retardation

Very convenient mnemonic!

Goldenhar

OLDENHAR

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
  - Crouzon syndrome
  - Goldenhar syndrome
  - Treacher Collins syndrome
  - Pierre Robin sequence
  - Fetal alcohol syndrome

Not Craniosynostoses
  - Apert syndrome
  - Pfeiffer syndrome
  - Saethre-Chotzen syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar’s non-eponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

What is the classic vertebral finding?
Hemivertebrae, aka butterfly vertebrae

Very convenient mnemonic!

Goldenhar
OAV syndrome
L
D
E
N
H
A
R

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

What is Goldenhar’s non-eponyymous name? Oculo-Auriculo-Vertebral (OAV) syndrome

What is the classic vertebral finding? Hemivertebrae, aka perty...

Goldenhar
- OAV syndrome

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

Crouzon syndrome

Goldenhar syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

What is Goldenhar’s non-systotic name?

Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?

-- Lid coloboma
-- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?

-- Ear abnormalities (pre-auricular appendages; aural fistulae)
-- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?

At the limbus

Are Goldenhar individuals cognitively impaired?

A minority (~10%) have mental retardation

Very convenient mnemonic!

Goldenhar syndrome

Hemivertebrae, aka butterfly vertebrae

What is the classic vertebral finding?

Hemivertebrae, aka butterfly vertebrae

Goldenhar syndrome

OAV syndrome

Treacher Collins syndrome

Pierre Robin sequence

Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar syndrome?
Craniofacial Malformations

Goldenhar syndrome: Butterfly vertebrae
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

Goldenhar syndrome
- Saethre-Chotzen syndrome
- Apert syndrome
- Pfeiffer syndrome
- Crouzon syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar's noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Goldenhar OAV syndrome

Very convenient mnemonic!

What is the classic vertebral finding?
Hemivertebrae, aka butterfly vertebrae

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
Alagille syndrome

What is the noneponymous name for Alagille syndrome?
Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
An infant with jaundice will present to the eye service as a 'rule out Alagille syndrome' consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
The presence of a posterior embryotoxon on the infant's corneas

What is a posterior embryotoxon?
An anteriorly displaced and thickened Schwalbe's line
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar
- Treacher Collins

What is Goldenhar's noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Goldenhar OAV syndrome

Very convenient mnemonic!

What is the classic vertebral finding?
Hemivertebrae, aka butterfly vertebrae

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
Alagille syndrome

What is the noneponymous name for Alagille syndrome?
Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
An infant with jaundice will present to the eye service as a 'rule out Alagille syndrome' consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
The presence of a posterior embryotoxon on the infant's corneas

What is a posterior embryotoxon?
An anteriorly displaced and thickened Schwalbe's line
Craniofacial Malformations

Not Craniosynostoses

Goldenhar syndrome
Treacher Collins syndrome
Pierre Robin sequence
Fetal alcohol syndrome

Craniosynostoses

Crouzon syndrome
Apert syndrome
Pfeiffer syndrome
Saethre-Chotzen syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins

What is Goldenhar's noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
Lid coloboma
Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
Ear abnormalities (pre-auricular appendages; aural fistulae)
Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
Alagille syndrome

What is the noneponymous name for Alagille syndrome?
Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
An infant with jaundice will present to the eye service as a 'rule out Alagille syndrome' consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
The presence of a posterior embryotoxon on the infant's corneas

What is a posterior embryotoxon?
An anteriorly displaced and thickened Schwalbe's line
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

Goldenhar's noneponymous name is Oculo-Auriculo-Vertebral (OAV) syndrome.

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

Are Goldenhar individuals cognitively impaired?
- A minority (~10%) have mental retardation

What is the classic vertebral finding?
- Hemivertebrae, aka butterfly vertebrae

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
- Alagille syndrome

What is the noneponymous name for Alagille syndrome?
- Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
- An infant with jaundice will present to the eye service as a 'rule out Alagille syndrome' consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
- The presence of a posterior embryotoxon on the infant's corneas

What is a posterior embryotoxon?
- An anteriorly displaced and thickened Schwalbe's line
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Goldenhar OAV syndrome

Very convenient mnemonic!

What is the classic vertebral finding?
Hemivertebrae, aka butterfly vertebrae

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
Alagille syndrome

What is the noneponymous name for Alagille syndrome?
Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
An infant with jaundice will present to the eye service as a 'rule out Alagille syndrome' consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
The presence of a posterior embryotoxon on the infant’s corneas

What is a posterior embryotoxon?
An anteriorly displaced and thickened Schwalbe’s line
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
Vertebral Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

What is the classic vertebral finding?
Hemivertebrae, aka butterfly vertebrae

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
Alagille syndrome

What is the noneponymous name for Alagille syndrome?
Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
An infant with jaundice will present to the eye service as a ‘rule out Alagille syndrome’ consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
The presence of a posterior embryotoxon on the infant’s corneas

What is a posterior embryotoxon?
An anteriorly displaced and thickened Schwalbe’s line
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
- Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

Are Goldenhar individuals cognitively impaired?
- A minority (~10%) have mental retardation

What is the classic vertebral finding?
- Hemivertebrae, aka “butterfly vertebrae”

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
- Alagille syndrome

What is the noneponymous name for Alagille syndrome?
- Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
- An infant with jaundice will present to the eye service as a ‘rule out Alagille syndrome’ consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
- The presence of a posterior embryotoxon on the infant’s corneas

What is a posterior embryotoxon?
- An anteriorly displaced and thickened Schwalbe’s line
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
Alagille syndrome

What is the noneponymous name for Alagille syndrome?
Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
An infant with jaundice will present to the eye service as a ‘rule out Alagille syndrome’ consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
The presence of a posterior embryotoxon on the infant’s corneas

Goldenhar syndrome

Treacher Collins syndrome

Pierre Robin sequence

Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Very convenient mnemonic!

What is the classic vertebral finding?
Hemivertebrae, aka butterfly vertebrae

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
Alagille syndrome

What is the noneponymous name for Alagille syndrome?
Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
An infant with jaundice will present to the eye service as a ‘rule out Alagille syndrome’ consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
The presence of a posterior embryotoxon on the infant’s corneas

What is a posterior embryotoxon?
An anteriorly displaced and thickened Schwalbe’s line
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar's noneponymous name?
- Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

Are Goldenhar individuals cognitively impaired?
- A minority (~10%) have mental retardation

What is the classic vertebral finding?
- Hemivertebrae, aka butterfly vertebrae

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
- Alagille syndrome

What is the noneponymous name for Alagille syndrome?
- Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt? An infant with jaundice will present to the eye service as a 'rule out Alagille syndrome' consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome? The presence of a posterior embryotoxon on the infant's corneas

What is a posterior embryotoxon?
- An anteriorly displaced and thickened Schwalbe's line

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
- Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

Are Goldenhar individuals cognitively impaired?
- A minority (~10%) have mental retardation

Very convenient mnemonic!

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?
- Alagille syndrome

What is the noneponymous name for Alagille syndrome?
- Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
- An infant with jaundice will present to the eye service as a ‘rule out Alagille syndrome’ consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
- The presence of a posterior embryotoxon on the infant’s corneas

What is a posterior embryotoxon?
- An anteriorly displaced and thickened Schwalbe’s line
Craniofacial Malformations

Craniosynostoses

Crouzon syndrome

Not Craniosynostoses

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it? Alagille syndrome

What is the noneponymous name for Alagille syndrome? Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt? An infant with jaundice will present to the eye service as a ‘rule out Alagille syndrome’ consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome? The presence of a posterior embryotoxon on the infant’s corneas

What is a posterior embryotoxon? An anteriorly displaced and thickened Schwalbe’s line

What is Goldenhar’s noneponymous name? Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common? Lid coloboma, Dermoids of the cornea, Duane syndrome

What nonocular findings are usually present? Ear abnormalities (pre-auricular appendages; aural fistulae), Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located? At the limbus

Are Goldenhar individuals cognitively impaired? A minority (~10%) have mental retardation

What is the classic vertebral finding? Hemivertebrae, aka butterfly vertebrae

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it? Alagille syndrome

What is the noneponymous name for Alagille syndrome? Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt? An infant with jaundice will present to the eye service as a ‘rule out Alagille syndrome’ consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome? The presence of a posterior embryotoxon on the infant’s corneas

What is a posterior embryotoxon? An anteriorly displaced and thickened Schwalbe’s line

Very convenient mnemonic!
Posterior embryotoxon
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Treacher Collins syndrome

Not Craniosynostoses
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation
204

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
-- Lid colobomas

What nonocular findings are usually present?
-- Ear abnormalities (pre-auricular appendages; aural fistulae)
-- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
DEHNAR
Craniofacial Malformations

Goldenhar syndrome: Lid coloboma
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence

Goldenhar’s non-eponymous name? Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
--Lid coloboma

Does the coloboma tend to be in the upper lid, or the lower?

Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
DENHAR
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

- Goldenhar syndrome
  - Treacher Collins syndrome
  - Pierre Robin sequence
  - Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?

Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?

- Lid coloboma

Does the coloboma tend to be in the upper lid, or the lower?

Depends on who you ask. The BCSC Cornea book says the upper, whereas the Plastics book indicates the lower. (The Peds book doesn’t address this issue.) Caveat emptor.

Goldenhar

OAV syndrome

Lid colobomas

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar
- Treacher Collins

What is Goldenhar’s nonenponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Very convenient mnemonic!

(Now this one--actually two, both of which start with ‘D’)

Goldenhar
OAV syndrome
Lid colobomas

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name? Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?

- Lid coloboma
- Dermoids of the cornea; Duane syndrome

Goldenhar

OAV syndrome

Lid colobomas

Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses

Goldenhar syndrome
- Oculo-Auriculo-Vertebral (OAV) syndrome
- Bilateral microphthalmia
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
-- Lid colobomas
-- Dermoids of the cornea

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

What is Goldenhar’s nonlenonynous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
-- Lid colobomas
-- Dermoids of the cornea

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea
- Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities

Note: There is another legit answer, so if you came up with that one, no worries (we’ll identify it shortly)
Craniofacial Malformations

Goldenhar syndrome: Epibulbar dermoid
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
  - Oculo-Auriculo-Vertebral (OAV) syndrome
  - Lid coloboma
    - Dermoids of the cornea
    - Duane syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid colobomas
- Dermoids of the cornea
- Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Very convenient mnemonic!

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Goldenhar OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins syndrome

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

In what noncosmetic way are they significant?

Very convenient mnemonic!

Goldenhar syndrome
- OAV syndrome
- Lid colobomas
- Dermoid; Duane syndrome
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Apert syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
-- Lid colobomas
-- Dermoids of the cornea

What nonocular findings are usually present?
-- Ear abnormalities (pre-auricular appendages; aural fistulae)
-- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

In what noncosmetic way are they significant?
They can be amblyogenic

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
- Saethre-Chotzen syndrome
- Apert syndrome
- Pfeiffer syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Oculo-Auriculo-Vertebral (OAV) syndrome

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea
- Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

Should epibulbar dermoids be excised if they seem to be amblyogenic?
Yes, although the success rate is not high (the post-op scar is often amblyogenic in and of itself)

In what noncosmetic way are they significant?
They can be amblyogenic

Very convenient mnemonic!

Goldenhar OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar's non-eponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

What is the 'full' name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

In what noncosmetic way are they significant?
They can be amblyogenic

Should epibulbar dermoids be excised if they seem to be amblyogenic?
Yes, although the success rate is not high (the post-op scar is often amblyogenic in and of itself)
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
- Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

Are Goldenhar individuals cognitively impaired?
- A minority (~10%) have mental retardation

What is the ‘full’ name of the dermoid in question?
- Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
- They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?

Very convenient mnemonic!
- Goldenhar
- OAV syndrome
- Lid colobomas
- Dermoid; Duane syndrome
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Goldenhar syndrome
- Oculo-Auriculo-Vertebral (OAV) syndrome
- Lid coloboma
- Dermoids of the cornea
- Duane syndrome

What is Goldenhar’s noneponymous name? Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea
- Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

Are Goldenhar individuals cognitively impaired?
- A minority (~10%) have mental retardation

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

What is the ‘full’ name of the dermoid in question?
- Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
- They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
- There is none; they are distinct and separate entities

Very convenient mnemonic!
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?

- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)? Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?

What ocular/periocular abnormalities are common?
-- Lid coloboma
-- Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities.
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)? Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located? The temporal fornix.

What ocular/periorbital abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question? Epibulbar dermoid.

Are epibulbar dermoids clinically significant for reasons other than cosmesis? They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts? There is none; they are distinct and separate entities.

Goldenhar syndrome

OAV syndrome

Lid colobomas

Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

Goldenhar syndrome: Dermolipoma
Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)? Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

What are the non-systotic conditions that are examples of what are called branchial arch syndromes? Goldenhar and Treacher Collins.

What is Goldenhar's noneponymous name? Oculo-Auriculo-Vertebral (OAV) syndrome.

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where are dermolipomas typically located? The temporal fornix.

With what similar-appearing but etiologically-distinct condition are dermolipomas confused? Prolapsed orbital fat.

How can the two be differentiated at the slit lamp?
- By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white)
- By texture (prolapsed fat is squishy, whereas a dermolipoma is firm)

What is the 'full' name of the dermoid in question? Epibulbar dermoid.

Are epibulbar dermoids clinically significant for reasons other than cosmesis? They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts? There is none; they are distinct and separate entities.
Craniofacial Malformations

**Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?**
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

**Where are dermolipomas typically located?**
The temporal fornix.

**With what similar-appearing but etiologically-distinct condition are dermolipomas confused?**
Prolapsed orbital fat.

- **What ocular/periocular abnormalities are common?**
  - Lid coloboma
  - Dermoids of the cornea; Duane syndrome

- **What is the ‘full’ name of the dermoid in question?**
  Epibulbar dermoid

- **Are epibulbar dermoids clinically significant for reasons other than cosmesis?**
  They can be, and usually are.

- **What is the relationship between epibulbar dermoids and dermoid cysts?**
  There is none; they are distinct and separate entities.
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar.

Where are dermolipomas typically located?
The temporal fornix

With what similar-appearing but etiologically-distinct condition are dermolipomas confused?
Prolapsed orbital fat

How can the two be differentiated at the slit lamp?
-- By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white)
-- By texture (prolapsed fat is squishy, whereas a dermolipoma is firm)

What ocular/periorcular abnormalities are common?
-- Lid coloboma
-- Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?
The temporal fornix.

With what similar-appearing but etiologically-distinct condition are dermolipomas confused?
Prolapsed orbital fat.

How can the two be differentiated at the slit lamp?
-- By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white).
-- By texture (prolapsed fat is squishy, whereas a dermolipoma is firm).

What ocular/periocular abnormalities are common?
-- Lid colobomas
-- Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid.

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities.

Goldenhar syndrome
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?
The temporal fornix.

With what similar-appearing but etiologically-distinct condition are dermolipomas confused?
Prolapsed orbital fat.

How can the two be differentiated at the slit lamp?
--By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white).
--By texture (prolapsed fat is squishy, whereas a dermolipoma is firm).

What ocular/periocular abnormalities are common?
--Lid coloboma
--Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid.

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities.

Goldenhar syndrome
Oculo-Auriculo-Vertebral (OAV) syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)? Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located? The temporal fornix.

What about on MRI/CT—how can they be differentiated?

- By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white)
- By texture (prolapsed fat is squishy, whereas a dermolipoma is firm)

On MRI/CT? Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

How can the two be differentiated at the slit lamp?—

What ocular/periocular abnormalities are common?

- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis? They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts? There is none; they are distinct and separate entities
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?
The temporal fornix.

What about on MRI/CT--how can they be differentiated?
On imaging, prolapsed orbital fat will be seen to be continuous with intraconal fat, whereas a dermolipoma will not.

With what similar-appearing but etiologically-distinct condition are dermolipomas confused?
Prolapsed orbital fat.

How can the two be differentiated at the slit lamp?
--By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white)
--By texture (prolapsed fat is squishy, whereas a dermolipoma is firm)

What ocular/periocular abnormalities are common?
--Lid colobomas
--Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?
The text is not legible.

What about on MRI/CT—how can they be differentiated?
On imaging, prolapsed orbital fat will be seen to be continuous with intraconal fat, whereas a dermolipoma will not.

With what similar-appearing but etiologically-distinct condition are dermolipomas confused?
Prolapsed orbital fat.

How can the two be differentiated at the slit lamp?
---By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white)
---By texture (prolapsed fat is squishy, whereas a dermolipoma is firm)

On imaging, prolapsed orbital fat will be seen to be continuous with intraconal fat, whereas a dermolipoma will not.

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities.
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?
The term refers to “lipodermoids” of the temporal fornix.

What about on MRI/CT—how can they be differentiated?
On imaging, prolapsed orbital fat will be seen to be continuous with intraconal fat, whereas a dermolipoma will not.

Do dermolipomas need to be excised?
Generally no— they should be left alone.

How can the two be differentiated?
-- By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white)
-- By texture (prolapsed fat is squishy, whereas a dermolipoma is firm)

What ocular/periocular abnormalities are common?
-- Lid colobomas
-- Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities.

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Apert syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses

What is Goldenhar’s non-eponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
-- Lid coloboma
-- Dermoids of the cornea

Duane syndrome

What is Duane syndrome’s middle name?
Duane’s syndrome

What nonocular findings are usually present?
-- Ear abnormalities (pre-auricular appendages; aural fistulae)
-- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Very convenient mnemonic!

Duane syndrome has a ‘middle’ name—what is it?
Duane’s syndrome

Treacher Collins syndrome

OAV syndrome

Pierre Robin sequence

Goldenhar syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

Apert syndrome

Fetal alcohol syndrome

Craniofacial Malformations
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Goldenhar syndrome

Not Craniosynostoses

- Treacher Collins syndrome
- Pierre Robin sequence

What is Goldenhar’s noneponymous name? Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea

Goldenhar syndrome

Duane syndrome

OAV syndrome

Duane syndrome has a ‘middle’ name—what is it? Duane’s retraction syndrome

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins syndrome

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
--Lid coloboma
--Dermoids of the cornea

Duane syndrome

Duane syndrome has a ‘middle’ name--what is it?
Duane’s retraction syndrome

Duane’s is an example of a ‘congenital cranial dysinnervation disorder.’ In Duane’s, which cranial nerve incorrectly innervates which muscle?

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Apert syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
- Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea
- Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

Are Goldenhar individuals cognitively impaired?
- A minority (~10%) have mental retardation

Duane syndrome has a ‘middle’ name--what is it?
- Duane’s retraction syndrome

Duane’s is an example of a ‘congenital cranial dysinnervation disorder.’ In Duane’s, which cranial nerve incorrectly innervates which muscle?
- CN3 innervates the lateral rectus

Very convenient mnemonic!

Duane’s retraction syndrome
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
- Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

Are Goldenhar individuals cognitively impaired?
- A minority (~10%) have mental retardation

Duane syndrome has a ‘middle’ name—what is it?
- Duane’s retraction syndrome

Duane’s is an example of a ‘congenital cranial dysinnervation disorder.’ In Duane’s, which cranial nerve incorrectly innervates which muscle?
- CN3 innervates the lateral rectus

What is the observed manifestation of Duane’s, ie, with what issue do pts present?

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Pierre Robin sequence
- Fetal alcohol syndrome

What is Goldenhar’s non-eponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
-- Lid coloboma
-- Dermoids of the cornea

Duane syndrome

Duane syndrome has a ‘middle’ name—what is it?
Duane’s retraction syndrome

Duane’s is an example of a ‘congenital cranial dysinnervation disorder.’ In Duane’s, which cranial nerve incorrectly innervates which muscle?
CN3 innervates the lateral rectus

Very convenient mnemonic!

What is the observed manifestation of Duane’s, ie, with what issue do pts present?
They have limited horizontal eye movement, and attempted adduction causes the eye to retract.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Crouzon syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar’s non-eponomous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
-- Lid coloboma
-- Dermoids of the cornea

What nonocular findings are usually present?
-- Ear abnormalities (pre-auricular appendages; aural fistulae)
-- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Duane syndrome has a ‘middle’ name--what is it?
Duane’s retraction syndrome

Duane’s is an example of a ‘congenital cranial dysinnervation disorder.’ In Duane’s, which cranial nerve incorrectly innervates which muscle?
CN3 innervates the lateral rectus

What is the observed manifestation of Duane’s, ie, with what issue do pts present?
They have limited horizontal eye movement, and attempted adduction causes the eye to retract
Craniofacial Malformations

Duane syndrome (Type 1, OS)
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar's non-eponym name?

Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?

- Lid coloboma
- Dermoids of the cornea
- Duane syndrome

What nonocular findings are usually present?

- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?

At the limbus

Are Goldenhar individuals cognitively impaired?

A minority (~10%) have mental retardation

Goldenhar OAV syndrome Lid coloboma Dermoid; Duane syndrome ENHA

Very convenient mnemonic!

Duane syndrome has a 'middle' name--what is it? (Note: It has nothing to do with the craniofacial malformations.)

Duane’s retraction syndrome

Duane’s is an example of a ‘congenital cranial dysinnervation disorder.’ In Duane’s, which cranial nerve incorrectly innervates which muscle?

CN3 innervates the lateral rectus

What is the observed manifestation of Duane’s, ie, with what issue do pts present?

They have limited horizontal eye movement, and attempted adduction causes the eye to retract

Another congenital cranial dysinnervation disorder should come readily to mind--what is it? (Note: It has nothing to do with the craniofacial malformations.)

Marcus-Gunn jaw wink

In MGJW, which cranial nerve incorrectly innervates which muscle?

CN7 innervates the levator

What is the observed manifestation of MGJW, ie, with what issue do pts present?

They have ptosis that resolves when they move their jaw a certain way
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar's noneponymous name?
- Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Another congenital cranial dysinnervation disorder should come readily to mind--what is it? (Note: It has nothing to do with the craniofacial malformations.)
- Marcus-Gunn jaw wink

Duane syndrome
- Duane's retraction syndrome

Duane's is an example of a 'congenital cranial dysinnervation disorder.' In Duane's, which cranial nerve incorrectly innervates which muscle?
- CN3 innervates the lateral rectus

What is the observed manifestation of Duane's, ie, with what issue do pts present?
- They have limited horizontal eye movement, and attempted adduction causes the eye to retract

Another convenient mnemonic!

Duane’s retraction syndrome has a ‘middle’ name--what is it?
- Duane's retraction syndrome

Duane's is an example of a 'congenital cranial dysinnervation disorder.' In Duane's, which cranial nerve incorrectly innervates which muscle?
- CN3 innervates the lateral rectus

What is the observed manifestation of Duane's, ie, with what issue do pts present?
- They have limited horizontal eye movement, and attempted adduction causes the eye to retract
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses
- Goldenhar syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

- Goldenhar
- Treacher Collins

What is Goldenhar's non-epponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Another congenital cranial dysinnervation disorder should come readily to mind--what is it? (Note: It has nothing to do with the craniofacial malformations.)

**Marcus-Gunn jaw wink**

In MGJW, which cranial nerve incorrectly innervates which muscle?

What is the observed manifestation of Duane's, ie, with what issue do pts present?
They have limited horizontal eye movement, and attempted adduction causes the eye to retract

Another congenital cranial dysinnervation disorder should come readily to mind--what is it? (Note: It has nothing to do with the craniofacial malformations.)

**Duane’s retraction syndrome**

Duane's is an example of a 'congenital cranial dysinnervation disorder.' In Duane's, which cranial nerve incorrectly innervates which muscle?
CN3 innervates the lateral rectus

What is the observed manifestation of Duane's, ie, with what issue do pts present?
They have limited horizontal eye movement, and attempted adduction causes the eye to retract.
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

- Goldenhar
- Treacher Collins

What is Goldenhar's noneponymous name?

Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?

- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?

- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?

At the limbus

Are Goldenhar individuals cognitively impaired?

A minority (~10%) have mental retardation

Another congenital cranial dysinnervation disorder should come readily to mind--what is it? (Note: It has nothing to do with the craniofacial malformations.)

Marcus-Gunn jaw wink

In MGJW, which cranial nerve incorrectly innervates which muscle?

CN7 innervates the levator

Duane syndrome

Duane’s retraction syndrome

Duane’s is an example of a ‘congenital cranial dysinnervation disorder.’ In Duane’s, which cranial nerve incorrectly innervates which muscle?

CN3 innervates the lateral rectus

What is the observed manifestation of Duane’s, ie, with what issue do pts present?

They have limited horizontal eye movement, and attempted adduction causes the eye to retract

Very convenient mnemonic!

Duane syndrome

Duane’s retraction syndrome

Duane’s is an example of a ‘congenital cranial dysinnervation disorder.’ In Duane’s, which cranial nerve incorrectly innervates which muscle?

CN3 innervates the lateral rectus

What is the observed manifestation of Duane’s, ie, with what issue do pts present?

They have limited horizontal eye movement, and attempted adduction causes the eye to retract

Marcus-Gunn jaw wink

In MGJW, which cranial nerve incorrectly innervates which muscle?

CN7 innervates the levator

What is the observed manifestation of MGJW, ie, with what issue do pts present?

They have ptosis that resolves when they move their jaw a certain way
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins

What is Goldenhar's non-eponymic name?
- Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

Are Goldenhar individuals cognitively impaired?
- A minority (~10%) have mental retardation

Goldenhar syndrome

Oculo-Auriculo-Vertebral (OAV) syndrome

Lid coloboma

Dermoid; Duane syndrome

Duane syndrome

Duane's retraction syndrome

Duane's is an example of a 'congenital cranial dysinnervation disorder.' In Duane's, which cranial nerve incorrectly innervates which muscle?
- CN3 innervates the lateral rectus

What is the observed manifestation of Duane's, ie, with what issue do pts present?
- They have limited horizontal eye movement, and attempted adduction causes the eye to retract

Another congenital cranial dysinnervation disorder should come readily to mind--what is it? (Note: It has nothing to do with the craniofacial malformations.)
- Marcus-Gunn jaw wink

In MGJW, which cranial nerve incorrectly innervates which muscle?
- CN7 innervates the levator

What is the observed manifestation of MGJW, ie, with what issue do pts present?
- They have ptosis that resolves when they move their jaw a certain way

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses

Crouzon syndrome

Not Craniosynostoses

Goldenhar syndrome

Crouzon syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

Goldenhar syndrome

Treacher Collins syndrome

Pierre Robin sequence

Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar's noneponymous name?

Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?

Lid coloboma

Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?

Ear abnormalities (pre-auricular appendages; aural fistulae)

Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?

At the limbus

Are Goldenhar individuals cognitively impaired?

A minority (~10%) have mental retardation

Very convenient mnemonic!

Duane syndrome has a 'middle' name--what is it? (Note: It has nothing to do with the craniofacial malformations.)

Duane's retraction syndrome

Duane's is an example of a "congenital cranial dysinnervation disorder." In Duane's, which cranial nerve incorrectly innervates which muscle?

CN3 innervates the lateral rectus

What is the observed manifestation of Duane's, ie, with what issue do pts present?

They have limited horizontal eye movement, and attempted adduction causes the eye to retract

Another congenital cranial dysinnervation disorder should come readily to mind--what is it?

Marcus-Gunn jaw wink

In MGJW, which cranial nerve incorrectly innervates which muscle?

CN7 innervates the levator

What is the observed manifestation of MGJW, ie, with what issue do pts present?

Ptosis that resolves when the pt moves their jaw a certain way

Note: It has nothing to do with the craniofacial malformations.
Craniofacial Malformations

MGJW
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

What is Goldenhar’s non-eponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
-- Lid coloboma
-- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
--

A minority (~10%) have mental retardation

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
  - Treacher Collins syndrome
  - Pierre Robin sequence
  - Fetal alcohol syndrome

**What is Goldenhar’s noneponymous name?**
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)

Very convenient mnemonic:
- **G**oldenhar
- **OAV** syndrome
- **L**id colobomas
- **D**ermoid; Duane syndrome
- **E**ar abnormalities
- **N**othing starts w/ ‘N’
- **HAR**
Craniofacial Malformations

Goldenhar syndrome: Ear abnormalities
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

What is Goldenhar’s noneponymous name? Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Ear abnormalities (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located? At the limbus

Are Goldenhar individuals cognitively impaired? A minority (~10%) have mental retardation

Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Ear abnormalities
Nothing starts w/ ‘N’

H
A
R
(next)
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
--Lid coloboma
--Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
--Ear abnormalities (pre-auricular appendages; aural fistulae)
--Hemifacial microsomia (maxillary/mandibular hypoplasia)

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Ear abnormalities
Nothing starts w/ ‘N’
Hemifacial microsomia
A
R

Very convenient mnemonic!
Craniofacial Malformations

Goldenhar syndrome: Hemifacial microsomia
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
-- Lid coloboma
-- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
-- Ear abnormalities (pre-auricular appendages; aural fistulae)
-- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Which side of the face is more likely to be affected?
The right

Why the right side?
I have no idea

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Ear abnormalities
Nothing starts w/ ‘N’
Hemifacial microsomia
A
R

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

- Goldenhar syndrome
  - Treacher Collins syndrome
  - Pierre Robin sequence
  - Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar’s non-eponymous name?

Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?

- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?

- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Ear abnormalities (maxillary/mandibular hypoplasia)
- Hemifacial microsomia

Which side of the face is more likely to be affected?

The right

Goldenhar

OAV syndrome

Lid colobomas

Dermoid; Duane syndrome

Ear abnormalities

Nothing starts w/ ‘N’

Hemifacial microsomia

A

R

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
-金氏综合征

Goldenhar syndrome

What is Goldenhar’s nonpymous name? Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Which side of the face is more likely to be affected?
The right

Why the right side?

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
- Goldenhar and Treacher Collins syndrome

What is Goldenhar’s non-eponymous name?
- Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Ear abnormalities (maxillary/mandibular hypoplasia)

- Hemifacial microsomia

Which side of the face is more likely to be affected?
The right

Why the right side?
I have no idea
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
-- Lid coloboma
-- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
-- Ear abnormalities (pre-auricular appendages; aural fistulae)
-- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are epibulbar dermoids commonly located?

(next)
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are epibulbar dermoids commonly located?
At the limbus

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Ear abnormalities
Nothing starts w/ ‘N’
Hemifacial microsomia
At the limbus
R

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are epibulbar dermoids commonly located?
At the limbus

Several slides ago I acknowledged that epibulbar dermoids had another legit name. At long last--what is it?

One word: dermoids

Goldenhar
- OAV syndrome
- Lid colobomas
- Dermoid; Duane syndrome
- Ear abnormalities
- Nothing starts w/ ‘N’
- Hemifacial microsomia
- At the limbus
- R

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
  - Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are epibulbar dermoids commonly located?
At the limbus

Several slides ago I acknowledged that epibulbar dermoids had another legit name. At long last—what is it?
Limbal dermoids

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Ear abnormalities
Nothing starts w/ ‘N’
Hemifacial microsomia
At the limbus
Goldenhar syndrome: Limbal (epibulbar) dermoids.
Note also the lid coloboma (arrow)
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are epibulbar dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
-- Lid coloboma
-- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
-- Ear abnormalities (pre-auricular appendages; aural fistulae)
-- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are epibulbar dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired? 
A minority (~10%) have mental retardation

Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Ear abnormalities
Nothing starts w/ ‘N’
Hemifacial microsomia
At the limbus
Retardation in ~10%
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Apert syndrome
- Pfeiffer syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins

What are the notable features of Treacher Collins syndrome (TCS)?

No, intelligence is normal in TCS
Two of the non-syndactylous conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins

What are the notable features of Treacher Collins syndrome (TCS)?
In addition to severe mandibular hypoplasia a la Goldenhar, TCS is marked by severe hypoplasia of the bony structures regions. Hypoplasia in these areas produces downsloping of the lateral orbital rims, which in turn leads to downsloping of the palpebral fissures.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins

What are the notable features of Treacher Collins syndrome (TCS)? In addition to severe mandibular hypoplasia a la Goldenhar, TCS is marked by severe hypoplasia of the malar eminence/zygoma regions. Hypoplasia in these areas produces downsloping of the lateral orbital rims, which in turn leads to downsloping of the palpebral fissures.

Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
Craniofacial Malformations

Treacher Collins syndrome
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Saethre-Chotzen syndrome
- Pfeiffer syndrome
- Apert syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

What are the notable features of Treacher Collins syndrome (TCS)?
In addition to severe mandibular hypoplasia a la Goldenhar, TCS is marked by severe hypoplasia of the malar eminence/zygoma regions. Hypoplasia in these areas produces downsloping of the lateral orbital rims, which in turn leads to downsloping of the palpebral fissures.

Is TCS inherited in sporadic fashion, like Goldenhar?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-cystotic conditions are examples of what are called **branchial arch syndromes**. Which two? Goldenhar and Treacher Collins

What are the notable features of Treacher Collins syndrome (TCS)?
In addition to severe mandibular hypoplasia a la Goldenhar, TCS is marked by severe hypoplasia of the malar eminence/zygoma regions. Hypoplasia in these areas produces downsloping of the lateral orbital rims, which in turn leads to downsloping of the palpebral fissures.

Is TCS inherited in sporadic fashion, like Goldenhar?
No, it is AD
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Apert syndrome
- Pfeiffer syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
Goldenhar and Treacher Collins

What are the notable features of Treacher Collins syndrome (TCS)?
In addition to severe mandibular hypoplasia a la Goldenhar, TCS is marked by severe hypoplasia of the malar eminence/zygoma regions. Hypoplasia in these areas produces downsloping of the lateral orbital rims, which in turn leads to downsloping of the palpebral fissures.

Is TCS inherited in sporadic fashion, like Goldenhar?
No, it is AD

Is cognitive impairment associated with TCS, like Goldenhar?
No, intelligence is normal in TCS
Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins.

What are the notable features of Treacher Collins syndrome (TCS)?
In addition to severe mandibular hypoplasia a la Goldenhar, TCS is marked by severe hypoplasia of the malar eminence/zygoma regions. Hypoplasia in these areas produces downsloping of the lateral orbital rims, which in turn leads to downsloping of the palpebral fissures.

Is TCS inherited in sporadic fashion, like Goldenhar?
No, it is AD

Is cognitive impairment associated with TCS, like Goldenhar?
No, intelligence is normal in TCS
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
Pronounced as PEA-err roe-BAHN
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)? PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence? It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.
How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
P EA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

*How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?*
PEA-err roe-BAHN

*With regard to congenital anomalies, what is meant by the term sequence?*
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues

*In PRS, what is the ‘single developmental malformation’ that triggers the sequence?*
Micrognathia
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin syndrome
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?

Micrognathia

What does micrognathia mean?
**Craniofacial Malformations**

**Craniosynostoses**
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

**Not Craniosynostoses**
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

---

*How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?*

PEA-err roe-BAHN

*With regard to congenital anomalies, what is meant by the term sequence?*

It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.

*In PRS, what is the ‘single developmental malfunction’ that triggers the sequence?*

**Micrognathia**

*What does micrognathia mean?*

It means ‘severe hypoplasia of the mandible’.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
Pronounce Pierre Robin as PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?
Micrognathia

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?
Micrognathia

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties
How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?
Micrognathia

And what is the ‘sequence,’ ie, the malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties

What does glossoptosis refer to?
The position of the tongue being too posterior.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
P EA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?
Micrognathia

And what is the ‘sequence,’ i.e., the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties

What does glossoptosis refer to?
The position of the tongue being too posterior.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?
Micrognathia

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties

What does glossoptosis refer to?
The position of the tongue being too posterior.
Craniofacial Malformations

Pierre-Robin sequence. Note the micrognathia.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues

Wait--there’s nothing remotely ophthalmic about anything in the sequence.
Given this, why are we eye dentists talking about PRS?

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties
How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues

Wait—there’s nothing remotely ophthalmic about anything in the sequence.
Given this, why are we eye dentists talking about PRS?
Because of its association with Stickler syndrome

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome
- Pierre Robin sequence

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.

Wait--there’s nothing remotely ophthalmic about anything in the sequence.
Given this, why are we eye dentists talking about PRS?
Because of its association with Stickler syndrome.

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
In two words (not counting ‘A’), what sort of condition is Stickler syndrome?
A hereditary hyaloideoretinopathy with optically empty vitreous (Note: This is the term used in the BCSC Peds book)

Wait—there’s nothing remotely ophthalmic about anything in the sequence. Given this, why are we eye dentists talking about PRS? Because of its association with Stickler syndrome.

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia \( \rightarrow \) glossoptosis \( \rightarrow \) cleft palate \( \rightarrow \) feeding difficulties
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome
- Pierre Robin sequence

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?

In two words (not counting ‘A’), what sort of condition is Stickler syndrome?
A ‘hereditary vitreoretinopathy’ (Note: This is the term used in the BCSC Peds book)

Wait—there’s nothing remotely ophthalmic about anything in the sequence. Given this, why are we eye dentists talking about PRS?
Because of its association with Stickler syndrome. 

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome
- Pierre Robin sequence

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?

In two words (not counting ‘A’), what sort of condition is Stickler syndrome?
A ‘hereditary vitreoretinopathy’ (Note: This is the term used in the BCSC Peds book)

OK, now in five words (not counting ‘A’ and ‘with’), what sort of condition is Stickler syndrome?
A hereditary hyaloideoretinopathy with optically empty vitreous (Note: This is the term used in the BCSC Retina book)

Wait—there’s nothing remotely ophthalmic about anything in the sequence.
Given this, why are we eye dentists talking about PRS?
Because of its association with Stickler syndrome

And what is the ‘sequence,’ i.e., the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome
- Pierre Robin sequence

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
In two words (not counting ‘A’), what sort of condition is Stickler syndrome?
A ‘hereditary vitreoretinopathy’ (Note: This is the term used in the BCSC Peds book)

OK, now in five words (not counting ‘A’ and ‘with’), what sort of condition is Stickler syndrome?
A ‘hereditary hyaloideoretinopathy with optically empty vitreous’ (Note: This is the term used in the BCSC Retina book)

Wait—there’s nothing remotely ophthalmic about anything in the sequence. Given this, why are we eye dentists talking about PRS?
Because of its association with Stickler syndrome

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome

Pierre Robin sequence

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?

In two words (not counting ‘A’), what sort of condition is Stickler syndrome?
A ‘hereditary vitreoretinopathy’ (Note: This is the term used in the BCSC Peds book)

OK, now in five words (not counting ‘A’ and ‘with’), what sort of condition is Stickler syndrome?
A ‘hereditary hyaloideoretinopathy with optically empty vitreous’ (Note: This is used in the BCSC Retina book)

Wait—there’s nothing remotely ophthalmic about anything in the sequence. Given this, why are we eye dentists talking about PRS?
Because of its association with Stickler syndrome

What is the status of the vitreous?
Optically empty vitreous

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?

In two words (not counting ‘A’), what sort of condition is Stickler syndrome?
A ‘hereditary vitreoretinopathy’ (Note: This is the term used in the BCSC Peds book)

OK, now in five words (not counting ‘A’ and ‘with’), what sort of condition is Stickler syndrome?
A ‘hereditary hyaloideoretinopathy with optically empty vitreous’ (Note: This is the term used in the BCSC Retina book)

Wait—there’s nothing remotely ophthalmic about anything in the sequence.
Given this, why are we eye dentists talking about PRS?
Because of its association with Stickler syndrome

What is the status of the vitreous?
It is liquefied

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?

PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?

It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?

Micrognathia

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?

Micrognathia → glossoptosis → cleft palate → feeding difficulties

Wait—there’s nothing remotely ophthalmic about anything in the sequence. Given this, why are we eye dentists talking about PRS?

Because of its association with Stickler syndrome.

In two words (not counting ‘A’), what sort of condition is Stickler syndrome?

A hereditary vitreoretinopathy (Note: This is the term used in the BCSC Peds book)

OK, now in five words (not counting ‘A’ and ‘with’), what sort of condition is Stickler syndrome?

A hereditary hyaloideoretinopathy with optically empty vitreous (Note: This is the term used in the BCSC Retina book)

What is the status of the vitreous?

It is liquefied.

Optically empty, liquefied vitreous—doesn’t sound so bad. Is Stickler syndrome associated with ocular manifestations that are clinically significant?

Indeed it is, including:

- Glaucoma
- High myopia
- Cataracts
- Lattice degeneration
- Retinal detachment
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?
Micrognathia

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties

Wait—there’s nothing remotely ophthalmic about anything in the sequence. Given this, why are we eye dentists talking about PRS?
Because of its association with Stickler syndrome.

In two words (not counting ‘A’), what sort of condition is Stickler syndrome?
A ‘hereditary vitreoretinopathy’ (Note: This is the term used in the BCSC Peds book)

OK, now in five words (not counting ‘A’ and ‘with’), what sort of condition is Stickler syndrome?
A ‘hereditary hyaloideoretinopathy with optically empty vitreous’ (Note: This is the term used in the BCSC Retina book)

What is the status of the vitreous? It is liquefied

Optically empty, liquefied vitreous—doesn’t sound so bad. Is Stickler syndrome associated with ocular manifestations that are clinically significant?
Indeed it is, including:
- -
- -
- -
- -
Craniofacial Malformations

Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Apert syndrome
- Crouzon syndrome

Not Craniosynostoses
- Fetal alcohol syndrome
- Pierre Robin sequence
- Goldenhar syndrome
- Treacher Collins syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Apert syndrome
- Crouzon syndrome

**How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?**

PEA-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?

It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?

Micrognathia

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?

Micrognathia → glossoptosis → cleft palate → feeding difficulties

**Wait—there’s nothing remotely ophthalmic about anything in the sequence. Given this, why are we eye dentists talking about PRS?**

Because of its association with Stickler syndrome.

In two words (not counting ‘A’), what sort of condition is Stickler syndrome?

A ‘hereditary vitreoretinopathy’ (Note: This is the term used in the BCSC Peds book)

OK, now in five words (not counting ‘A’ and ‘with’), what sort of condition is Stickler syndrome?

A ‘hereditary hyaloideoretinopathy with optically empty vitreous’ (Note: This is the term used in the BCSC Retina book)

Wait—there’s nothing remotely ophthalmic about anything in the sequence.

Given this, why are we eye dentists talking about PRS?

Because of its association with Stickler syndrome.

What is the status of the vitreous?

It is liquefied

**Optically empty, liquefied vitreous—doesn’t sound so bad. Is Stickler syndrome associated with ocular manifestations that are clinically significant?**

Indeed it is, including:

--Glaucoma
--High myopia
--Cataracts
--Lattice degeneration
--Retinal detachment
What are the notable ophthalmic features of fetal alcohol syndrome (FAS)?

- Ptosis
- Shortened fissures
- Epicanthal folds
- Telecanthus

The craniosynostoses are associated with exotropia. Is FAS?

No, it is associated with esotropia.
What are the notable ophthalmic features of fetal alcohol syndrome (FAS)?
A number are lid-related:

- Telecanthus

The craniosynostoses are associated with exotropia. Is FAS?
No, it is associated with esotropia.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
What are the notable ophthalmic features of fetal alcohol syndrome (FAS)?
A number are lid-related:
-- Ptosis
-- Shortened fissures
-- Epicanthal folds
-- Telecanthus

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
Craniofacial Malformations

Fetal alcohol syndrome. Note ptosis, shortened fissures, epicanthal folds, and telecanthus
What are the notable ophthalmic features of fetal alcohol syndrome (FAS)?
A number are lid-related:
--Ptosis
--Shortened fissures
--Epicanthal folds
--Telecanthus

The craniosynostoses are associated with exotropia. Is FAS?
What are the notable ophthalmic features of fetal alcohol syndrome (FAS)?
A number are lid-related:
--Ptosis
--Shortened fissures
--Epicanthal folds
--Telecanthus

The craniosynostoses are associated with exotropia. Is FAS?
No, it is associated with esotropia