Craniofacial Malformations

Basic distinction used in the BCSC Peds book
Craniofacial Malformations

Basic distinction used in the BCSC Peds book

Craniosynostoses

Not Craniosynostoses
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

To what does the term craniosynostosis refer?
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

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

Cranial sutures of the newborn
Craniofacial Malformations

Craniosynostoses

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

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.
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Craniofacial Malformations

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Craniofacial Malformations

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Craniofacial Malformations

Encephalocele
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

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

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Craniofacial Malformations

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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?

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OK then smart guy, what should be done to differentiate between the two?
Neuroimaging.
If an -ocele comes to the attention of an ophthalmologist, where is it most likely to be located?
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Given this location, what entity is it likely to be mistaken for?
A dacryoccele

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Craniofacial Malformations

Nasal encephalocele

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**Craniofacial Malformations**

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

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Craniofacial Malformations

Craniosynostoses

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Craniofacial Malformations

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---If the mass is a dacryocele, it is palpable below the medial canthal tendon.
---If it contains the contents of the upper eyelid, it is palpable below the canthal tendon.
---If it contains the contents of the upper eyelid, it is palpable above the canthal tendon.
---If it is pulsatile, it is a dacryocele.
---If it is not pulsatile, it is a meningoencephalocele.

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

#### Craniosynostoses

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  - To the premature closing of cranial suture(s)

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#### Not Craniosynostoses

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

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- **What would such an outpouching be called?**
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### Craniofacial Malformations

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How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
**Craniofacial Malformations**

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Craniofacial Malformations

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What is syndactyly?
Partial fusion of the digits
Craniofacial Malformations

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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?
Partial fusion of the digits
Craniofacial Malformations

Craniosynostoses

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

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What is brachydactyly?
Craniofacial Malformations

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Syndactyly and brachydactyly

What is syndactyly?
Partial fusion of the digits

What is brachydactyly?
Abnormally short 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?
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The hands and feet

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

Craniosynostoses

Not Craniosynostoses

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To the premature closing of cranial suture(s)

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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 ophthalmology 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
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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
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Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
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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*
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
- 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, ie what is the cause?
The orbits are abnormally shallow
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
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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?
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What serious sequelae can result from the proptosis/shallow orbits?
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
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Not Craniosynostoses

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Exposure keratopathy
Craniofacial Malformations

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

Is the proptosis accompanied by lid retraction?
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
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Not Craniosynostoses

Three have similar facies—\textit{which ones?}

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

\textit{What serious sequelae can result from the proptosis/shallow orbits?}
Exposure keratopathy

\textit{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

What orbital features are common to all three conditions?

- Proptosis
- Hypertelorism

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

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

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Craniofacial Malformations

Craniosynostoses

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

Craniosynostoses
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- Apert syndrome
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Craniofacial Malformations

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Proptosis and hypertelorism

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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, i.e., the PD
In addition to an increased interpupillary distance, hypertelorism will result in an increased distance between what other orbital structures?

**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

*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)
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 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?
The interpupillary distance

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

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
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?

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

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

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Yes
Craniofacial Malformations

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The medial canthi

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

Proptosis and 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?
The interpupillary distance

Is the proptosis accompanied by lid retraction?
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Telecanthus

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

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

The interpupillary distance
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The medial canthi

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

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

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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

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, 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

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
- 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.

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 vs extorted, and diverge vs converge excessively from one another

The interpupillary distance, ie, the PD...
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 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?

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

They are extorted, and diverge excessively from one another

And telecanthus
Craniofacial Malformations

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

Crouzon syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

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

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?

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

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

**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 and telecanthus and...extorted.

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

**What is hypertelorism?**
An abnormally large distance between the 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).

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**The orbits in Crouzon, Apert, and Pfeiffer syndromes have other important abnormalities. What are these?**
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**What is hypertelorism?**
An abnormally large distance between the orbital walls. What easily obtainable measurement is used as a proxy?
The interpupillary distance, ie, the PD.
Craniofacial Malformations

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**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 orbital features are common to all three conditions?**

*Proptosis* and *hypertelorism* and *telecanthus* and... *extorted*, excessively divergent.

Is the proptosis accompanied by lid retraction?

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

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

What is hypertelorism?

An abnormally large distance between the orbital medial 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 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

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

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What determines the amount of divergence between the orbits?

What orbital features are common to all three conditions?

Proptosis and hypertelorism

What is hypertelorism?

An abnormally large distance between the medial orbital walls.

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Exposure keratopathy

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They are extorted, and diverge excessively from one another

What determines the amount of divergence between the orbitals?

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 Crouzon-facies craniosynostoses?

It varies, but is often well above 90
Craniofacial Malformations

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

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Exposure keratopathy.

Is the proptosis accompanied by lid retraction?
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Three have similar facies--which ones?
Crouzon syndrome, Apert syndrome, Pfeiffer syndrome.

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

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Craniofacial Malformations

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

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Craniofacial Malformations

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Craniofacial Malformations

Normal orbital divergence
Craniofacial Malformations

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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 is the cause of proptosis?
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 orbital features are common to all three conditions?
Proptosis and hypertelorism.

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Craniofacial Malformations

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Craniofacial Malformations

Normal orbital divergence

Excessive orbital divergence in Crouzon
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

**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)

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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.

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*. What determines the amount of divergence between the orbits?

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

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Craniofacial Malformations

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proptosis/shallow orbits Exposure keratopathy

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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

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Why do they have proptosis, ie what is the cause?
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Putting it all together, we can see that these pts have a V-pattern exotropia.
Craniofacial Malformations

Crouzon syndrome: Exotropia
Craniofacial Malformations

**Craniosynostoses vs. 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?

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**What determines the amount of divergence between the orbits?**

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

**Craniosynostoses Not Craniosynostoses**

- Crouzon syndrome
- Apert syndrome
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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?

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- Proptosis
- Hypertelorism
- Telecanthus and...extorted, excessively divergent

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

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?

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- Proptosis
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- Exorbitism

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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’

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Craniofacial Malformations

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

Not Craniosynostostoses

*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 do **not** 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
These three have common hand/feet findings:

Crouzon syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

These three have common hand/feet findings:

Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

two terms mentioned previously
Craniofacial Malformations

Craniosynostoses

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

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

Not Craniosynostoses
Craniofacial Malformations

Craniosynostoses

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

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
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Not Craniosynostoses

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Apert syndrome? Apert is known for severe
Craniofacial Malformations

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

Not Craniosynostoses

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Craniofacial Malformations

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

Not Craniosynostoses

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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
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- **Apert syndrome**
- Pfeiffer syndrome
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Not Craniosynostoses

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

Got a mnemonic for this important factoid?
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
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Not Craniosynostoses

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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…

*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

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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’)
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
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Craniofacial Malformations

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foot feature

short vs long

narrow vs broad
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
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- Pfeiffer syndrome
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Craniofacial Malformations

Pfeiffer syndrome
Craniofacial Malformations

Craniosynostoses

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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*
Craniofacial Malformations

Craniosynostoses

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

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

Craniosynostoses

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

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Craniofacial Malformations

Saethre-Chotzen syndrome: Mild syndactyly
Craniofacial Malformations

Craniosynostoses

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

Craniosynostoses

Crouzon syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

Not Craniosynostoses

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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
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Not Craniosynostoses

These three have hand/feet involvement

TLDR
Craniofacial Malformations

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Not Craniosynostoses

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These three have hand/feet involvement

TLDR
Craniofacial Malformations

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

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

Craniosynostoses
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Craniofacial Malformations

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Not Craniosynostoses
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By what other name are branchial arches called?
Craniofacial Malformations

Craniosynostoses

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**Pharyngeal** arches (in fact, this name is preferred, but the *Peds* book uses ‘branchial,’ so we will too)
Craniofacial Malformations
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Craniosynostoses
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How many arches are there, and which are we concerned with?
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

- Goldenhar syndrome
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Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
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Not Craniosynostoses
- Goldenhar syndrome
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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).
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Craniofacial Malformations

Craniosynostoses

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Goldenhar and Treacher Collins

Not Craniosynostoses

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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.
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Craniofacial Malformations

Craniosynostoses

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- Pfeiffer syndrome
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Craniofacial Malformations

Craniosynostoses

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Craniofacial Malformations

Craniosynostoses

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### Craniofacial Malformations

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Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
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Not Craniosynostoses

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Craniosynostoses Not Craniosynostoses

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Craniofacial Malformations

Craniosynostoses

Crouzon syndrome

Saethre-Chotzen syndrome

Goldenhar syndrome

Treacher Collins syndrome

Pharyngeal arch syndromes (branches of branchial arches)

Goldenhar and Treacher Collins

Not Craniosynostoses

Goldenhar syndrome

Treacher Collins syndrome

Pierre Robin sequence

Fetal alcohol syndrome

Craniosynostoses

Crouzon syndrome

Goldenhar syndrome

Treacher Collins syndrome

Saethre-Chotzen syndrome

Pfeiffer syndrome

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Craniofacial Malformations

Craniosynostoses
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What are the muscles of mastication? --Medial (aka internal) pterygoid
--Lateral (aka external) pterygoid
--Masseter
--Temporalis

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

The first and the second branchial arch.

Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome

Not Craniosynostoses
- Pierre Robin sequence
- Fetal alcohol syndrome

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.

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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

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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? --Medial (aka internal) pterygoid
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Craniofacial Malformations

Craniosynostoses

Crouzon syndrome

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Not Craniosynostoses

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Pierre Robin sequence

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Craniosynostoses Not Craniosynostoses

Crouzon syndrome

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--M

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- Mylohyoid
- Anterior belly of the
- Tensor
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Craniosynostoses

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Craniosynostoses Not Craniosynostoses

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Pfeiffer syndrome

Goldenhar syndrome

Goldenhar and Treacher Collins

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--A nterior belly of the digastric

--T ector tympani

--T ector veli palatini

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Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

- Goldenhar syndrome
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**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 syndrome

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).

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**Not Craniosynostoses**

- Goldenhar syndrome
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Craniofacial Malformations

Craniosynostoses

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Craniosynostoses
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Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Goldenhar and Treacher Collins
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There are two non-syndactylic conditions that are examples of what are called branchial arch syndromes. Which two?
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The hyoid arch.

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

Craniofacial Malformations

Craniosynostoses
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- Treacher Collins syndrome
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- Goldenhar syndrome
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- Pierre Robin sequence
- Fetal alcohol syndrome
- Trisomy 13
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**Craniofacial Malformations**

**Craniosynostoses**

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**Not Craniosynostoses**

- Goldenhar syndrome
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- Craniosynostoses
- Not Craniosynostoses

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Craniofacial Malformations

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Craniofacial Malformations

Craniosynostoses

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Pierre Robin sequence

Craniosynostoses Not Craniosynostoses

Crouzon syndrome

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Craniofacial Malformations

Craniosynostoses

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Craniofacial Malformations

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At long last: What is a branchial arch syndrome?
Craniofacial Malformations

Craniosynostoses
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Craniofacial Malformations

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- Crouzon syndrome
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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

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What is the incidence of Goldenhar? About 1/4000 live births.
Craniofacial Malformations

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It is sporadic in the majority of cases
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Is there a sex predilection?
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Yes, M > F are # x as likely to be affected
Craniofacial Malformations

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What is the incidence of Goldenhar?
About 1/4000 live births

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It is sporadic in the majority of cases

Is there a sex predilection?
Yes, males are twice as likely to be affected
Craniofacial Malformations

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- Crouzon syndrome

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- Goldenhar syndrome
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- Pierre Robin sequence
- Goldenhar syndrome

What is Goldenhar’s noneponymous name? (the answer will start with an ‘O’)

Goldenhar
- O
- L
- D
- E
- N
- H
- A

Very convenient mnemonic!
Craniofacial Malformations

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What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

Goldenhar
OAV syndrome

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Craniofacial Malformations

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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!
Craniofacial Malformations

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Goldenhar
OAV syndrome
LODENHAR
Craniofacial Malformations

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Goldenhar syndrome: Butterfly vertebrae
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Vertebral

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Craniosynostoses

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Craniosynostoses Not Craniosynostoses

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two words
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Goldenhar OAV syndrome

Lid colobomas

Very convenient mnemonic!
Craniofacial Malformations

Goldenhar syndrome: Lid coloboma
Craniofacial Malformations

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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 coloboma
Craniofacial Malformations

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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.
Craniofacial Malformations

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- （now this one—actually two, both of which start with ‘D’）

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Goldenhar
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Lid colobomas
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Goldenhar's noneponymous name is 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

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

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

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

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Goldenhar OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

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
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Not Craniosynostoses
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Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

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Craniofacial Malformations

Craniosynostoses

Crouzon syndrome

Not Craniosynostoses

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Goldenhar syndrome

Lid colobomas

Dermoid; Duane syndrome

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They can be, and usually are

Very convenient mnemonic!
Craniofacial Malformations

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In what noncosmetic way are they significant?

Very convenient mnemonic!
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In what noncosmetic way are they significant?
They can be amblyogenic
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Should epibulbar dermoids be excised if they seem to be amblyogenic?

Very convenient mnemonic!
Craniofacial Malformations

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Not Craniosynostoses

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What is Goldenhar’s non-epibulbar dermoid?
Epibulbar dermoid

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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
Craniofacial Malformations

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Very convenient mnemonic!

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OAV syndrome
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Craniofacial Malformations

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OAV syndrome
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Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?

Goldenhar OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Very convenient mnemonic!

What ocular/periorbital abnormalities are common?
-- Lid coloboma
-- Dermoids of the cornea; Duane syndrome

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Goldenhar syndrome
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
ENHAR

Very convenient mnemonic!

What ocular/periocular abnormalities are common?
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- Dermoids of the cornea; Duane syndrome

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Where are dermolipomas typically located?

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Craniofacial Malformations

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Yes. The relationship is that, like dermoids, lipoderoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?
The temporal fornix

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question?
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Goldenhar syndrome
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

Goldenhar syndrome: Dermolipoma
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.

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There is none; they are distinct and separate entities.

Goldenhar syndrome
- Oculo-Auriculo-Vertebral (OAV) syndrome
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Very convenient mnemonic!
Craniofacial Malformations

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OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Craniofacial Malformations

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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.

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Craniofacial Malformations

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Where are dermolipomas typically located?
The temporal fornix

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How can the two be differentiated at the slit lamp?
--By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white)
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What ocular/periocular abnormalities are common?
--Lid coloboma
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Where are dermolipomas typically located?
The temporal fornix.

What about on MRI/CT--how can they be differentiated?
On MRI/CT, prolapsed orbital fat will be seen to be continuous with intraconal fat, whereas a dermolipoma will not.

What ocular/periocular abnormalities are common?
- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question?
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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 term dermolipoma is a controversial one; there is no evidence that these lesions arise from lipomatous tissue. The lesions are usually found in the temporal region.

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.

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Craniofacial Malformations

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Where are dermolipomas typically located?
The term "lipodermoid" refers to a tumor that contains fat and skin, which is not listed among the craniosynostoses. Unlike dermoids, dermolipomas are not associated with branchial arch syndromes. The term "lipodermoid" was coined specifically for this entity. It is not associated with branchial arch syndromes.

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How can the two be differentiated at the slit lamp?

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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.

What is the ‘full’ name of the dermoid in question?
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- Goldenhar and Treacher Collins

What is Goldenhar’s non-eponymous name?
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What ocular/periocular abnormalities are common?
- Lid coloboma
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What nonocular findings are usually present?
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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 syndrome

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
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- Pierre Robin sequence
- Fetal alcohol syndromes

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Duane’s retraction syndrome

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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?
**Craniofacial Malformations**

**Craniosynostoses**
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- Goldenhar syndrome
  - Oculo-Auriculo-Vertebral (OAV) syndrome
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- Pierre Robin sequence
- Fetal alcohol syndrome
- Goldenhar
- OAV syndrome
- Lid retraction syndrome

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**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?**
CN3 innervates the lateral rectus

**Very convenient mnemonic!**
Craniofacial Malformations

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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

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

Goldenhar's non-eponym 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 are dermoids commonly located?
- At the limbus

Are Goldenhar individuals cognitively impaired?
- A minority (~10%) have mental retardation

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

Not Craniosynostoses
- Goldenhar syndrome
- Oculo-Auriculo-Vertebral (OAV) 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

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Where specifically are dermoids commonly located?
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Craniofacial Malformations

Duane syndrome (Type 1, OS)
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

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?

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Where specifically are dermoids commonly located?

At the limbus

Are Goldenhar individuals cognitively impaired?

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Another congenital cranial dysinnervation disorder should come readily to mind--what is it? (Note: It has nothing to do with the craniofacial malformations.)

Duane syndrome

Duane’s retraction syndrome

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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
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Apert 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

Goldenhar's non-eponymous name?
- Oculo-Auriculo-Vertebral (OAV) syndrome

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Where specifically are dermoids commonly located?
- At the limbus

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Very convenient mnemonic!

Duane syndrome
Duane’s retraction syndrome

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Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
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Not Craniosynostoses

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Goldenhar and Treacher Collins

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Where specifically are dermoids commonly located?

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Craniofacial Malformations

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Craniofacial Malformations

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**Duane’s retraction syndrome**

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Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
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- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
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Very convenient mnemonic!
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

Goldenhar syndrome

What is Goldenhar’s non-eponymous name?
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Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Craniofacial Malformations

Craniosynostoses
  - Crouzon syndrome

Not Craniosynostoses
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Goldenhar
  OAV syndrome
  Lid colobomas
  Dermoid; Duane syndrome
  Ear abnormalities
  Nothing starts w/ ‘N’

Very convenient mnemonic!
Goldenhar syndrome: Ear abnormalities

Craniofacial Malformations
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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Goldenhar syndrome
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Ear abnormalities
Nothing starts w/ 'N'
H
A
R

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
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Not Craniosynostoses

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Goldenhar syndrome

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
- 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 and Treacher Collins

What is Goldenhar’s noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?
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What nonocular findings are usually present?
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Which side of the face is more likely to be affected?
The right

Why the right side?
I have no idea

Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Ear abnormalities
Nothing starts w/ ‘N’
Hemifacial microsomia
A
R

Lid coloboma
Dermoids of the cornea; Duane syndrome
Ear abnormalities
Hemifacial microsomia
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

- Goldenhar syndrome
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Goldenhar syndrome

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Craniofacial Malformations

Craniosynostoses
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Not Craniosynostoses
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Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?

Goldenhar and Treacher Collins

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Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

What is Goldenhar’s noneponymous name?
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What nonocular findings are usually present?
-- Ear abnormalities (pre-auricular appendages; aural fistulae)
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Where specifically are dermoids commonly located?
At the limbus

Are Goldenhar individuals cognitively impaired?
A minority (~10%) have mental retardation

Which side of the face is more likely to be affected?
The right

Why the right side?
I have no idea

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

Goldenhar syndrome

Treacher Collins syndrome

Pierre Robin sequence

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Goldenhar’s non-eponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

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- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are epibulbar dermoids commonly located?
(next)
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?

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Where specifically are epibulbar dermoids commonly located?

At the limbus
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

Goldenhar syndrome

What is Goldenhar’s nononymous name?
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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

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
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What is Goldenhar’s noneponymous name?
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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?

**Limb**al dermoids

**Goldenhar**
- OAV syndrome
- Lig colobomas
- Dermoid; Duane syndrome
- Ear abnormalities
- Nothing starts w/ ‘N’
- Hemifacial microsomia
- At the limbus

Very convenient mnemonic!
Goldenhar syndrome: Limbal (epibulbar) dermoids.
Note also the lid coloboma (arrow)
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

Craniosynostoses
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Goldenhar syndrome

Treacher Collins syndrome
- Pierre Robin sequence

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Where specifically are epibulbar dermoids commonly located?
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Are Goldenhar individuals cognitively impaired? (finally)
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

Not Craniosynostoses
- 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 are the notable features of Treacher Collins syndrome (TCS)?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome
- Pfeiffer syndrome
- Apert 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 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 of the lateral orbital rims, which in turn leads to downsloping of the palpebral fissures.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
- Saethre-Chotzen syndrome

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- Pfeiffer syndrome
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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
- Goldenhar syndrome
- Saethre-Chotzen syndrome
- Pfeiffer syndrome
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Is TCS inherited in sporadic fashion, like Goldenhar?
Craniofacial Malformations

Treacher Collins syndrome
Craniofacial Malformations

Craniosynostoses
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Not Craniosynostoses
- 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 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
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome

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*Is TCS inherited in sporadic fashion, like Goldenhar?*
No, it is AD

*Is cognitive impairment associated with TCS, like Goldenhar?*
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
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)?
Craniofacial Malformations

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

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
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How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
PEA-err roe-BAHN
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?
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)?
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?
Craniofacial Malformations

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

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
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How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
PEA-err roe-BAHN

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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 sequence
- Fetal alcohol syndrome

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
Pronounced as: PEA-err roe-BAHN

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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?

It means ‘severe hypoplasia of the mandible’.
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.

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What does micrognathia mean?
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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
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Not Craniosynostoses
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PEA-err roe-BAHN

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**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
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)?
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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

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The position of the tongue being too posterior
Craniofacial Malformations

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- Crouzon syndrome
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Not Craniosynostoses
- Goldenhar syndrome
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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
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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
Craniofacial Malformations

Craniosynostoses
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- Apert syndrome
- Pfeiffer syndrome
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Because of its association with *Stickler syndrome* eponym

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
**Micrognathia → glossoptosis → cleft palate → feeding difficulties**
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Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

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Not Craniosynostoses
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- 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 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
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- Goldenhar syndrome
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- Pierre Robin sequence

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In two words (not counting ‘A’), what sort of condition is Stickler syndrome?
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Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

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Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

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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
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And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties

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?

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)

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What is the status of the vitreous? It is liquefied

And what is the ‘sequence,’ i.e., the subsequent malformations and functional issues?

Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
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- Fetal alcohol syndrome
- Pierre Robin sequence

Not Craniosynostoses

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 is the status of the vitreous? It is liquefied.

Optically empty vitreous--doesn't sound so bad. Is Stickler syndrome associated with ocular manifestations that are clinically significant?

Indeed it is, including:

- "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.

In two words (not counting 'A'), what sort of condition is Stickler syndrome?

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Craniofacial Malformations

Craniosynostoses

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- Apert syndrome
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Not Craniosynostoses

- Goldenhar syndrome
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- Fetal alcohol syndrome

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Micrognathia

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?

Micrognathia → glossoptosis → cleft palate → feeding difficulties

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

OK, now in five words (not counting ‘A’ and ‘with’), what sort of condition is Stickler syndrome?

A ‘hereditary vitreoretinopathy’ (Note: This is the term used in the BCSC Peds book)

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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 Stickler syndrome associated with ocular manifestations that are clinically significant?

- 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:
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--
--
What are the notable ophthalmic features of fetal alcohol syndrome (FAS)?

A number are lid-related:
-- Ptosis
-- Shortened fissures
-- Epicanthal folds
-- Telecanthus
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

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