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

Cranial sutures of the newborn
Craniofacial Malformations

Craniosynostostoses

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 \( \text{closing of cranial suture(s)} \) 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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Not Craniosynostoses
Craniofacial Malformations

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Craniofacial Malformations

Encephalocele
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?

A dacryocele

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

Fine needle biopsy, perhaps?

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

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

Neuroimaging
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Craniofacial Malformations

Nasal encephalocele

Dacryocele
Craniofacial Malformations

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A dacryocele.

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

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

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Craniofacial Malformations

Craniosynostoses  Not Craniosynostoses

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Neuroimaging
**Craniofacial Malformations**

**Craniosynostoses**

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

- What results from premature suture closing?
  - Premature closure produces abnormal growth patterns of the skull and face.
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**Not Craniosynostoses**

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

- What clinically significant finding might result?
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- What would such an outpouching be called?
  - That depends upon its contents…

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- If the answer to relation is below it, the pulsatile should be ?
- If the answer to relation is above it, the pulsatile should also be ?

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Craniofacial Malformations

Craniosynostoses

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### Craniofacial Malformations

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

  - **Neuroimaging**

#### Table: Dacryocele vs. Meningoencephalocele

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

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

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

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

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

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What is syndactyly?
Partial fusion of the digits
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

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What is brachydactyly?
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There are four classic craniofacial synostosis syndromes. What are they?
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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
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Not Craniosynostoses

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Craniofacial Malformations

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

- Not Craniosynostoses

Three have similar facies—which ones?
Craniofacial Malformations

Craniosynostoses

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

Not Craniosynostoses

Three have similar facies—which ones?
Craniofacial Malformations

Crouzon syndrome  Apert syndrome  Pfeiffer syndrome
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
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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
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Not Craniosynostoses

Three have similar facies— which ones?

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

Crouzon syndrome: Proptosis
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
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Not Craniosynostoses

Three have similar facies—which ones?

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

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

The orbits are abnormally shallow
Craniofacial Malformations

Craniosynostoses
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Not Craniosynostoses

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Craniosynostoses

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Not Craniosynostoses

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Exposure keratopathy
Craniofacial Malformations

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Craniofacial Malformations

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Craniofacial Malformations

Crouzon syndrome: Ptosis + inferior scleral show
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
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Craniosynostoses

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

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

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

Proptosis and hypertelorism

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Exposure keratopathy

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The interpupillary distance

What orbital features are common to all three conditions?

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
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The medial canthi

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

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Proptosis and hypertelorism
Craniofacial Malformations

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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?

What is the cause?
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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, 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.

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

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
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Not Craniosynostoses

Three have similar facies—which ones?

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Exposure keratopathy

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

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

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The interpupillary distance, ie, the PD
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
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Not Craniosynostoses

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They are extorted, and diverge excessively from one another

The interpupillary distance, ie, the PD is used as a proxy for measuring the distance between the medial orbital walls.
Craniofacial Malformations

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

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

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The orbits are abnormally shallow

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

**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.

**And telecanthus and...extorted, excessively divergent**
Craniofacial Malformations

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Craniofacial Malformations

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What orbital features are common to all three conditions? Proptosis and hypertelorism...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 medial orbital walls. What easily obtainable measurement is used as a proxy? The interpupillary distance, ie, the PD.

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

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

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

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

What orbital features are common to all three conditions?

Proptosis and hypertelorism and telecanthus and…extorted, excessively divergent

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

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Craniofacial Malformations

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

Three have similar facies—which ones?

What orbital features are common to all three conditions?

Proptosis and hypertelorism

What is hypertelorism?

An abnormally large distance between the orbitals

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What is the distance between the medial orbital walls used as a proxy?

The interpupillary distance, ie, the PD
Craniofacial Malformations

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The interpupillary distance, ie, the PD
Craniofacial Malformations

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

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

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- Proptosis
- Hypertelorism

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Craniofacial Malformations

Craniosynostoses Not Craniosynostoses

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

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What determines the amount of divergence between the orbits?

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

What is the normal angle formed by these walls?

Ninety degrees

What is it in the Crouzon-facies craniosynostoses?

It varies, but is often well above 90.
**Craniofacial Malformations**

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They are extorted, and diverge excessively.

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Exposure keratopathy

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Craniofacial Malformations

Normal orbital divergence
Craniofacial Malformations

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?
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Craniofacial Malformations

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Craniofacial Malformations

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Craniofacial Malformations

Normal orbital divergence

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

They are extorted, and diverge excessively from one another.

Exposure keratopathy

Is the proptosis accompanied by lid retraction?

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

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What is proptosis/shallow orbits?
Exposure keratopathy

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What easily obtainable measurement is used as a proxy?
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What serious sequelae can result from the proptosis/shallow orbits?

Craniosynostoses Not Craniosynostoses
Crouzon syndrome
Apert syndrome
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Craniofacial Malformations

Crouzon syndrome: Exotropia
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Craniofacial Malformations

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**Craniofacial Malformations**

**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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**Craniofacial Malformations**

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Crouzon syndrome: V-pattern exotropia
Why do they have proptosis, ie what is the cause?

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

Exposure keratopathy

Is the proptosis accompanied by lid retraction?

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Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Apert syndrome
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Not Craniosynostoses

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Proptosis, hypertelorism

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An abnormally large distance between the medial orbital walls

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?

Craniosynostoses

Not Craniosynostoses

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

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What is the proptosis accompanied by lid retraction?

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Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
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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’

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

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Craniofacial Malformations

Craniosynostoses

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

Not Craniosynostoses

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

Saethre-Chotzen syndrome
These three have common hand/feet findings:

- Syndactyly and/or brachydactyly

Craniosynostoses

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

Not Craniosynostoses:

These three have common hand/feet findings:

- Syndactyly
- Brachydactyly

two terms mentioned previously
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
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Not Craniosynostoses

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Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
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Not Craniosynostoses

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

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

Craniosynostoses

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

Not Craniosynostoses

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

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

Apert syndrome? Apert is known for severe...
Craniofacial Malformations

Craniosynostoses

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

Not Craniosynostoses

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Craniofacial Malformations

Craniosynostoses
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Craniofacial Malformations

Apert syndrome
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
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- 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, likening most or all of the toes with...**

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, likening most or all of the toes with Apert syndrome can’t pull their fingers ‘apert’ (i.e., ‘apart’)

Got a mnemonic for this important factoid?

Pts with Apert syndrome can’t pull their fingers ‘apert’ (i.e., ‘apart’)

Got a mnemonic for this important factoid?
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- **Apert syndrome**
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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**
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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?_ Mitten deformity (‘A’ for effort if you said ‘boxing glove deformity’)_
Craniofacial Malformations

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

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**Pfeiffer syndrome?**
Craniofacial Malformations

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

Not Craniosynostoses

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

Craniosynostoses

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Craniofacial Malformations

Craniosynostoses

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

Craniosynostoses

Crouzon syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

Not Craniosynostoses

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Craniofacial Malformations

Pfeiffer syndrome
Craniofacial Malformations

Craniosynostoses

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

Not Craniosynostoses

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

Got a mnemonic for remembering this?

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

Craniosynostoses

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

Not Craniosynostoses

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Pfeiffer syndrome? The syndactyly is much less severe than in Apert. The defining feature in Pfeiffer syndrome is that the thumbs and great toes are very short and broad. I remember it because the actress Michelle Pfeiffer is well-known for having short/fat thumbs and great toes—that's why her hands and feet are never seen on camera*

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

Craniosynostoses

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

Not Craniosynostoses

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

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

Craniosynostoses

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

Not Craniosynostoses

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

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

Saethre-Chotzen syndrome: Mild syndactyly
Craniofacial Malformations

Craniosynostoses

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

Not Craniosynostoses

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

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

Craniosynostoses

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

Not Craniosynostoses

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

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

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

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

Not Craniosynostoses

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

TLDR
Craniofacial Malformations

Craniosynostoses

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

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

Not Craniosynostoses

This one does not

TLDR
Craniofacial Malformations

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

Not Craniosynostoses

These three have hand/feet involvement

TLDR
Craniofacial Malformations

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

Not Craniosynostoses

These three have hand/feet involvement

This one does not

TLDR
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
  
  This one does not

- Apert syndrome

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

- Pfeiffer syndrome

  These three have hand/feet involvement

- Saethre-Chotzen syndrome

  This one does not

Not Craniosynostoses

TLDR

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

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

Not Craniosynostoses
- ?
- ?
- ?
Craniofacial Malformations

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

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

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

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

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

Craniosynostoses
- Crouzon syndrome
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Not Craniosynostoses
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Craniofacial Malformations

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

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

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

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
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- Fetal alcohol syndrome

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

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

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

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

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

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

**By what other name are branchial arches called?**
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
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Before we go any further: What is a branchial arch?
Arch-like features of the early embryo; several give rise to the head and neck

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

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
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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
  - Treacher Collins syndrome
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  - Fetal alcohol syndrome

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

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

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

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

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Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

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

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

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

The mandibular arch

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

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

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

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome
- Pharyngeal arch syndromes
  - Goldenhar syndrome
  - Treacher Collins syndrome
  - Pfeiffer syndrome
  - Saethre-Chotzen syndrome
  - Goldenhar syndrome
  - Treacher Collins syndrome
  - Crouzon syndrome
  - Apert syndrome
  - Pfeiffer syndrome
  - Saethre-Chotzen syndrome
  - Goldenhar syndrome
  - Treacher Collins syndrome

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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The mandibular arch

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Craniofacial Malformations
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Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

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Goldenhar and Treacher Collins

Not Craniosynostoses

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

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

The ear (both internal and external)

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

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

Craniosynostoses

Crouzon syndrome

Saethre-Chotzen syndrome

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Goldenhar and Treacher Collins

Not Craniosynostoses

Goldenhar syndrome
Treacher Collins syndrome
Pierre Robin sequence
Fetal alcohol syndrome

Craniosynostoses

Not Craniosynostoses

Goldenhar syndrome
Treacher Collins syndrome
Pierre Robin sequence
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How many arches are there, and which are we concerned with?
There are five or six (depends on who you ask), but we’re only concerned with the first and the second arch.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
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Craniofacial Malformations

Craniosynostoses

Crouzon syndrome

Saethre-Chotzen syndrome

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

Not Craniosynostoses

Goldenhar syndrome

Treacher Collins syndrome

Pierre Robin sequence

Fetal alcohol syndrome

Craniosynostoses Not Craniosynostoses

Goldenhar syndrome

Treacher Collins syndrome

Pierre Robin sequence

Fetal alcohol syndrome

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The mandibular arch

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

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)
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Apert syndrome
- Pfeiffer syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Crouzon syndrome
- Pauli syndrome
- Pierre Robin sequence
- Fetal alcohol syndrome
- Other syndromes

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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The mandibular division of the trigeminal (V3)

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

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

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

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Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

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

Not Craniosynostoses

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

Craniofacial Malformations

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).

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Not Craniosynostoses
- Goldenhar syndrome
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- Fetal alcohol syndrome

What are the muscles of mastication?

What are the muscles of the mandibular arch?

muscles of mastication

the first
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The ear (both internal and external)

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

Goldenhar and Treacher Collins

What are the muscles of mastication?

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

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

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

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Craniofacial Malformations

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Goldenhar and Treacher Collins

Not Craniosynostoses

Goldenhar syndrome

Treacher Collins syndrome

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Craniosynostoses Not Craniosynostoses

Goldenhar syndrome

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Goldenhar and Treacher Collins

What are these MATT muscles of which you speak?

M, A, T, T

What are the muscles of the mandibular arch?

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

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Goldenhar and Treacher Collins.

What are these MATT muscles of which you speak?

- Mylohyoid
- Anterior belly of the
- Tensor
- Tensor
  two Latin words

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**Craniofacial Malformations**

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

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

What are these MATT muscles of which you speak? -- Mylohyoid
-- Anterior belly of the digastric
-- Tensor tympani
-- Tensor veli palatini

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The first
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Craniofacial Malformations

Craniosynostoses
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- Saethre-Chotzen syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
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Craniosynostoses

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Goldenhar and Treacher Collins

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Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Goldenhar and Treacher Collins

Not Craniosynostoses

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- Fetal alcohol syndrome
- Craniosynostoses
- Not Craniosynostoses

Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
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- Craniosynostoses
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Goldenhar and Treacher Collins

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

The hyoid arch

What nerve innervates the musculature of the hyoid arch?

The facial nerve

What are the muscles of the hyoid arch?

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

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

It does indeed.
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Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
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- Fetal alcohol syndrome
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
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Craniofacial Malformations

First (mandibular) arch
Second (hyoid) arch
Third
Fourth
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Craniofacial Malformations

Craniosynostoses

Crouzon syndrome

Craniosynostoses

Goldenhar syndrome

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Goldenhar syndrome

Pierre Robin sequence

Pfeiffer syndrome

Saethre-Chotzen syndrome

Goldenhar syndrome

Treacher Collins syndrome

Crouzon syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

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

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Craniofacial Malformations

Craniosynostoses

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Craniofacial Malformations

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Craniosynostoses Not Craniosynostoses

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Craniofacial Malformations

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- Crouzon syndrome

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Craniofacial Malformations

Craniosynostoses

Crouzon syndrome

Not Craniosynostoses

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Treacher Collins syndrome

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Craniosynostoses

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

It does indeed.

What are these PASS muscles?

--P

--A

--S

PASS

--S

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Craniofacial Malformations

Craniosynostoses
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It does indeed

What are these PASS muscles?
-- Posterior belly of the digastric
-- Auricular muscles
-- Stylohyoid
-- Stapedius

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Craniofacial Malformations

Craniosynostoses

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Craniosynostoses Not Craniosynostoses

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Craniofacial Malformations

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The second
Craniofacial Malformations

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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

*At long last: What is a branchial arch syndrome?*
Two of the non-syndactylic conditions are examples of what are called \textit{branchial arch syndromes}. Which two? Goldenhar and Treacher Collins.

\textit{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.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
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Not Craniosynostoses
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At long last: What is a branchial arch syndrome? As one might expect, it is a constellation of craniofacial abnormalities secondary to maldevelopment or dysgenesis of structures arising from the mandibular and/or hyoid branchial arches.

Which structures are those?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

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

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

At long last: **What is a branchial arch syndrome?**
As one might expect, it is a constellation of craniofacial abnormalities secondary to maldevelopment or dysgenesis of structures arising from the mandibular and/or hyoid branchial arches.

Which structures are those?
For starters: The mandible, the maxilla, the muscles of facial expression and mastication, and the ears.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

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

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

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

What is its inheritance pattern? It is sporadic.

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

Craniosynostoses
- Crouzon syndrome
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Craniofacial Malformations

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- Pfeiffer syndrome
- Apert syndrome
- Crouzon syndrome

Not Craniosynostoses
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Craniofacial Malformations

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Not Craniosynostoses
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Yes, $M \times F$ are $\#x$ as likely to be affected
Craniofacial Malformations

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Craniosynostoses

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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? (the answer will start with an ‘O’)

What ocular/periocular abnormalities are common?

- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?

- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?

At the limbus

Are Goldenhar individuals cognitively impaired?

A minority (~10%) have mental retardation

Goldenhar

OLDENHAR

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

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

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

Goldenhar syndrome
- OAV syndrome

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

- Goldenhar syndrome
  - Oculo-Auriculo-Vertebral (OAV) syndrome

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

What is the classic vertebral finding? GOLDENHAR

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

What is the classic vertebral finding? Hemivertebrae, aka butterfly vertebrae

Goldenhar syndrome
OAV syndrome

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
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What is Goldenhar's non-eponymous name? Oculo-Auriculo-Vertebral (OAV) syndrome

What is the classic vertebral finding? Hemivertebrae, aka **butterfly vertebrae**

Goldenhar OAV syndrome

Very convenient mnemonic!
Craniofacial Malformations

Goldenhar syndrome: Butterfly vertebrae
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?

Goldenhar syndrome
- Oculo-Auriculo-Vertebral (OAV) syndrome

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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Where specifically are dermoids commonly located?
At the limbus

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Goldenhar (OAV) syndrome

Very convenient mnemonic!

What is the classic vertebral finding?
Hemivertebrae, aka butterfly vertebrae

Another syndrome of ophthalmic concern includes butterfly vertebrae as a finding. What is it?

Alagille syndrome
- Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
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What is a posterior embryotoxon?
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Craniofacial Malformations

Craniosynostoses
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Not Craniosynostoses

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L D E N H A R

Very convenient mnemonic!

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Alagille syndrome

Rule out
Craniofacial Malformations

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Not Craniosynostoses

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Goldenhar OAV syndrome
L D E N H A R

Very convenient mnemonic!

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Alagille syndrome

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Craniosynostoses
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Goldenhar and Treacher Collins

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Oculo-Auriculo-Vertebral (OAV) syndrome

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Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

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- Treacher Collins syndrome
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What is Goldenhar’s noneponymous name?

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At the limbus

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What is the classic vertebral finding?

Hemivertebrae, aka *butterfly vertebrae*

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Alagille syndrome

What is the noneponymous name for Alagille syndrome?

Arterohepatic dysplasia

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Craniofacial Malformations

Craniosynostoses

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Where specifically are dermoids commonly located? At the limbus

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Very convenient mnemonic! V-Vertebral, L-Lid, D-Dermoids, E-Ear, N-Hemifacial, A-Auricular, R-Microsomia

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What is the classic vertebral finding? Hemivertebrae, aka butterfly vertebrae
Posterior embryotoxon
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
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Goldenhar
O
A
V syndrome

Goldenhar
Craniofacial Malformations

Craniosynostoses
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A minority (~10%) have mental retardation

Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
DENHAR

Very convenient mnemonic!
Craniofacial Malformations

Goldenhar syndrome: Lid coloboma
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

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

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

What ocular/periocular abnormalities are common?
--Lid coloboma

Does the coloboma tend to be in the upper lid, or the lower?

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

Goldenhar syndrome

Treacher Collins syndrome

Pierre Robin sequence

Goldenhar syndrome

Oculo-Auriculo-Vertebral (OAV) syndrome

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

What ocular/periocular abnormalities are common? -- Lid coloboma

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

Craniosynostoses
- Crouzon syndrome

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

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

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

What ocular/periocular abnormalities are common?
- Lid coloboma
- (now this one--actually two, both of which start with ‘D’)

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
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

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

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

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

What is the ‘full’ name of the dermoid in question?
- Epibulbar dermoid

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

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

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

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Note: There is another legit answer, so if you came up with that one, no worries (we’ll identify it shortly)

Very convenient mnemonic!
Goldenhar syndrome: Epibulbar dermoid
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

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

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

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?

Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
E
D
H
A
R
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

Goldenhar and Treacher Collins syndrome

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

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

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

Very convenient mnemonic!

Goldenhar
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

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

Not Craniosynostoses

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

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

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

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

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

What is the ‘full’ name of the dermoid in question?
- Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
- They can be, and usually are

In what noncosmetic way are they significant?

Goldenhar

OAV syndrome

Lid colobomas

Dermoid; Duane syndrome

Very convenient mnemonic!
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 noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

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

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

In what noncosmetic way are they significant?
They can be amblyogenic
Craniofacial Malformations

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

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

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

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

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

Very convenient mnemonic!

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically other than cosmesis?
They can be, and usually are

In what noncosmetic way are they significant?
They can be amblyogenic

Should epibulbar dermoids be excised if they seem to be amblyogenic?
Yes, although the success rate is not high (the post-op scar is often amblyogenic in and of itself)
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

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

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

Goldenhar and Treacher Collins

What is Goldenhar’s noneponymous name?

Oculo-Auriculo-Vertebral (OAV) syndrome

What ocular/periocular abnormalities are common?

- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?

- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?

At the limbus

Are Goldenhar individuals cognitively impaired?

A minority (~10%) have mental retardation

What is the ‘full’ name of the dermoid in question?

Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?

They can be, and usually are

Should epibulbar dermoids be excised if they seem to be amblyogenic?

Yes, although the success rate is not high (the post-op scar is often amblyogenic in and of itself)

In what noncosmetic way are they significant?

They can be amblyogenic

Very convenient mnemonic!

Goldenhar

OAV syndrome

Lid colobomas

Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome

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

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

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

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

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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Goldenhar and Treacher Collins

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- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

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

What is the 'full' name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities

Very convenient mnemonic!

Goldenhar OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
ENHAR

Very convenient mnemonic!
Craniofacial Malformations

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

- Lid coloboma
- Dermoids of the cornea; Duane syndrome

What ocular/periocular abnormalities are common?

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!
Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

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

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

What nonocular findings are usually present?
--Ear abnormalities (pre-auricular appendages; aural fistulae)
--Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

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

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities.
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)? Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?

Goldenhar syndrome
OAV (Oculo-Auriculo-Vertebral) syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis? They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts? There is none; they are distinct and separate entities.
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar.

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?
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

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.

What is the relationship between epibulbar dermoids and dermoid cysts? There is none; they are distinct and separate entities.

Goldenhar syndrome
Oculo-Auriculo-Vertebral (OAV) syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?
The temporal fornix.

With what similar-appearing but etiologically-distinct condition are dermolipomas confused?
Prolapsed orbital fat.

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid.

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities.

Goldenhar syndrome
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?
The temporal fornix.

With what similar-appearing but etiologically-distinct condition are dermolipomas confused?
Prolapsed orbital fat.

How can the two be differentiated at the slit lamp?
- By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white)
- By texture (prolapsed fat is squishy, whereas a dermolipoma is firm)

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

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid.

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities.
Is there a relationship between epibulbar dermoids and lipodermoid (aka dermolipomas)? Yes. The relationship is that, like dermoids, lipodermoid are associated with Goldenhar syndrome.

Where are dermolipomas typically located? The temporal fornix.

With what similar-appearing but etiologically-distinct condition are dermolipomas confused? Prolapsed orbital fat.

How can the two be differentiated at the slit lamp? By color and texture.

What ocular/periocular abnormalities are common? Lid colobomas, dermoids of the cornea, Duane syndrome.

What is the ‘full’ name of the dermoid in question? Epibulbar dermoid.

Are epibulbar dermoids clinically significant for reasons other than cosmesis? They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts? There is none; they are distinct and separate entities.

Goldenhar syndrome
Oculo-Auriculo-Vertebral (OAV) syndrome
Lid colobomas
Dermoid; Duane syndrome
Very convenient mnemonic!
Craniofacial Malformations

**Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?**
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

**Where are dermolipomas typically located?**
The temporal fornix.

**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** (prolated fat is yellow, whereas a dermolipoma is pinkish-white),
--By **texture**

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

**What is the ‘full’ name of the dermoid in question?**
Epibulbar dermoid.

**Are epibulbar dermoids clinically significant for reasons other than cosmesis?**
They can be, and usually are.

**What is the relationship between epibulbar dermoids and dermoid cysts?**
There is none; they are distinct and separate entities.
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?
The 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

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid.

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities.

Goldenhar
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

Where are dermolipomas typically located?
The temporal fornix

With what similar-appearing but etiologically-distinct condition are dermolipomas confused?
Prolapsed orbital fat

How can the two be differentiated at the slit lamp?
-- By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white)
-- By texture (prolapsed fat is squishy, whereas a dermolipoma is firm)

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities
Craniofacial Malformations

**Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?**
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

**Where are dermolipomas typically located?**
The temporal fornix.

**With what similar-appearing but etiologically-distinct condition are dermolipomas confused?**
Prolapsed orbital fat.

**How can the two be differentiated at the slit lamp?**
--By **color** (prolated fat is yellow, whereas a dermolipoma is pinkish-white)
--By **texture** (prolated fat is squishy, whereas a dermolipoma is firm)

--Lid coloboma

**What is the ‘full’ name of the dermoid in question?**
Epibulbar dermoid

**Are epibulbar dermoids clinically significant for reasons other than cosmesis?**
They can be, and usually are.

**What is the relationship between epibulbar dermoids and dermoid cysts?**
There is none; they are distinct and separate entities.

Goldenhar syndrome
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
ever convenient mnemonic!
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located?
The temporal fornix.

How can the two be differentiated on MRI/CT?
-- By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white);
-- By texture (prolapsed fat is squishy, whereas a dermolipoma is firm).

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities.

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

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 tendrils of the lipodermoids enter the orbit and, once inside, become continuous with intraconal fat.

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

With what condition are dermolipomas confused? Prolapsed orbital fat.

How can the two be differentiated at the slit lamp? -- By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white)
-- By texture (prolapsed fat is squishy, whereas a dermolipoma is firm)

On 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 colobomas
-- Dermoids of the cornea; Duane syndrome

What is the ‘full’ name of the dermoid in question? Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis? They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts? There is none; they are distinct and separate entities.

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!
**Craniofacial Malformations**

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 tegumentary covering of the eyelids and the temporal fornix.

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

Do dermolipomas need to be excised?
Generally no--they should be left alone.

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are.

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities.
Craniofacial Malformations

Is there a relationship between epibulbar dermoids and lipodermoids (aka dermolipomas)?
Yes. The relationship is that, like dermoids, lipodermoids are associated with Goldenhar.

Where are dermolipomas typically located?
The te

Do dermolipomas need to be excised?
Generally no—they should be left alone.

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

With what similar-appearing but etiologically-distinct condition are dermolipomas confused?
Prolapsed orbital fat

How can the two be differentiated at the slit lamp?
--By color (prolapsed fat is yellow, whereas a dermolipoma is pinkish-white)
--By texture (prolapsed fat is squishy, whereas a dermolipoma is firm)

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

What is the ‘full’ name of the dermoid in question?
Epibulbar dermoid

Are epibulbar dermoids clinically significant for reasons other than cosmesis?
They can be, and usually are

What is the relationship between epibulbar dermoids and dermoid cysts?
There is none; they are distinct and separate entities

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

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

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

What nonocular findings are usually present?
- Ear abnormalities (pre-auricular appendages; aural fistulae)
- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
- At the limbus

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

Duane syndrome has a ‘middle’ name—what is it?
Duane’s retraction syndrome

Very convenient mnemonic!

Duane syndrome
- Lid retraction
- Hypothenar hypoplasia
- Dactylyolysis

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

Goldenhar syndrome has a 'middle' name--what is it?

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What is Goldenhar's non-eponymous name?

Oculo-Auriculo-Vertebral (OAV) syndrome

Goldenhar's non-eponymous name:

OAV syndrome

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CN3 innervates the lateral rectus

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Craniofacial Malformations

Duane syndrome (Type 1, OS)
Craniofacial Malformations

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Marcus-Gunn jawk 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?
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**Marcus-Gunn jaw wink**

In MGJW, which cranial nerve incorrectly innervates which muscle?
CN5 innervates the levator

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OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Ear abnormalities
Nothing starts w/ ‘N’
H
A
R
Craniofacial Malformations

Goldenhar syndrome: Ear abnormalities
Craniofacial Malformations

Craniosynostoses

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Goldenhar syndrome: Hemifacial microsomia
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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

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

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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?
- Limbal dermoids

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

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

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

What nonocular findings are usually present?
-- Ear abnormalities (pre-auricular appendages; aural fistulae)
-- Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are epibulbar dermoids commonly located?
At the limbus

Several slides ago I acknowledged that epibulbar dermoids had another legit name. At long last—what is it?
Limbal dermoids

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

Craniosynostoses
- Crouzon syndrome

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

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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Where specifically are epibulbar dermoids commonly located?
At the limbus

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

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Where specifically are epibulbar dermoids commonly located?
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Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
Ear abnormalities
Nothing starts w/ ‘N’
Hemifacial microsomia
At the limbus
Retardation in ~10%
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
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- Treacher Collins syndrome
- Pierre Robin sequence
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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)?

No, intelligence is normal in TCS
Craniofacial Malformations

Craniosynostoses
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- Saethre-Chotzen syndrome

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

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

What are the notable features of Treacher Collins syndrome (TCS)? In addition to severe mandibular hypoplasia a la Goldenhar, TCS is marked by severe hypoplasia of the bony structures.
Craniofacial Malformations

Craniosynostoses
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- Goldenhar syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses
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- Pfeiffer syndrome
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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.
Craniofacial Malformations

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- Pierre Robin sequence
- Fetal alcohol syndrome

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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
- Saethre-Chotzen syndrome

Not Craniosynostoses

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Craniofacial Malformations

Treacher Collins syndrome
Craniofacial Malformations

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With regard to congenital anomalies, what is meant by the term sequence?
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Micrognathia

What does micrognathia mean?
Craniofacial Malformations

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Micrognathia

What does micrognathia mean?
It means ‘severe hypoplasia of the mandible’
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And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties
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What does glossoptosis refer to?
The position of the tongue being too posterior
Craniofacial Malformations

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The position of the tongue being too posterior.
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Craniofacial Malformations

Pierre-Robin sequence. Note the micrognathia.
Craniofacial Malformations

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Micrognathia→glossoptosis→cleft palate→feeding difficulties
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How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?

In two words (not counting ‘A’), what sort of condition is Stickler syndrome?
A 'hereditary vitreoretinopathy' (Note: This is the term used in the BCSC Peds book)

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OK, now in five words (not counting ‘A’ and ‘with’), what sort of condition is Stickler syndrome?
A [two words] with [three words] (Note: This is the term used in the BCSC Retina book)

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Craniofacial Malformations

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Not Craniosynostoses
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- Pierre Robin sequence

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Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

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What is the status of the vitreous?
Optically empty vitreous

Wait—there’s nothing remotely ophthalmic about anything in the sequence. Given this, why are we eye dentists talking about PRS?
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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
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
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Not Craniosynostoses

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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

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?

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What is the status of the vitreous? It is liquefied

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Craniofacial Malformations

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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:

- Micrognathia
- Glossoptosis
- Cleft palate
- Feeding difficulties

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Not Craniosynostoses

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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

Optically empty, liquefied vitreous—doesn’t sound so bad.

Is Stickler syndrome associated with ocular manifestations that are clinically significant?

Indeed it is, including:

-- Glaucoma
-- High myopia
-- Cataracts
-- Lattice degeneration
-- Retinal detachment

What is the status of the vitreous?

It is liquefied

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

A ‘hereditary hyaloideoretinopathy with optically empty vitreous’ (Note: This is the term used in the BCSC Retina book)

Wait—there’s nothing remotely ophthalmic about anything in the sequence. Given this, why are we eye dentists talking about PRS?

Because of its association with Stickler syndrome.

For more on Stickler syndrome, see set R3
What are the notable ophthalmic features of fetal alcohol syndrome (FAS)?

- Ptosis
- Shortened fissures
- Epicanthal folds
- Telecanthus

Craniofacial Malformations

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

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- **Fetal alcohol syndrome**
What are the notable ophthalmic features of fetal alcohol syndrome (FAS)? A number are lid-related:

- Telecanthus

The craniosynostoses are associated with exotropia. Is FAS?

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Fetal alcohol syndrome. Note ptosis, shortened fissures, epicanthal folds, and telecanthus
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Fetal alcohol syndrome: Esotropia
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A globe malformation is common in FAS, so much so as to help cinch the dx. What is it?

Microphthalmia

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What is microphthalmia?

To a small, disorganized, and malformed globe.
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What is the term for a (way) small eye that is otherwise relatively normal anatomically?

Nanophthalmos

How short does an eye have to be to qualify as nanophthalmic?

There is no set number; most studies use anywhere from 19 to 21 mm. The BCSC Path book uses 20.5, but the Peds book 18. EyeWiki uses 20. Caveat emptor.

What is the refractive status of these eyes?

They are almost always severely hyperopic

What two (often intertwined) pathologies are nanophthalmic eyes prone to?

Uveal effusion and angle-closure glaucoma
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Uveal effusion and angle-closure glaucoma are two (often intertwined) pathologies prone to nanophthalmic eyes.

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**Microphthalmia**

To what does the term *microphthalmia* refer?

To a small, disorganized, and malformed globe but organized/well-formed.

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