Craniofacial Malformations

Basic distinction used in the BCSC Peds book
Craniofacial Malformations

Basic distinction used in the BCSC Peds book

Craniosynostoses  Not Craniosynostoses
To what does the term craniosynostosis refer?
To what does the term craniosynostosis refer?
To the premature closing of cranial suture(s)
Craniofacial Malformations

Craniosynostoses

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

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

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

Craniosynostoses

Not Craniosynostoses

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

The opposite of craniosynostosis--ie, the failure of sutures to completely close--is that 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.

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

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

Craniosynostoses

Not Craniosynostoses

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Craniofacial Malformations

Craniosynostoses

Not Craniosynostoses

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

If the outpouching contains meninges, it’s called a **meningocele**.
If it contains brain tissue, it’s called an **encephalocele**.
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Craniofacial Malformations

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At the medial canthus

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Craniofacial Malformations

Craniosynostoses

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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?

--If the outpouching contains meninges, it's called a meningoceles
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Craniofacial Malformations

Craniosynostoses

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

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

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

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If the mass is pulsatile...
--If it comes...
--If it comes...

Relation to medial canthal tendon

Dacryocele

Below it

Mening/enceph-ocele

Above it
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Neuroimaging

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Relation to medial canthal tendon

Pulsatile?

What causes a mening/enceph-ocele to pulsate?

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Craniofacial Malformations

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

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Neuroimaging

---If the \( -ocele \) comes to the attention of an ophthalmologist, where is it most likely to be located?
---If it comes to the attention of an otorhinolaryngologist, where is it most likely to be located?
---If it comes to the attention of a radiologist, where is it most likely to be located?

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What causes a mening/enceph-ocele to pulsate?

Direct transmission of the intracranial pulse pressure
Craniofacial Malformations

Craniosynostoses

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

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

How does craniosynostosis present (ie, isolated/sporadic; syndromic; etc)?
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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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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 ophtho residents can name two of these, if not all three

You a baller if you can name this one
Craniofacial Malformations

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Not Craniosynostoses

How is this pronounced?
Craniofacial Malformations

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Not Craniosynostoses

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

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All four have the same inheritance pattern--what is it?
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Why is hypertelorism always associated with telecanthus?

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

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They are extorted, and diverge excessively from one another

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

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

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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 determines the amount of divergence between the orbits? They are extorted, and diverge excessively from one another.

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

What important abnormalities are common to all three conditions? Exposed sclera, telecanthus and...extorted, excessively divergent.

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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 degrees.

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

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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?
They are extorted, and diverge excessively from one another

What is it in the Pfeiffer syndrome?
They have other important abnormalities. What are these?

What is it in the Saethre-Chotzen syndrome?
They also have other abnormalities. What are these?

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

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

What easily obtainable measurement is used as a proxy?
The interpupillary distance, ie, the PD

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

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

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

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

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

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

What 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°

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

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

What orbital features are common to all three conditions? Proptosis

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

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

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

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Craniofacial Malformations

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

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

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

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. As a result, the angle formed by the lateral orbital walls with respect to one another is greater than normal. This produces a horizontal deviation (arcuate deviation) and telecanthus and…

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

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

**What orbital features are common to all three conditions?** Proptosis/shallow orbits, exposure keratopathy

**What easily obtainable measurement is used as a proxy for the distance between the medial orbital walls?** The interpupillary distance, ie, the PD

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

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

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

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

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

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

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

Well, the excessive divergence produces exotropia, and the extorsion causes the exotropia to be worse in upgaze. Putting it all together, we can see that these pts have a V-pattern exotropia.

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

**Exposure keratopathy**

They are extorted, and diverge excessively from one another.

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

Extruded, and excessively divergent.

**What is hypertelorism?**

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

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

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

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

They are extorted, and diverge excessively from one another. As expected, these conditions?

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

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

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

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

What is the cause of proptosis, ie what is it? The orbits are abnormally shallow
Craniofacial Malformations

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

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

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

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

Craniosynostoses Not Craniosynostoses

Crouzon syndrome
Apert syndrome
Pfeiffer syndrome
Saethre-Chotzen syndrome
Three have similar facies--which ones?

What orbital features are common to all three conditions?

Proptosis and hypertelorism

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

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

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

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

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

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

OK, the orbits are extorted. Does this lead to clinical sequelae of consequence?
Indeed it does. Consider: Because the orbits 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.

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Well, the excessive divergence produces exotropia, and the extorsion causes the extrophia to be worse in upgaze. Putting it all together we can see that these pts have a V-pattern exotropia.

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

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

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

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

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

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

Three have similar facies--which ones?

What orbital features are common to all three conditions?

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

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

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

What 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)

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

Craniosynostoses

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

Not Craniosynostoses

Three have similar facies--which ones?

What orbital features are common to all three conditions?

Proptosis, hypertelorism, telecanthus

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

The orbits are abnormally shallow

What is hypertelorism?

An abnormally large distance between the medial orbital walls

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’
Craniofacial Malformations

Craniosynostoses

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

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

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

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

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

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

Not Craniosynostoses

*These three have common hand/feet findings:*

two terms mentioned previously
Craniofacial Malformations

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

Not Craniosynostoses

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

Craniosynostoses

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

Not Craniosynostoses

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

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

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

Not Craniosynostoses

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

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Craniofacial Malformations

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

Not Craniosynostoses

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Craniofacial Malformations

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

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

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

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

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

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

*Got a mnemonic for this important factoid?*
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

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In that regard, how do they tend to manifest in... Apert syndrome? **Apert is known for severe syndactyly**, which typically involves complete fusion of the index through pinky fingers, only the thumb is free. Likewise, most or all of the toes will

Got a mnemonic for this important factoid?

Pts with Apert syndrome can’t pull their fingers ‘apert’ (ie, ‘apart’)
Craniofacial Malformations

Craniosynostoses

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

Not Craniosynostoses

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

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

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

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

Not Craniosynostoses

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

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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**
- Pfeiffer syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

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

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

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

**Pfeiffer syndrome?**
Craniofacial Malformations

Craniosynostoses

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

Not Craniosynostoses

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

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

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

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

Not Craniosynostoses

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

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

Crouzon syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

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

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

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

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

Craniosynostoses

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

Not Craniosynostoses

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

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

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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.
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…

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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

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

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

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

Pfeiffer syndrome?
The syndactyly is much less severe than in Apert. The defining feature in Pfeiffer syndrome is that the thumbs and great toes are very short and broad—thats why her hands and feet are never seen on camera*

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

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

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

Craniosynostoses

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

Not Craniosynostoses

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

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

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

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

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

Craniosynostoses

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

Not Craniosynostoses

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

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

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

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

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

Craniosynostoses

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

Not Craniosynostoses

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

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

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

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

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

Craniosynostoses

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

Not Craniosynostoses

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

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

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

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

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

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

Not Craniosynostoses

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

TLDR
Craniofacial Malformations

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

Not Craniosynostoses

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

This one does not

TLDR
Craniofacial Malformations

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

Not Craniosynostoses

These three have hand/feet involvement

TLDR
Craniofacial Malformations

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

Not Craniosynostoses
- This one does not
- These three have hand/feet involvement

TLDR
Craniofacial Malformations

Craniosynostoses

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

Not Craniosynostoses

- This one does not
- These three present with exorbitism, ie, ‘bug eyed’
- These three have hand/feet involvement
- This one does not

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

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

Not Craniosynostostoses:
- 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?
Two of the non-syndactylic conditions are examples of what are called branchial arch syndromes. Which two? Goldenhar and Treacher Collins
Craniofacial Malformations

Craniosynostostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostostoses
- Goldenhar syndrome
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Before we go any further: What is a branchial arch?
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

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
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- 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?
Craniofacial Malformations

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- Saethre-Chotzen syndrome

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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

Craniosynostoses
- Crouzon syndrome
- Saethre-Chotzen syndrome

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- Goldenhar syndrome
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- Fetal alcohol syndrome

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

Craniosynostoses

- Crouzon syndrome

- Goldenhar syndrome

- Saethre-Chotzen syndrome

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

Goldenhar and Treacher Collins

Not Craniosynostoses

- Goldenhar syndrome

- Treacher Collins syndrome

- Pierre Robin sequence

- Fetal alcohol syndrome

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

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

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- Saethre-Chotzen syndrome

Not Craniosynostoses
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Craniofacial Malformations

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

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Craniofacial Malformations

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Craniofacial Malformations

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

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

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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

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Goldenhar and Treacher Collins

What are the muscles of mastication?

muscles of mastication

the first
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What are the muscles of the mandibular arch?

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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)

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

Goldenhar and Treacher Collins

What are the muscles of mastication?

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

muscles of mastication

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Craniofacial Malformations

Not Craniosynostoses

Goldenhar syndrome

Treacher Collins syndrome

Pierre Robin sequence

Fetal alcohol syndrome

Craniosynostoses

Crouzon syndrome

Saethre-Chotzen syndrome

Goldenhar syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

Goldenhar syndrome

Treacher Collins syndrome

Pierre Robin sequence

Fetal alcohol syndrome

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--Masseter
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muscles of mastication

the first
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Craniofacial Malformations

Not Craniosynostoses

Crouzon syndrome

Goldenhar syndrome

Saethre-Chotzen syndrome

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Pfeiffer syndrome

Goldenhar syndrome

Apert syndrome

Pierre Robin sequence

Fetal alcohol syndrome

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

Goldenhar and Treacher Collins

What are these MATT muscles of which you speak?

M

A

T

T

What are the muscles of the mandibular arch?

MATT

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

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

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

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

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

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

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

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

Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome
- Saethre-Chotzen syndrome

Not Craniosynostoses

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

Two of the non-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

What are the muscles of the mandibular arch?

The muscles of mastication, and... what's a mnemonic, BTW. MATT (the first)
Before we go any further: What is a branchial arch?
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The ear (both internal and external)

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Goldenhar and Treacher Collins

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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

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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

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? 

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

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

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

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

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

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

Does the hyoid arch contribute to the ear like the first arch? It does indeed.

Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

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

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

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

Craniofacial Malformations

Craniosynostostoses

- Crouzon syndrome

Not Craniosynostostoses

- Goldenhar syndrome
- Treacher Collins syndrome

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

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Here 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?

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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).

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Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

- Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Oligohydramnios
- 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 the non-numeric name of the second arch? The hyoid arch.

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

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?

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By what other name are branchial arches called?

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Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

Goldenhar and Treacher Collins

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

Hyoid arch

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

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

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

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

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

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Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Goldenhar syndrome
- Treacher Collins syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Crouzon syndrome

Not Craniosynostoses

Craniosynostoses
- Crouzon syndrome

Goldenhar syndrome
- Treacher Collins syndrome
- Apert syndrome
- Pfeiffer syndrome
- Saethre-Chotzen syndrome
- Goldenhar syndrome
- Treacher Collins syndrome
- Crouzon syndrome

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

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

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

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

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

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

Craniofacial Malformations

Craniosynostostoses

- Crouzon syndrome

- Goldenhar syndrome

- Treacher Collins syndrome

- Pfeiffer syndrome

- Saethre-Chotzen syndrome

- Goldenhar syndrome

- Treacher Collins syndrome

Not Craniosynostostoses

- Craniosynostoses

- Not Craniosynostoses

Craniosynostoses

- Crouzon syndrome

- Goldenhar syndrome

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- Pfeiffer syndrome

- Saethre-Chotzen syndrome

Goldenhar syndrome

Treacher Collins syndrome

Crouzon syndrome

Apert syndrome

Pfeiffer syndrome

Saethre-Chotzen syndrome

Goldenhar syndrome

Goldenhar syndrome

Treacher Collins syndrome

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

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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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?

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

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Craniofacial Malformations

Craniosynostoses

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- Fetal alcohol syndrome

Craniosynostoses

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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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What are these PASS muscles?

- P
- A
- S
- S

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- Auricular muscles
- Stylohyoid
- Stapedius

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Crouzon syndrome

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Goldenhar and Treacher Collins

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

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

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

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
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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 is Goldenhar’s noneponymous name? (the answer will start with an ‘O’)
- Oculo-Auriculo-Vertebral (OAV) syndrome

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

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

Where specifically are dermoids commonly located?
- At the limbus

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

Very convenient mnemonic!

Goldenhar
- O
- L
- D
- E
- N
- H
- A
- R
Craniofacial Malformations

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

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What is Alagille’s noneponymous name?
Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
An infant with jaundice will present to the eye service as a ‘rule out Alagille syndrome’ consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
The presence of a posterior embryotoxon on the infant’s corneas

What is a posterior embryotoxon?
An anteriorly displaced and thickened Schwalbe’s line
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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 is Goldenhar's noneponymous name?
Oculo-Auriculo-Vertebral (OAV) syndrome

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

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

Where specifically are dermoids commonly located?
At the limbus

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

Goldenhar OAV syndrome

Very convenient mnemonic!

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

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

What is the noneponymous name for Alagille syndrome?
Arterohepatic dysplasia

Under what circumstance is an ophthalmologist likely to encounter an Alagille pt?
An infant with jaundice will present to the eye service as a 'rule out Alagille syndrome' consult

What eye finding is the ophthalmologist looking for in order to rule in/out Alagille syndrome?
The presence of a posterior embryotoxon on the infant's corneas

What is a posterior embryotoxon?
An anteriorly displaced and thickened Schwalbe's line
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

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Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

What ocular/periocular abnormalities are common? -- (answer will start with an ‘L’)

Goldenhar syndrome
OAV syndrome

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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- At the limbus

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Goldenhar
OAV syndrome
Lid colobomas
DENHAR

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Treacher Collins syndrome
- Pierre Robin sequence

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- Goldenhar and Treacher Collins

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

What ocular/periocular abnormalities are common?
- Lid coloboma

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

Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
D
E
N
H
A
R

Very convenient mnemonic!

Goldenhar syndrome

Ear abnormalities (pre-auricular appendages; aural fistulae)

Hemifacial microsomia (maxillary/mandibular hypoplasia)

Where specifically are dermoids commonly located?
At the limbus

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

Depends on who you ask. The BCSC Cornea book says the upper, whereas the Plastics book indicates the lower. (The Peds book doesn’t address this issue.) Caveat emptor.
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome

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- Craniosynostoses
  - Crouzon syndrome
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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’)

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

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Very convenient mnemonic!

Goldenhar
OAV syndrome
Lid colobomas
D
d
E
H
A
R

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Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
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- Saethre-Chotzen syndrome

Goldenhar syndrome

Oculo-Auriculo-Vertebral (OAV) syndrome

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

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

Craniosynostoses
- Crouzon syndrome

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

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

- Common ocular/periocular abnormalities:
  -- Lid coloboma
  -- Dermoids of the cornea; Duane syndrome

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

- Where are dermoids commonly located?
  - At the limbus

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- What is the non-eponymous name of Goldenhar? 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

- 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!
  - OAV syndrome
  - Lid colobomas
  - Dermoid; Duane syndrome
  - E
  - N
  - H
  - A
  - R
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Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

Goldenhar syndrome
- Ocular abnormalities
  - Lid coloboma
  - Dermoids of the cornea
  - Duane syndrome

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)
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
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Two of the non-systotic conditions are examples of what are called branchial arch syndromes. Which two?
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Craniofacial Malformations

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Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

Very convenient mnemonic!

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

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At the limbus

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They can be, and usually are

In what noncosmetic way are they significant? 
Enhar
```
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
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Not Craniosynostoses
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Lid colobomas
Dermoid; Duane syndrome

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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

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

Craniosynostoses
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Not Craniosynostoses

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Where specifically are dermoids commonly located?
At the limbus

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

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

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

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
  - Treacher Collins syndrome
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Goldenhar syndrome
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Lid colobomas
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Craniofacial Malformations

Craniosynostoses
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Craniofacial Malformations

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Very convenient mnemonic!

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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)?

- Lid coloboma
- Dermoids of the cornea; Duane syndrome

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

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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.

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

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

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

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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.

Where are dermolipomas typically located?

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

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

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

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

Very convenient mnemonic:

Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
E
N
H
A
R
Is there a relationship between epibulbar dermoids and lipoderminoids (aka dermolipomas)? Yes. The relationship is that, like dermoids, lipoderminoids are associated with Goldenhar syndrome.

Where are dermolipomas typically located? The temporal fornix.

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.

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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.

Very convenient mnemonic!

Goldenhar
- OAV syndrome
- Lid colobomas
- Dermoid; Duane syndrome

ENHAR
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 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 (prolapsed fat is squishy, whereas a dermolipoma is firm)

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

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

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

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

Goldenhar
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) and 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.

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Craniofacial Malformations

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

Where are dermolipomas typically located? The text is unclear.

What about on MRI/CT--how can they be differentiated? With what? Prolapsed orbital fat

What ocular/periocular abnormalities are common? Lid coloboma

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

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

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Where are dermolipomas typically located?
The temporal fornix.

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

With what similar-appearing but etiologically-distinct condition are dermolipomas confused?
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OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
E
N
H
A
R
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.

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

How can the two be differentiated on MRI/CT? On imaging, prolapsed orbital fat will be seen to be continuous with intraconal fat, whereas a dermolipoma will not.

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

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

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Where specifically are dermoids commonly located? At the limbus.

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

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.

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Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
- Goldenhar syndrome
  - Oculo-Auriculo-Vertebral (OAV) syndrome
  - Lid coloboma
  - Dermoids of the cornea
  - Duane syndrome

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

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

Duane syndrome has a 'middle' name--what is it?
Duane's retraction syndrome

Very convenient mnemonic!
Duane's retraction syndrome

Duane's is an example of a 'congenital cranial dysinnervation syndrome.' In Duane's, which cranial nerve incorrectly innervates which muscle?
CN3 innervates the lateral rectus

What is the observed manifestation of MGJW, ie, with what issue do pts present?
They have limited horizontal eye movement, and attempted abduction causes the eye to retract
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?
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Very convenient mnemonic!

Duane’s retraction syndrome
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses
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- Pierre Robin sequence
- Goldenhar syndrome
- Apert syndrome
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- Pfeiffer syndrome
- Goldenhar syndrome
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Craniofacial Malformations

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Craniofacial Malformations

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- Pierre Robin sequence
- Fetal alcohol syndrome

Not Craniosynostoses

Goldenhar syndrome

What is Goldenhar's non-eponymous name?
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What ocular/periocular abnormalities are common?
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Goldenhar OAV syndrome

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

Marcus-Gunn jaw winkle

In MGJW, which cranial nerve incorrectly innervates which muscle?
CN7 innervates the levator

What is the observed manifestation of MGJW, ie, with what issue do pts present?
They have ptosis that resolves when they move their jaw a certain way
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
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Goldenhar OAV syndrome
Lid colobomas
Dermoid; Duane syndrome
ENHAR
Very convenient mnemonic!

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Goldenhar OAV syndrome
Lid colobomas
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ENAH

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Goldenhar
OAV syndrome
Lid colobomas
Dermoid; Duane syndrome

(EN)
Craniofacial Malformations

Craniosynostoses

- Crouzon syndrome

Not Craniosynostoses

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Goldenhar

OAV syndrome

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Dermoid; Duane syndrome

Ear abnormalities

Nothing starts w/ ‘N’

H

A

R

Very convenient mnemonic!
Craniofacial Malformations

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Craniofacial Malformations

Craniosynostoses
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Goldenhar syndrome
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- Nothing starts w/ ‘N’
- Hemifacial microsomia
- A
- R

Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

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

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

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

I have no idea

Very convenient mnemonic!

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

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Very convenient mnemonic!
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome

Not Craniosynostoses

Goldenhar syndrome
- Treacher Collins syndrome
- Pierre Robin sequence
- Facial clefts
- Encephalocele
- Fetal alcohol syndrome
- Aneuploidies

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Why the right side?

Very convenient mnemonic!

Goldenhar
OAV syndrome
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Craniofacial Malformations

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Craniofacial Malformations

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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
Craniofacial Malformations

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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

Very convenient mnemonic!
Craniofacial Malformations

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Craniofacial Malformations

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

Very convenient mnemonic!
Goldenhar
OAV syndrome
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Nothing starts w/ ‘N’
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At the limbus
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Goldenhar syndrome

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Goldenhar and Treacher Collins
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Where specifically are epibulbar dermoids commonly located?
At the limbus

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

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

Very convenient mnemonic!
Craniofacial Malformations

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

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

What are the notable features of \textit{Treacher Collins syndrome (TCS)}?

No, it is \textit{AD}

No, intelligence is normal in TCS
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

What are the notable features of Treacher Collins syndrome (TCS)? In addition to severe mandibular hypoplasia a la Goldenhar, TCS is marked by severe hypoplasia of the bony structures of the lateral orbital rims, which in turn leads to downsloping of the palpebral fissures.
Craniofacial Malformations

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- Pfeiffer syndrome
- Apert syndrome

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- Pierre Robin sequence
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Goldenhar and Treacher Collins

What are the notable features of Treacher Collins syndrome (TCS)?
In addition to severe mandibular hypoplasia a la Goldenhar, TCS is marked by severe hypoplasia of the malar eminence/zygoma regions. Hypoplasia in these areas produces downsloping of the lateral orbital rims, which in turn leads to downsloping of the palpebral fissures.
Craniofacial Malformations

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Is TCS inherited in sporadic fashion, like Goldenhar?
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Goldenhar syndrome
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Is cognitive impairment associated with TCS, like Goldenhar?
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How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?
**Craniofacial Malformations**

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**Pierre Robin sequence**

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

With regard to congenital anomalies, what is meant by the term sequence?
**Craniofacial Malformations**

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

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

**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.
Craniofacial Malformations

Craniosynostostoses

- Crouzon syndrome
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Not Craniosynostostoses

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With regard to congenital anomalies, what is meant by the term sequence?
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In PRS, what is the ‘single developmental malformation’ that triggers the sequence?
Craniofacial Malformations

Craniosynostostoses
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Not Craniosynostostoses
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P.E.A-err roe-BAHN

With regard to congenital anomalies, what is meant by the term sequence?
It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?
Micrognathia
Craniofacial Malformations

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

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

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Micrognathia

What does micrognathia mean?
Craniofacial Malformations

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It means that a single developmental malformation initiates a ‘domino effect’ which leads to other malformations, which in turn lead to significant functional issues.

In PRS, what is the ‘single developmental malformation’ that triggers the sequence?
Micrognathia

What does micrognathia mean?
It means ‘severe hypoplasia of the mandible’
Craniofacial Malformations

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
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And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
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And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia → glossoptosis → cleft palate → feeding difficulties
Craniofacial Malformations

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Not Craniosynostoses
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Micrognathia → glossoptosis → cleft palate → feeding difficulties

What does glossoptosis refer to?
The position of the tongue being too posterior.
Craniofacial Malformations

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Wait--there’s nothing remotely ophthalmic about anything in the sequence.
Given this, why are we eye dentists talking about PRS?
And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
Micrognathia→glossoptosis→cleft palate→feeding difficulties
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Because of its association with Stickler syndrome.

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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 ‘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?
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Micrognathia → glossoptosis → cleft palate → feeding difficulties
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Not Craniosynostoses
- Goldenhar syndrome
- Treacher Collins syndrome
- Fetal alcohol syndrome
- Pierre Robin sequence

How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?

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

OK, now in five words (not counting ‘A’ and ‘with’), what sort of condition is Stickler syndrome?
A ‘hereditary hyaloideoretinopathy with optically empty vitreous’ (Note: This is the term used in the BCSC Retina book)

Wait—there’s nothing remotely ophthalmic about anything in the sequence. Given this, why are we eye dentists talking about PRS? Because of its association with Stickler syndrome.

What is the status of the vitreous?
Optically empty vitreous

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

Craniosynostoses
- Crouzon syndrome
- Apert syndrome
- Pfeiffer syndrome
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How do you pronounce Pierre Robin in the context of the term Pierre Robin sequence (PRS)?

PEA-err roe-BAHN

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

It means that a single developmental malformation initiates a 'domino effect' which leads to other malformations, which in turn lead to significant functional issues.

In PRS, what is the 'single developmental malformation' that triggers the sequence?

Micrognathia

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

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What is the status of the vitreous?

It is liquefied

And what is the ‘sequence,’ ie, the subsequent malformations and functional issues?
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What is the status of the vitreous? It is liquefied.

Optically empty, liquefied vitreous—doesn’t sound so bad. Is Stickler syndrome associated with ocular manifestations that are clinically significant?
Indeed it is, including:
- Glaucoma
- High myopia
- Cataracts
- Lattice degeneration
- Retinal detachment

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

Hereditary vitreoretinopathy

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

A ‘hereditary hyaloideoretinopathy with optically empty vitreous’ (Not in the BCSC Peds book)

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What is the status of the vitreous? It is liquefied

optically empty vitreous
What are the notable ophthalmic features of fetal alcohol syndrome (FAS)?

- Ptosis
- Shortened fissures
- Epicanthal folds
- Telecanthus

Craniosynostoses:
- Crouzon syndrome
- Apert syndrome
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Not Craniosynostoses:
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- **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 esotropia. Is FAS? No, it is associated with esotropia.
What are the notable ophthalmic features of fetal alcohol syndrome (FAS)?
A number are lid-related:
-- Ptosis
-- Shortened fissures
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The craniosynostoses are associated with exotropia. Is FAS?
What are the notable ophthalmic features of fetal alcohol syndrome (FAS)?
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The craniosynostoses are associated with exotropia. Is FAS?
No, it is associated with esotropia

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