Congenital Neurologic Abnormalities

for USMLE Step One

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UMass Class of 2021

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Roadmap

(*Congenital Abnormalities for the Boards*)

- Neural Tube Defects
  - Anencephaly
  - Spina Bifida
    - 1) Occulta
    - 2) Meningocele
    - 3) Meningomyelocele
- Posterior Fossa
  - 1) Cerebral Aqueduct Stenosis
  - 2) Dandy Walker
  - 3) Chiari I/II
- Anterior Fossa
  - Holoprosencephaly
- Spinal Cord
  - Syringomyelia

Neural Tube Defects

Amniotic Fluid:

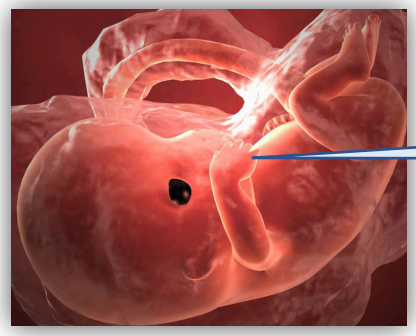
↑↑↑ AFP
↑↑↑ AChE

Neural Tube Defects

2	3	4	5	6	7	8
9	10	11	12	13	14	15

Embryo To Do List

Week 4: **Close Neural Tube**



Don’t forget your Folate!

Neural Tube Defects

Amniotic Fluid:

↑↑↑ AFP
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Neural Tube Defects

Anencephaly

Amniotic Fluid:

↑↑↑ AFP
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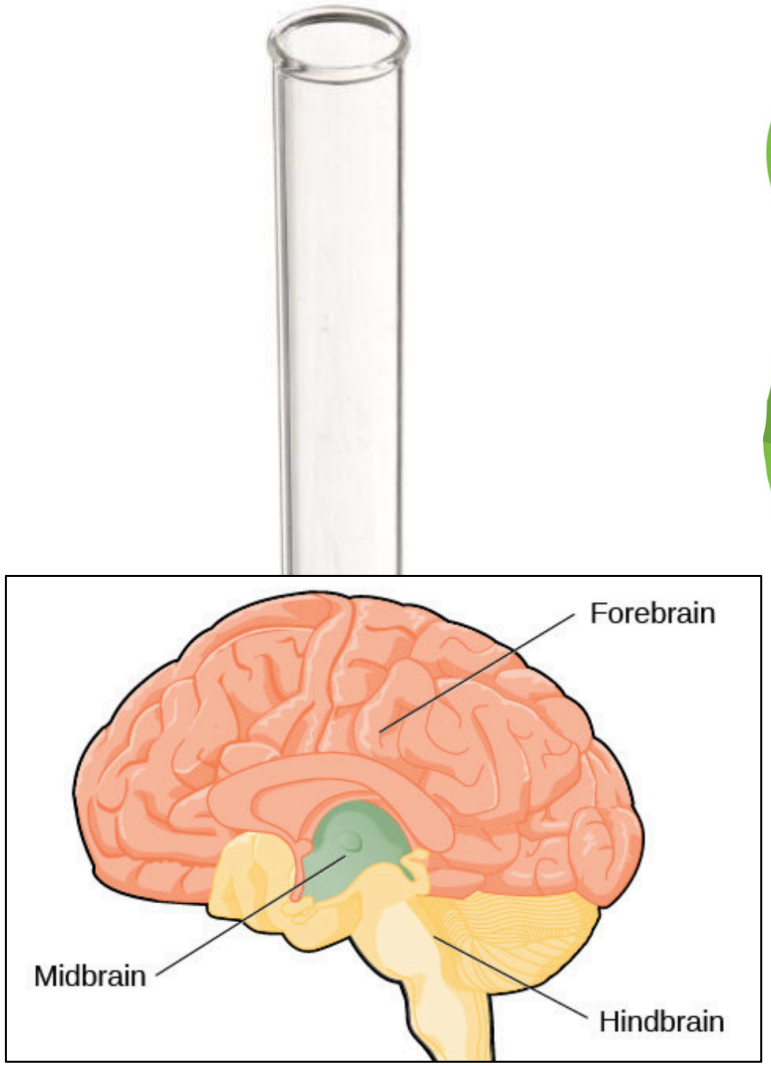
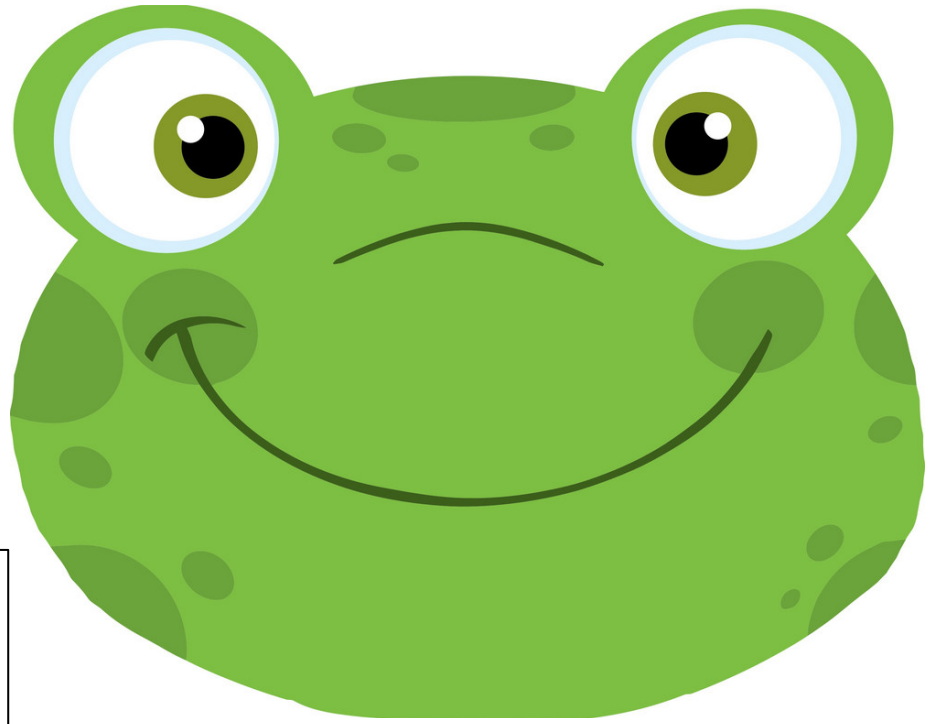
Spina Bifida

Anencephaly





Amniotic Fluid:

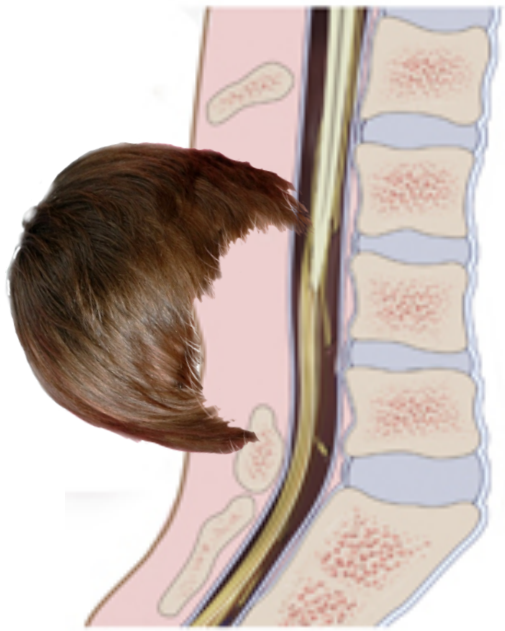
Polyhydramnios

↑↑↑ AFP

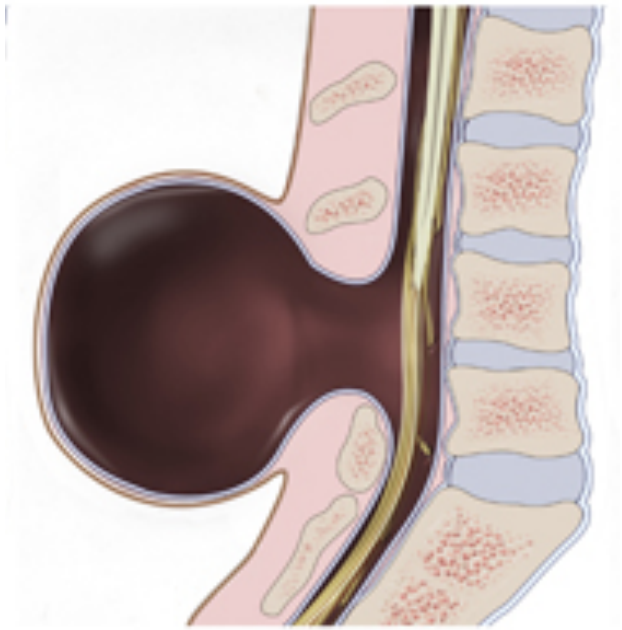
↑↑↑ AChE

Spina Bifida

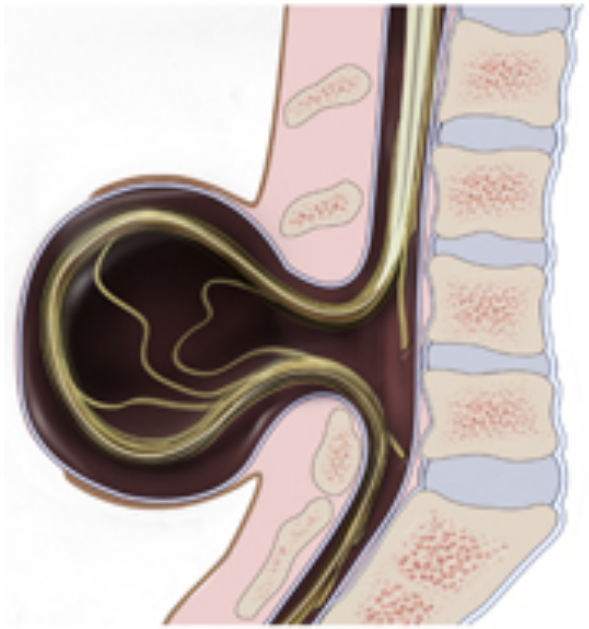
Occulta



Meningocele

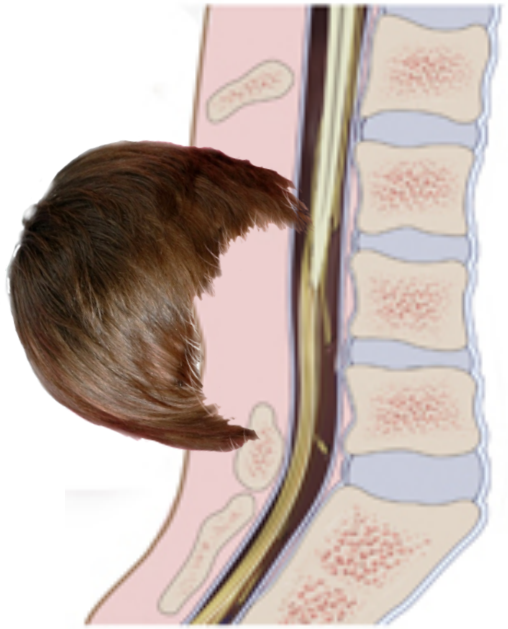


Meningomyelocele



Spina Bifida

Occulta



Amniotic Fluid:

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Spina Bifida

Amniotic Fluid:

↑↑↑ AFP
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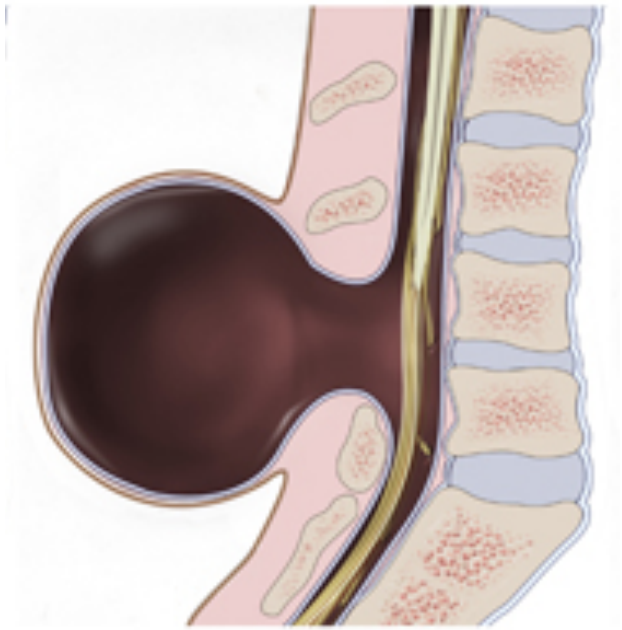
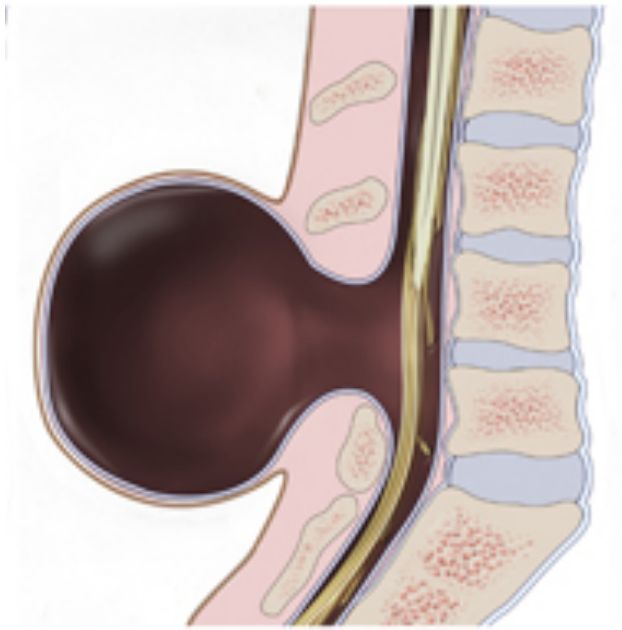
Meningocele



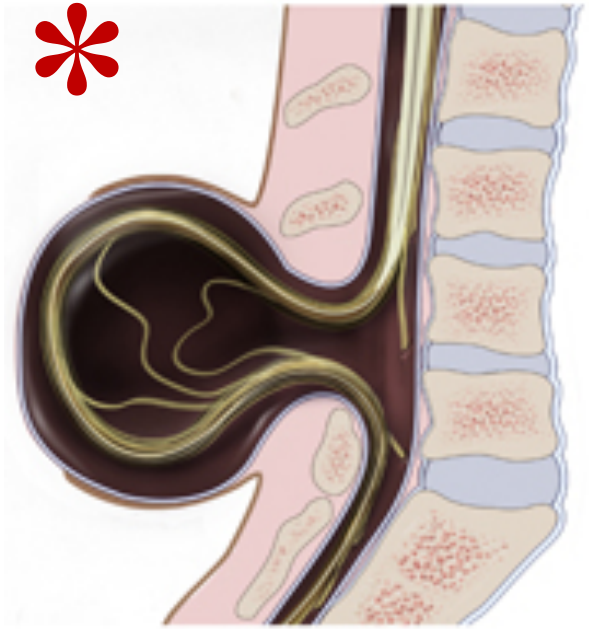
Meningomyelocele

Spina Bifida

Meningocele



Meningomyelocele



Incontinence, paralysis, areflexia

Amniotic Fluid:

↑↑↑ AFP
↑↑↑ AChE

Neural Tube Defects

Anencephaly

Spina Bifida

Valproic Acid: Decreases maternal folate absorption

Methotrexate: Inhibits Dihydrofolate Reductase

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  3) **Chiari I/II**
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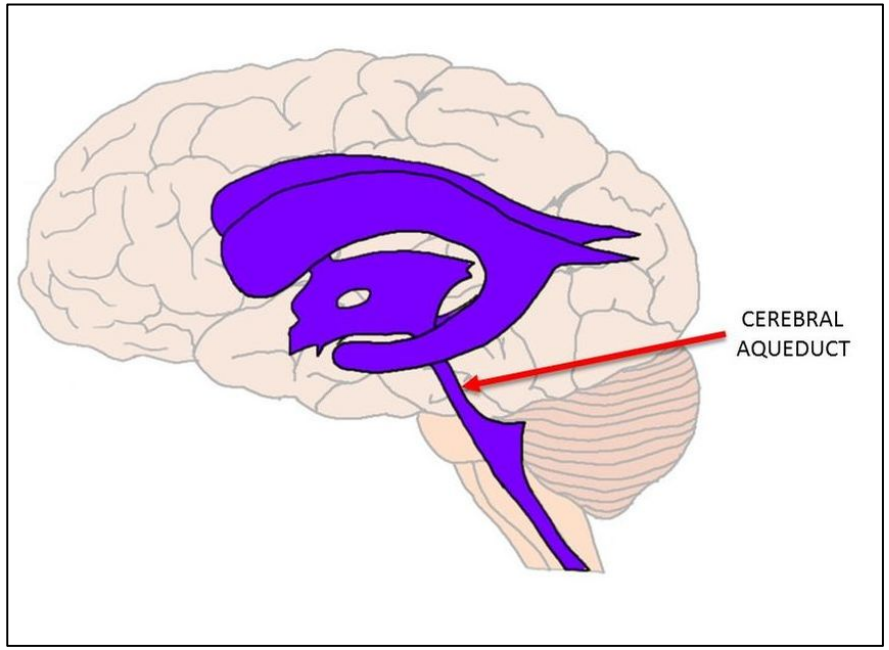
Cerebral Aqueduct Stenosis

Normal

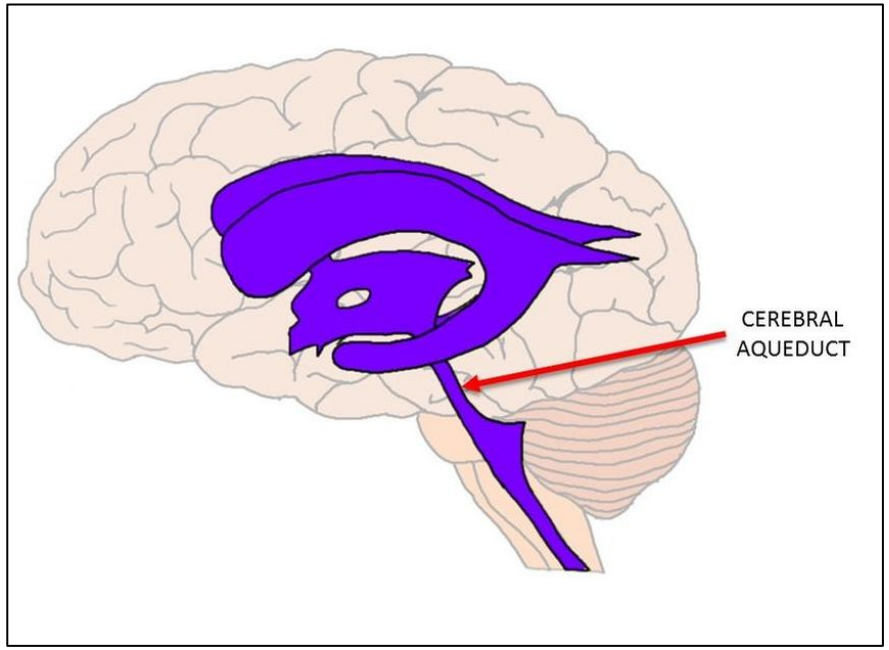


Connects ventricles 3 and 4

Stenosis

Cerebral Aqueduct Stenosis

Normal



Connects ventricles 3 and 4

Stenosis

Hydrocephalus:

- **Increased head size**
- **Bulging fontanelle**
- **Increased ICP= Papilledema**
- Seizures
- Fussy/vomiting/poor feeding

Cerebral Aqueduct Stenosis

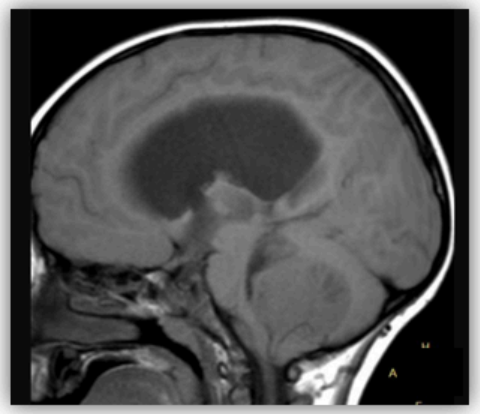
Pediatric Hydrocephalus

Congenital

1) Cerebral Aqueduct Stenosis
2) Chiari II malformation
3) Dandy Walker malformation

Acquired

1) Medulloblastoma
2) Traumatic Injury
3) Meningitis



Hydrocephalus:

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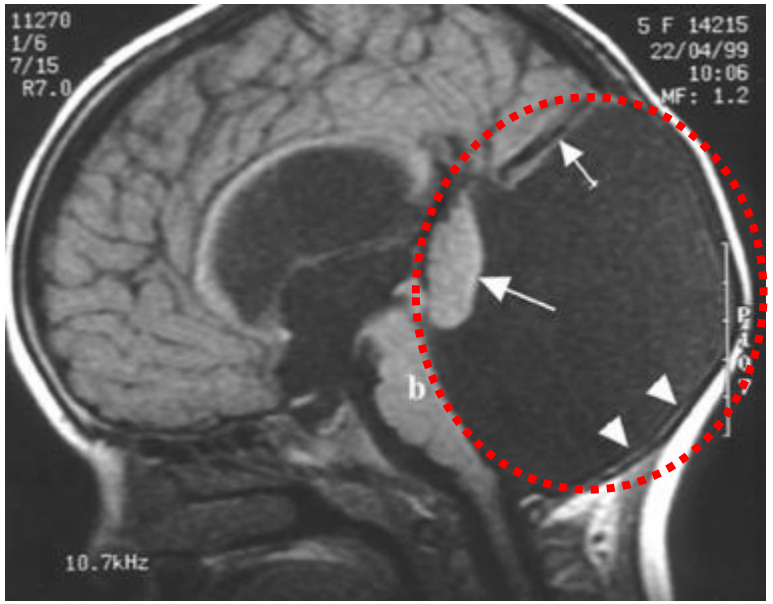
Dandy Walker Malformation



Cerebellar vermis coordinates proximal muscles

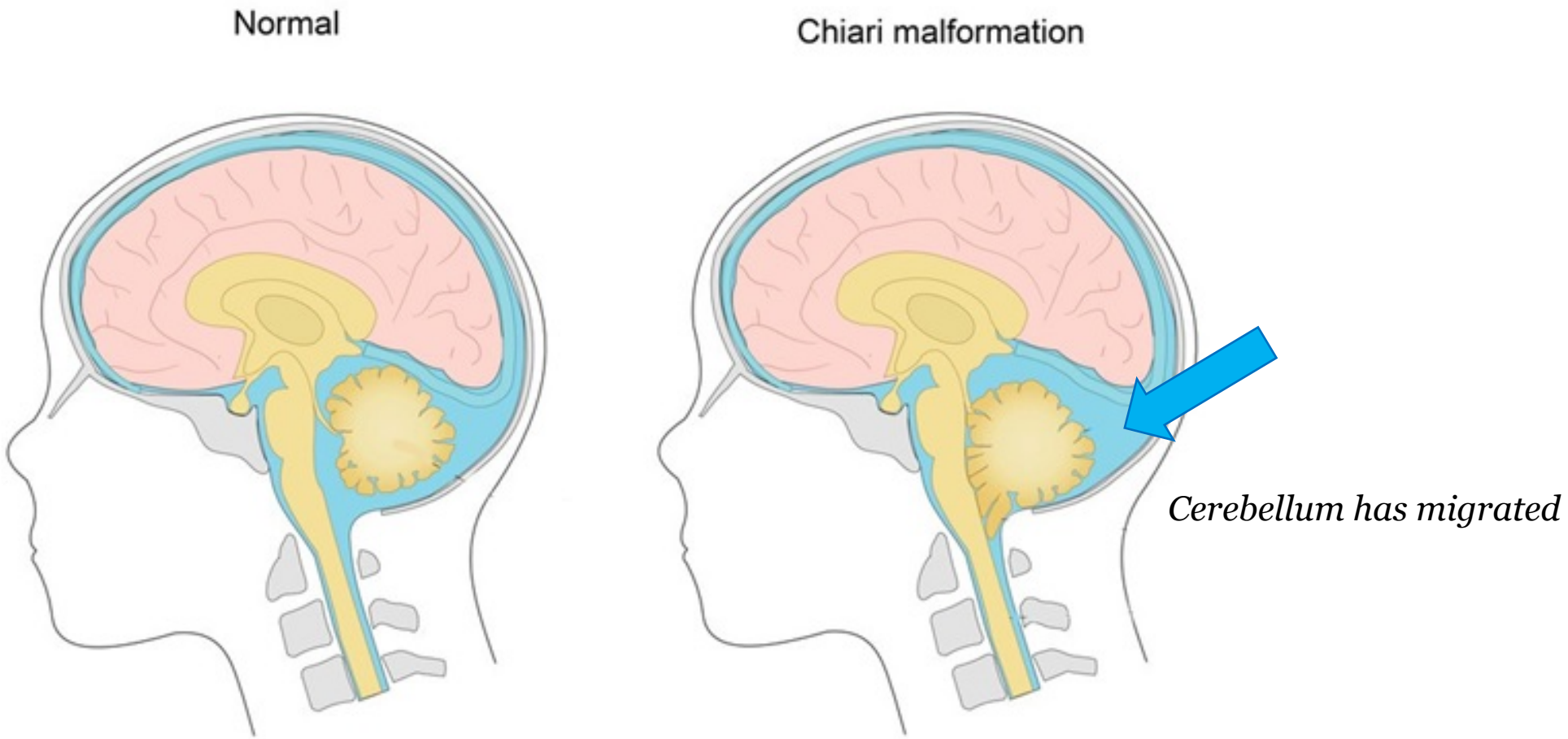
Dandy Walker Malformation

That's not a cerebellum!!

Chiari Malformations

Posterior Fossa

1) **Cerebral Aqueduct Stenosis**
2) **Dandy Walker**
3) **Chiari I/II**



Cerebellum has migrated

Chiari Malformations

Type 1

- Not as severe
- Syringomyelia association (‘*Type 1 fits inside syrinx*’)

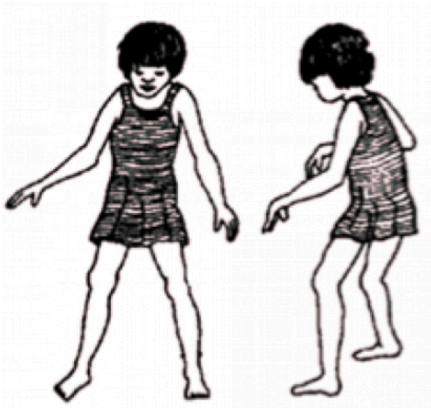
Type 2

Chiari Malformations

Type 1

- Not as severe
- **Syringomyelia** association (‘*Type 1 fits inside syrinx*’)

Headache

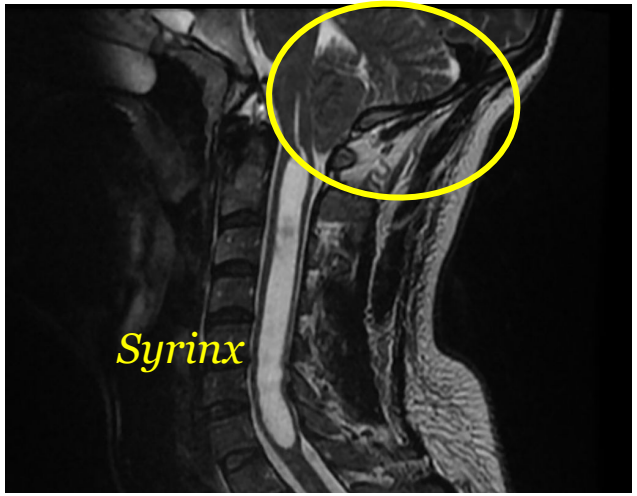


Ataxia

Chiari Malformations

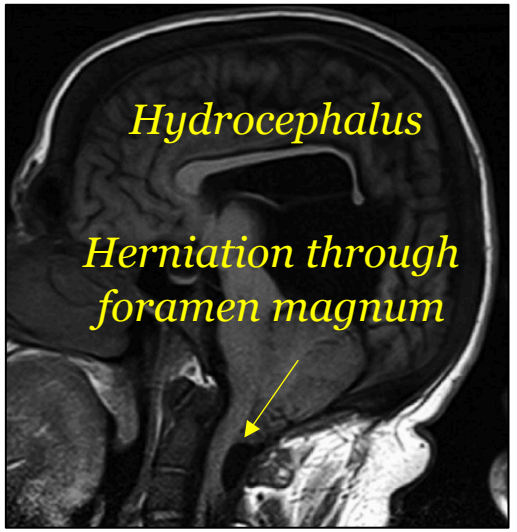
Type 1

- Not as severe
- **Syringomyelia** association (‘*Type 1 fits inside syrinx*’)



Type 2

- **BAD BAD BAD!!!**
- A/w **Meningomyelocele** (2 things herniate though spinal cord=dura and spinal cord)
- Causes hydrocephalus



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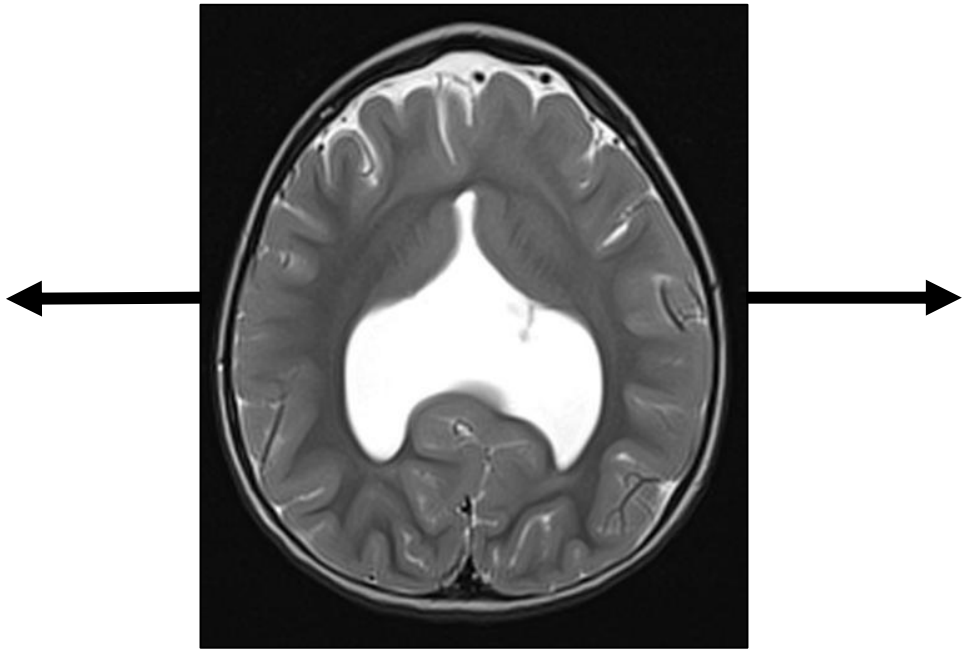
Holoprosencephaly

Left and right hemispheres
fail to separate



Holoprosencephaly

Associations:

- **Sonic Hedghehog** signaling mutations
- **Fetal Alcohol Syndrome**
- **Trisomy 13 AKA Patau Syndrome**=midline defects (Omphalocele, cleft lip, holoprosencephaly)+ rocker-bottom feet

Holoprosencephaly

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Holoprosencephaly

I'm almost 40?!?! Where did the years go!?

Holopros (pictured left), the cyclops, is 39 years old (3*13=trisomy 13) and an alcoholic. He just plays sonic all day!

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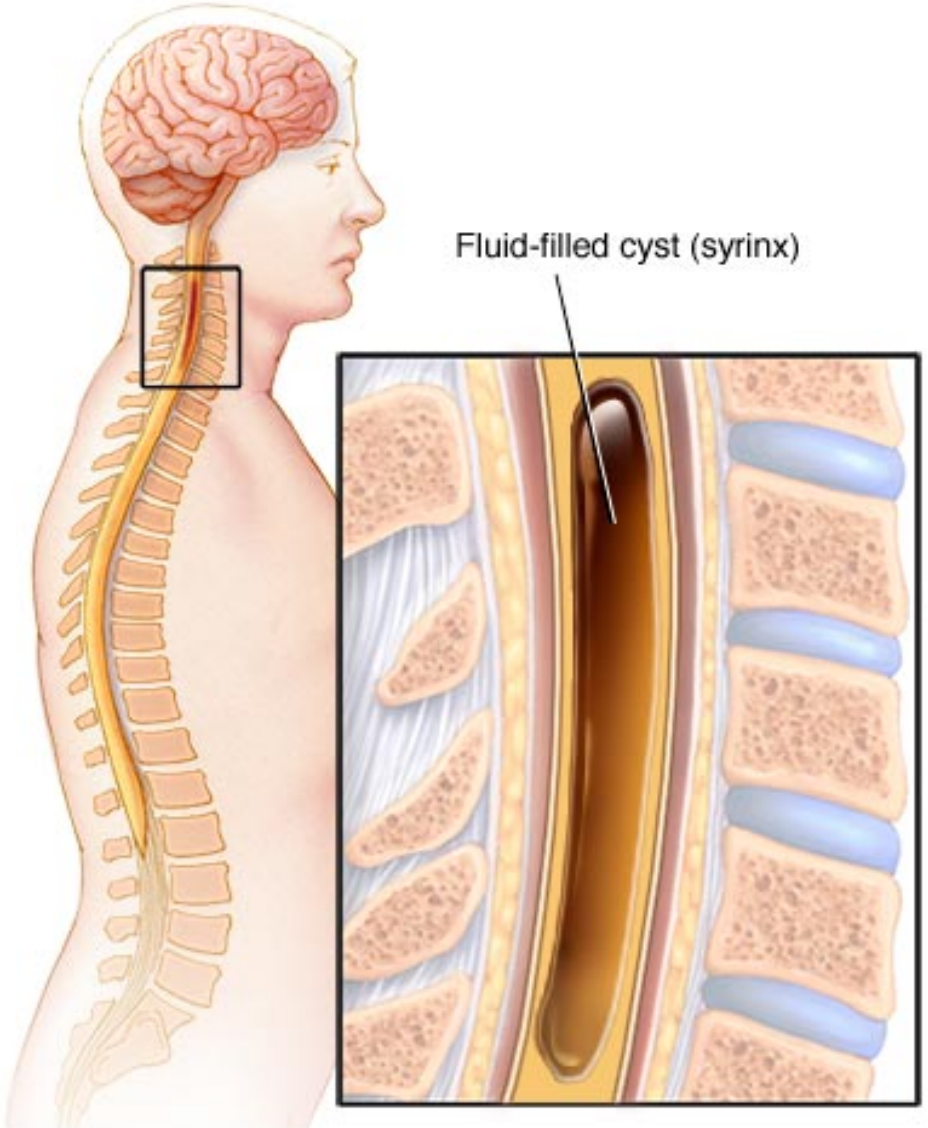
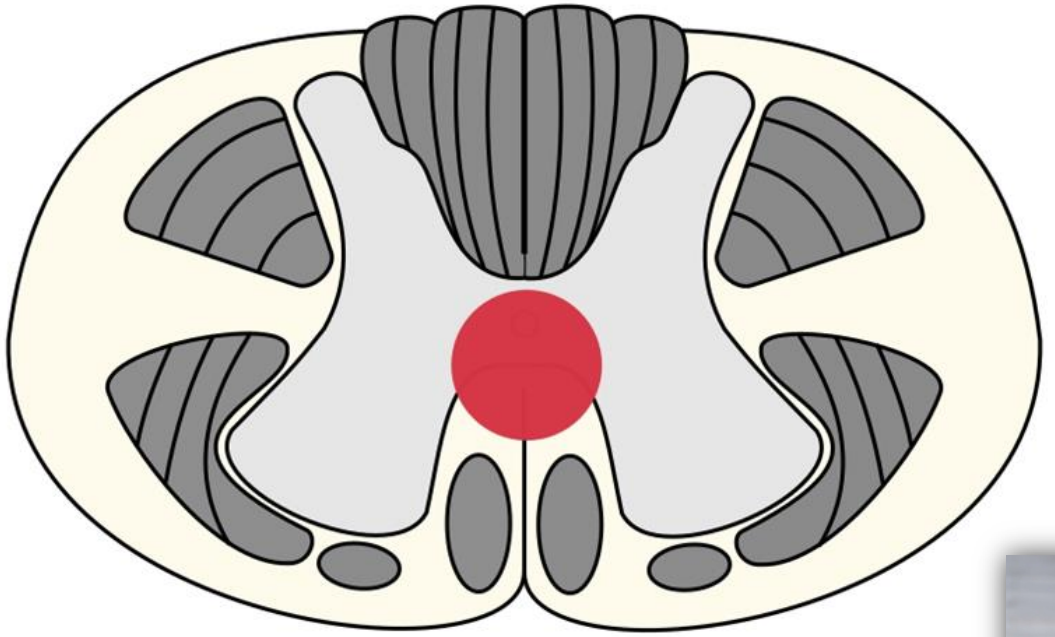
Sample questions to follow…

Syringomyelia

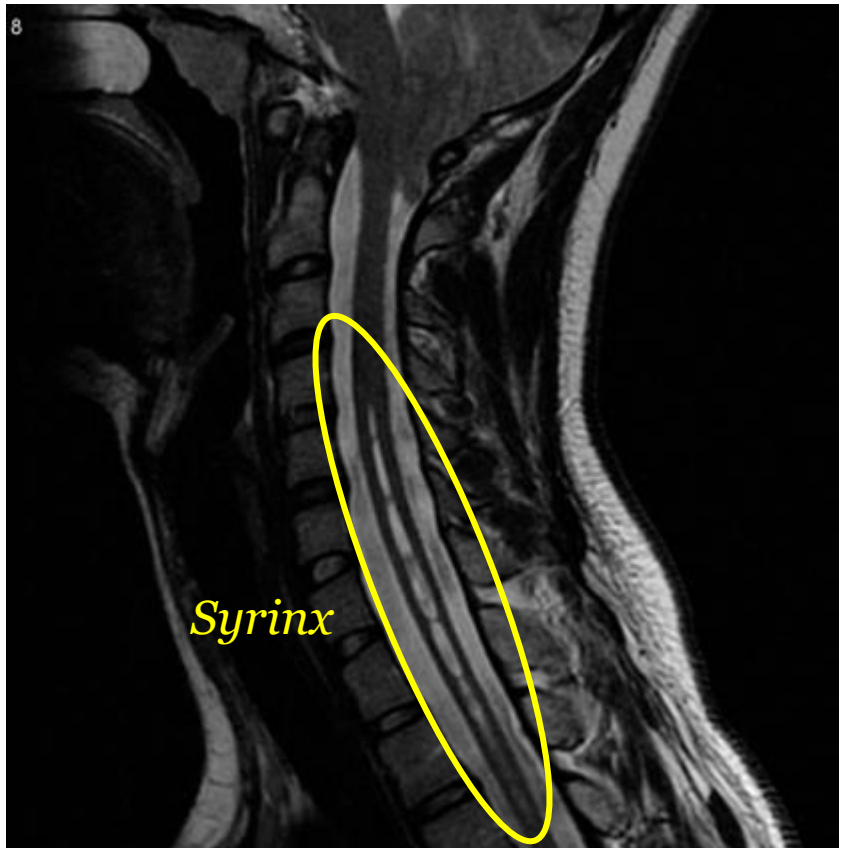


Impaired Pain and Temperature



Syringomyelia

Numbness to pain/temperature in upper back/arms



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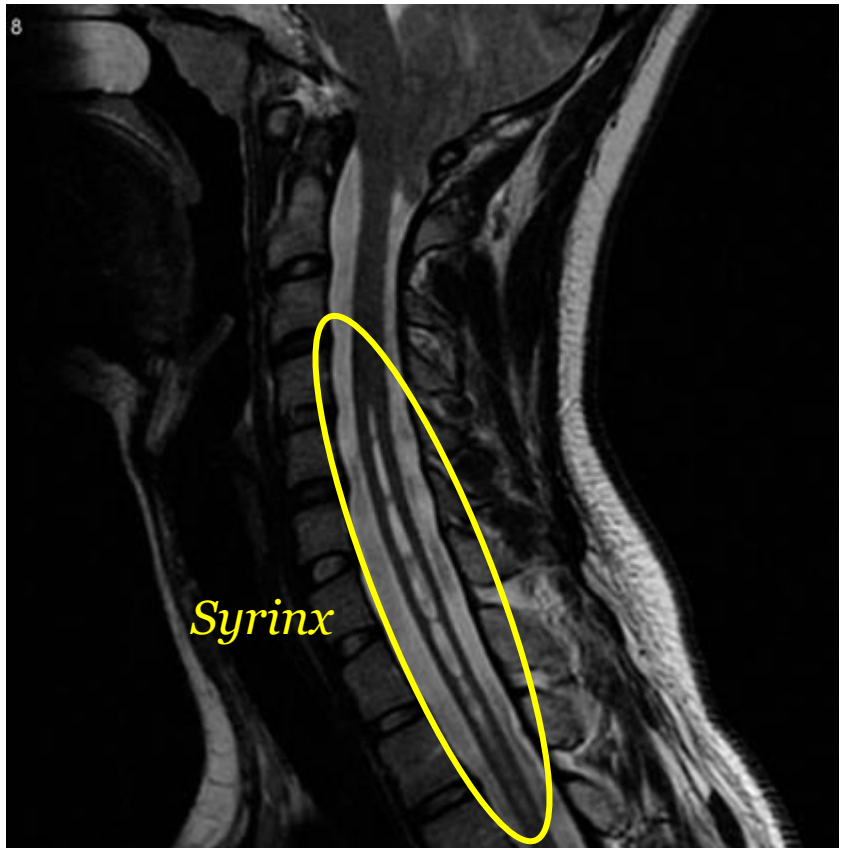

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