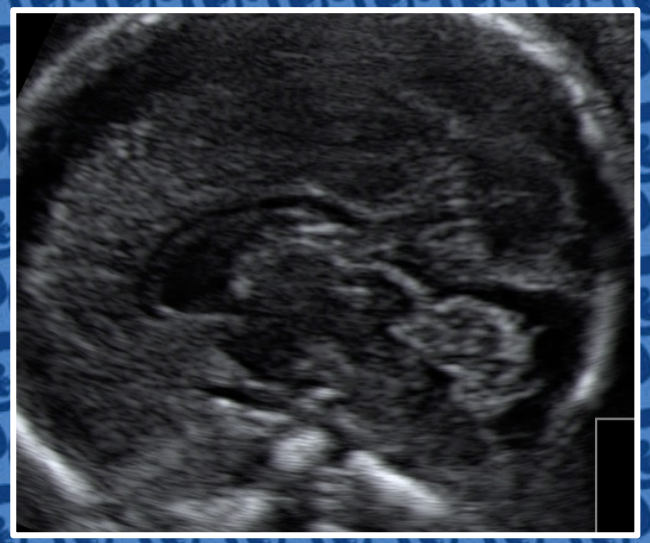
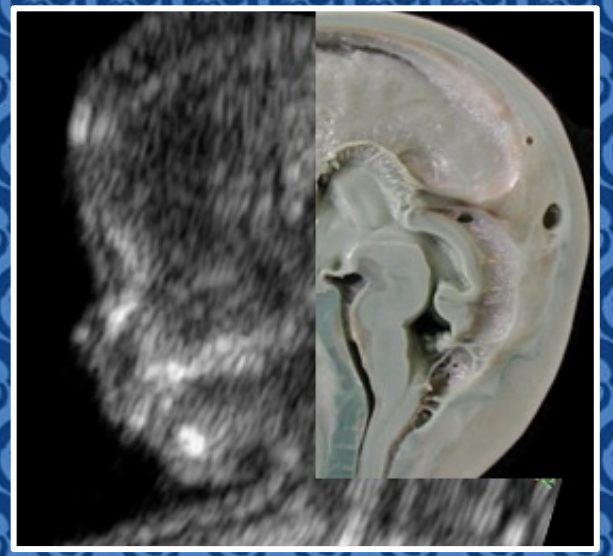
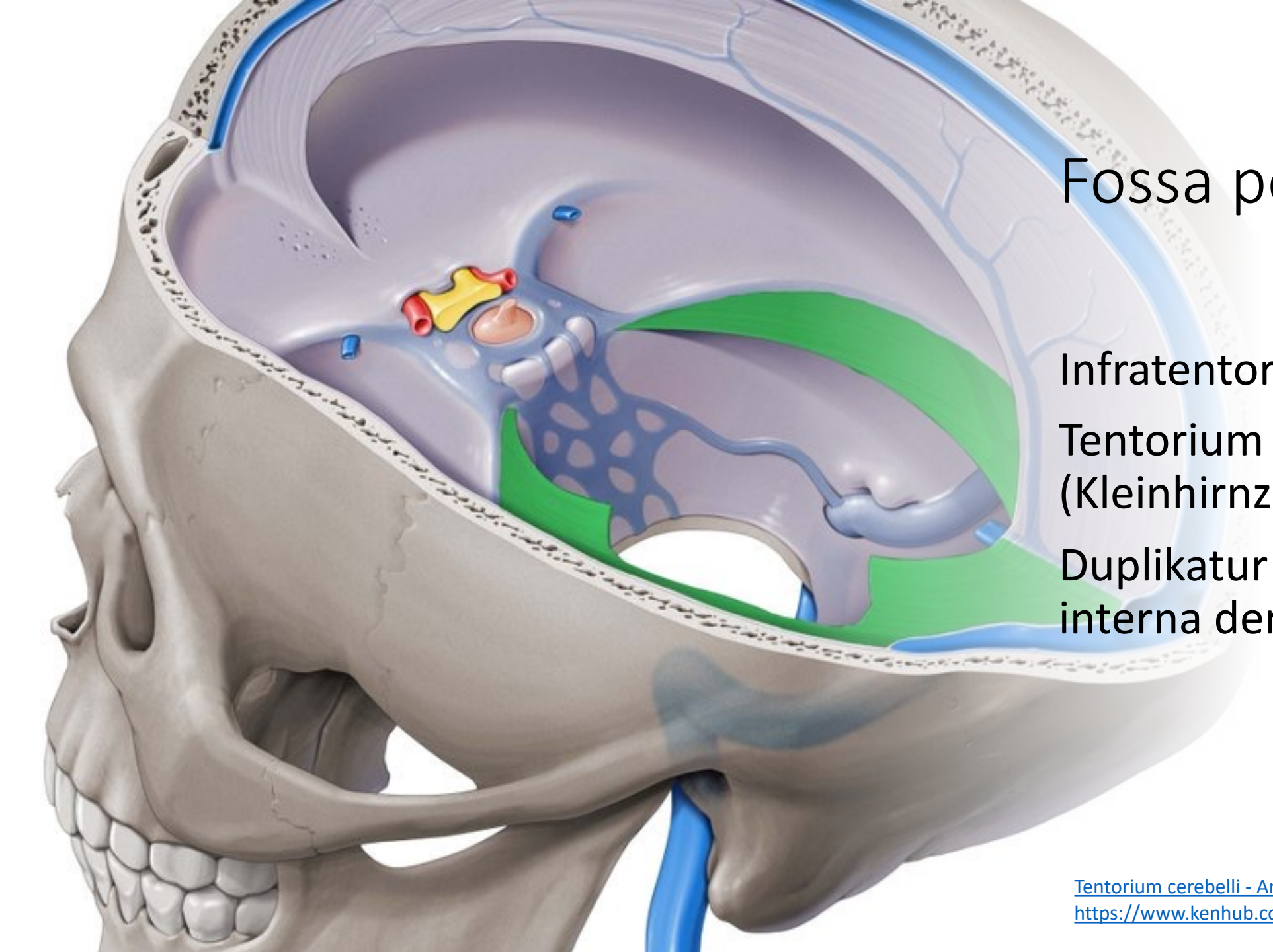
Sonographie der hinteren Schädelgrube

Univ.-Doz. Dr. Elisabeth Krampl-Bettelheim
Universitätsklinik für Frauenheilkunde
und
FetoMed
Wien







Fossa posterior

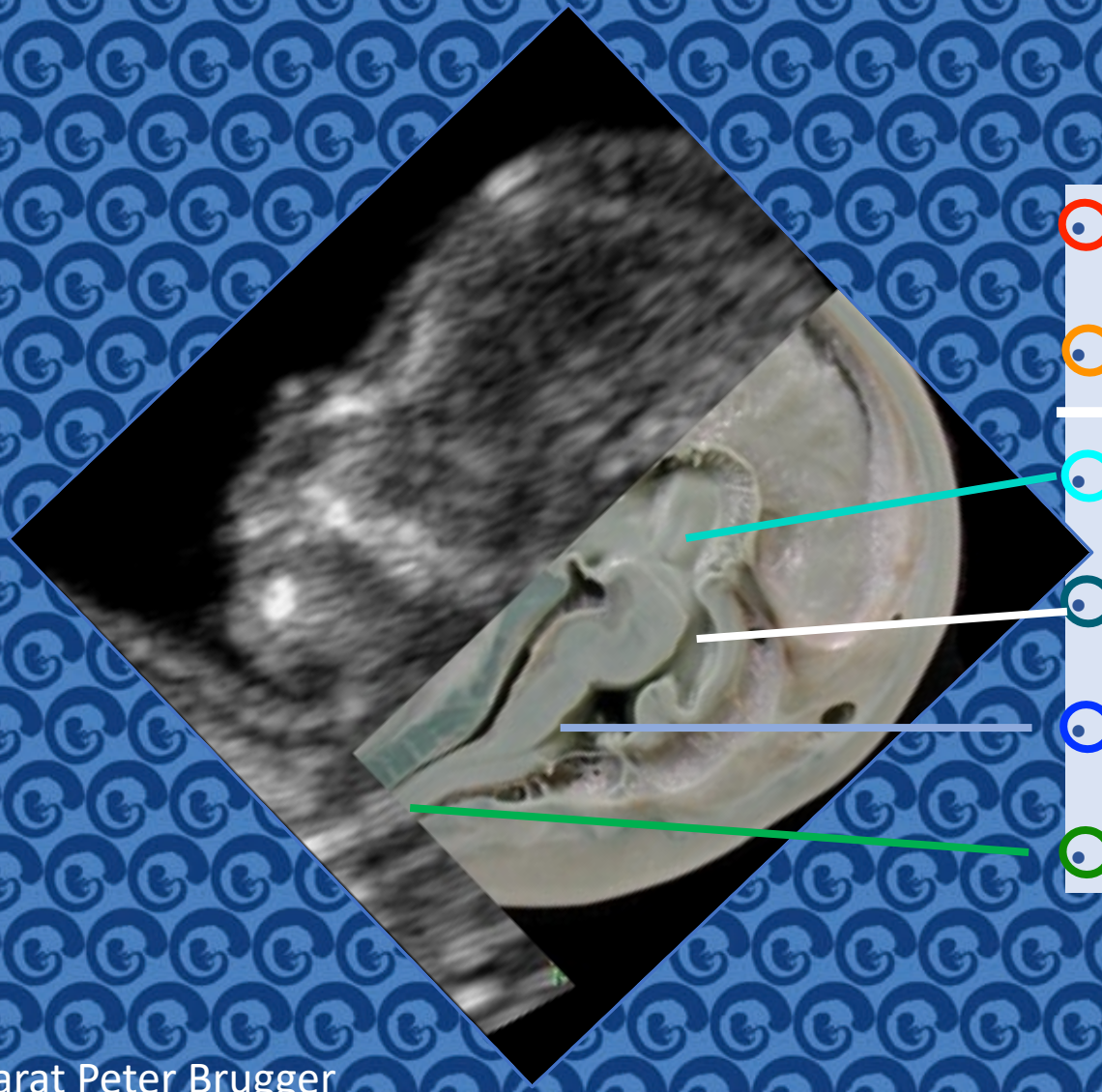
Infratentorieller Raum

Tentorium cerebelli (Kleinhirnzelt)

Duplikatur der Lamina interna der Dura mater

Tentorium cerebelli - Anatomie, Lage, Funktion | Kenhub
https://www.kenhub.com › library › tentorium-cerebelli

Normalbefund im ersten Trimenon



- Telencephalon
- Diencephalon

Prosencephalon

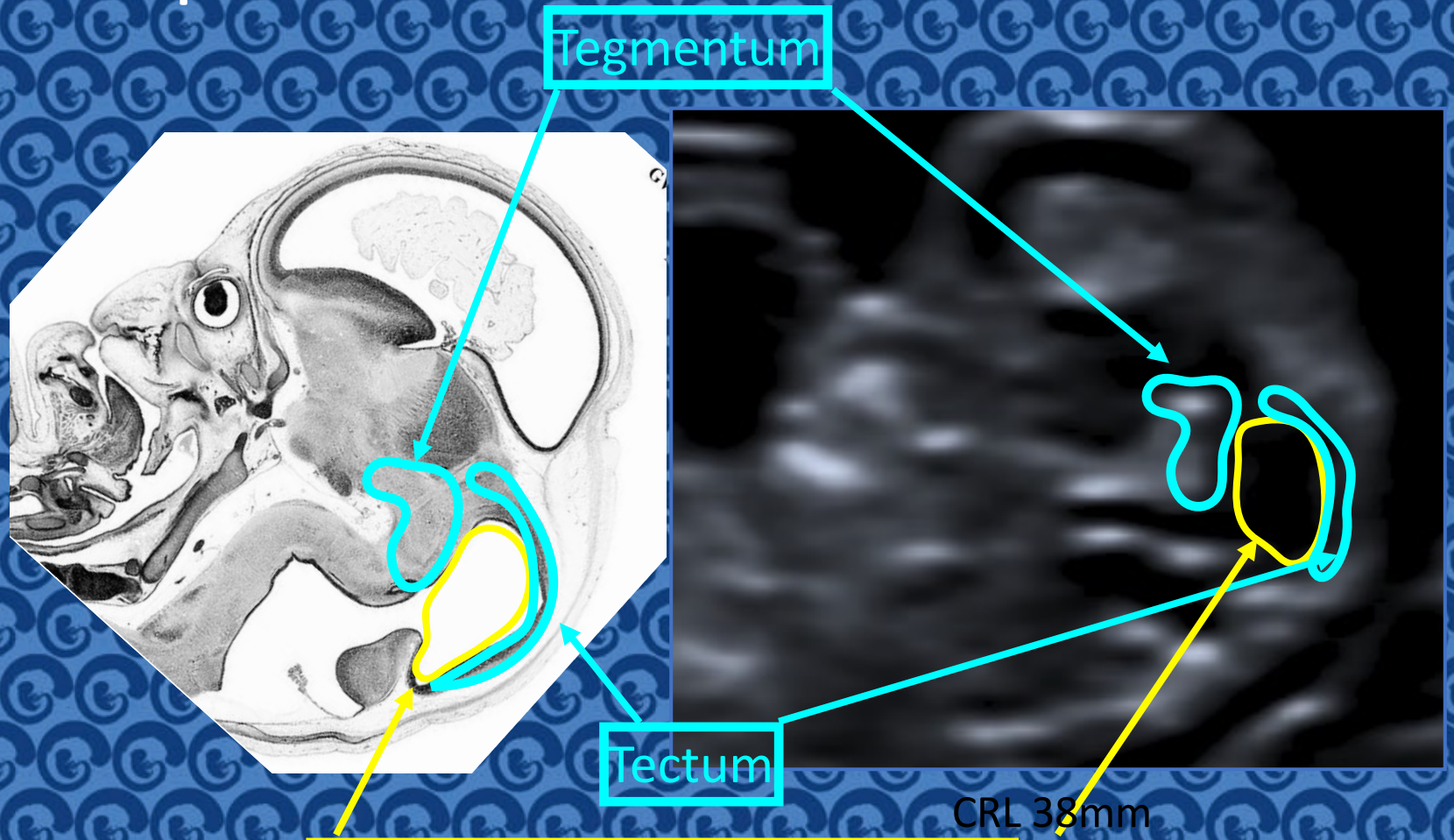
- Mesencephalon
- Metencephalon
- Myelencephalon

Rhombencephalon

- Rückenmark

Präparat Peter Brugger

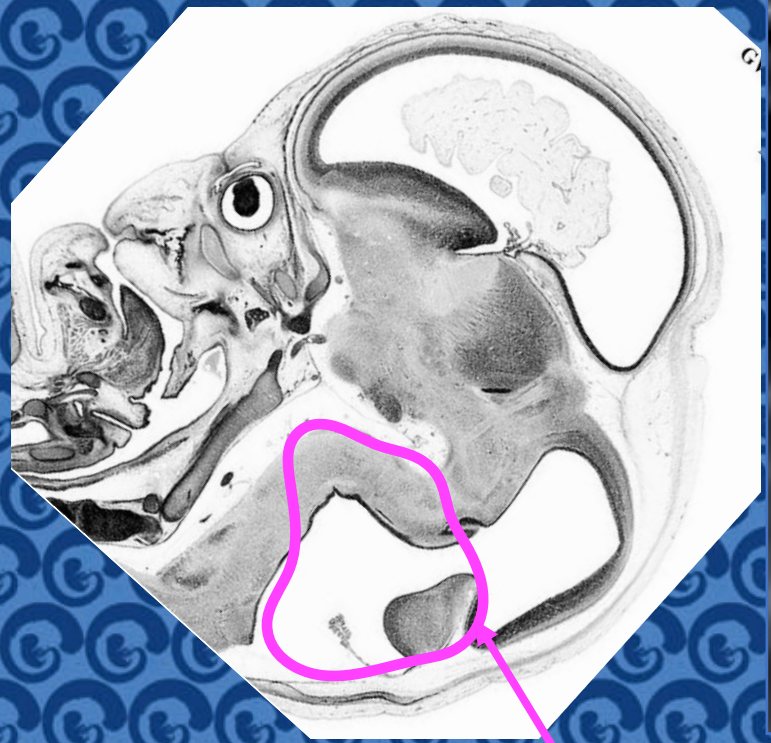
Mesencephalon

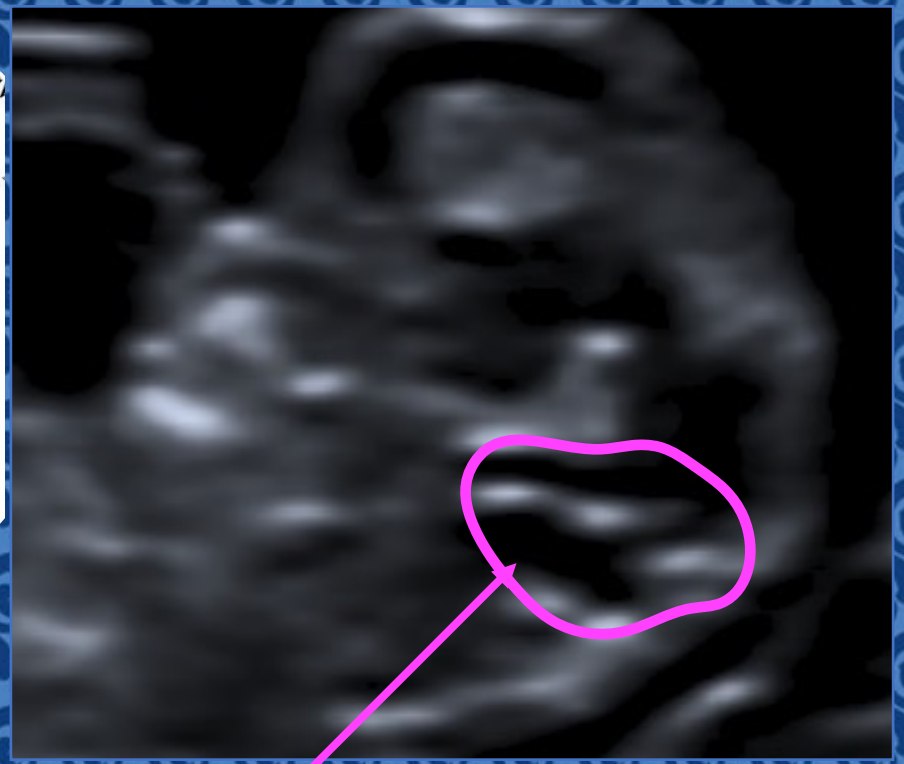


The Human Brain During the Late First Trimester, Bayer

Metencephalon

CRL 38mm

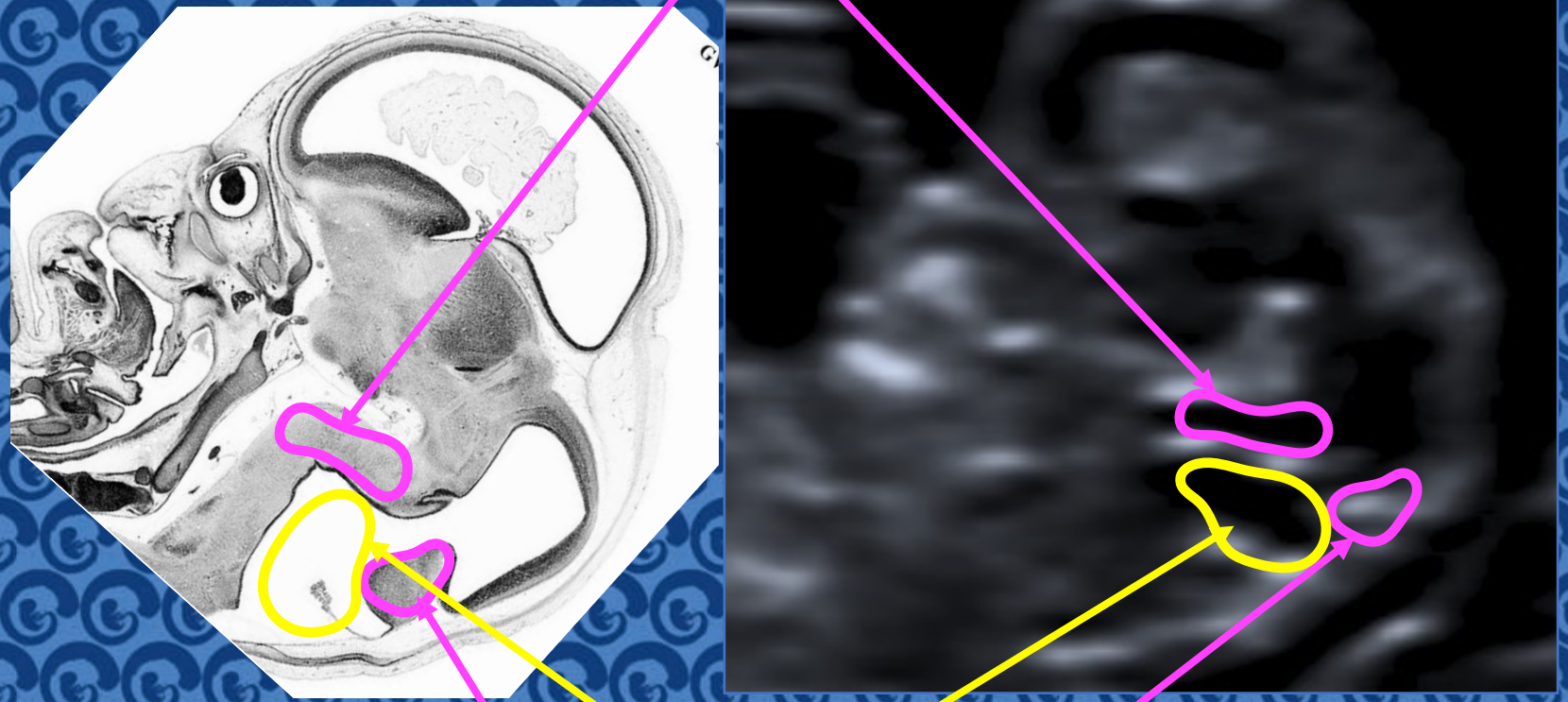




Metencephalon

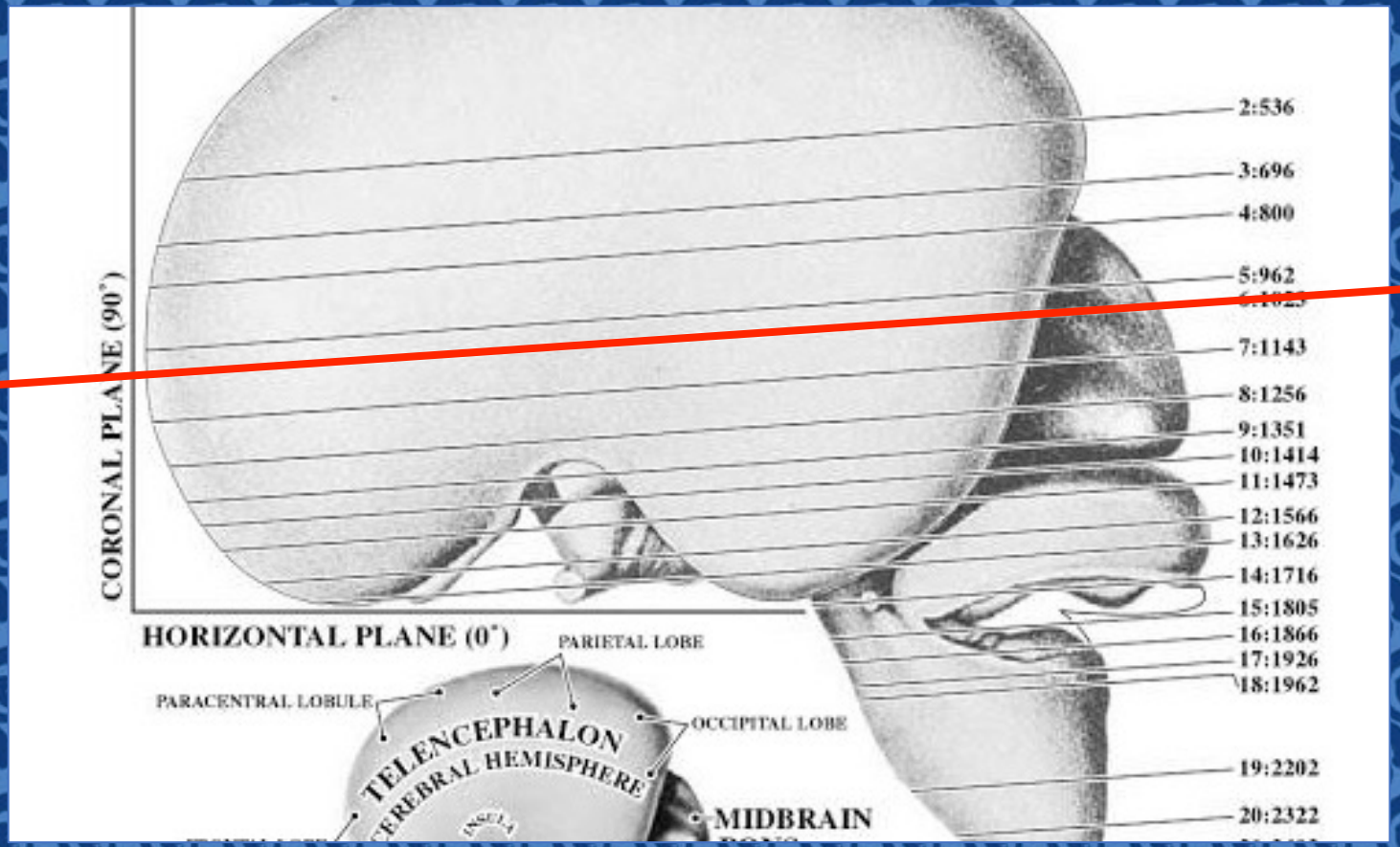
The Human Brain During the Late First Trimester, Bayer

Metencephalon



The Human Brain During the Late First Trimester, Bayer

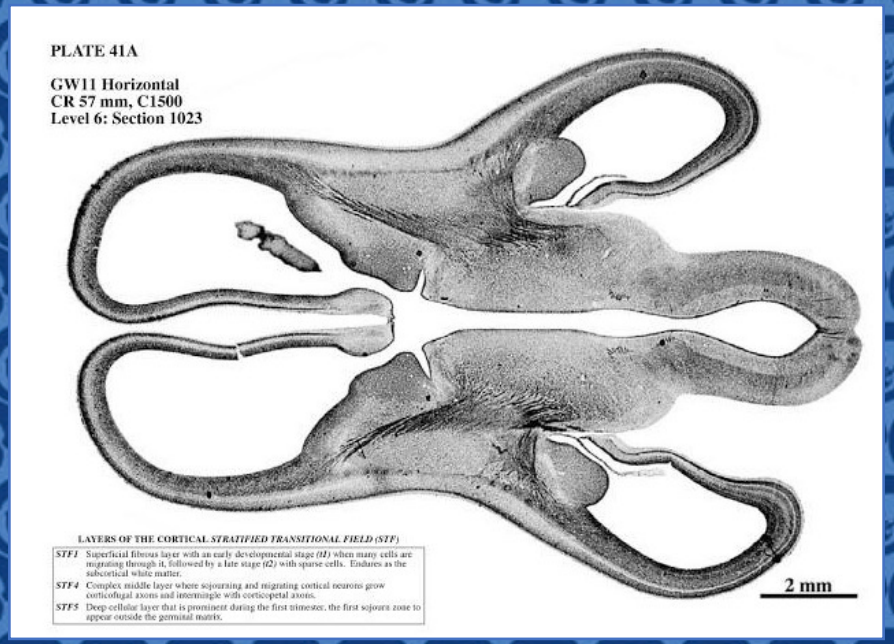
Axial - transthalamisch

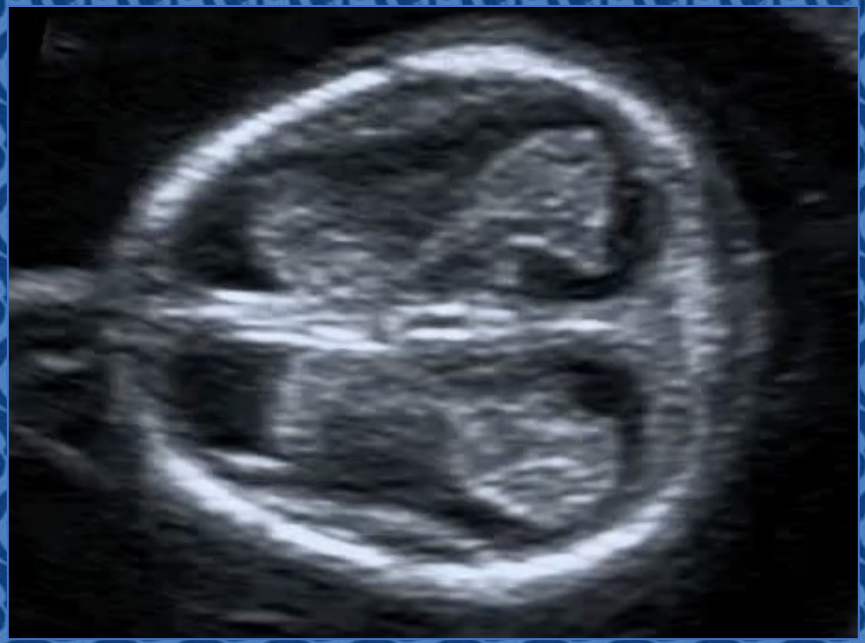


The Human Brain During the Late First Trimester, Bayer

Axial - transthalamisch

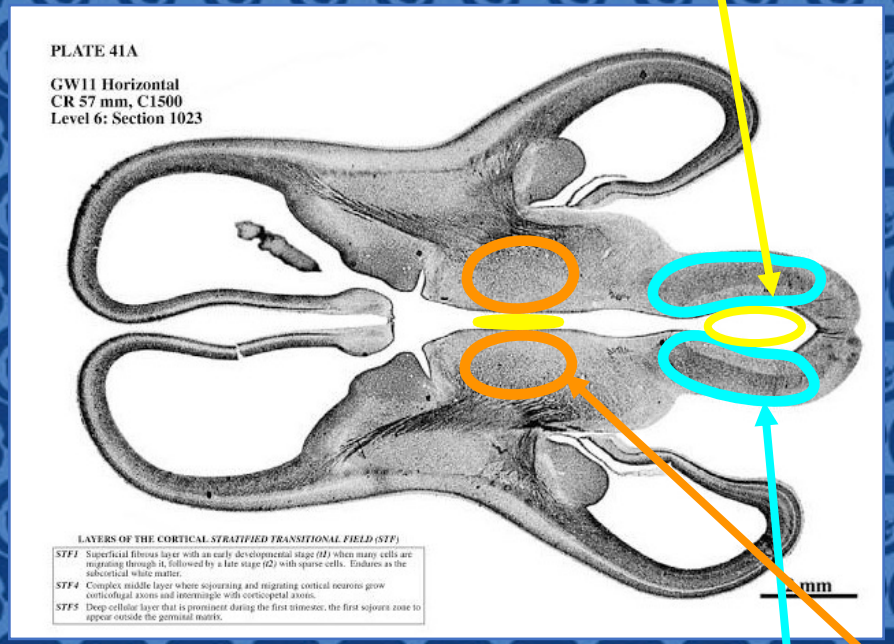
CRL 57mm

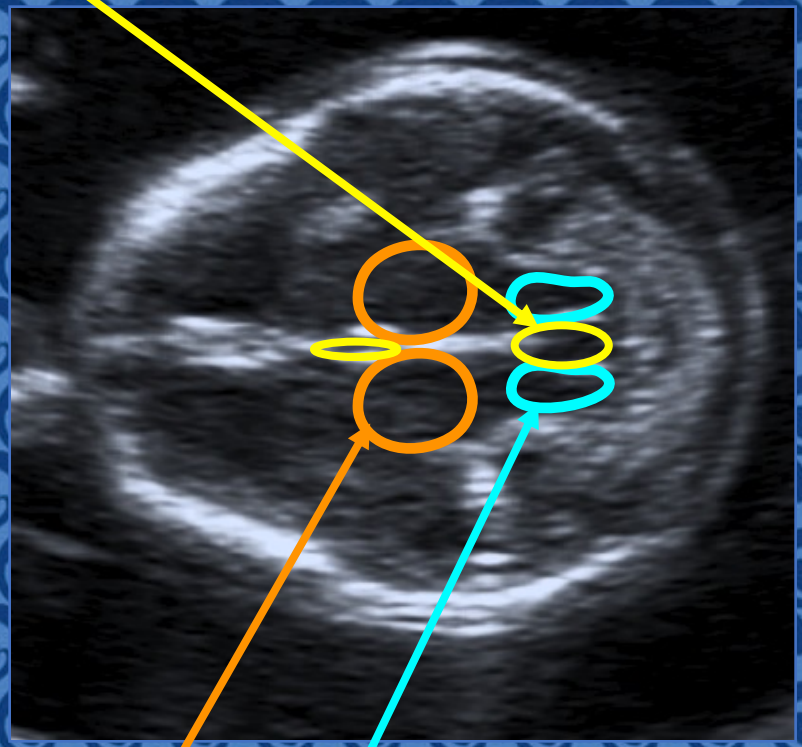




The Human Brain During the Late First Trimester, Bayer

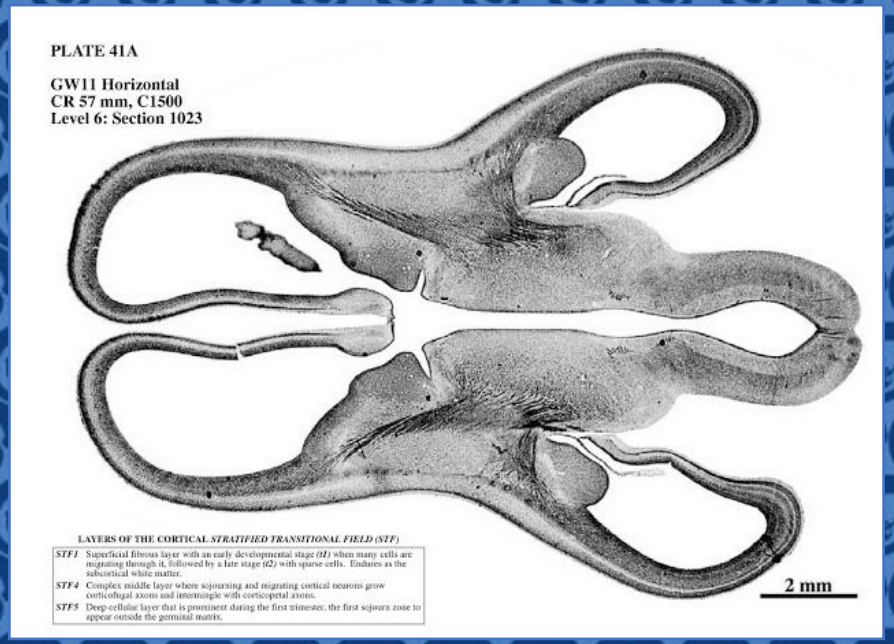
Axial - transthalamisch

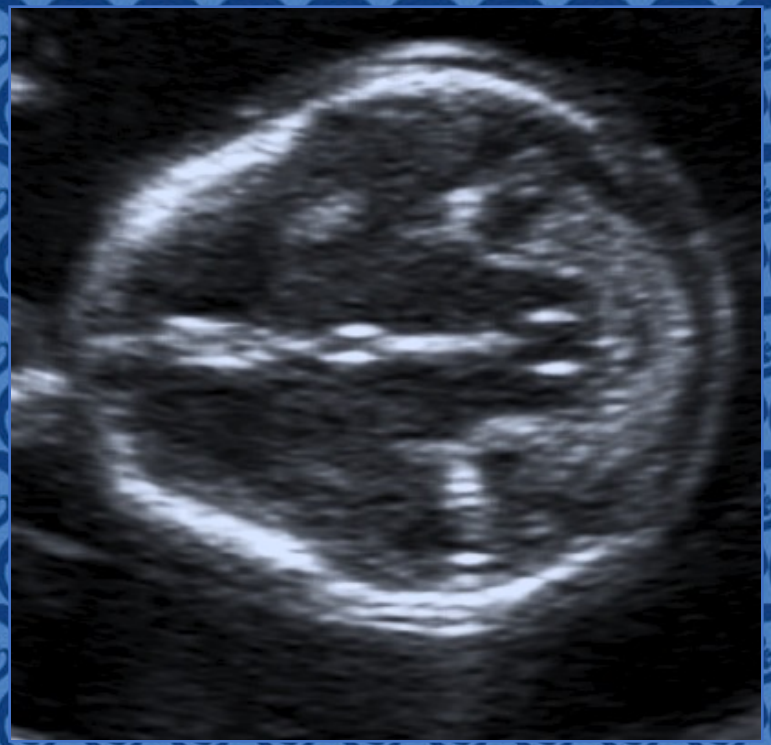




The Human Brain During the Late First Trimester, Bayer

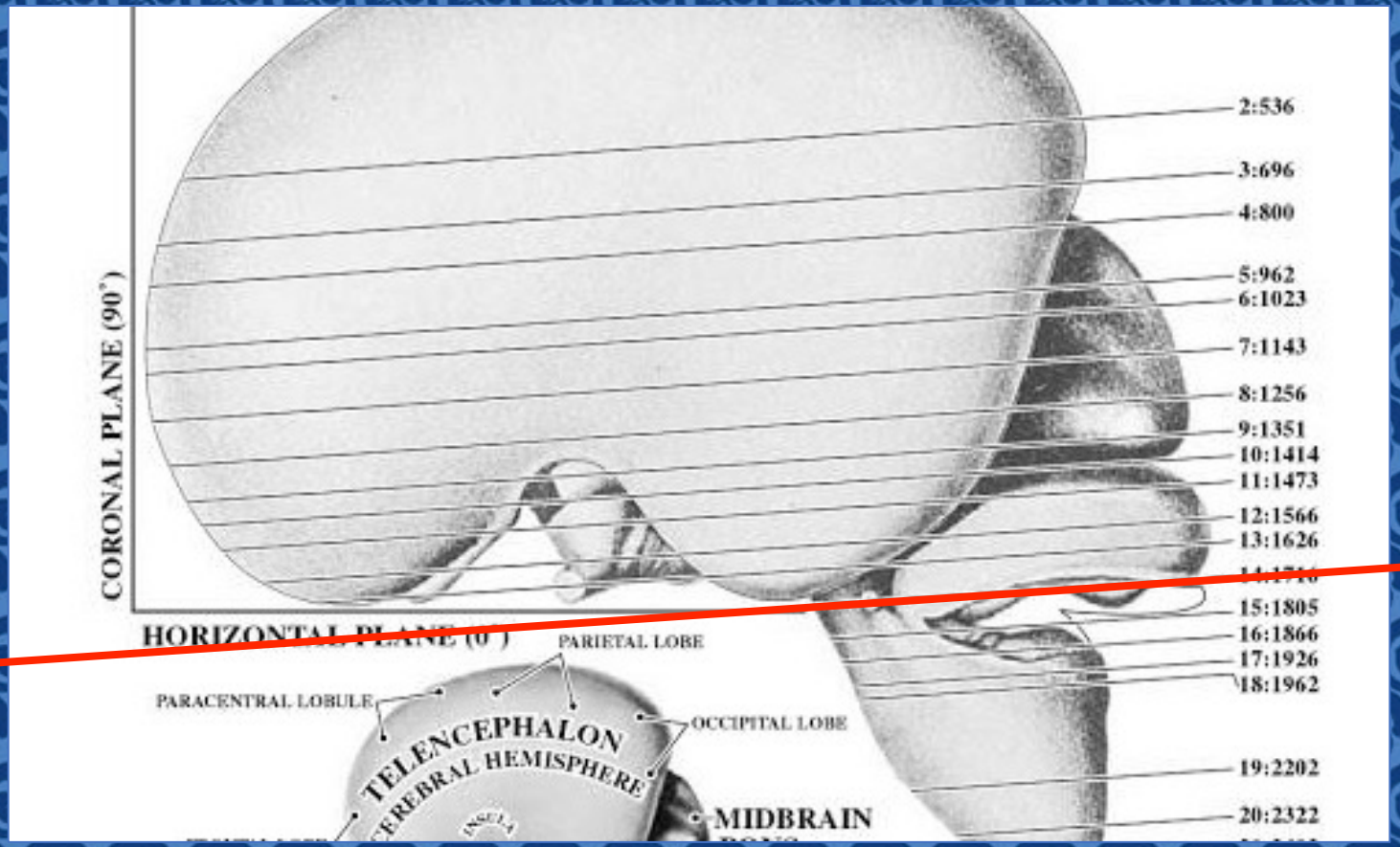
Axial - transthalamisch





The Human Brain During the Late First Trimester, Bayer

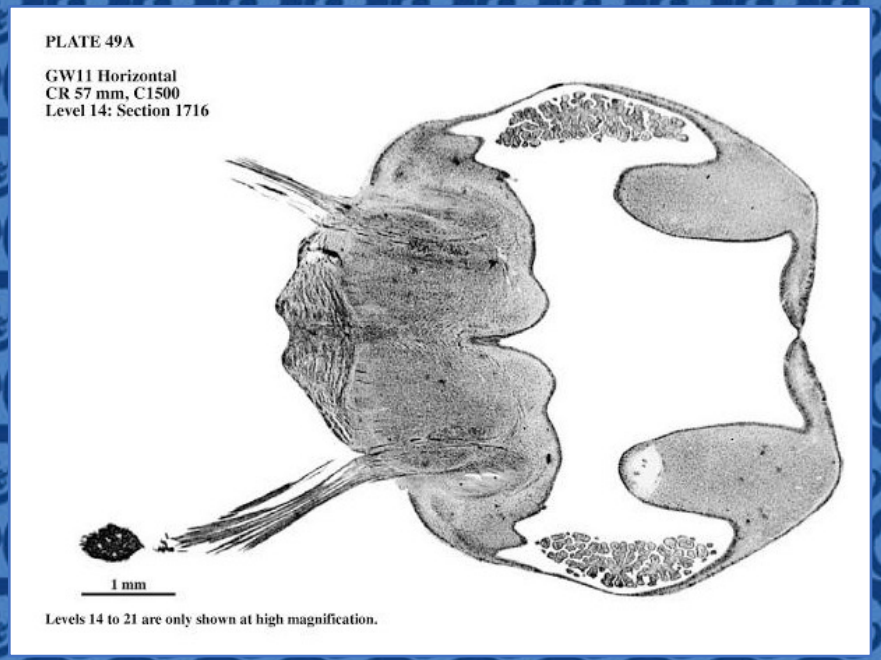
Axial – metencephale Ebene

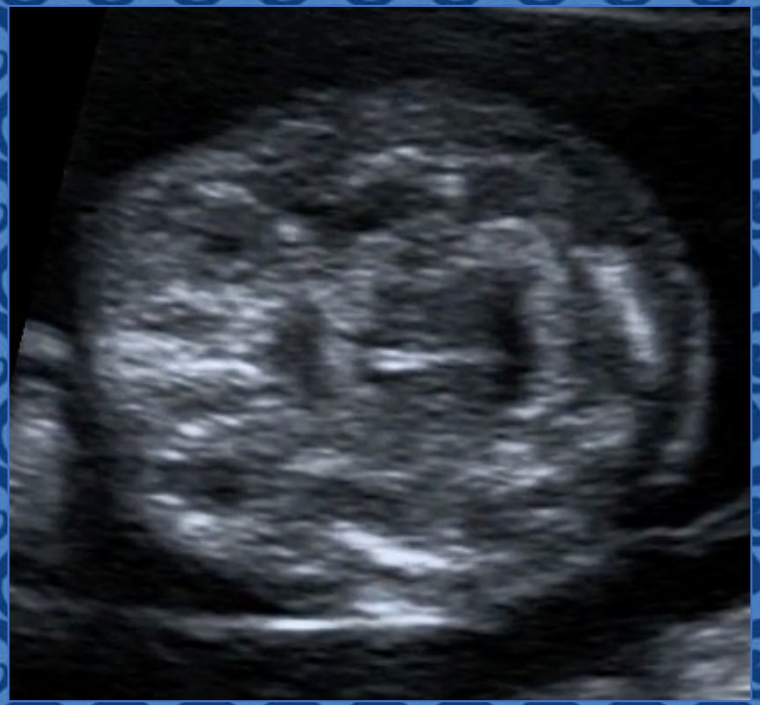


The Human Brain During the Late First Trimester, Bayer

Axial – metencephale Ebene

CRL 57mm

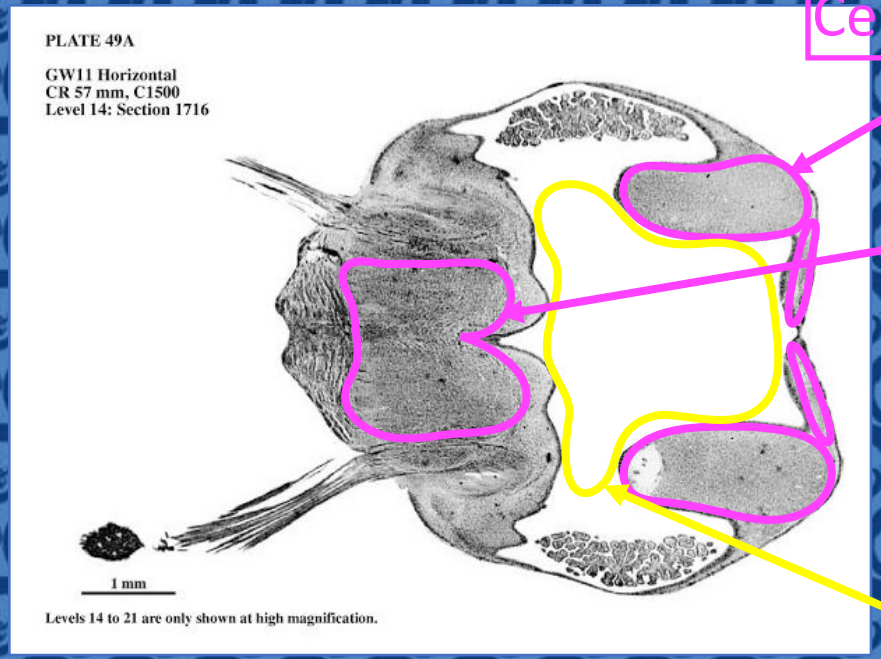


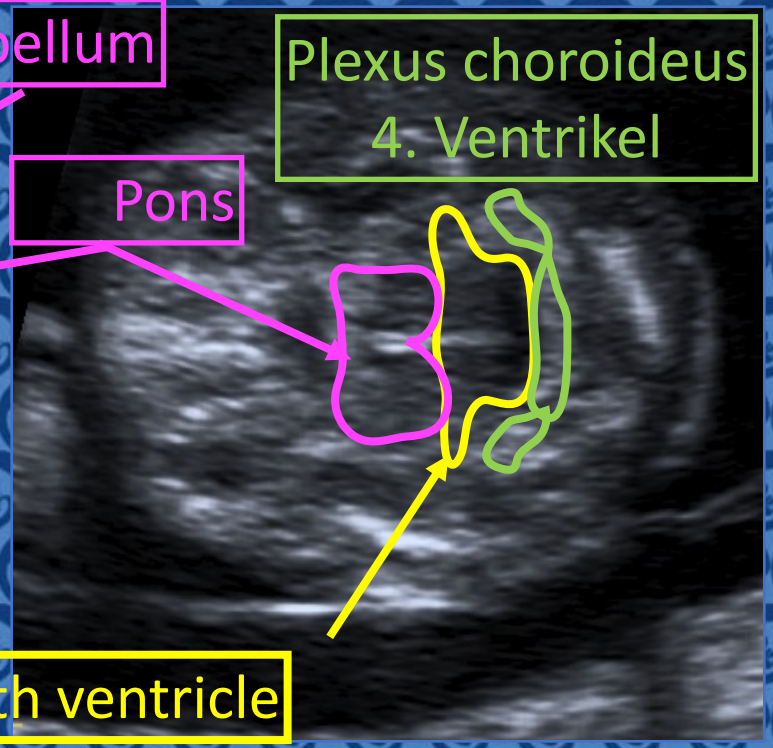


The Human Brain During the Late First Trimester, Bayer

Axial – metencephale Ebene

CRL 57mm





The Human Brain During the Late First Trimester, Bayer

Ultraschallbefunde

Erstes Trimenon

- Verbreiterter Hirnstamm
- Schmaler Hirnstamm

Zweites/drittes Trimenon

- Verminderte Flüssigkeit
- Vermehrte Flüssigkeit
- Vermindertes Volumen
- Strukturelle Veränderungen

Ultraschallbefunde

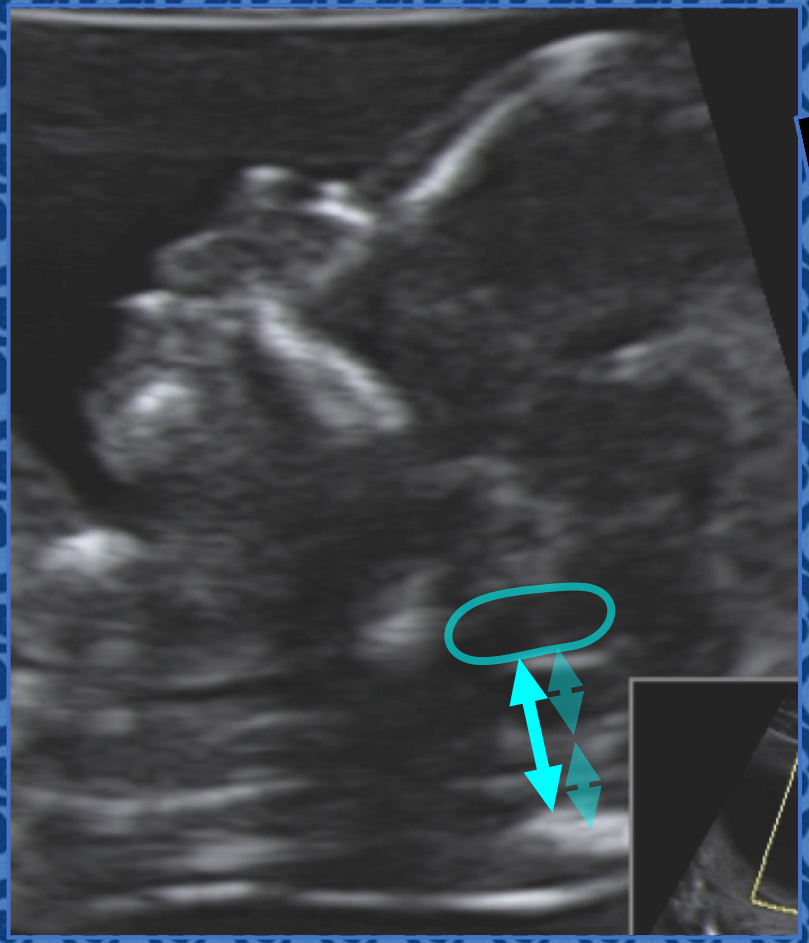
Erstes Trimenon

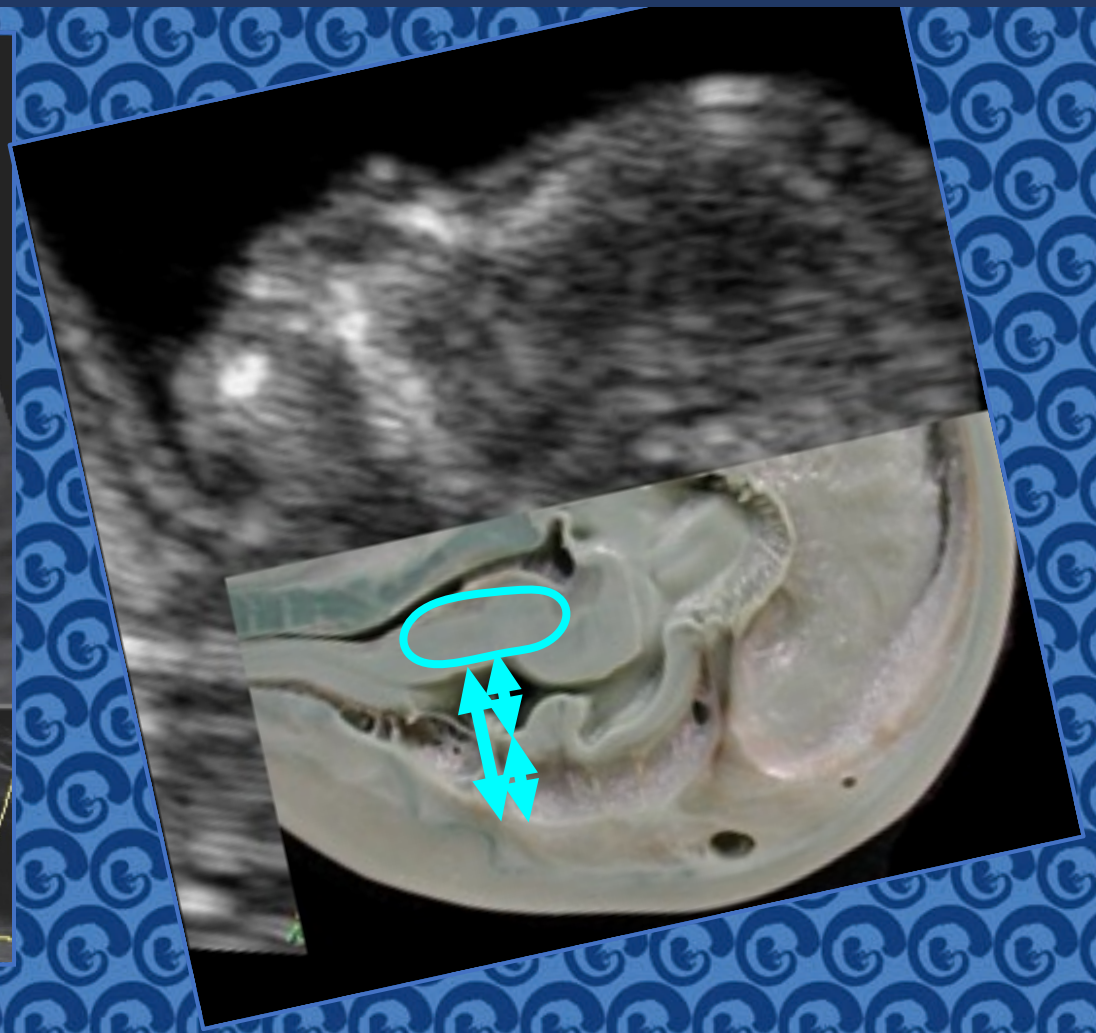
- **Verbreiterter Hirnstamm**
- Schmaler Hirnstamm

Zweites/drittes Trimenon

- Verminderte Flüssigkeit
- Vermehrte Flüssigkeit
- Vermindertes Volumen
- Strukturelle Veränderungen

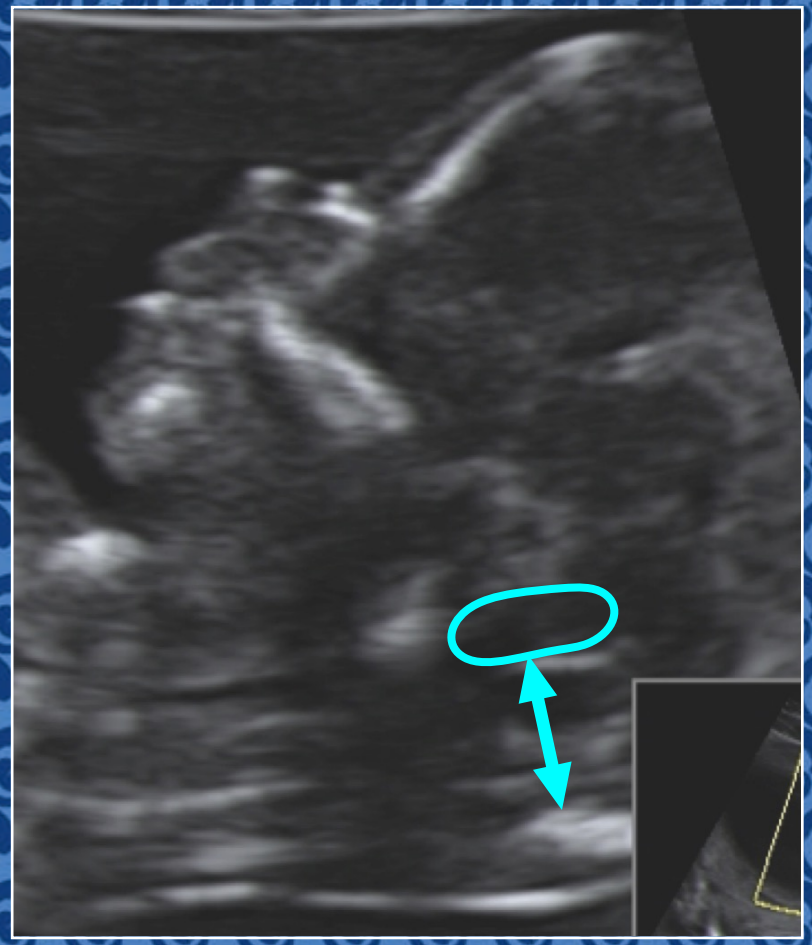
Normalbefund BS/BSOB SSW 12

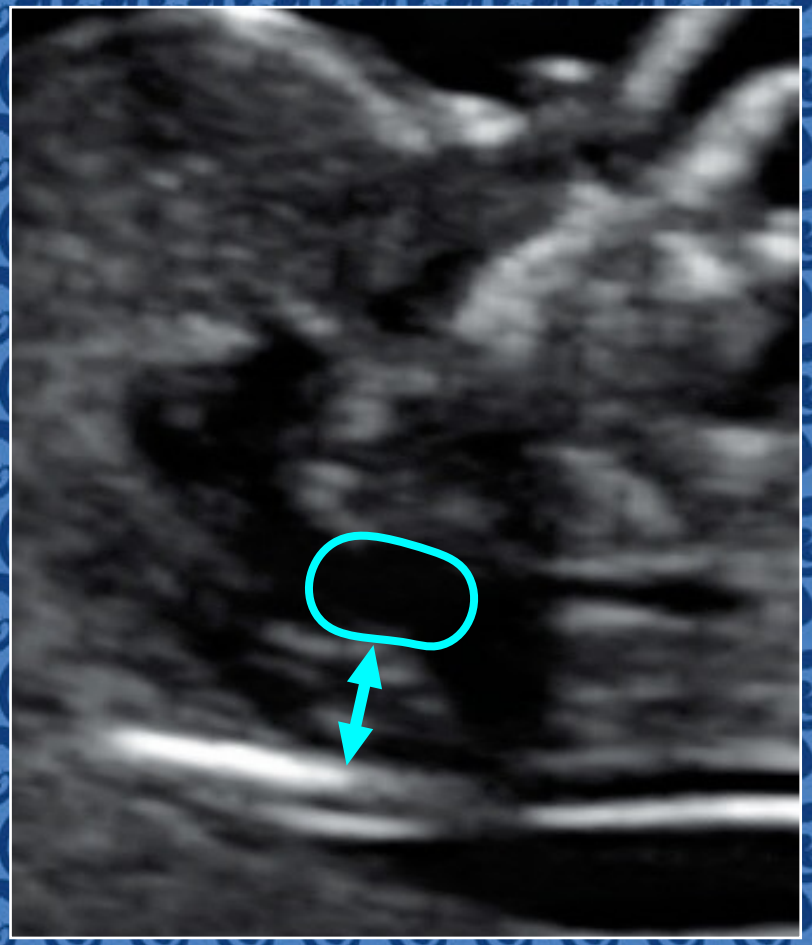




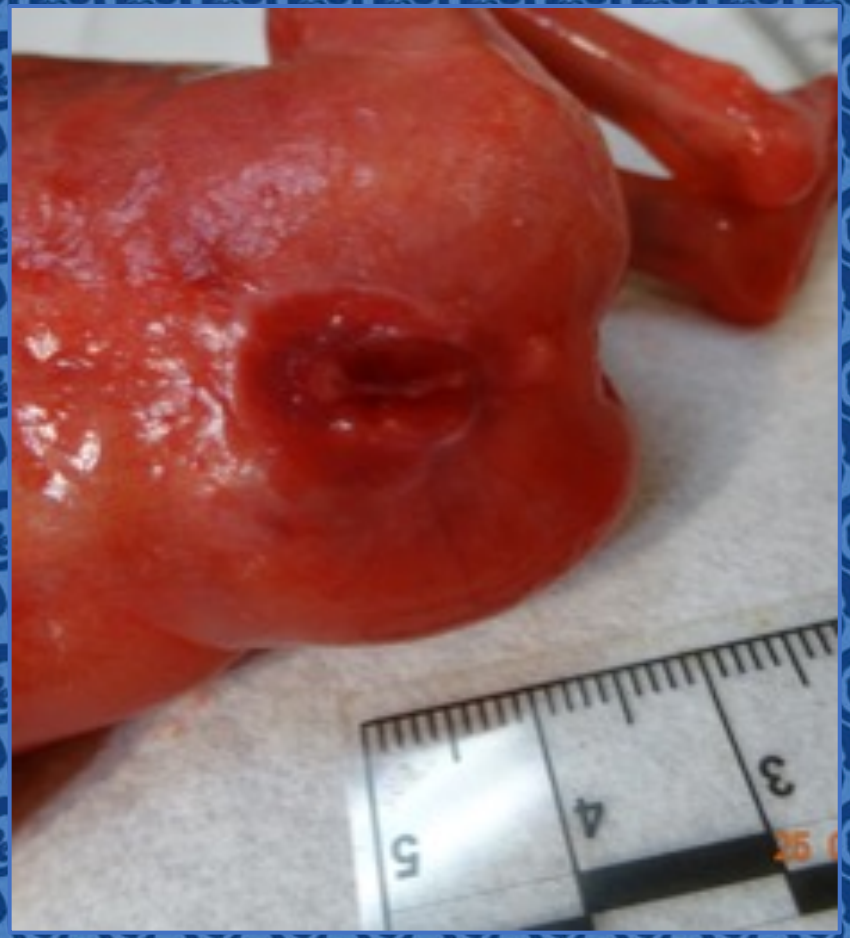
Präparat Peter Brugger

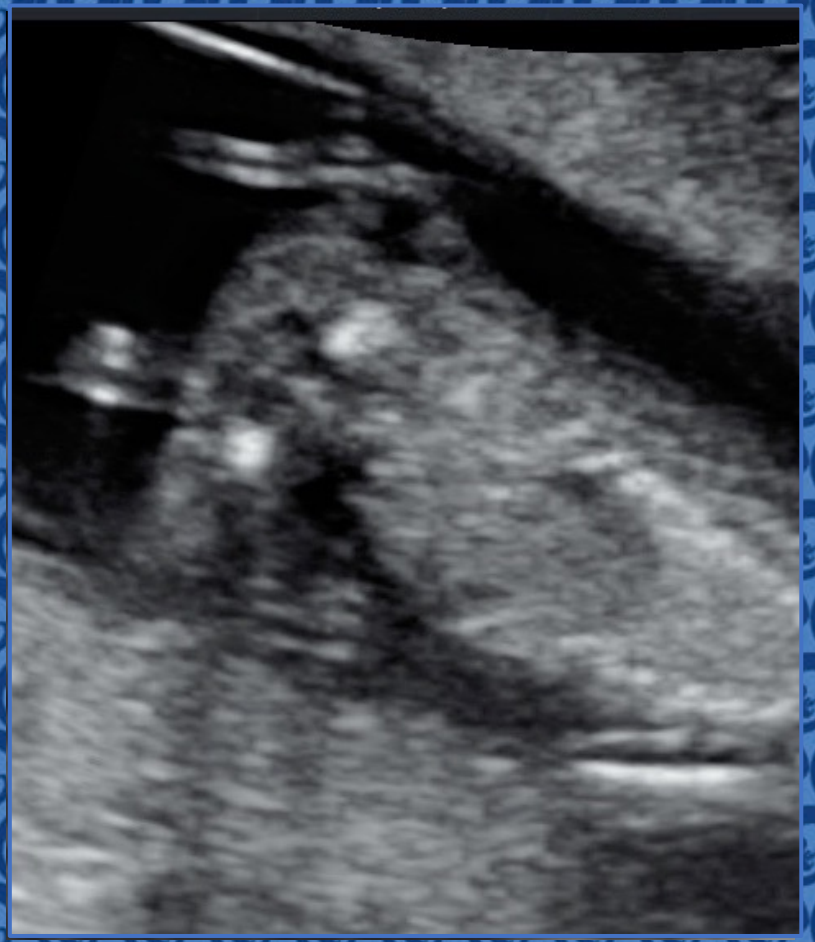
Spina bifida SSW 12





Spina bifida SSW 15





Sichtbare infratentorielle Fehlentwicklungen

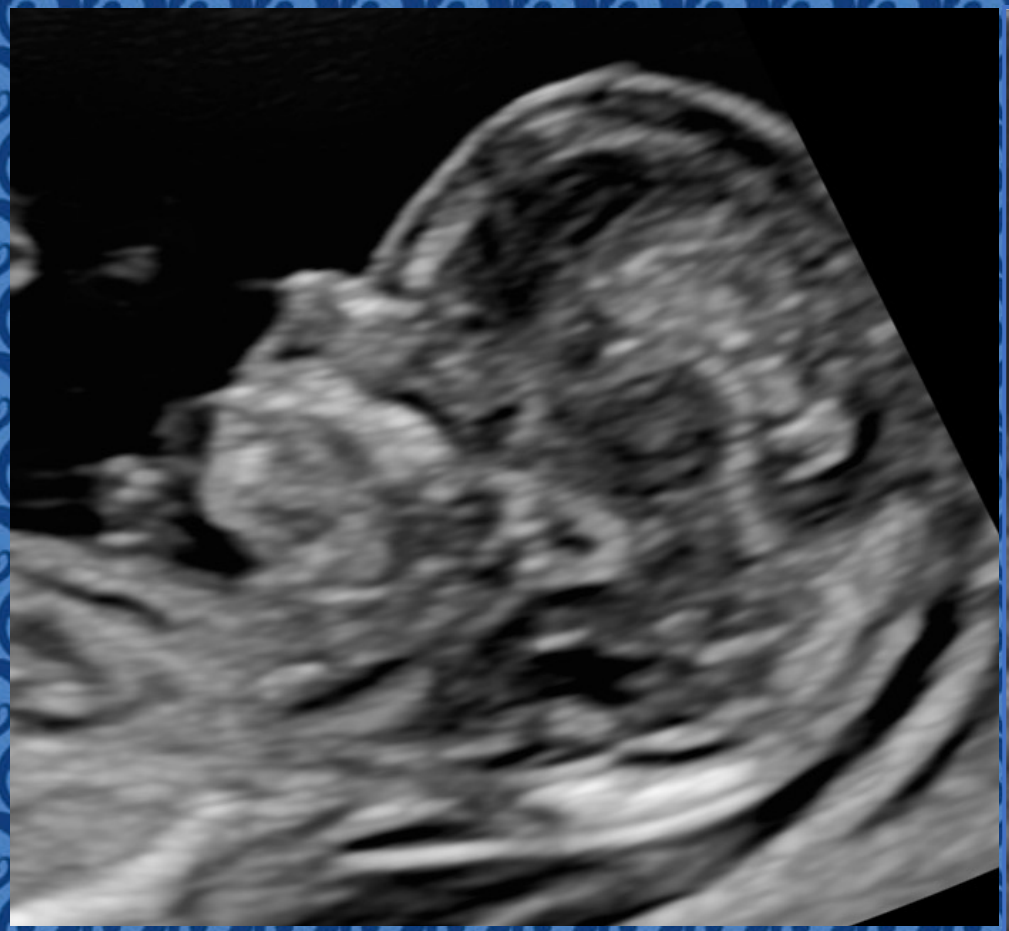
Erstes Trimenon

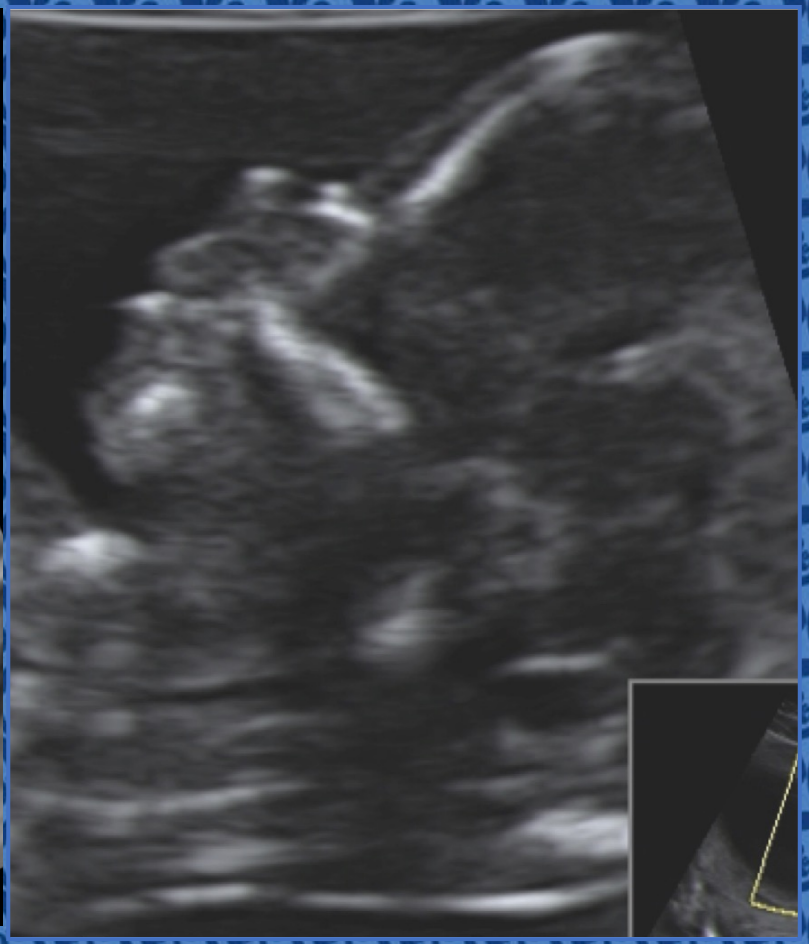
- Breiter Hirnstamm
- **Schmaler Hirnstamm**

Zweites/drittes Trimenon

- Verminderte Flüssigkeit
- Vermehrte Flüssigkeit
- Vermindertes Volumen
- Strukturelle Veränderungen

Schmaler Hirnstamm im 1. Trimenon





Sichtbare infratentorielle Fehlentwicklungen

Erstes Trimenon	Zweites/drittes Trimenon
• Spina bifida aperta • Zystische Läsionen	• Verminderte Flüssigkeit • Vermehrte Flüssigkeit • Vermindertes Volumen • Strukturelle Veränderungen

Sichtbare infratentorielle Fehlentwicklungen

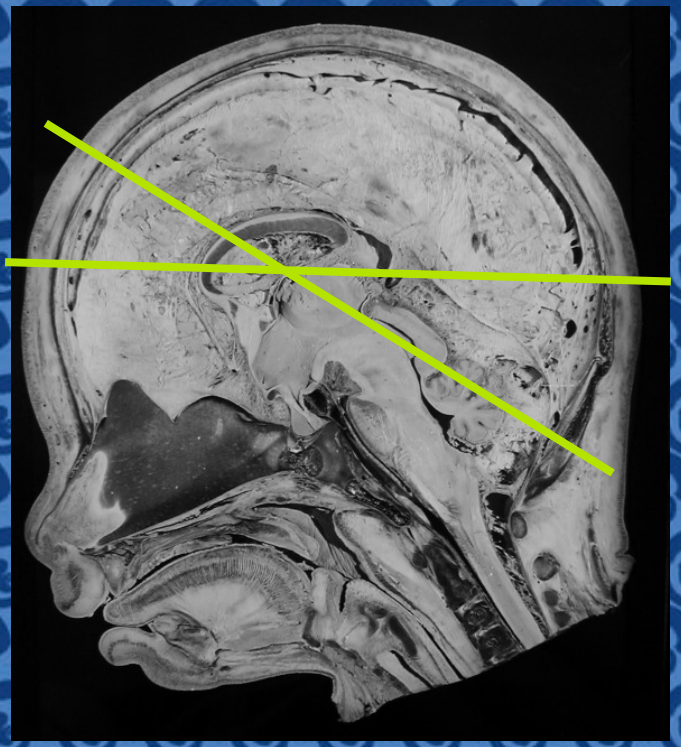
Erstes Trimenon

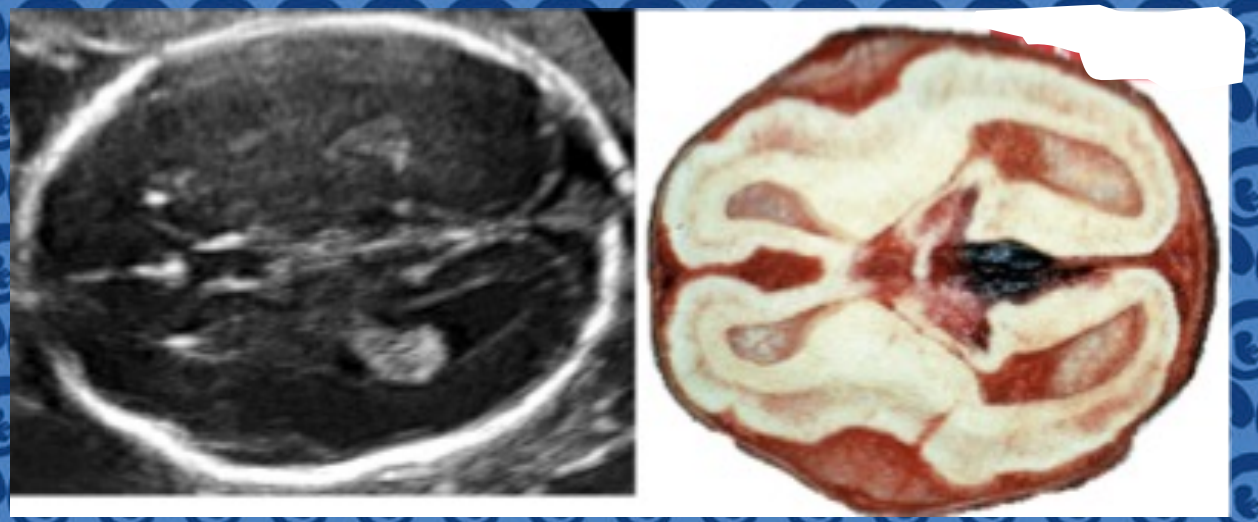
- Spina bifida aperta
- Zystische Läsionen

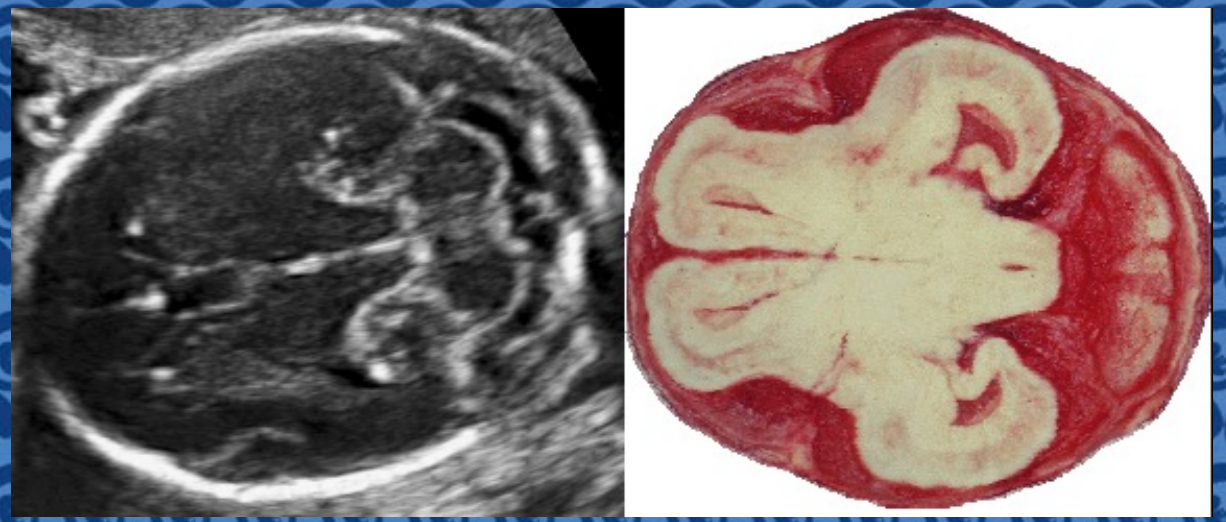
Zweites/drittes Trimenon

- Verminderte Flüssigkeit
- Vermehrte Flüssigkeit
- Vermindertes Volumen
- Strukturelle Veränderungen

Normalbefund

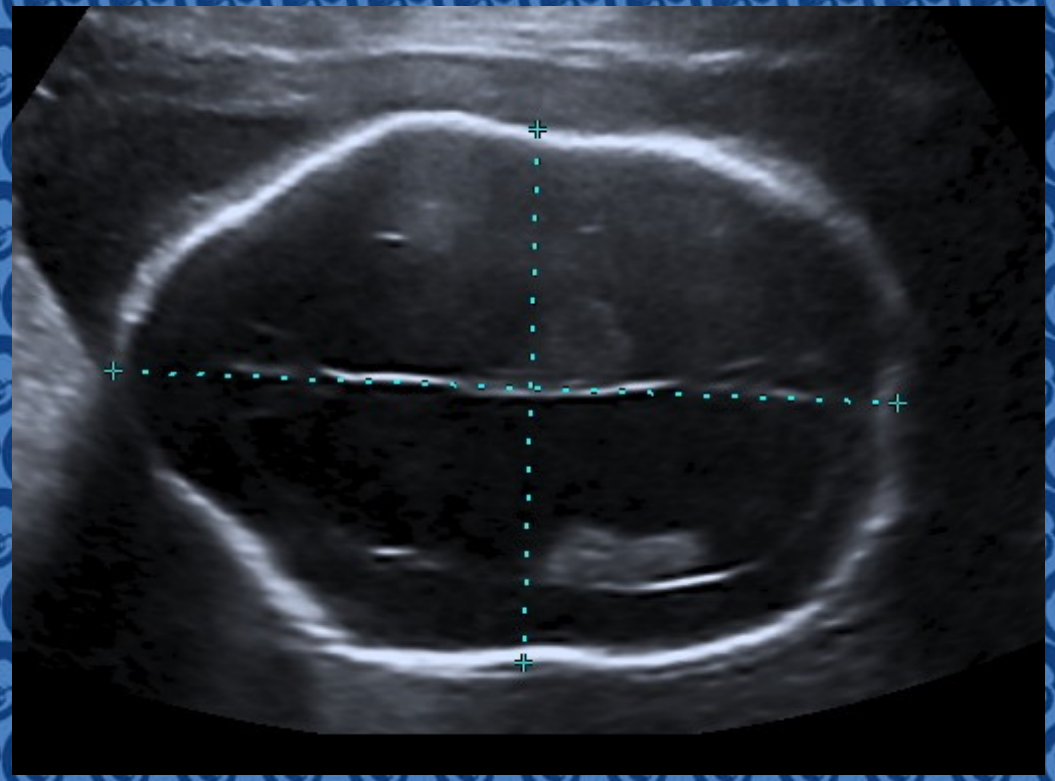


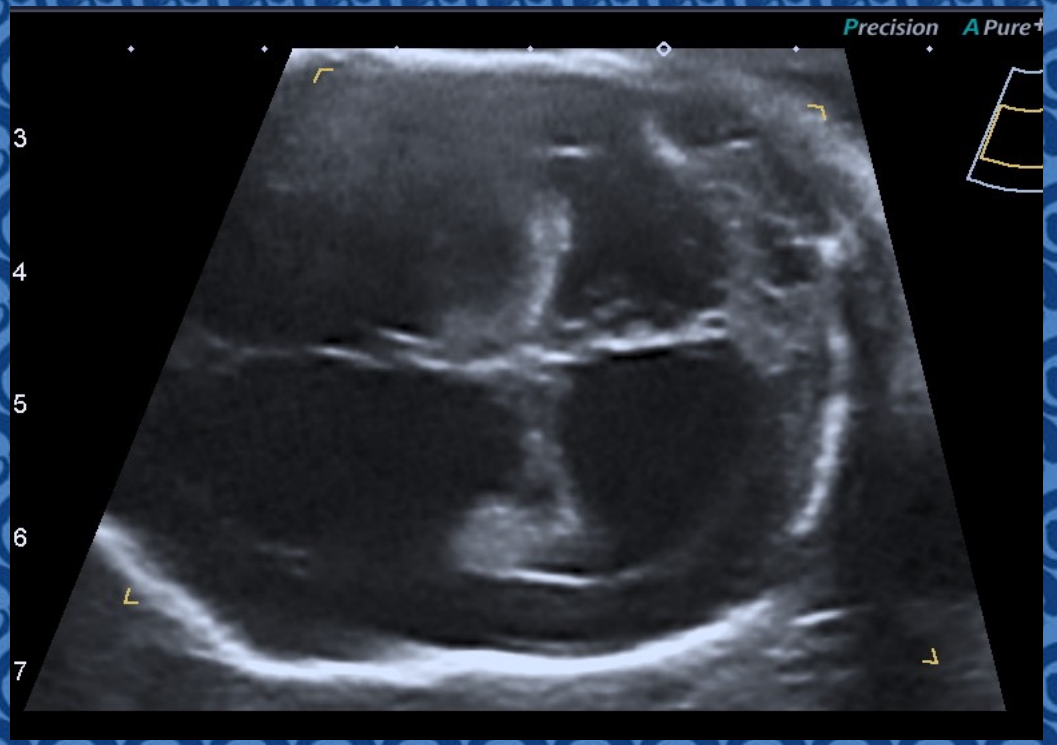




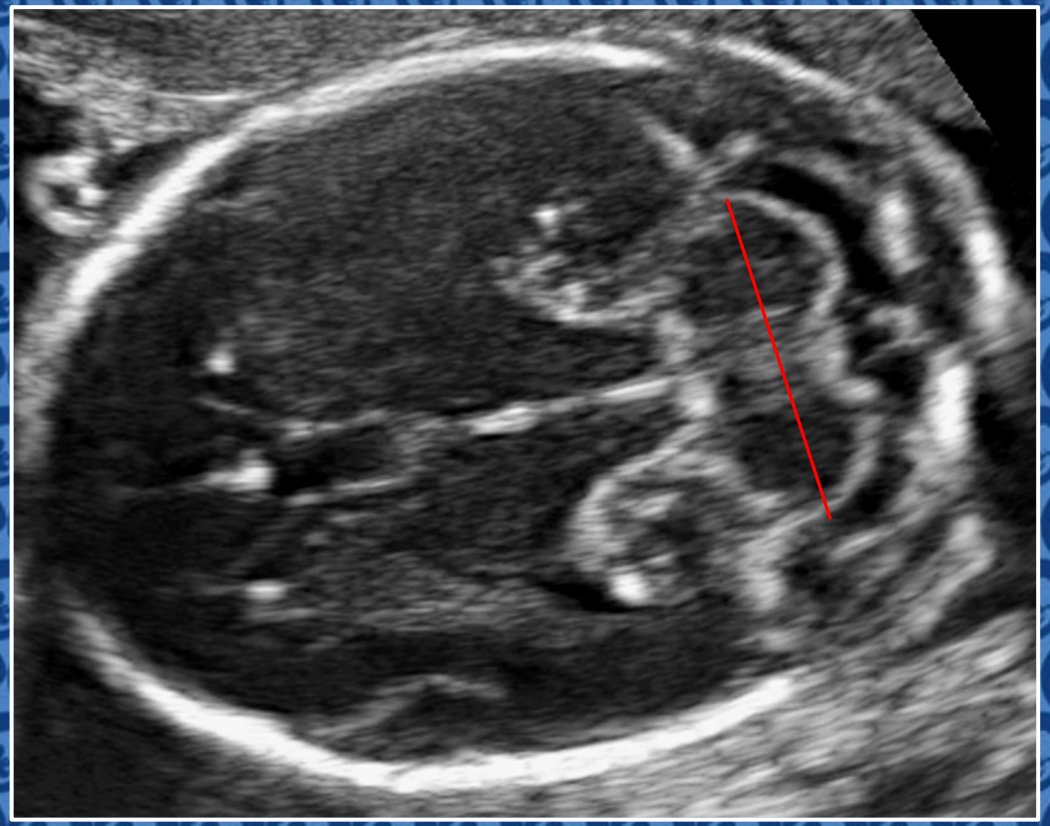
Quelle: ISUOG

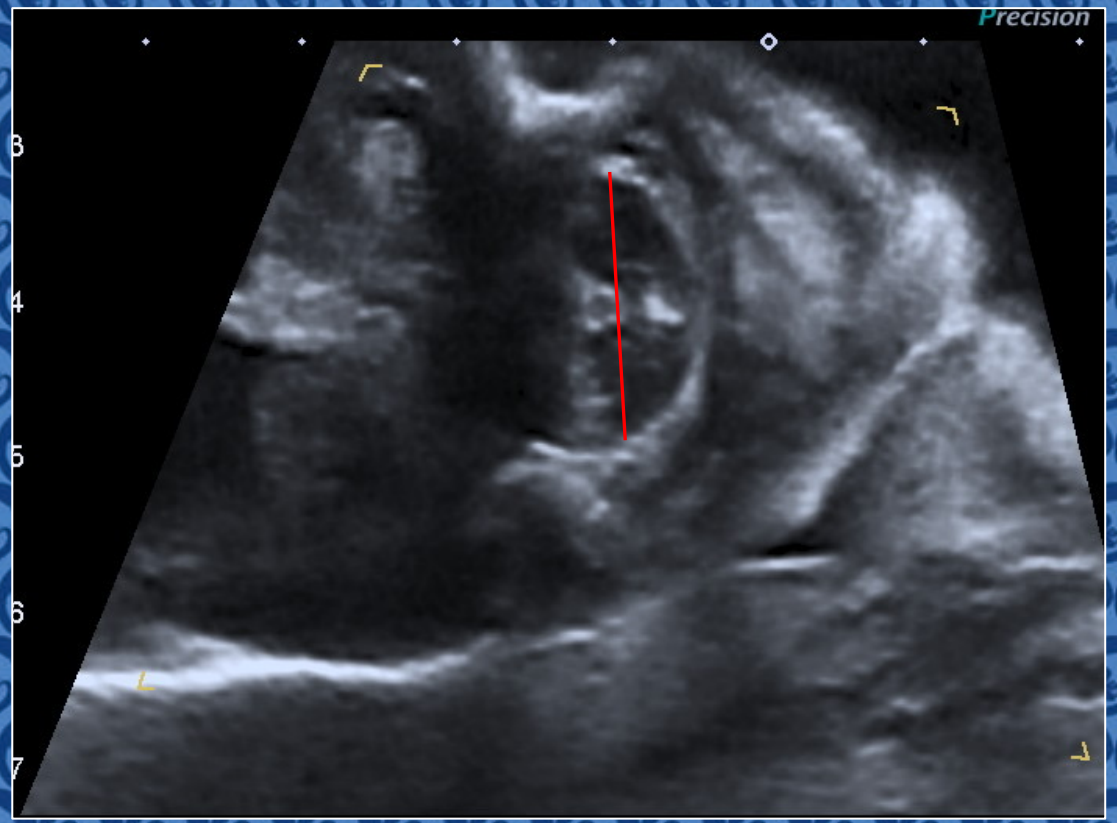
Kopfform - Ventrikulomegalie



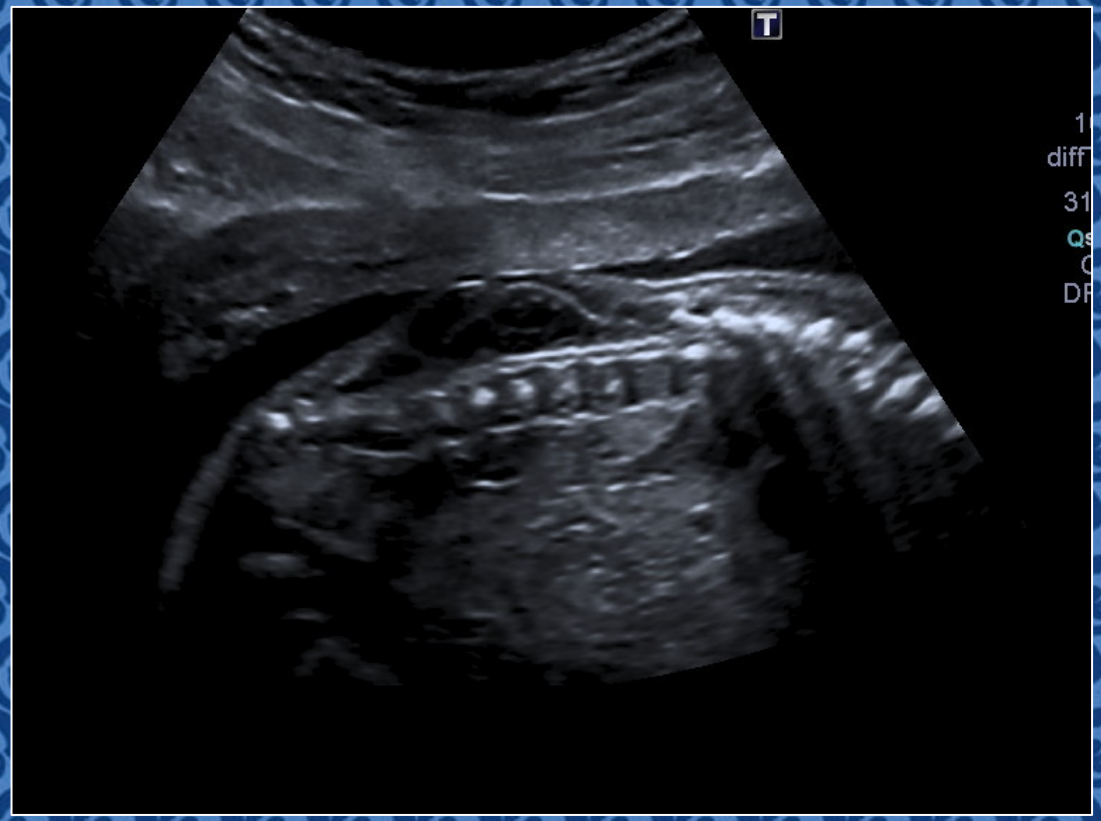


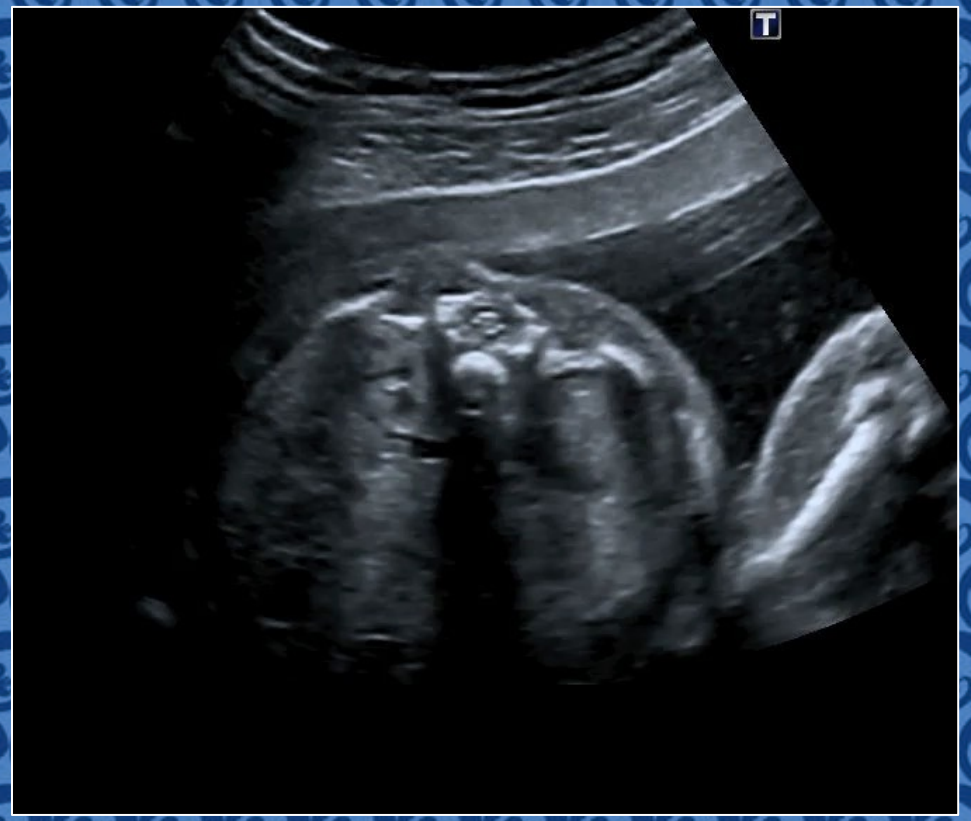
Normalbefund vs Chiari Malformation





Spina bifida - Myelomeningocele

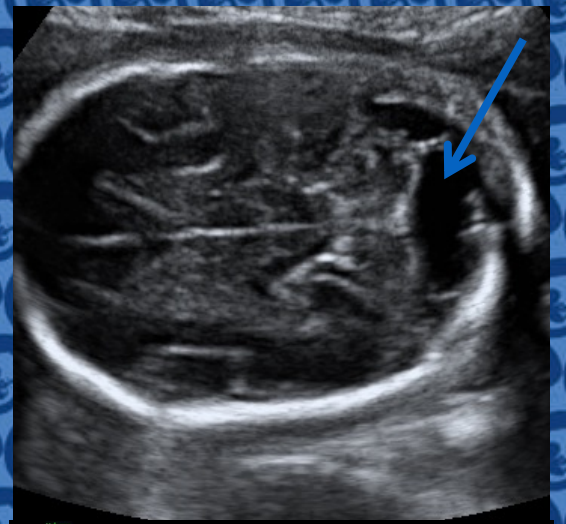


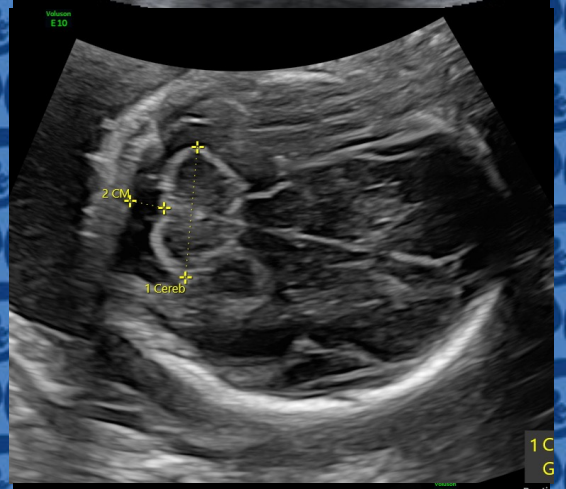


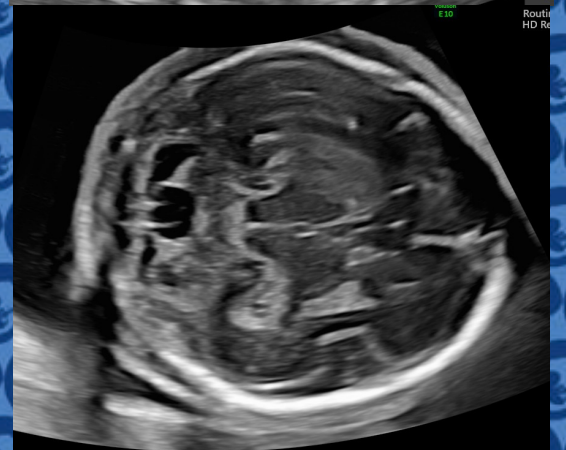
Hinweis auf Kleinhirnanomalien

- Großer Raum hinter dem Zerebellum
- Kleiner TCD
- Echogenität

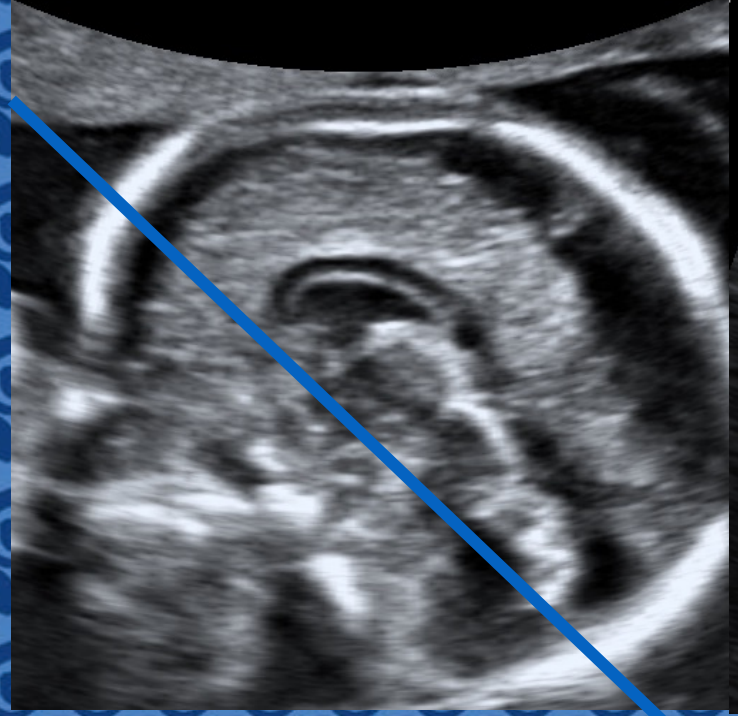
- IST DER VERMIS NORMAL?

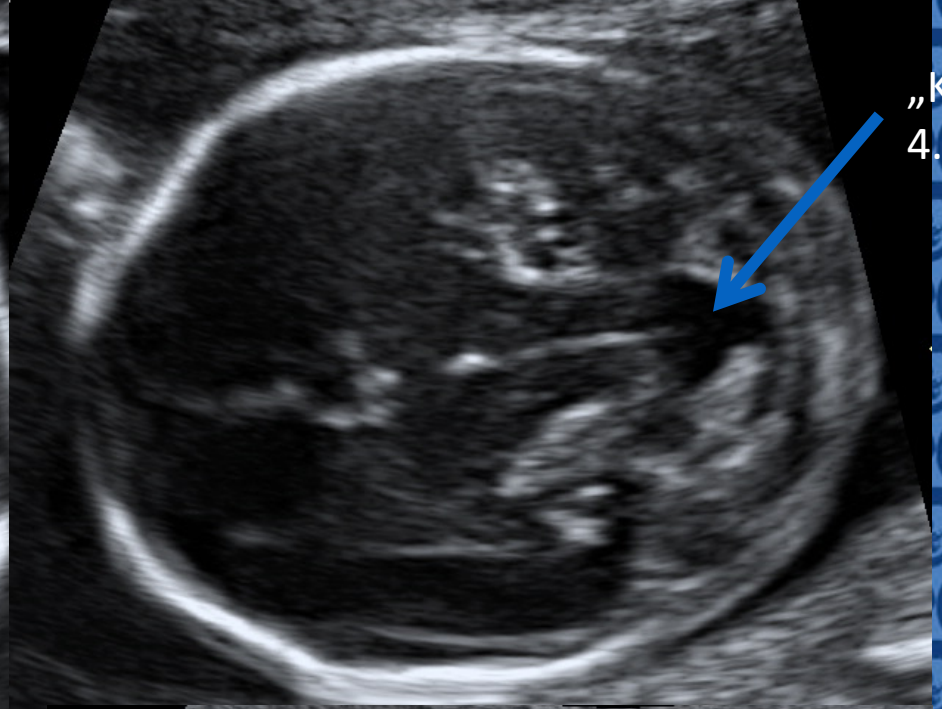






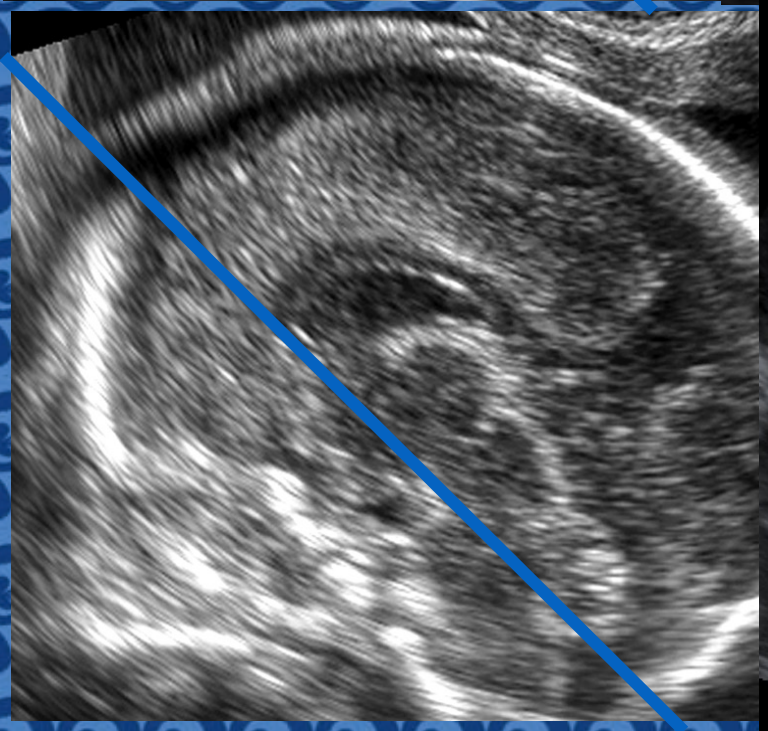
< 20 SSW

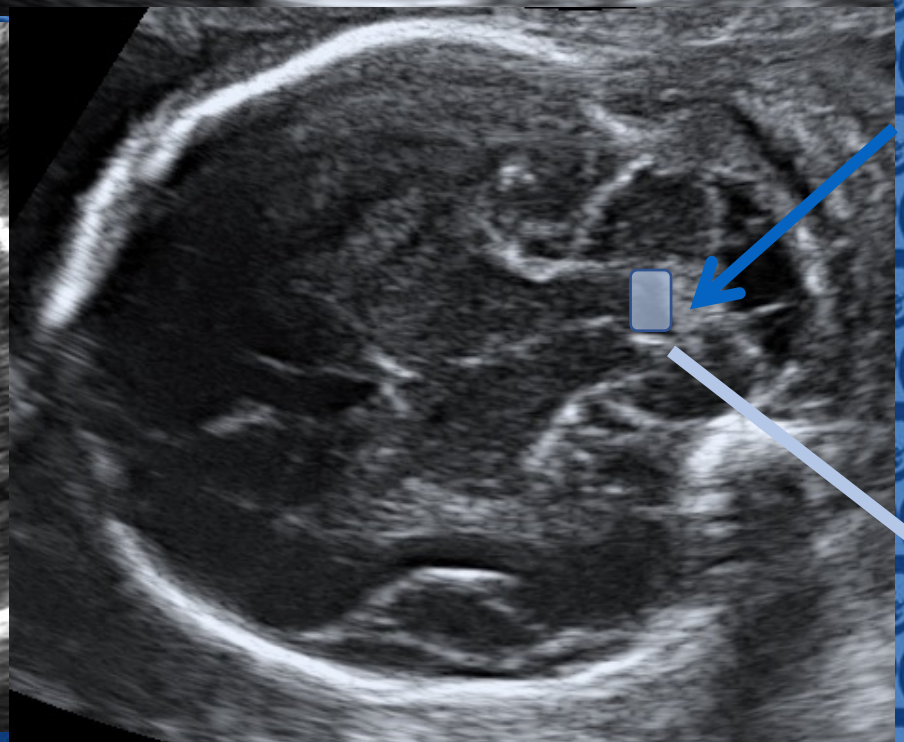




„Kommunikation“
4. Ventrikel / CM

≥ 20 SSW





Vermis

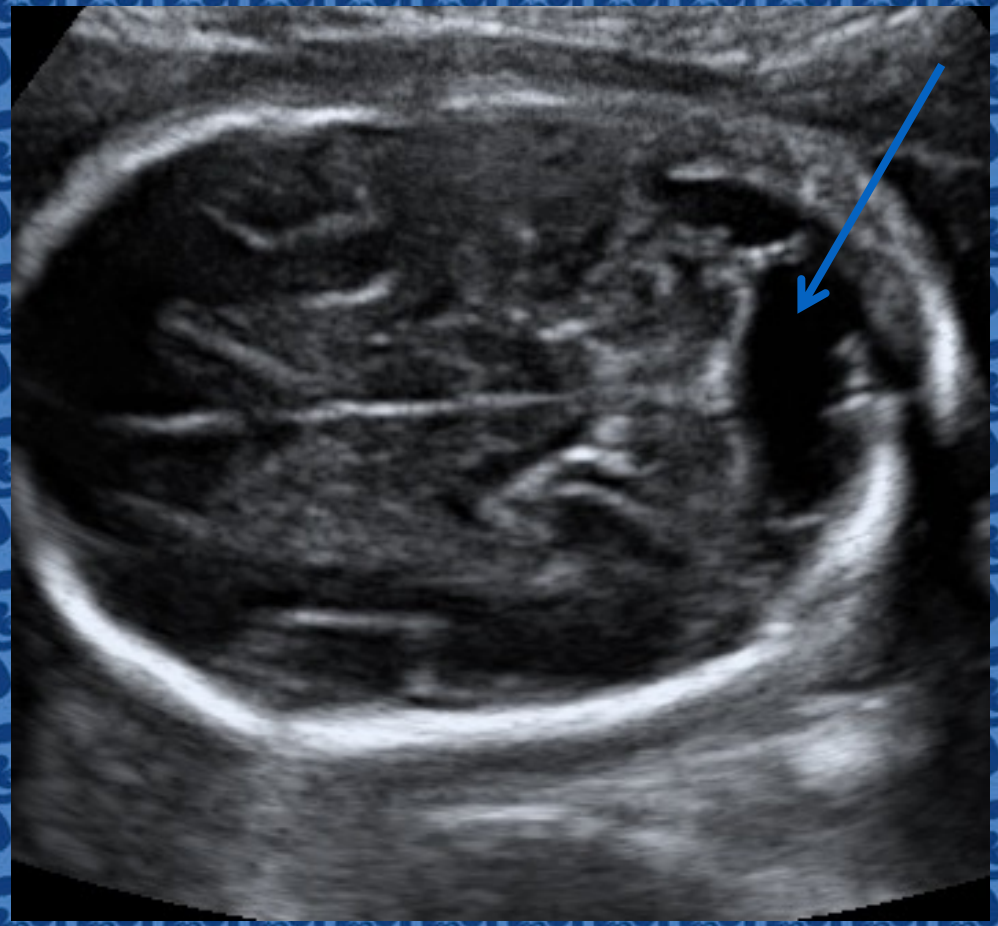
4. Ventrikel

Quelle: ISUOG

Hinweis auf Kleinhirnanomalien

- **Großer Raum hinter dem Zerebellum**
- Kleiner TCD
- Echogenizität

- IST DER VERMIS NORMAL?



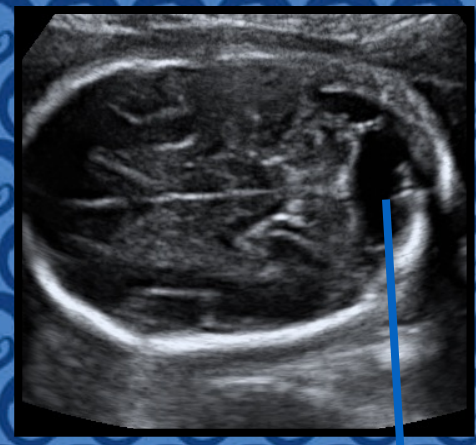
Cisterna magna > 10 mm

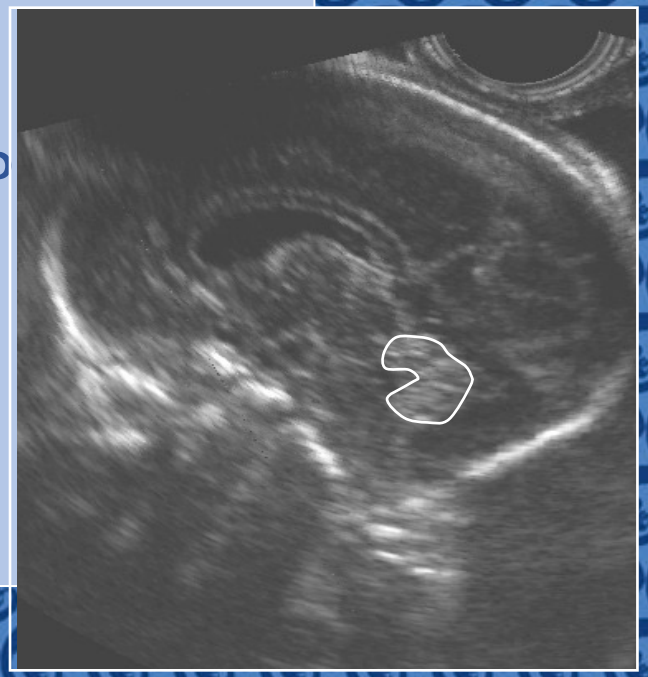
Hinweis auf Kleinhirnanomalien

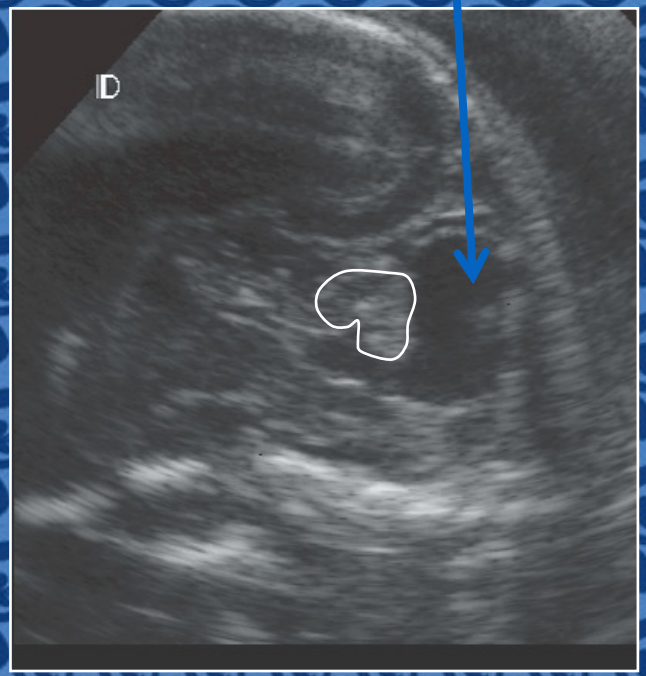
- **Großer Raum hinter dem Zerebellum, CM > 10mm**
- Kleiner TCD
- Echogenität

- IST DER VERMIS NORMAL?

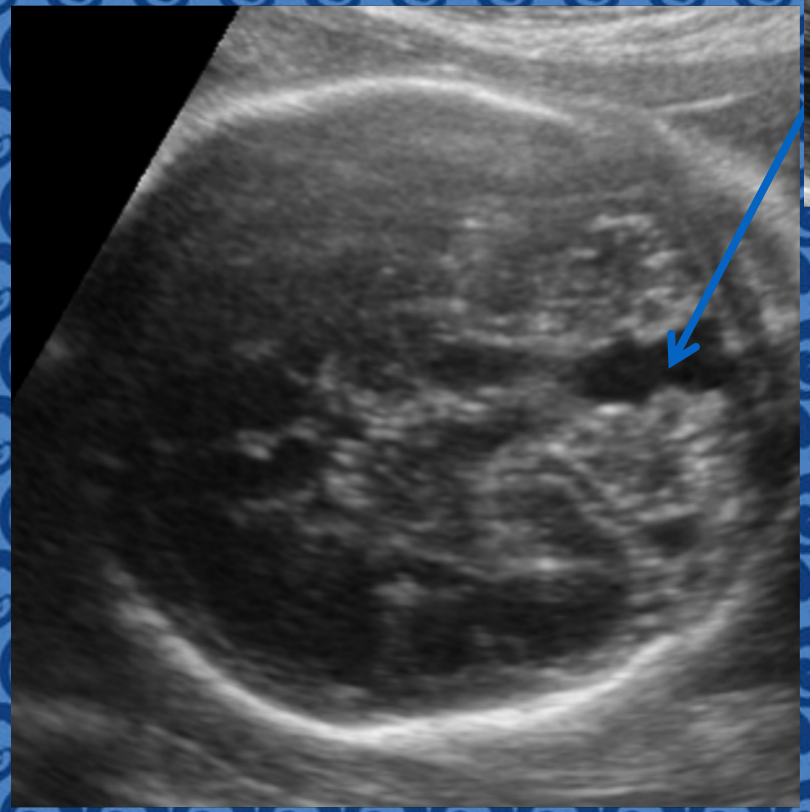
Megazisterna magna (MM)
Arachnoidalzyste (AC)

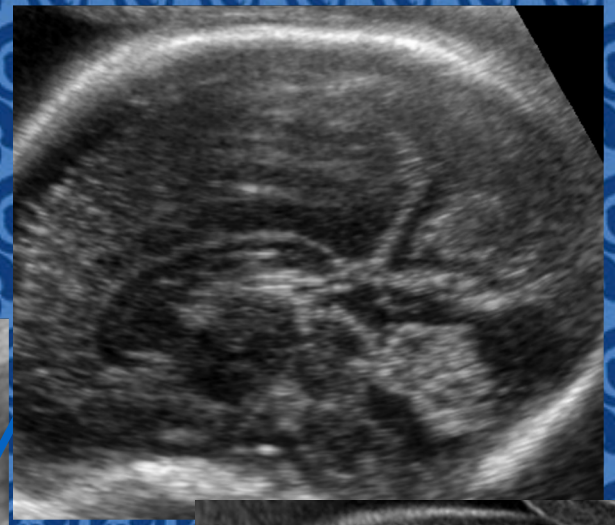




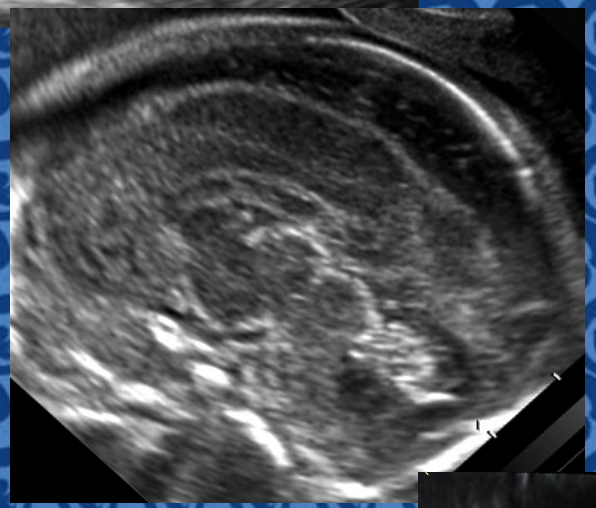


~~Cerebellar 'cleft'~~

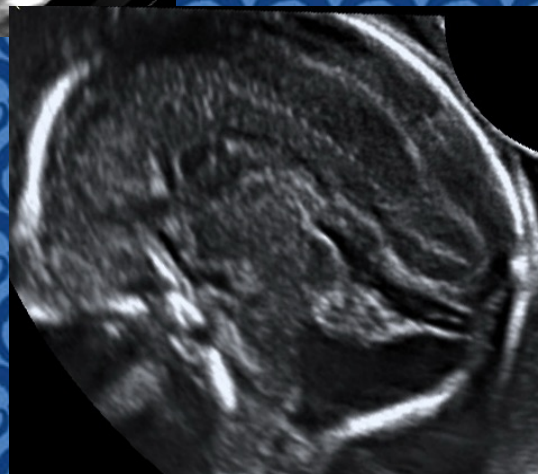




Blake's pouch cyst (BPC)



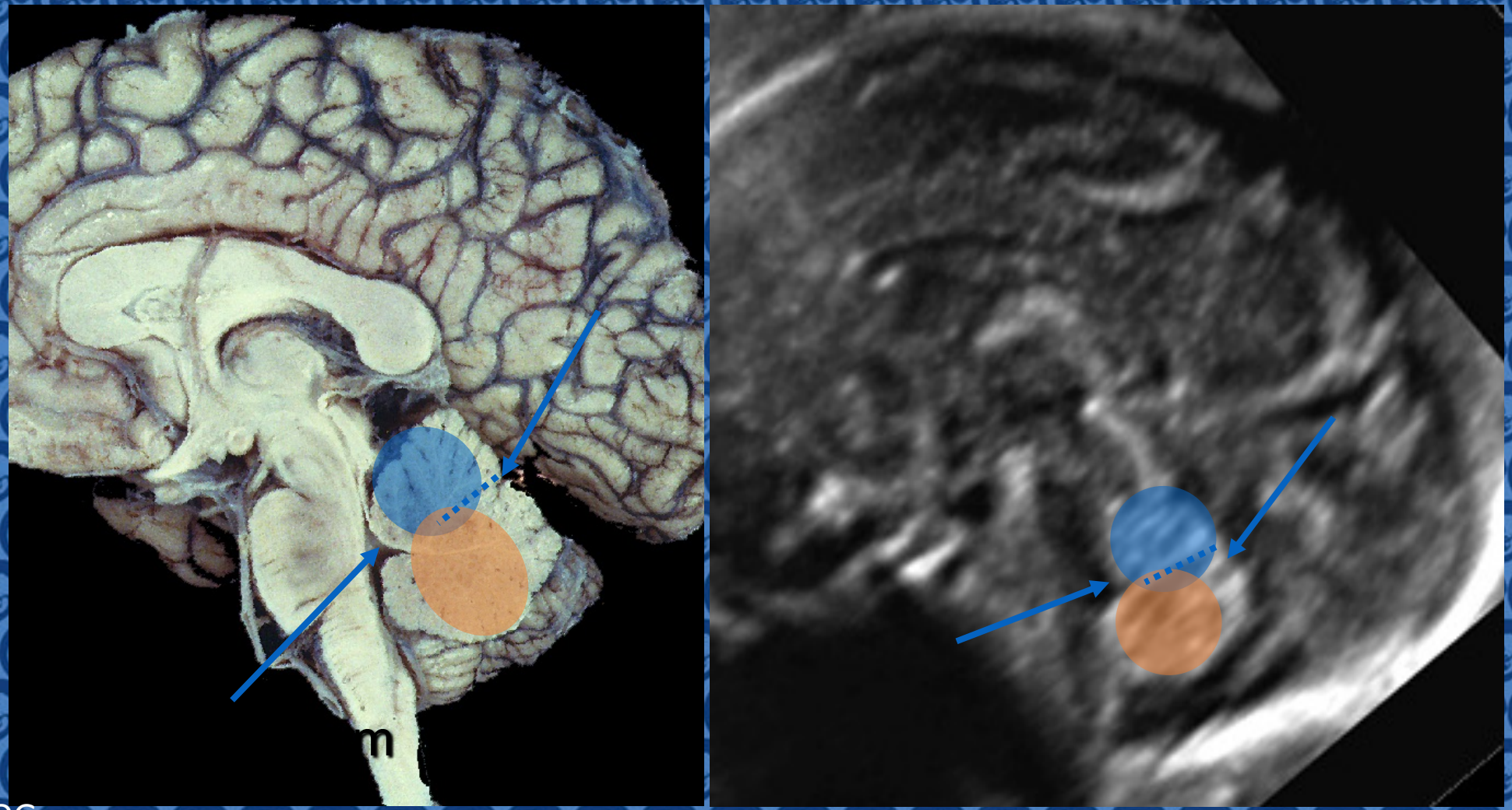
Vermian hypoplasia (VH)



Dandy-Walker malformation DWM

Quelle: ISUOG

Ist der vermis intakt?

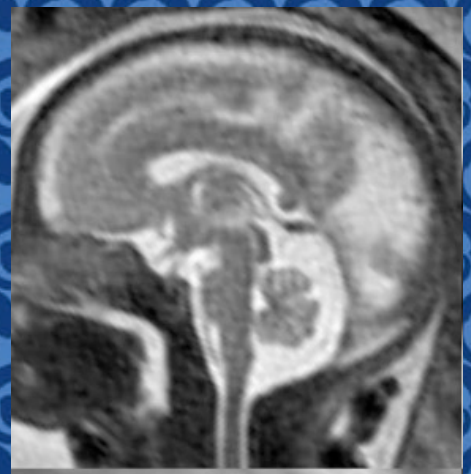


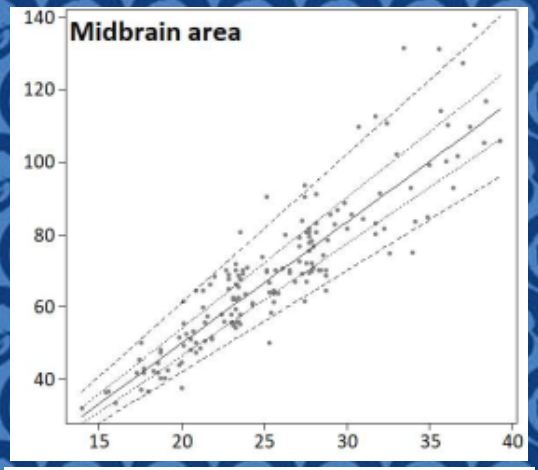
Quelle: ISUOG

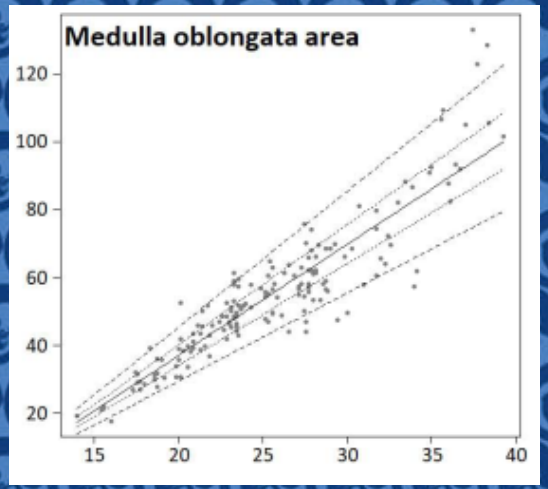
Twice as much vermis below the primary fissure than above

Fossa posterior

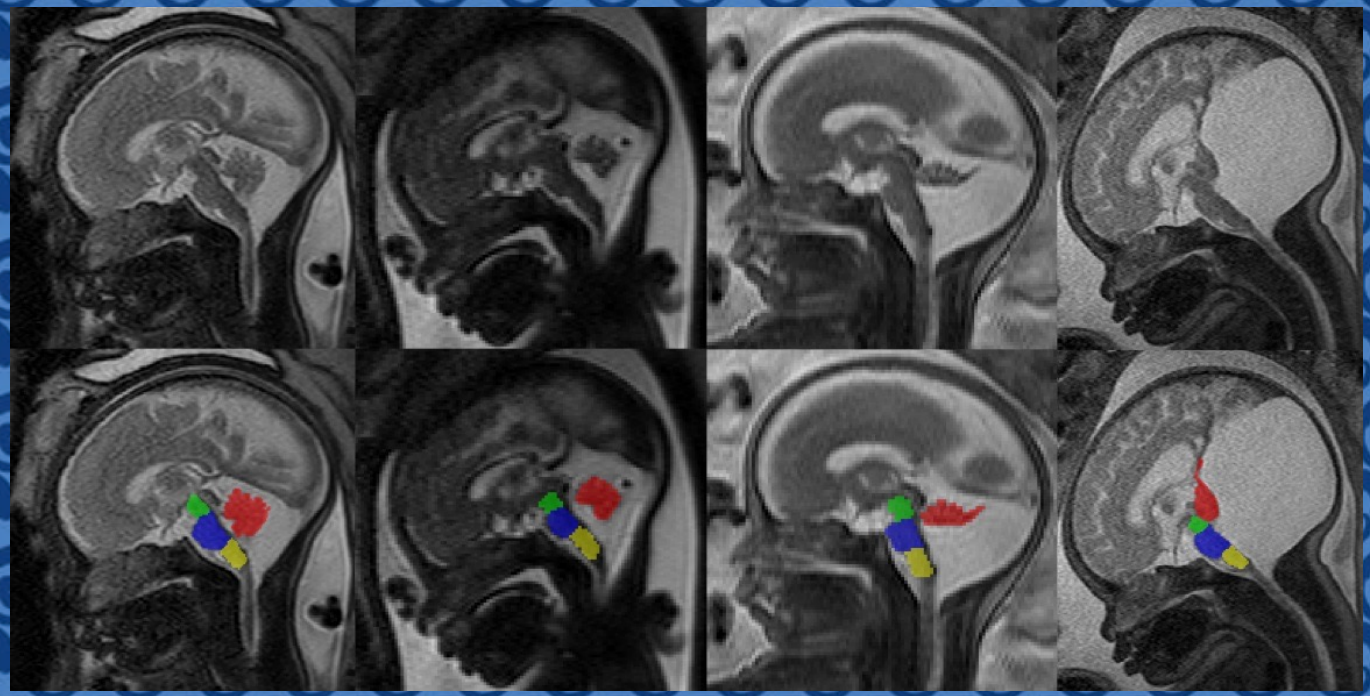
Dovjak GO, et al. UOG 2020.







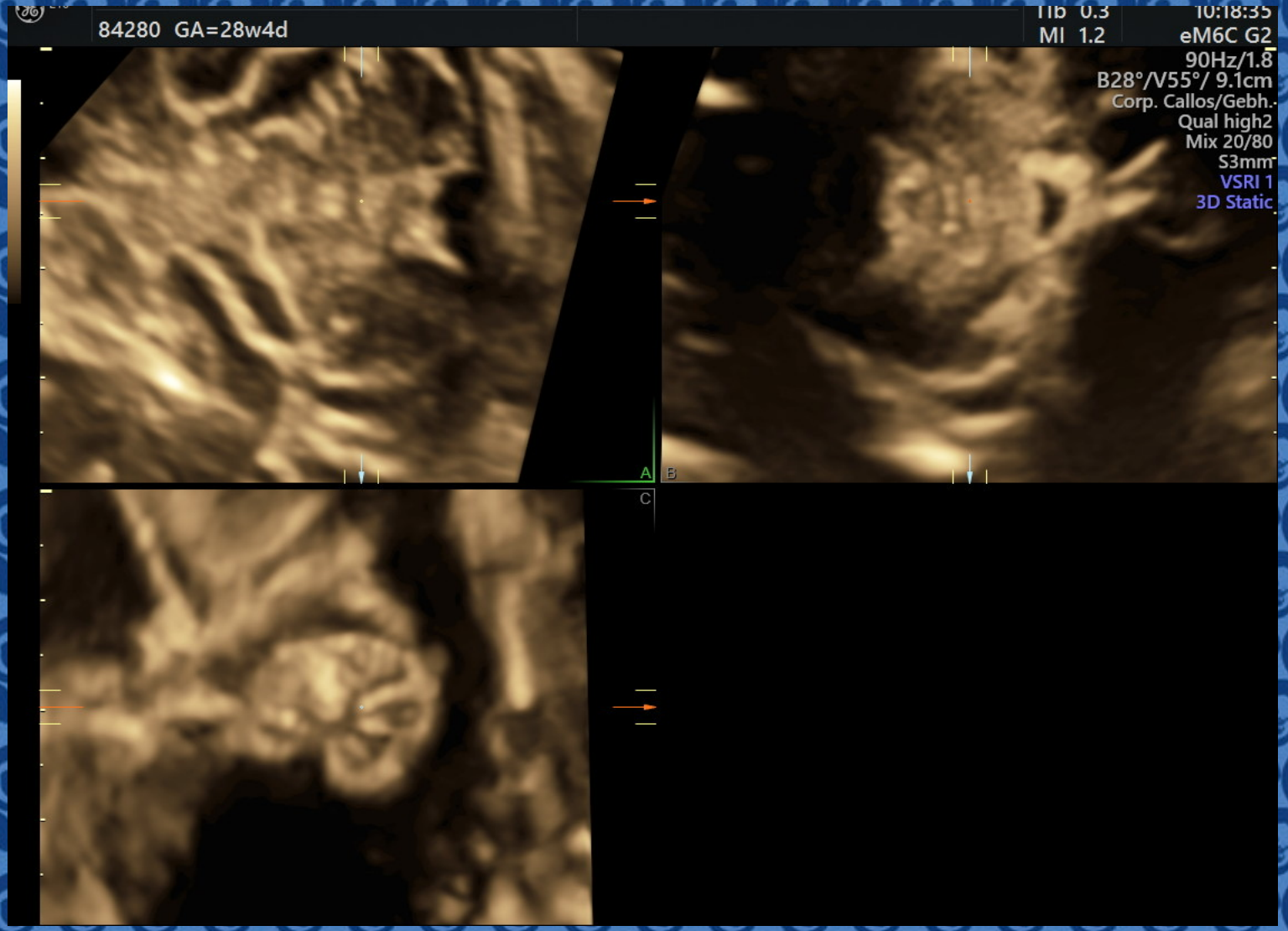
Group 1 Group 2 Group 3 Group 3

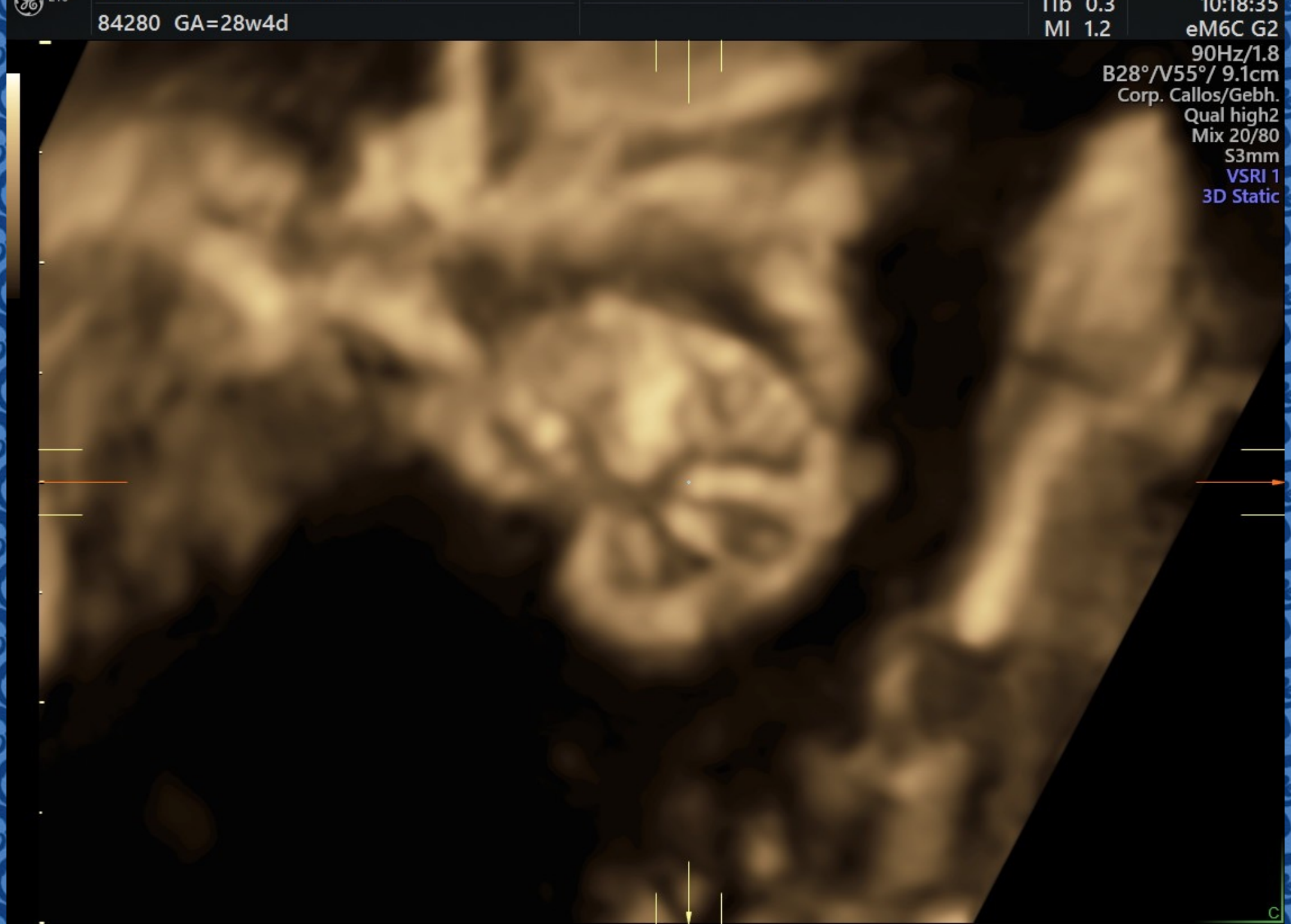


MM, BPC, AC – VH BPC - DWM

Dovjak GO, et al. UOG 2019;8:21890

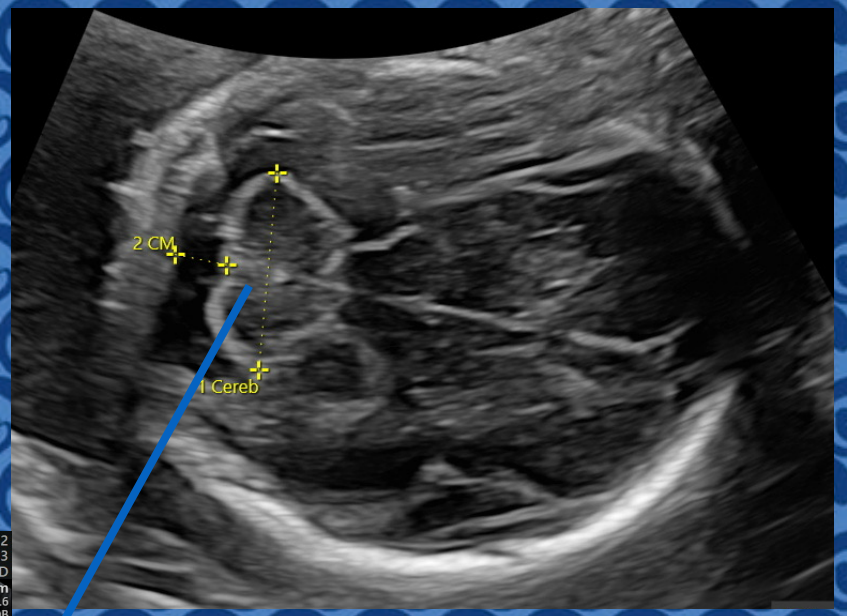
Ist der vermis intakt?

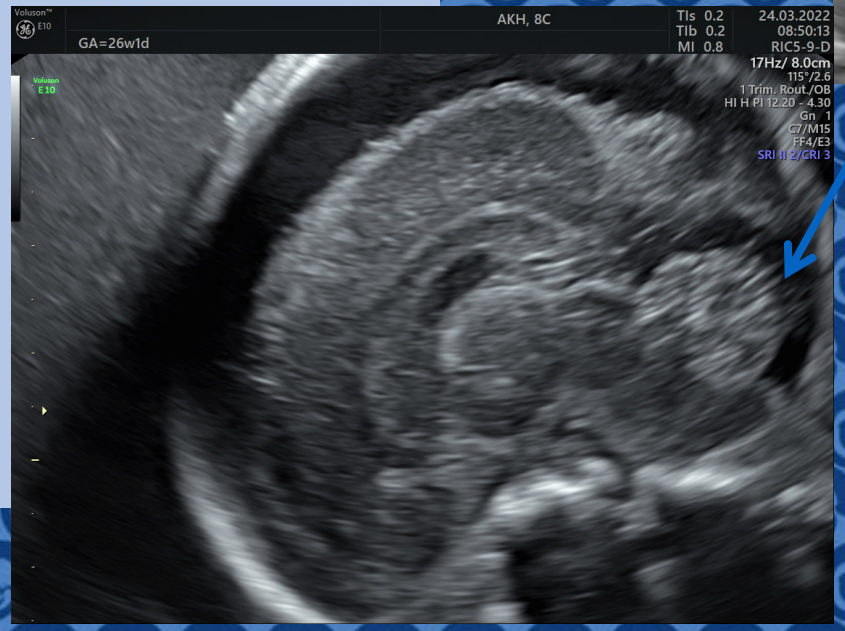



84280 GA=28w4d
TIb 0.3
MI 1.2
10:18:35
eM6C G2
90Hz/1.8
B28°/V55°/ 9.1cm
Corp. Callos/Gebh.
Qual high2
Mix 20/80
S3mm
VSRI 1
3D Static

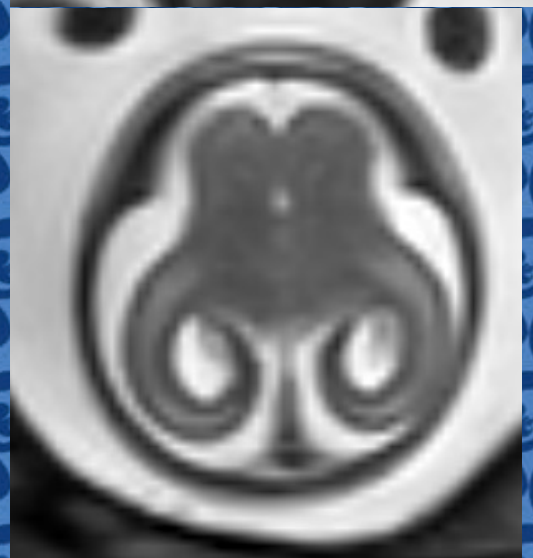
Hinweis auf Kleinhirnanomalien

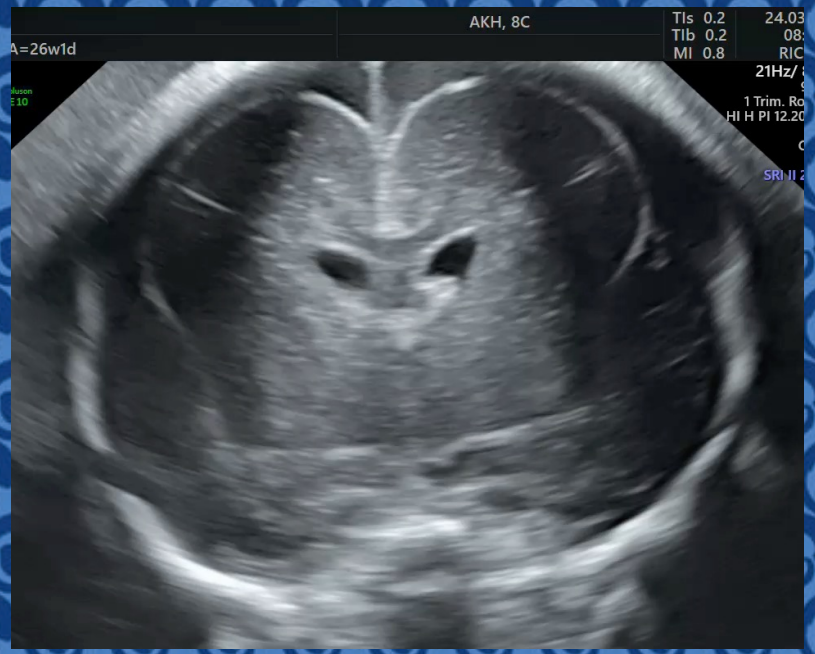
- Großer Raum hinter dem Zerebellum
- **Kleiner TCD**
- Echogenität

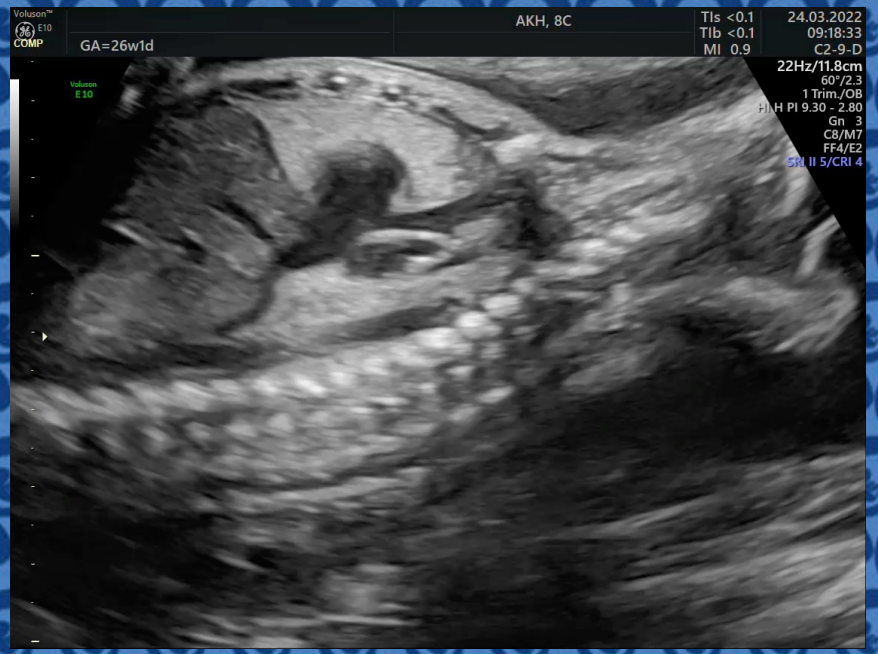




Mandibulo-faziales Dysostose-Mikrozephalie Syndrom

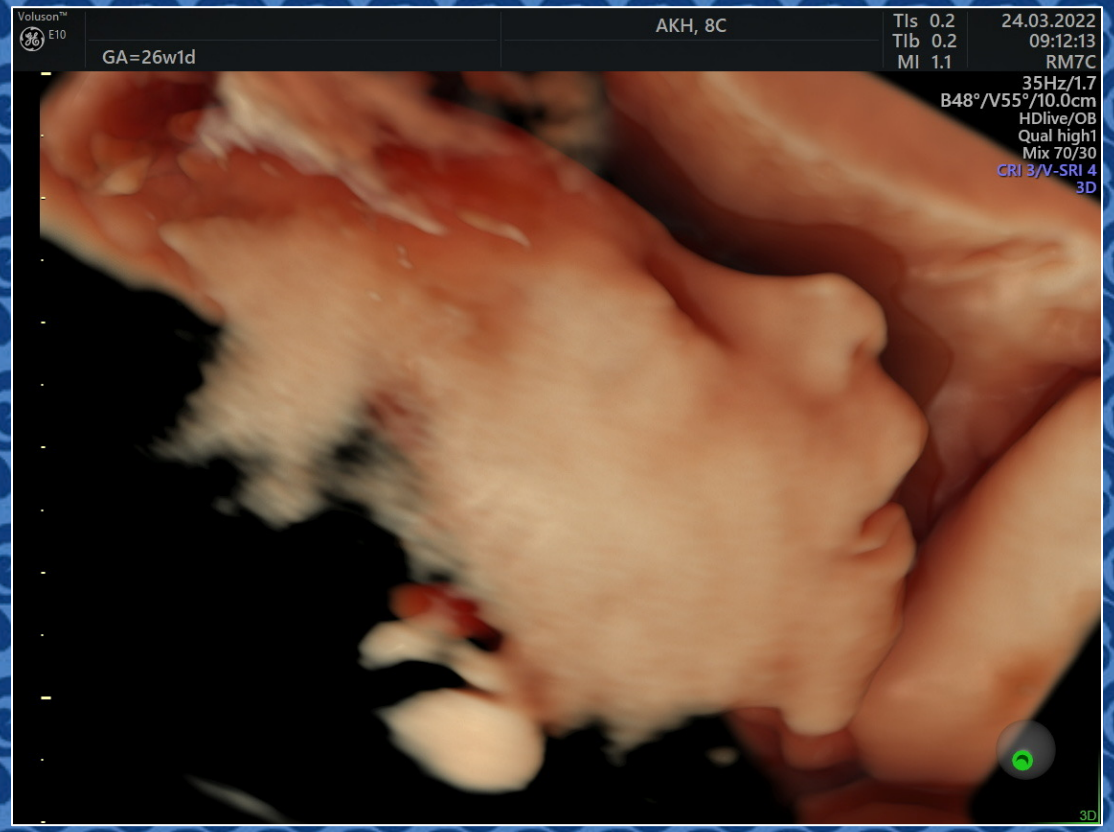


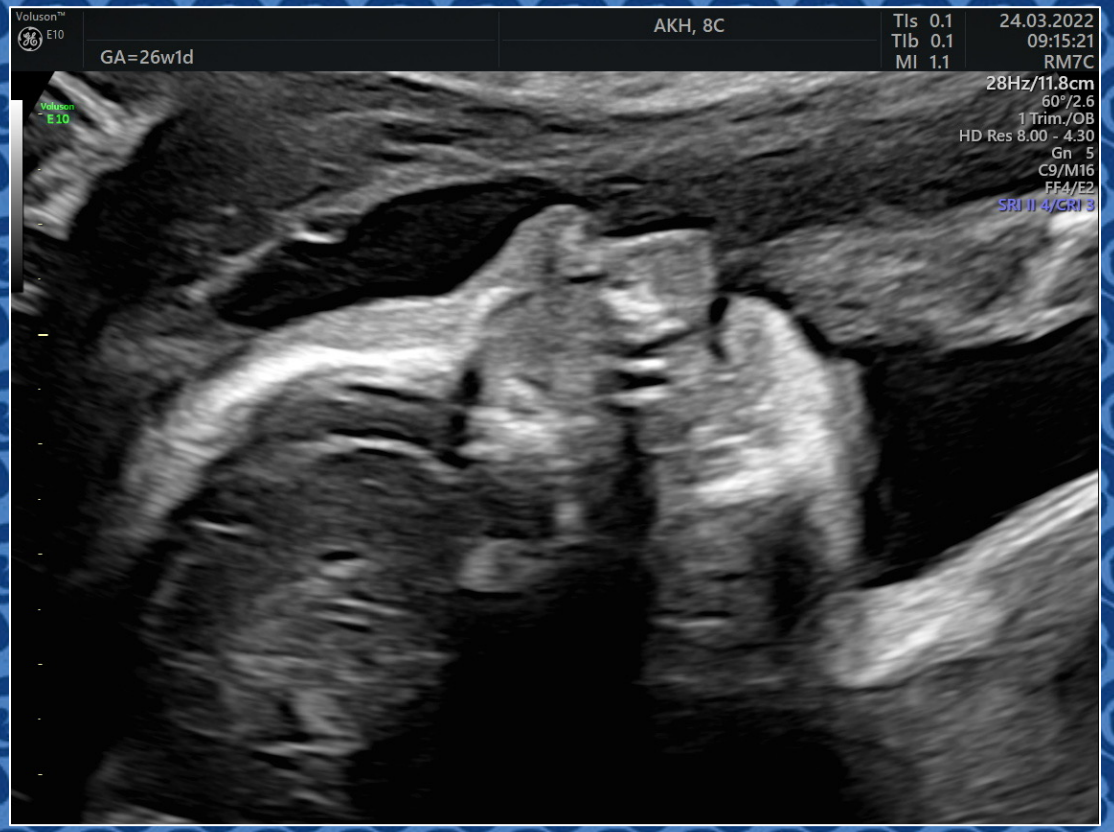




SSW 26 Flacher sulcus lateralis, Mikrozephalie, Kleinhirnhypoplasie
ösophagotracheale Fistel, Retrognathie
WES: Pathogene Veränderung am EFTUD2 Gen

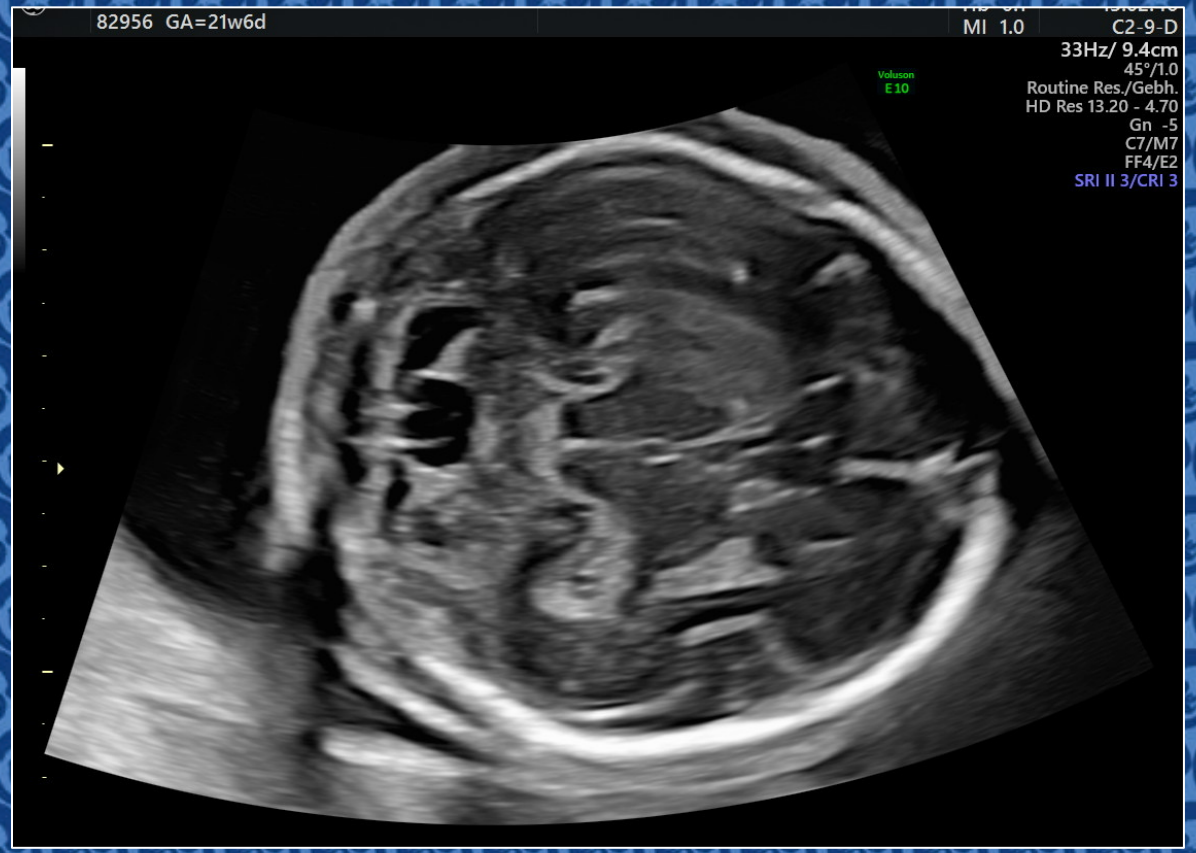
Mandibulo-faziales Dysostose-Mikrozephalie Syndrom





Hinweis auf Kleinhirnanomalien

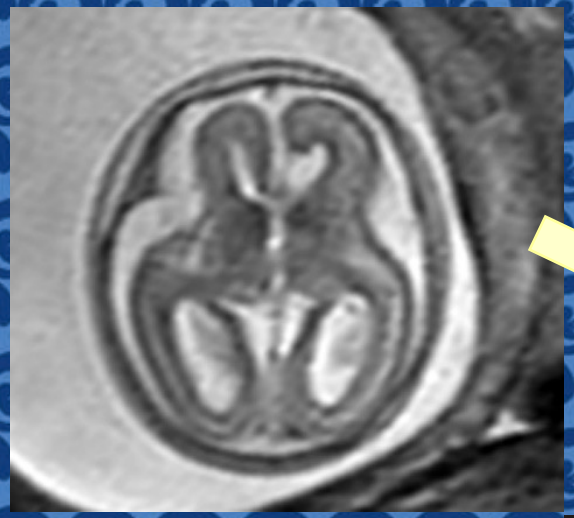
- Großer Raum hinter dem Zerebellum
- Kleiner TCD
- **Echogenität und Kontur**

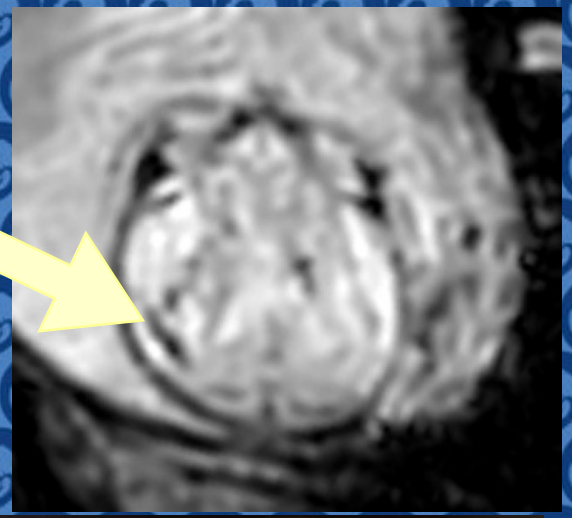


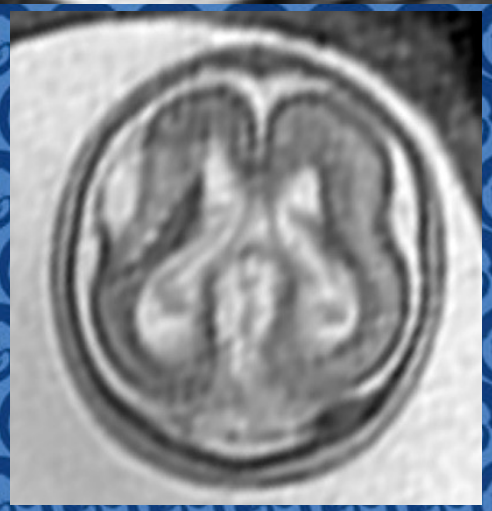
Wiederholte Gehirnblutung

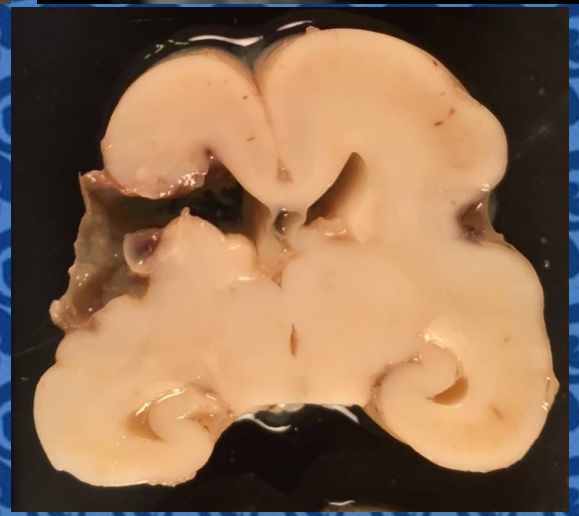
COL4A2 Mutation

1st pregnancy, 21GW

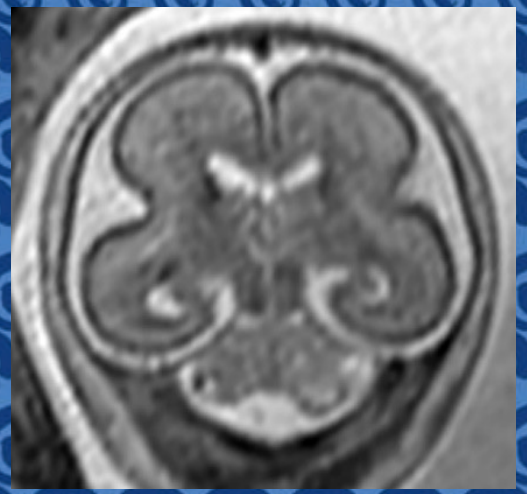


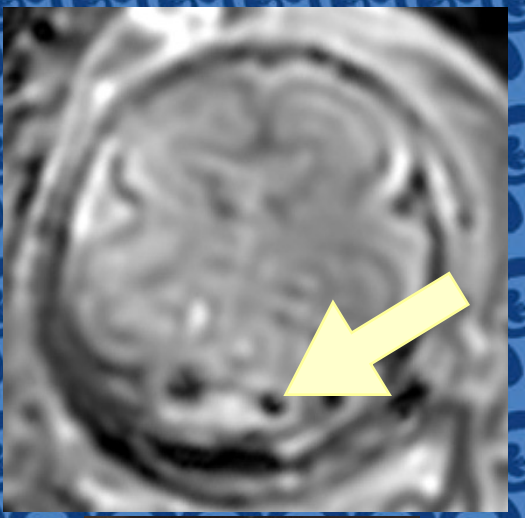


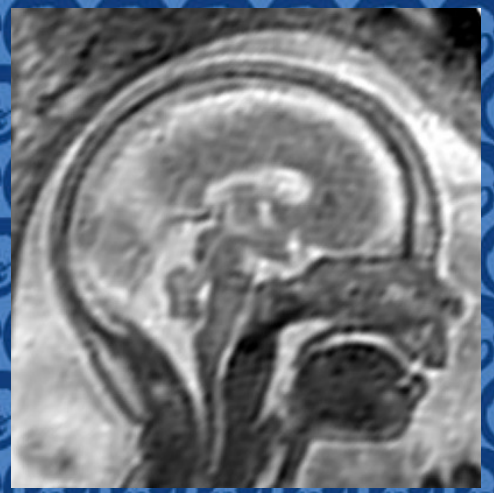


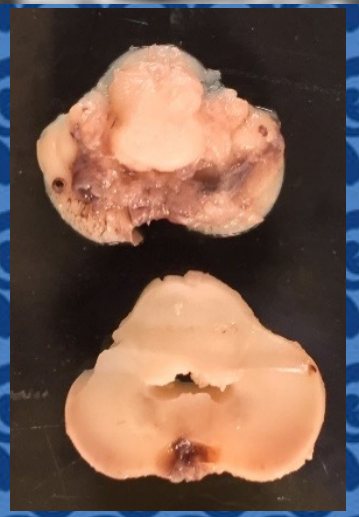


2nd pregnancy, 23GW

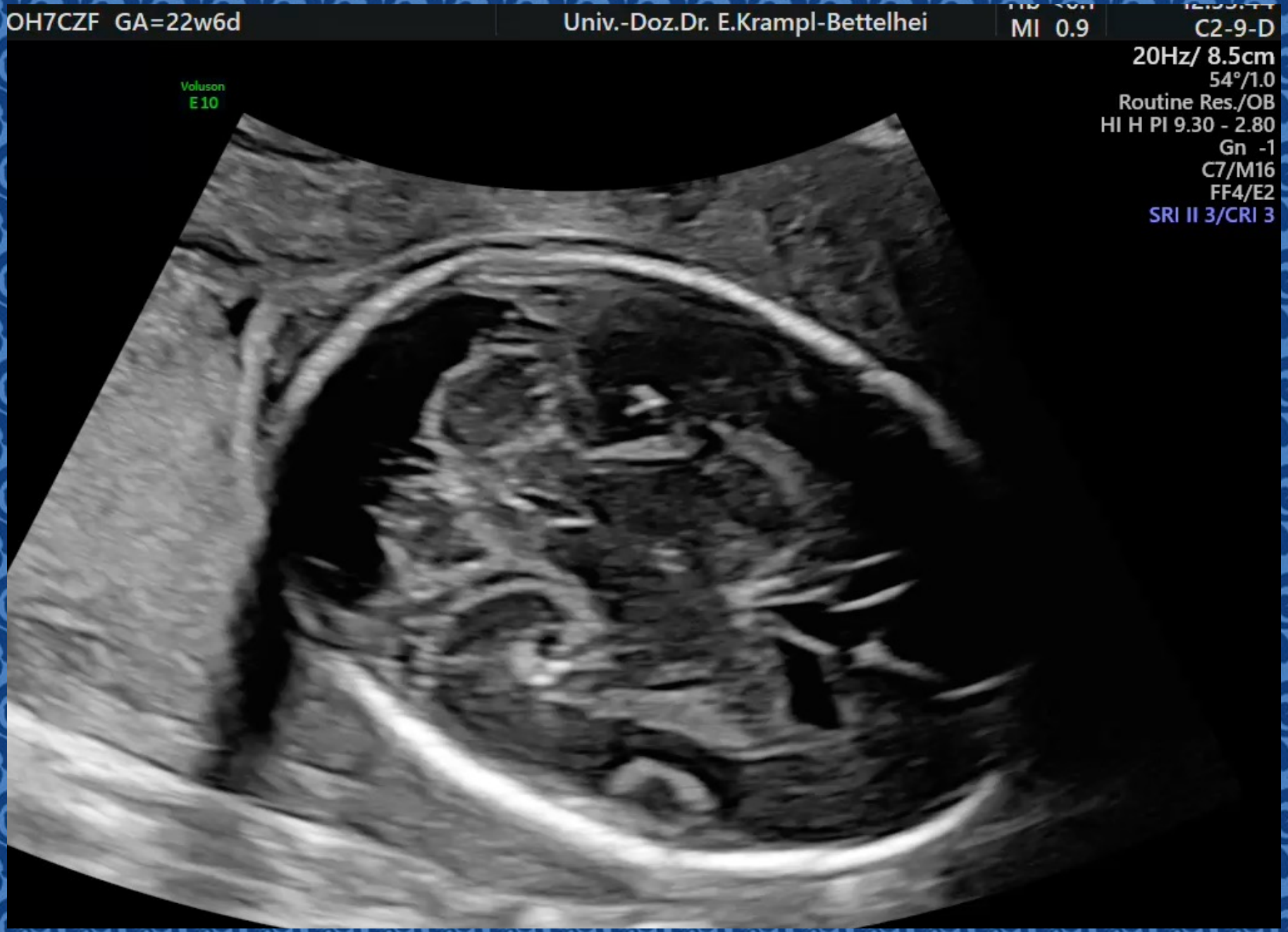




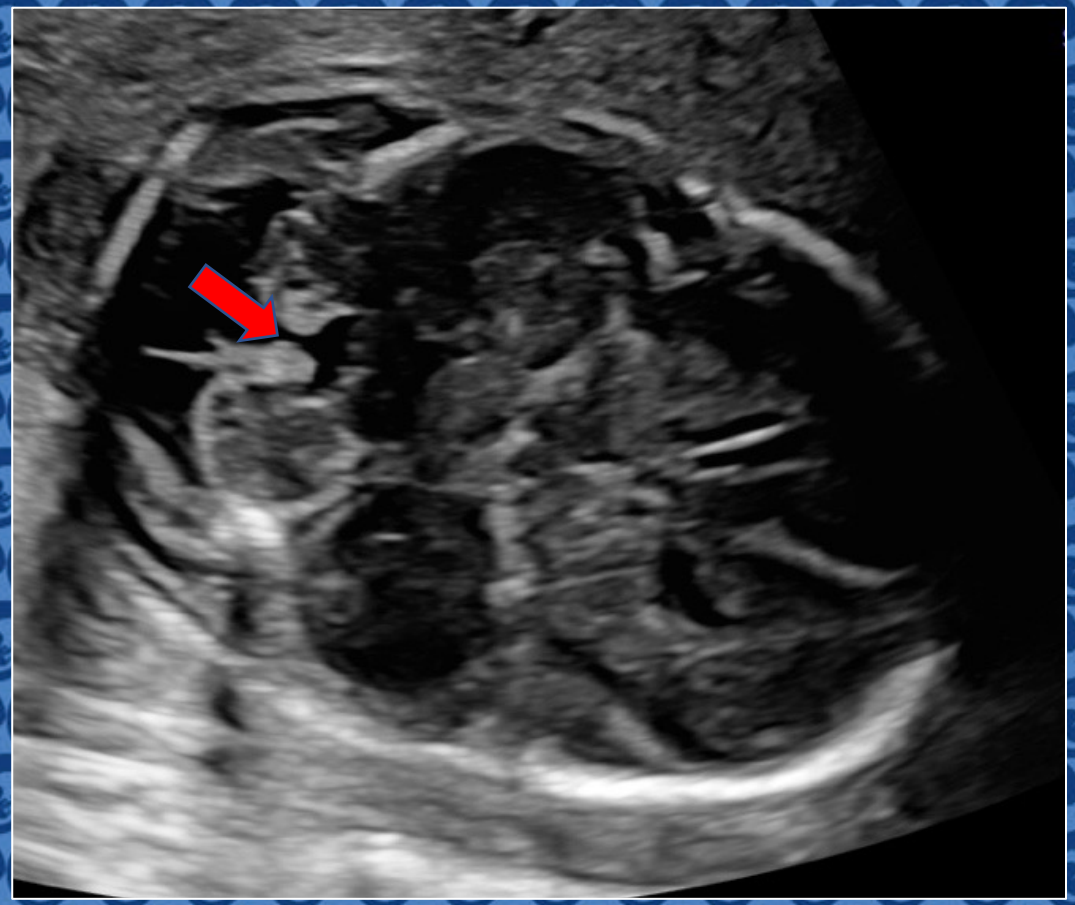


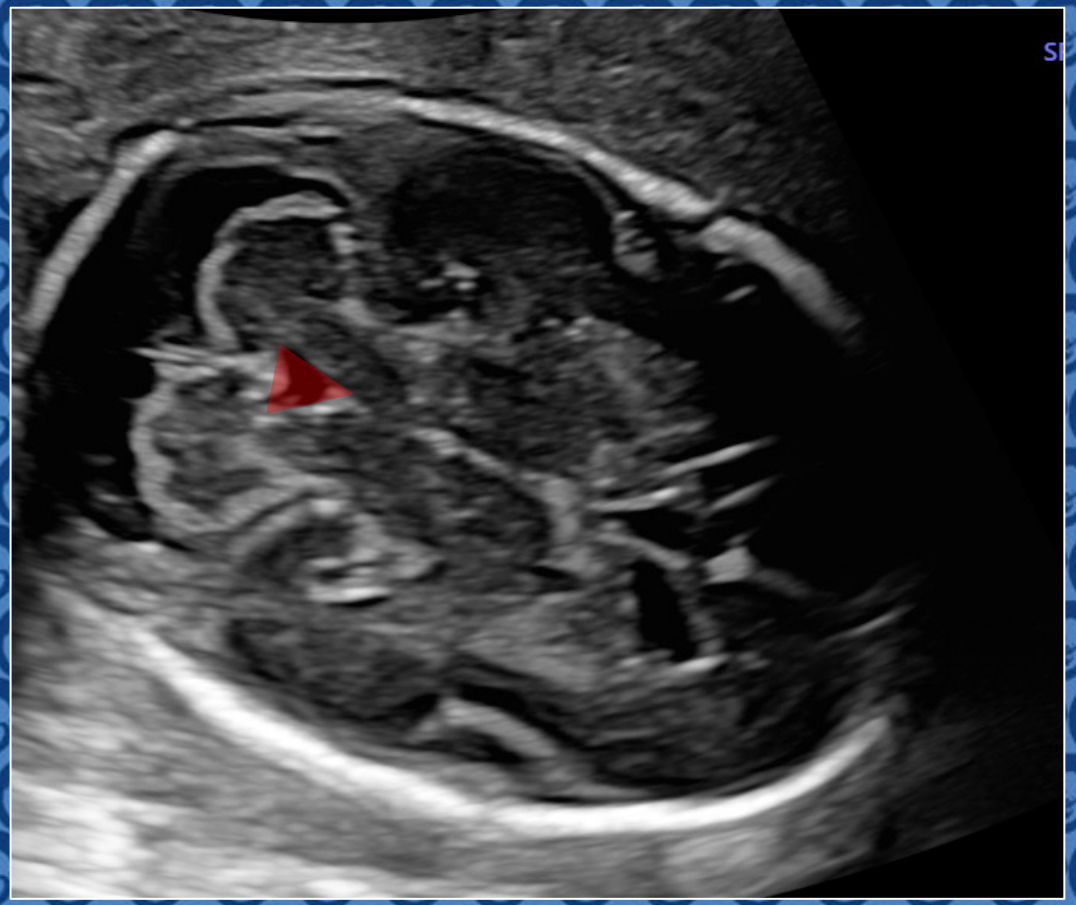


Entdeckung → Diagnose

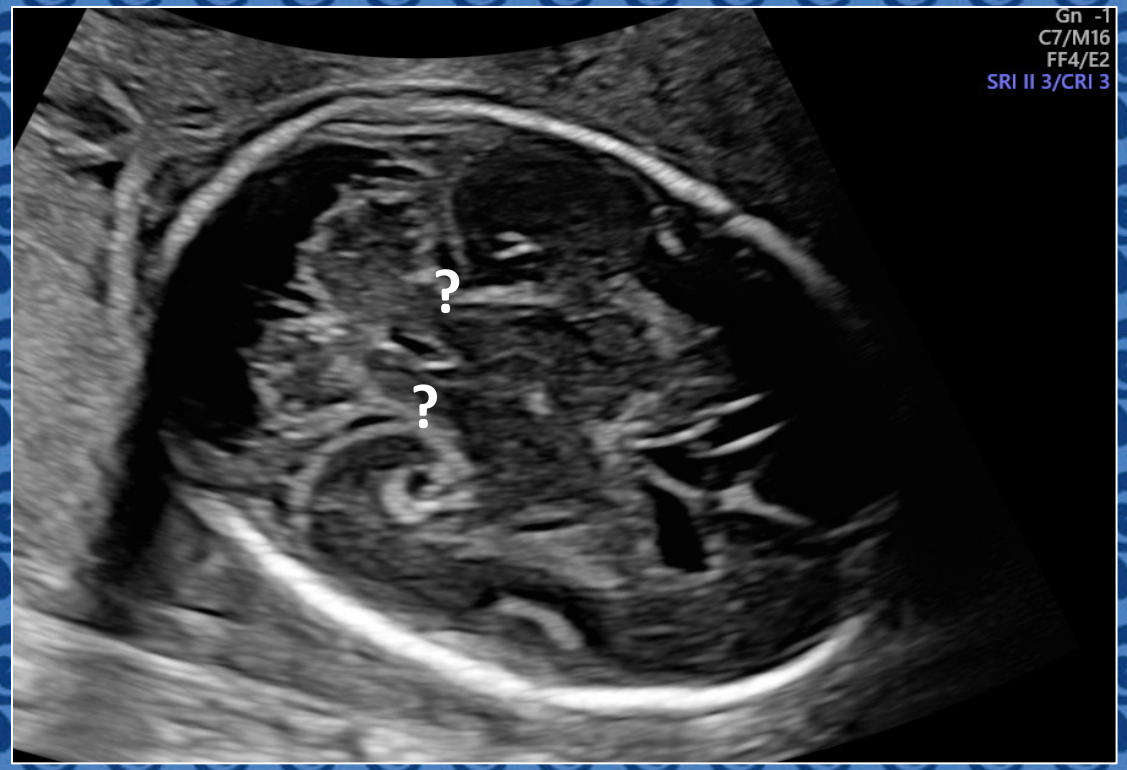

OH7CZF GA=22w6d
Univ.-Doz.Dr. E.Krampl-Bettelhei
MI 0.9
C2-9-D
20Hz/ 8.5cm
54°/1.0
Routine Res./OB
HI H PI 9.30 - 2.80
Gn -1
C7/M16
FF4/E2
SRI II 3/CRI 3
Voluson
E 10

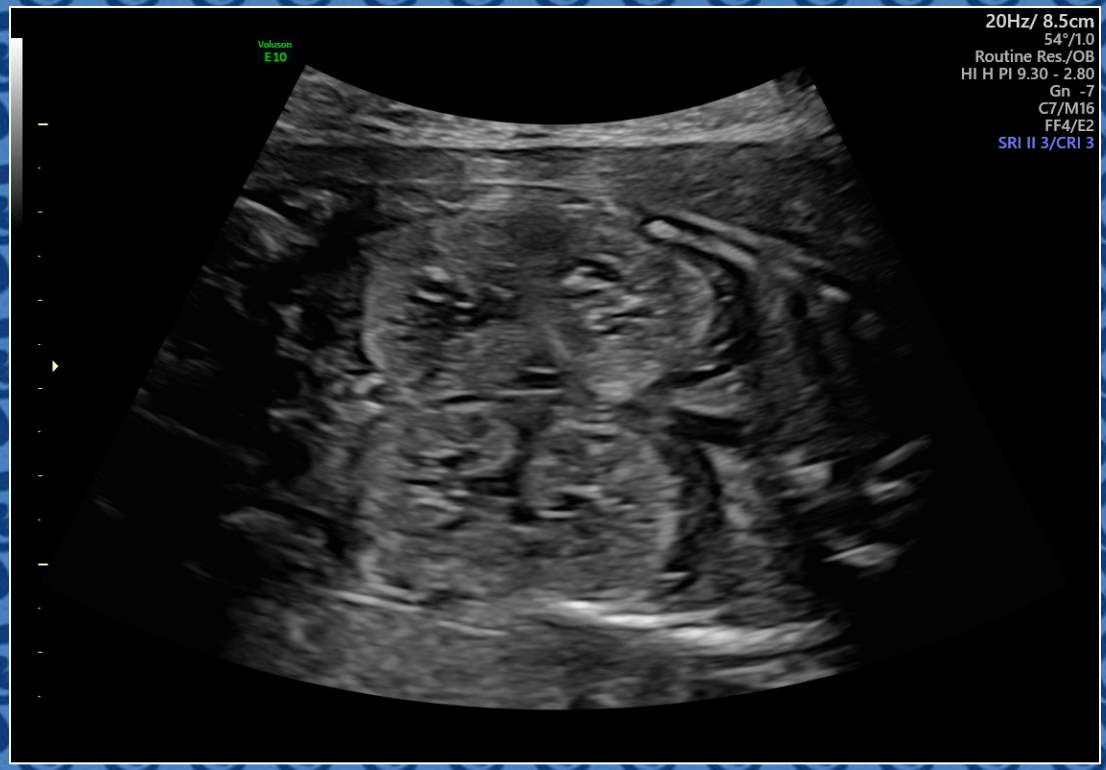
Was fällt auf? – SSW 22+6



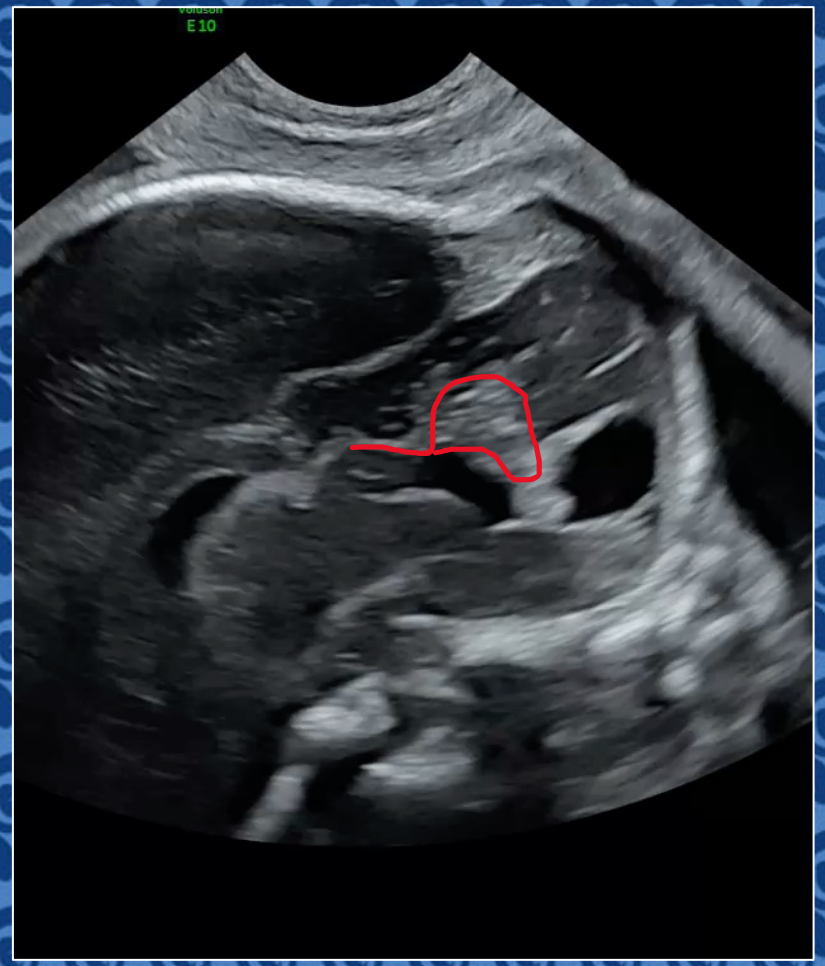


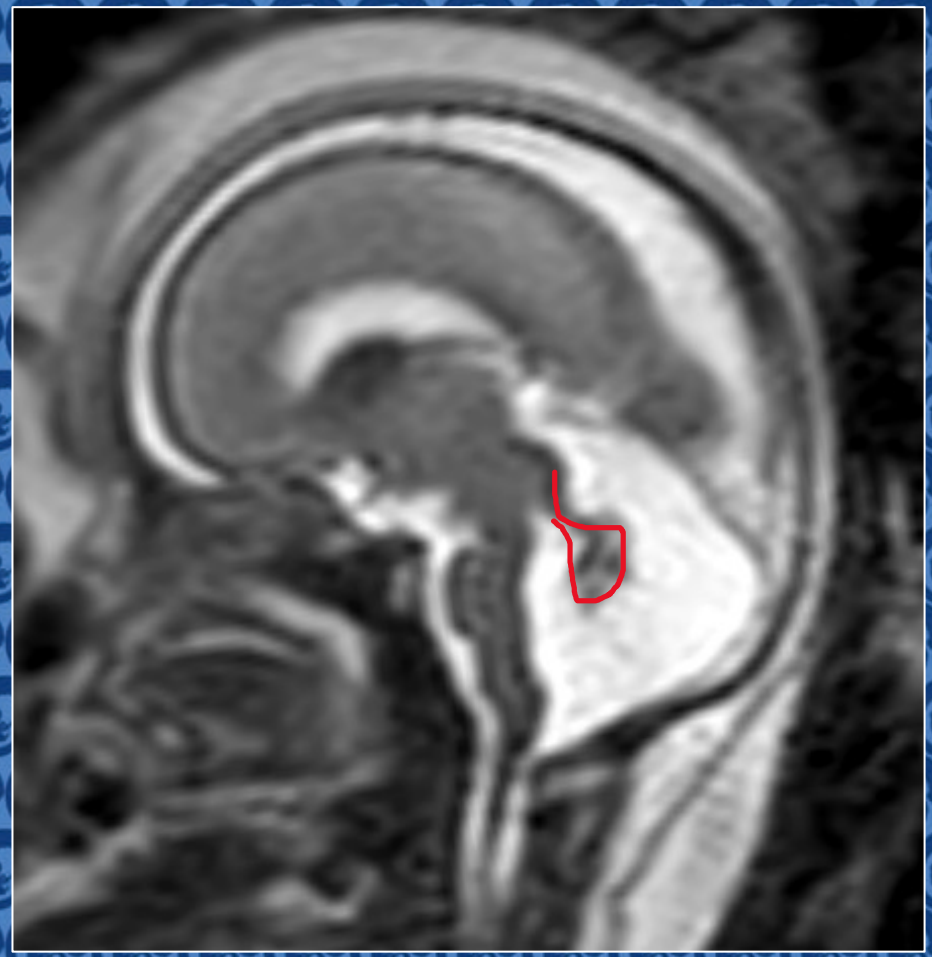
What next?

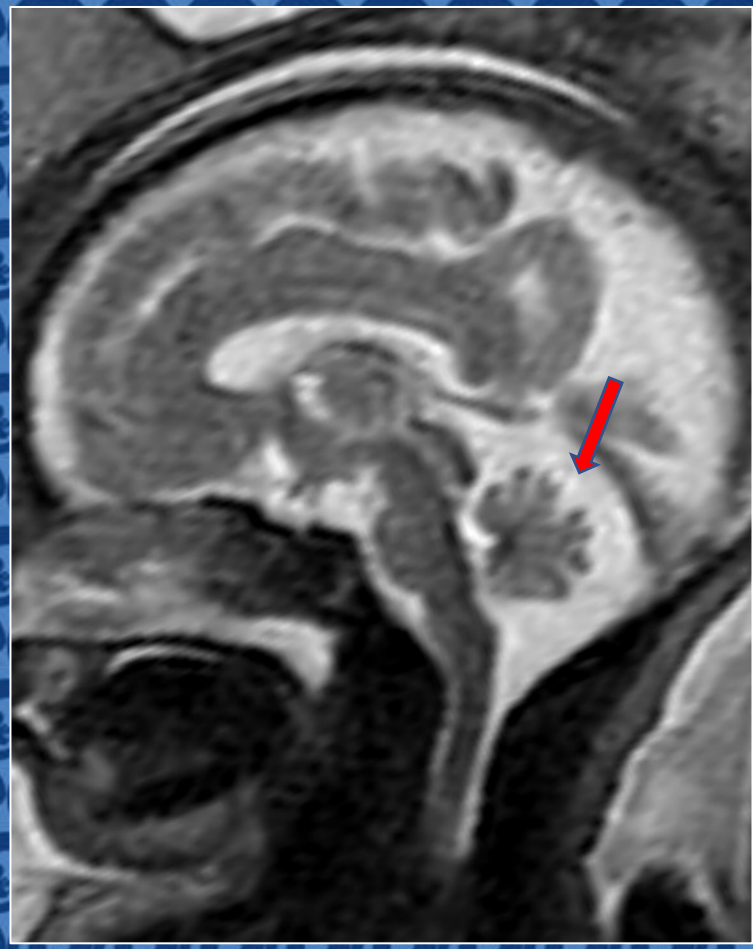

Gn -1
C7/M16
FF4/E2
SRI II 3/CRI 3
?
?


Voluson
E10
20Hz/ 8.5cm
54°/1.0
Routine Res./OB
HI H PI 9.30 - 2.80
Gn -7
C7/M16
FF4/E2
SRI II 3/CRI 3

Transvaginale Neurosonographie und MRI

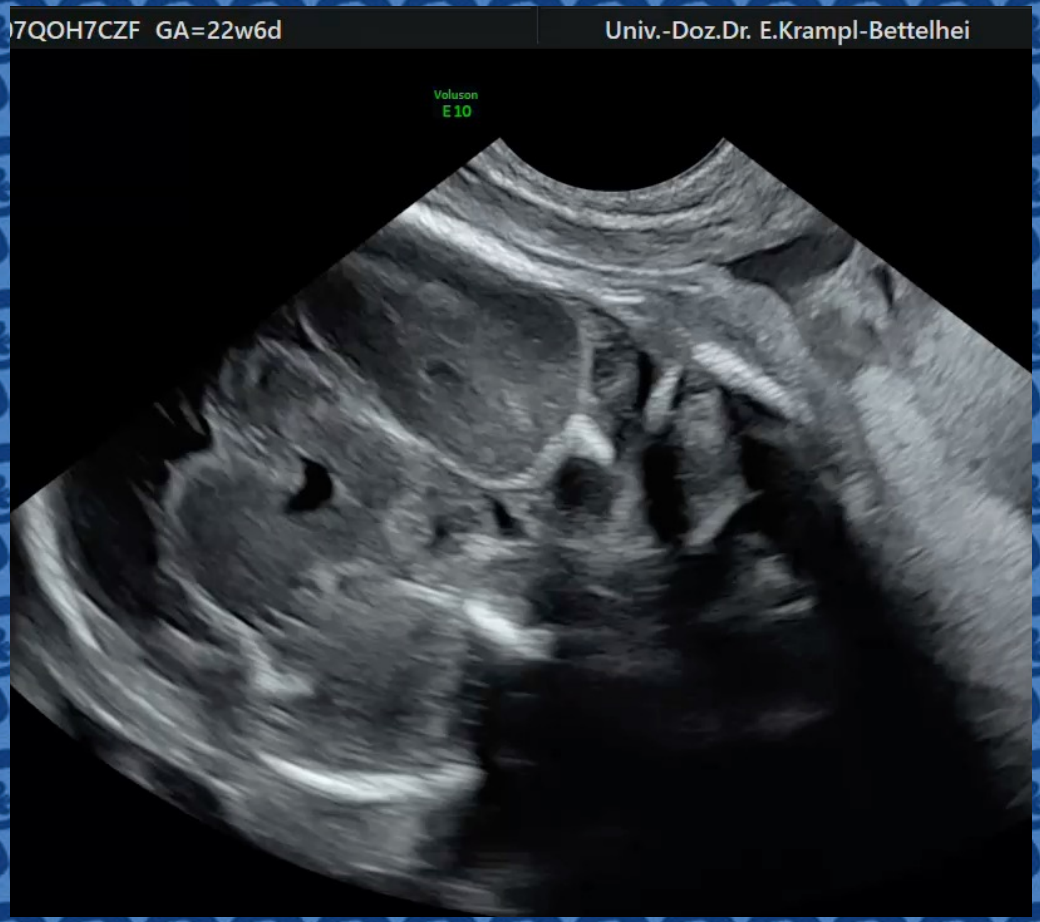




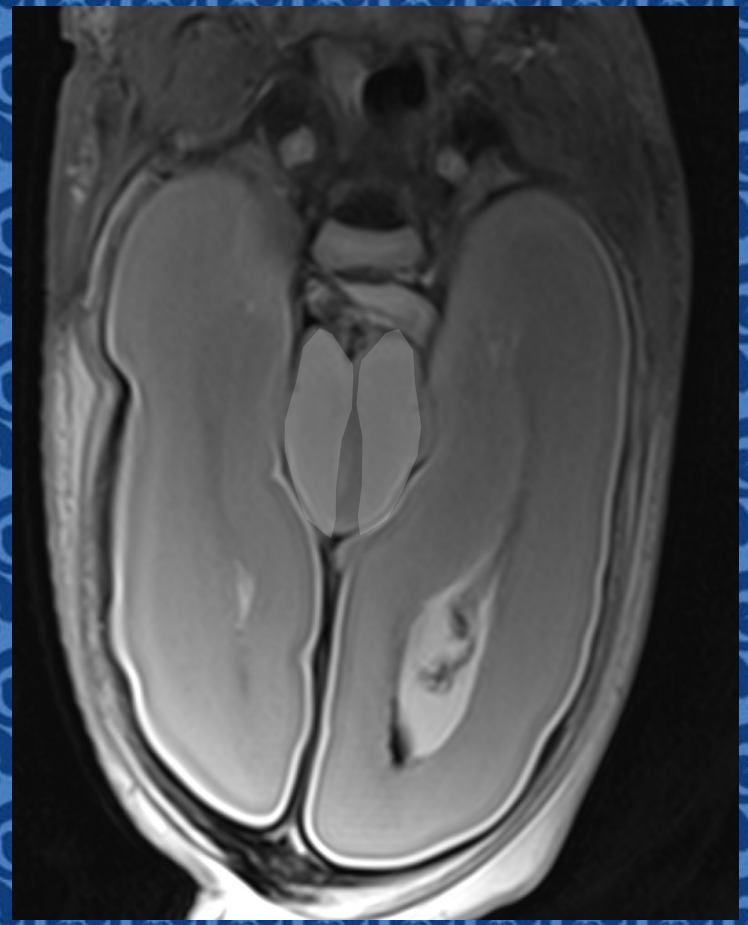


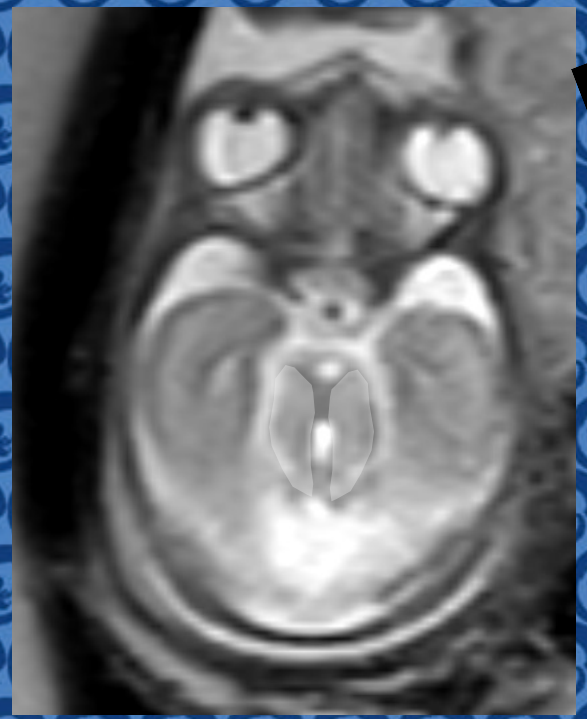
Die Lösung

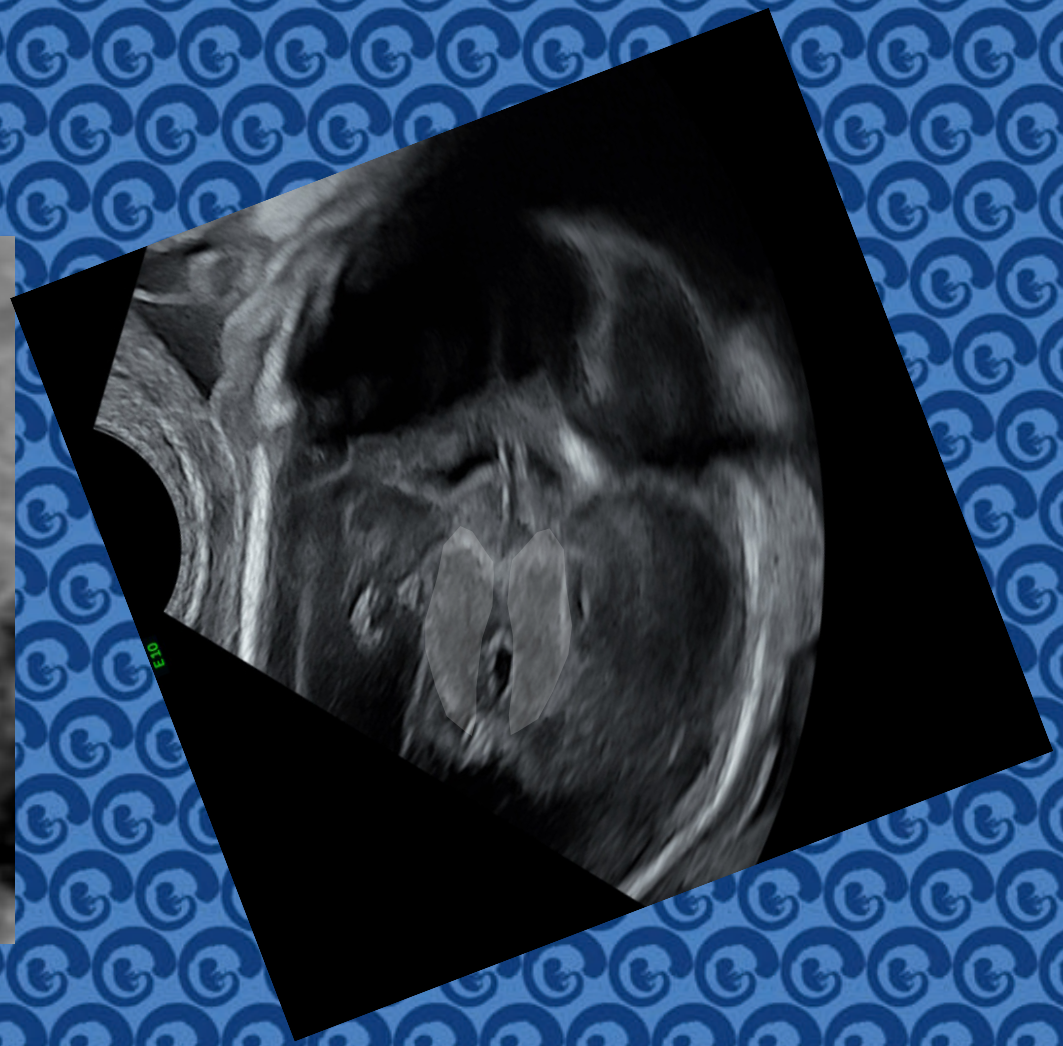
- Tiefe fossa interpeduncularis
- Verlängerte, verdickte und fehlpositionierte obere Kleinhirnstiele
- Hypoplastischer oder fehlender Kleinhirnwurm
- Molar Tooth Sign (MTS)
- Joubert Syndrom
- Trio WES *CEP290, 2013 MKS1*



Molar Tooth Sign







Post mortem MR

Joubert Syndrom

- autosomal rezessiv vererbte Erkrankung
- Vermishypoplasie (CVH cerebellar vermis hypoplasia), MTS
- Hypotonie - Ataxie
- Generelle Entwicklungseinschränkung
- Episoden von Hyperpnoe und Apnoe in der Neugeborenenperiode
- Okulomotorische Apraxie

Joubert Syndrom

- Pathogene Veränderungen in über 40 Genen sind bekannt, die als ursächlich für Joubert Syndrom gelten.
- Diese haben ihrerseits variable phänotypische Ausprägungen
- Können mit anderen Syndrome assoziiert sein
- Der pathogenetische Mechanismus ist eine Veränderung am primären Zilium = Ziliopathie
- Zellorganelle vieler Zelltypen

Prognose der Gehirnfehlbildungen

- Ultraschall
- Fetale Magentresonanztomographie
- Genetik – Whole Exome Sequencing (Trio WES)

- Der Phänotyp bestimmt die Prognose

DANKE