CERPO
Centro de Referencia Perinatal Oriente
Facultad de Medicina, Universidad de Chile

Examen del SNC en el 1° trimester y neurosonografía básica y extendida

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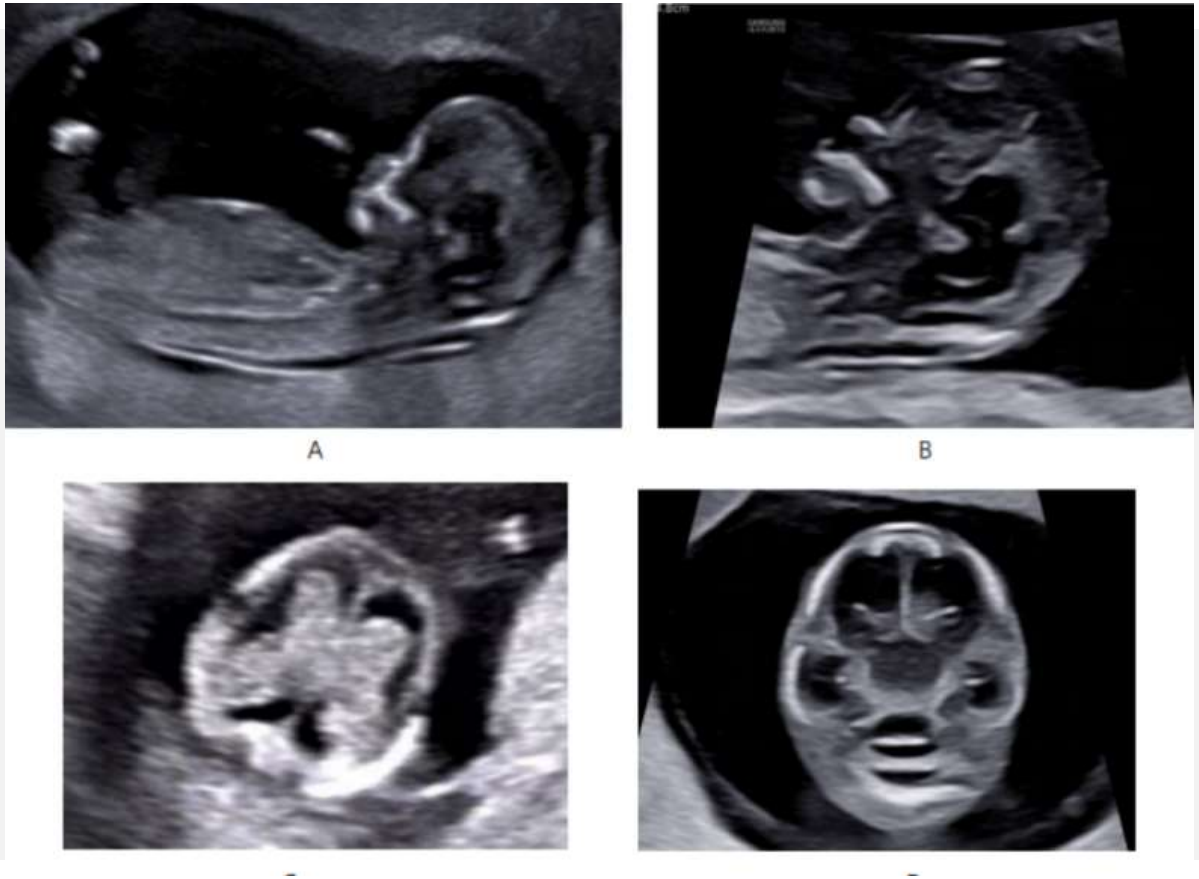
Examen del SNC en el 1° trimestre

INTRODUCCIÓN

- Las malformaciones del SNC están dentro de las más comunes 1:1000
- El desarrollo de la tecnología en ultrasonido y la evaluación temprana de la anatomía han permitido el diagnóstico temprano de anomalías del SNC.



Morris, J. K., Wellesley, D. G., Barisic, I., Addor, M. C., Bergman, J. E. H., Braz, P., Cavero-Carbonell, C., Draper, E. S., Gatt, M., Haeusler, M., Klungsoyr, K., Kurinczuk, J. J., Lelong, N., Luyt, K., Lynch, C., O'Mahony, M. T., Mokoroa, O., Nelen, V., Neville, A. J., Garne, E. (2019). Epidemiology of congenital cerebral anomalies in Europe: A multicentre, population-based EUROCAT study. *Archives of Disease in Childhood*, *104*(12), 1181–1187.

PREVALENCIA

Table 1 Number of cases with a congenital cerebral anomaly (ICD-10 code Q04), prevalence and proportion prenatally diagnosed in 29 EUROCAT registries in the period 2005–2014

Registry, country	Years of data	Population births (1000)	Total cases	Prevalence per 10 000 births	Proportion prenatally diagnosed (%)
South Portugal	2006–2014	161	44	2.7	68
South East Ireland	2005–2014	75	27	3.6	59
Zagreb, Croatia	2005–2014	58	24	4.1	71
Wessex, UK	2005–2014	298	156	5.2	86
East Midlands and South Yorkshire, UK	2005–2012	587	311	5.3	83
Tuscany, Italy	2005–2014	300	167	5.6	81
Norway	2005–2012	487	273	5.6	61
Malta	2005–2014	41	25	6.1	36
Cork and Kerry, Ireland	2005–2014	99	60	6.1	48
Hainaut, Belgium	2005–2014	126	85	6.7	72
Emilia Romagna, Italy	2005–2014	400	276	6.9	70
Valencia Region, Spain	2005–2014	403	278	6.9	59
Northern England, UK	2005–2014	331	231	7.0	71
Mainz, Germany	2005–2014	32	23	7.2	78
Ukraine	2005–2014	304	219	7.2	60
Thames Valley, UK	2005–2014	300	221	7.4	77
Northern Netherlands	2005–2014	174	130	7.5	62
Odense, Denmark	2005–2013	41	34	8.4	65
Wales, UK	2005–2014	347	305	8.8	64
Saxony-Anhalt, Germany	2005–2014	172	153	8.9	50
Antwerp, Belgium	2005–2014	206	185	9.0	56
Styria, Austria	2005–2012	83	77	9.3	75
Basque Country, Spain	2007–2014	205	201	9.8	77
South West England, UK	2005–2014	496	491	9.9	51
Isle de Reunion, France	2005–2014	146	176	12.1	78
Brittany, France	2011–2014	145	182	12.5	89
Vaud, Switzerland	2005–2014	79	122	15.4	73
Paris, France	2005–2012	214	352	16.5	94
French West Indies, France	2009–2014	60	99	16.6	92
Total	2005–2014	6368	4927	9.78 (95% CI: 8.50 to 11.16)*	70

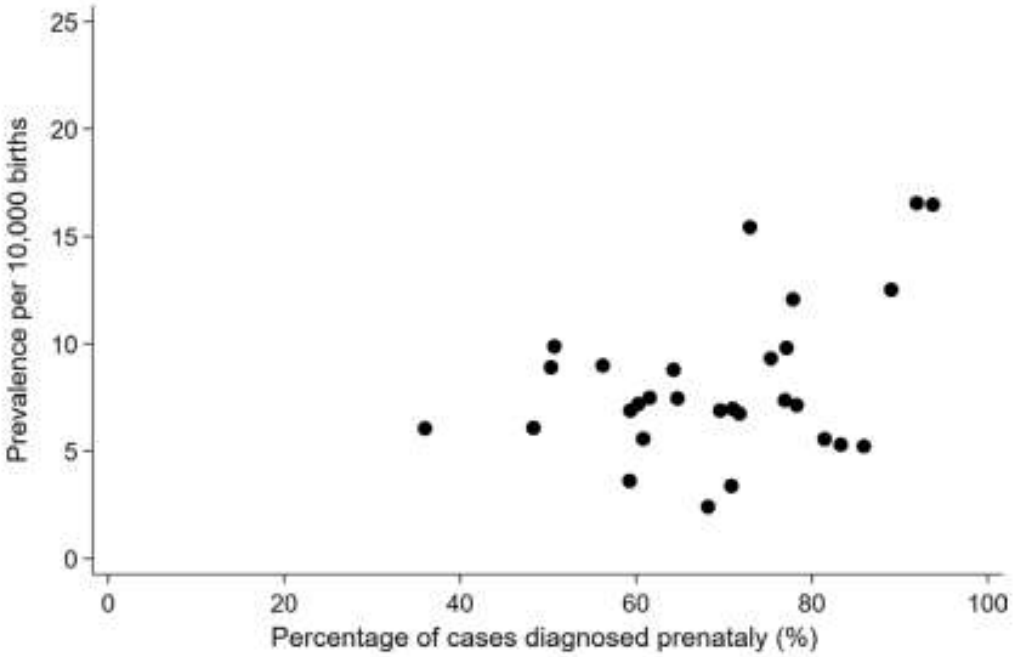


Figure 2 Association between the prevalence of cerebral anomalies (Q04) in EUROCAT registries and the percentage of cases diagnosed prenatally.

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Table 2 Epidemiology data for congenital cerebral anomaly cases in 29 EUROCAT registries from 2005 to 2014 with cerebral anomaly cases classified according to one main cerebral anomaly category with the diagnoses present on the left taking precedence over the ones on the right

ICD-10 code	Q04.2	Q04.1	Q04.4	Q04.5	Q04.3	Q04.0	Q04.8	Q04.6	Q04.9	Q04
Anomaly	Holoprosencephaly	Arhinencephaly	Septo-optic dysplasia	Megalencephaly	Other reduction deformities of brain	Congenital malformations of corpus callosum	Other specified congenital malformations of brain	Congenital cerebral cysts	Congenital malformation of brain, unspecified	All cases
No of cases	865	33	94	49	1409	1476	383	375	243	4927
No of diagnoses*	865	46	99	49	1464	1748	550	555	273	5649
Prevalence per 10 000 births (95% CI)†	1.55 (1.37 to 1.77)	0.04 (0.01 to 0.07)	0.19 (0.11 to 0.26)	0.08 (0.05 to 0.11)	2.92 (2.51 to 3.35)	3.25 (2.72 to 3.82)	0.75 (0.53 to 1.01)	0.69 (0.49 to 0.93)	0.39 (0.29 to 0.52)	9.78 (8.5 to 11.16)
Live births, n (%)	155 (18)	2 (6)	90 (96)	33 (67)	792 (56)	975 (66)	259 (68)	302 (81)	112 (46)	2720 (55)
Fetal deaths, n (%)	34 (4)	1 (3)	0 (0)	1 (2)	55 (4)	37 (3)	12 (3)	6 (2)	18 (7)	164 (3)
TOPFA, n (%)	676 (78)	30 (91)	4 (4)	15 (31)	562 (40)	464 (31)	112 (29)	67 (18)	113 (47)	2043 (41)
Non-genetic, n (%)	539 (62)	12 (36)	91 (97)	43 (88)	1119 (79)	1156 (78)	307 (80)	325 (87)	192 (79)	3784 (77)
Average maternal age (years)	30 (30–31)	32 (30–34)	23 (22–24)	30 (28–32)	30 (30–30)	30 (30–31)	30 (30–31)	29 (28–30)	29 (29–30)	30 (30–30)
Preterm birth (GA <37 weeks) live births, n (%)	62 (40)	1 (50)	17 (19)	5 (17)	229 (29)	208 (22)	79 (31)	113 (38)	34 (32)	748 (28)
Prenatal diagnosis, n (%)	811 (94)	32 (97)	32 (34)	27 (55)	962 (68)	1038 (70)	207 (54)	182 (49)	157 (65)	3448 (70)
Males, n (%)	316 (37)	14 (42)	52 (55)	29 (59)	684 (49)	749 (51)	198 (52)	196 (52)	114 (47)	2352 (48)

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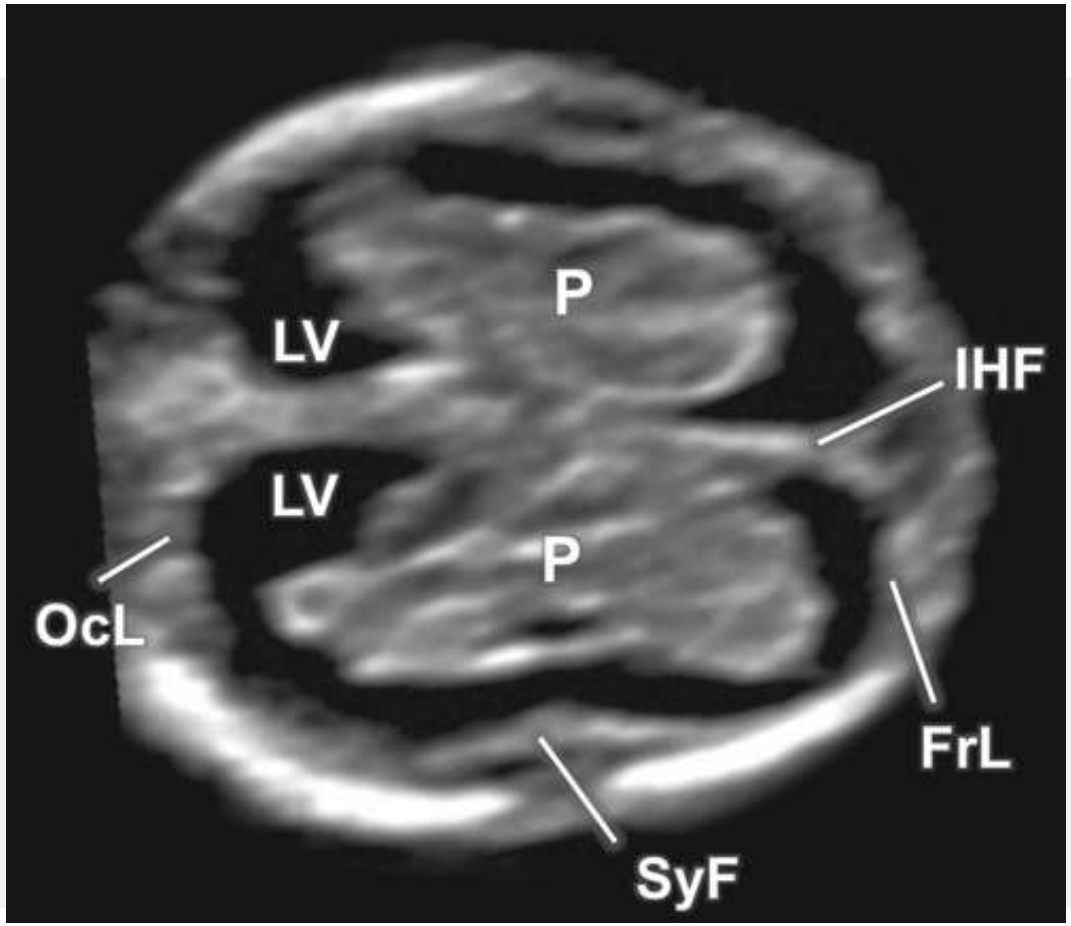
ANATOMÍA NORMAL EN EL 1° TRIMESTRE

- Idealmente debe realizarse con un transductor transvaginal.
- Evaluar cortes axiales transventricular y transtalámicos
- Evaluar corte mediosagital
- Reconstrucción 3D de plano coronal a partir de plano axial

Paladini D, Malinger G, Birnbaum R, Monteagudo A, Pilu G, Salomon LJ, Timor-Tritsch IE. ISUOG Practice Guidelines (updated): sonographic examination of the fetal central nervous system. Part 2: performance of targeted neurosonography. Ultrasound Obstet Gynecol 2021

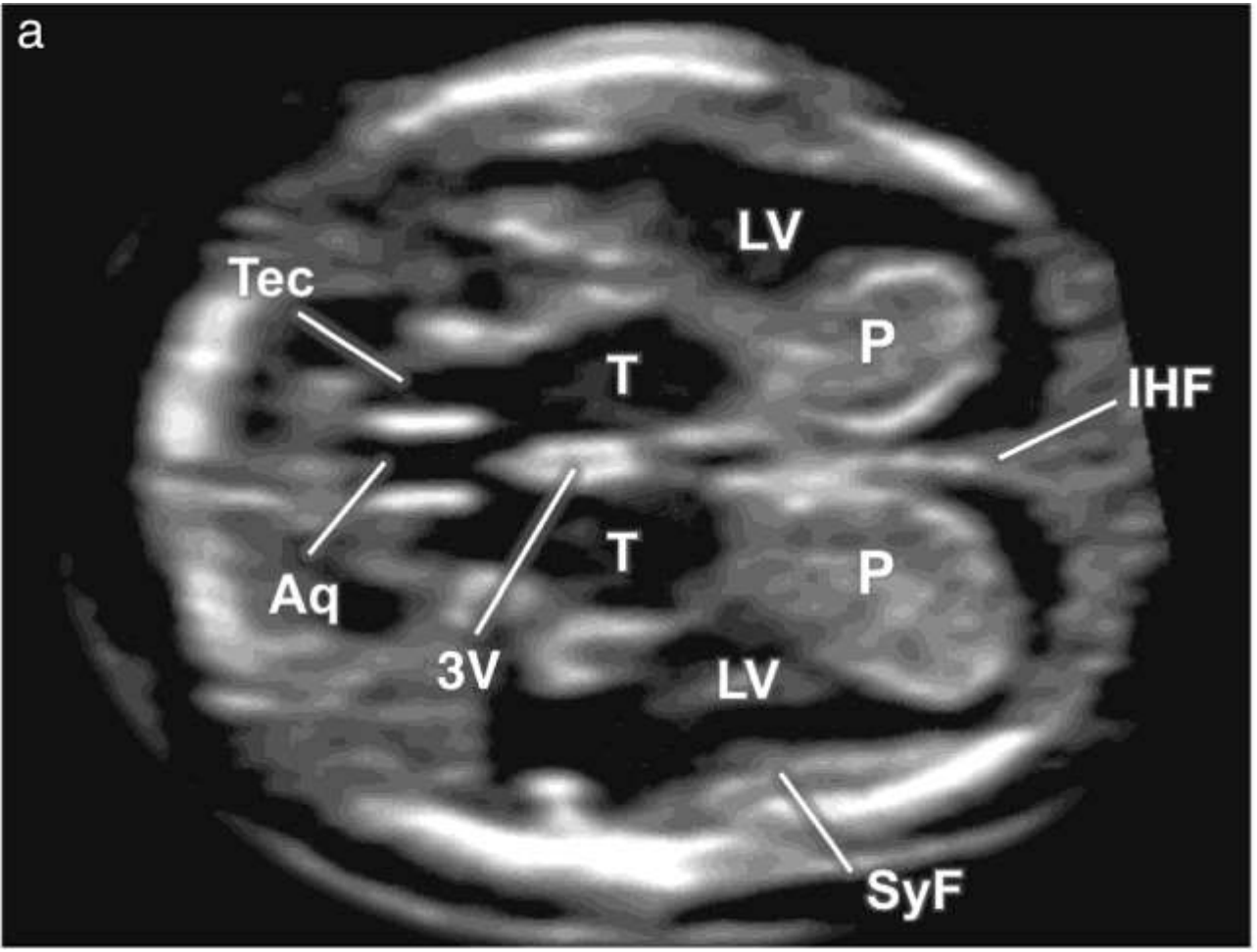
PLANO AXIAL

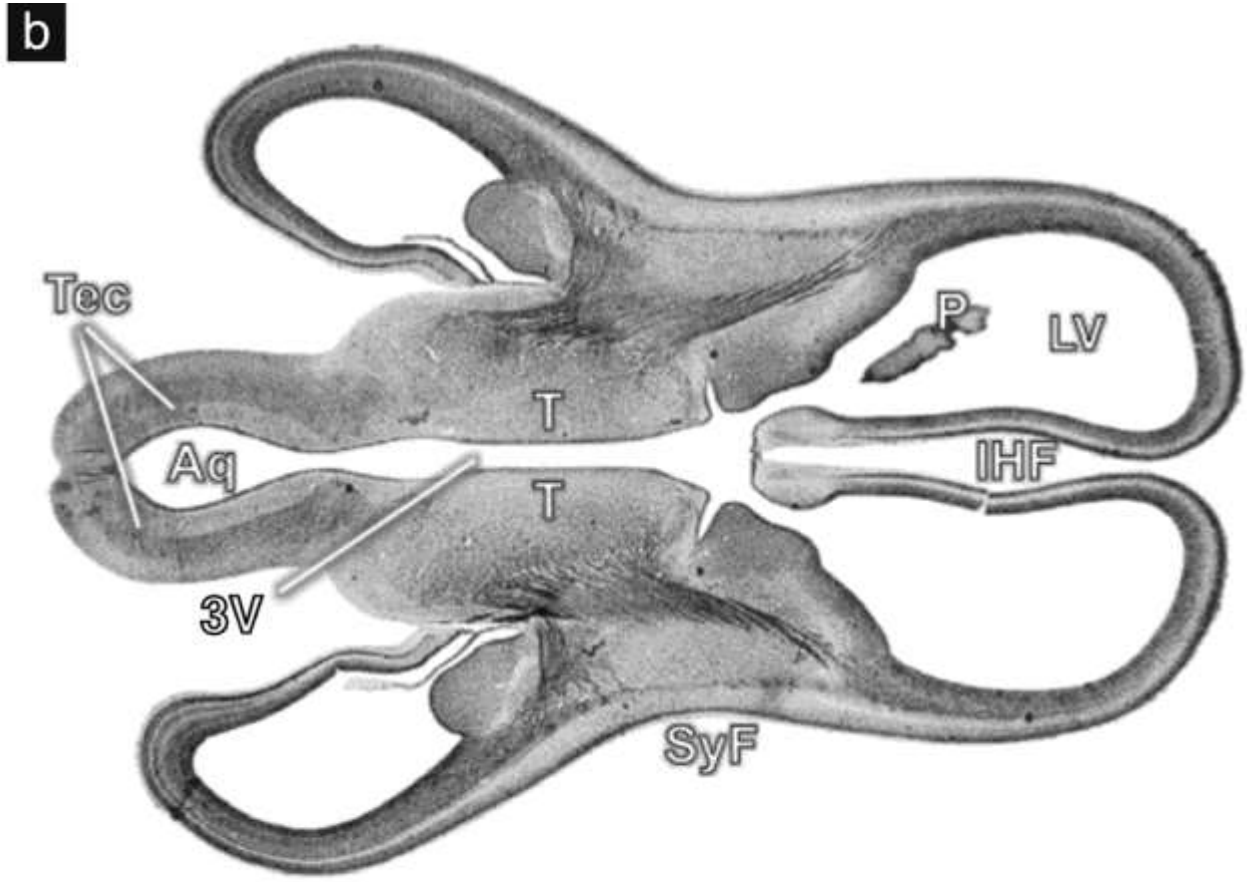


- Forma del cráneo, osificación de la calota.
- Eco medio como línea recta ininterrumpida
- Ventrículos laterales ocupados por los plexos coroideos
- Ausencia de cisuras o giros, con excepción del receso medio-lateral que dará origen a la cisura de Silvio

Volpe, N., Dall'Asta, A., di Pasquo, E., Frusca, T., & Ghi, T. (2021). First-trimester fetal neurosonography: technique and diagnostic potential. *Ultrasound in Obstetrics & Gynecology*, *57*(2), 204–214. https://doi.org/10.1002/UOG.23149

PLANO TRANSTALÁMICO





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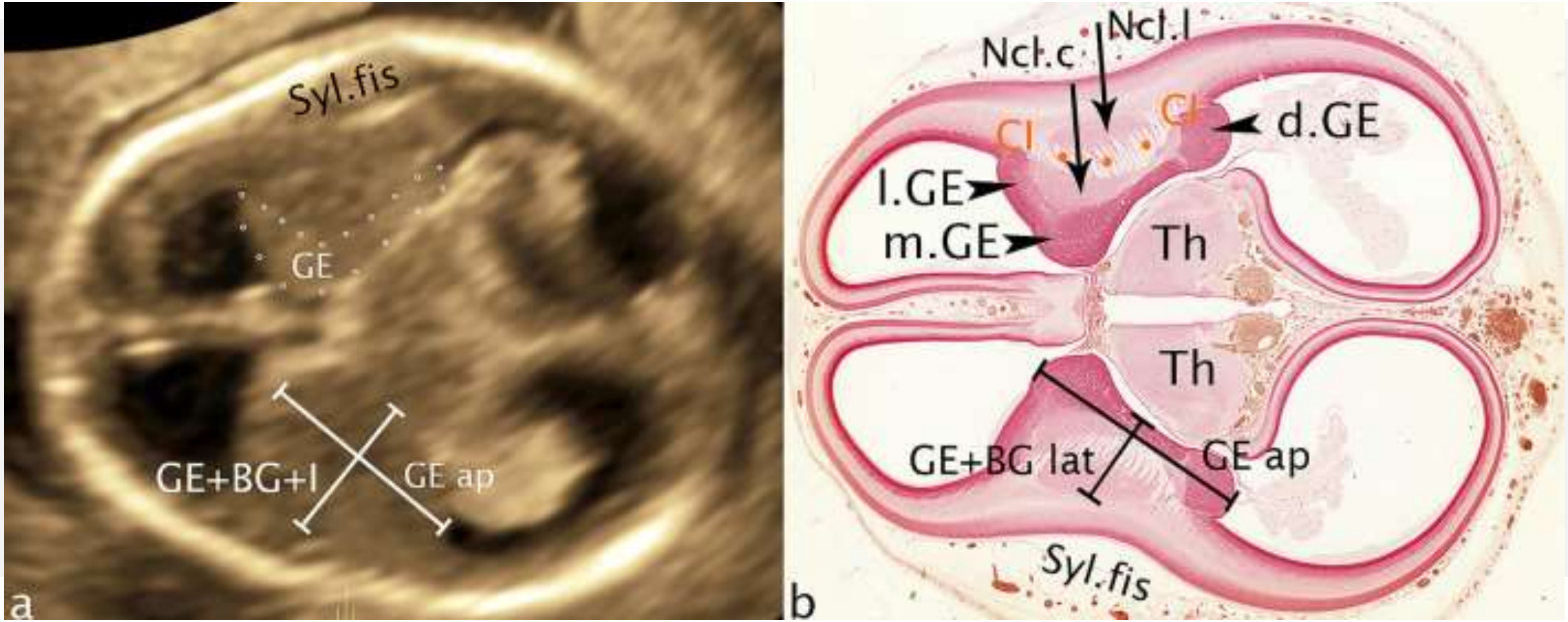
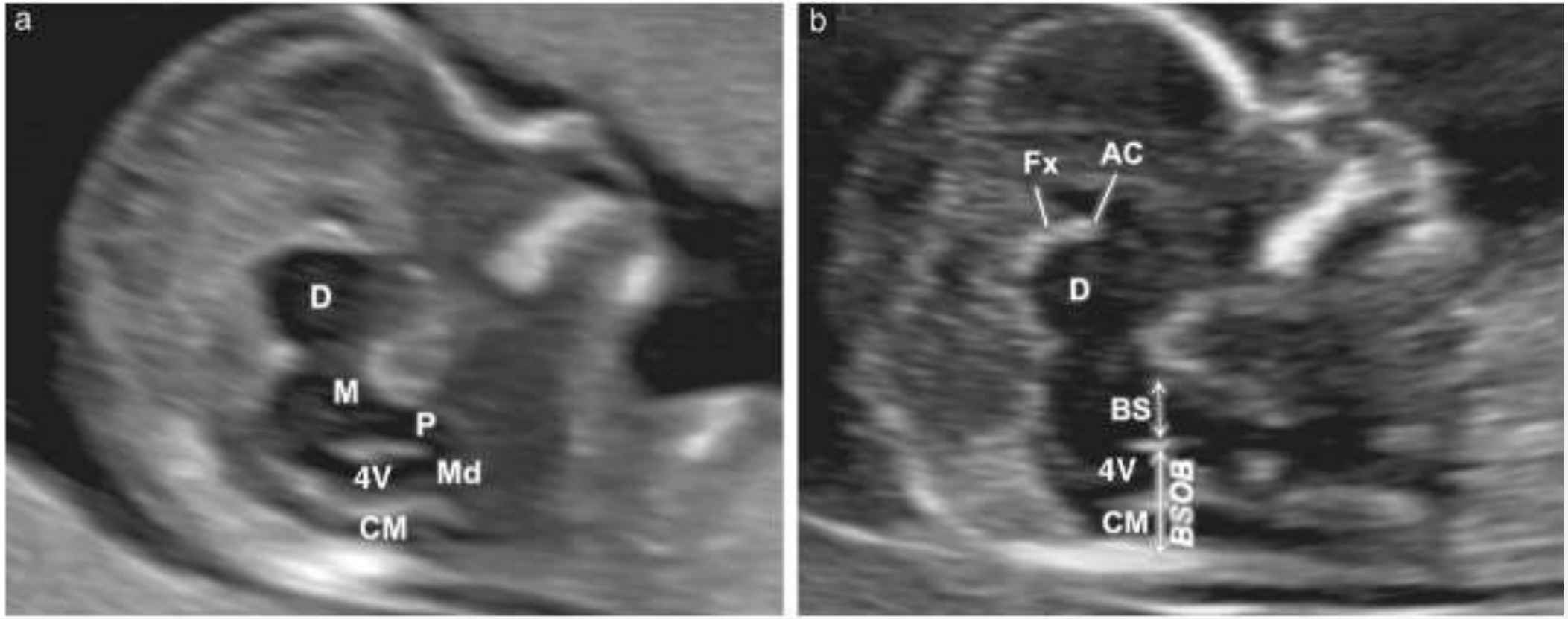
PLANO TRANSTALÁMICO



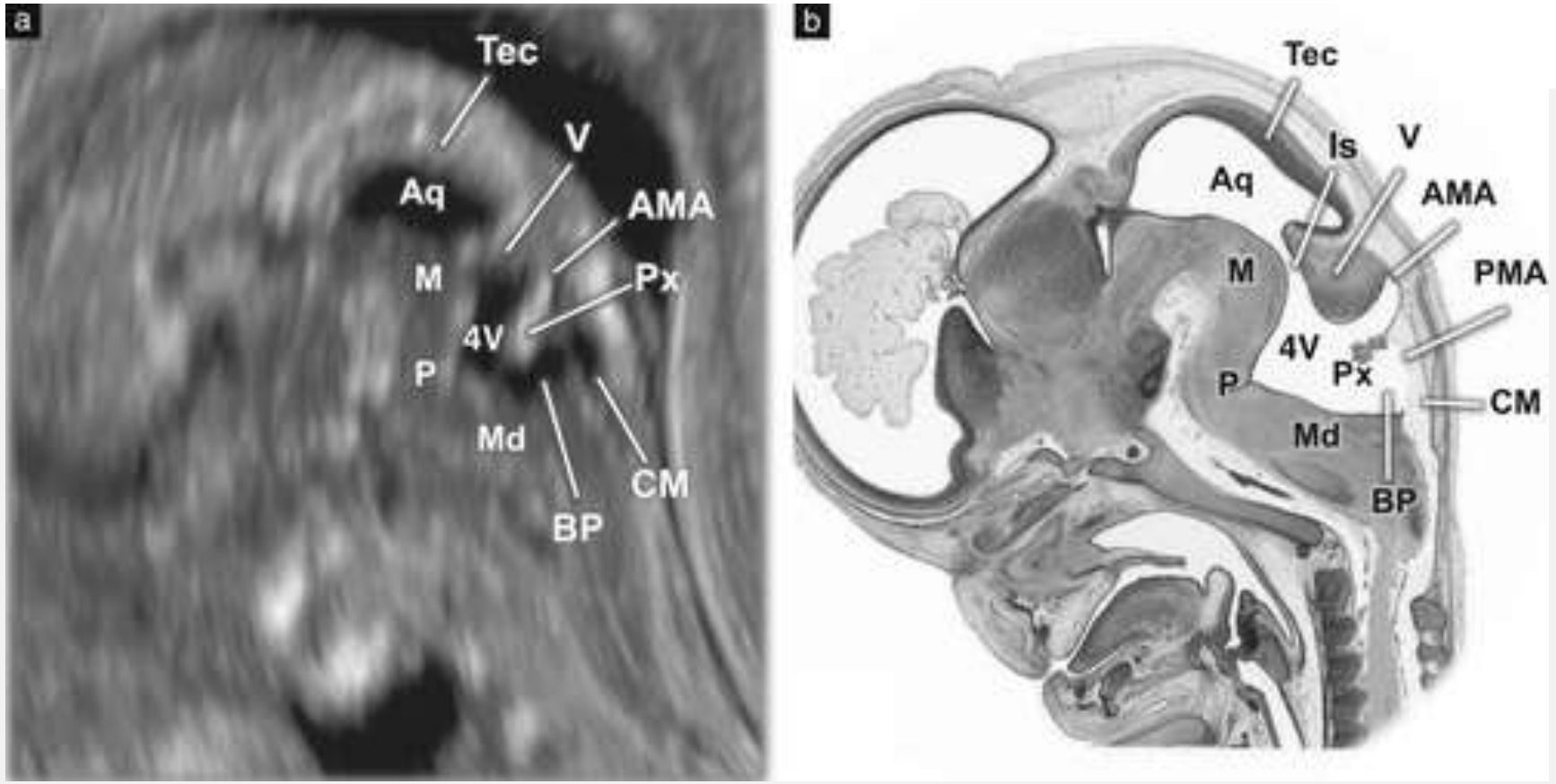
Altmann, R., Rechberger, T., Altmann, C., Hirtler, L., Scharnreitner, I., Stelzl, P., & Enengl, S. (2023). Development of the prosencephalic structures, ganglionic eminence, basal ganglia and thalamus at 11 + 3 to 13 + 6 gestational weeks on 3D transvaginal ultrasound including normative data. *Brain Structure & Function*, *228*(9), 2089. https://doi.org/10.1007/S00429-023-02679-Y

PLANO SAGITAL



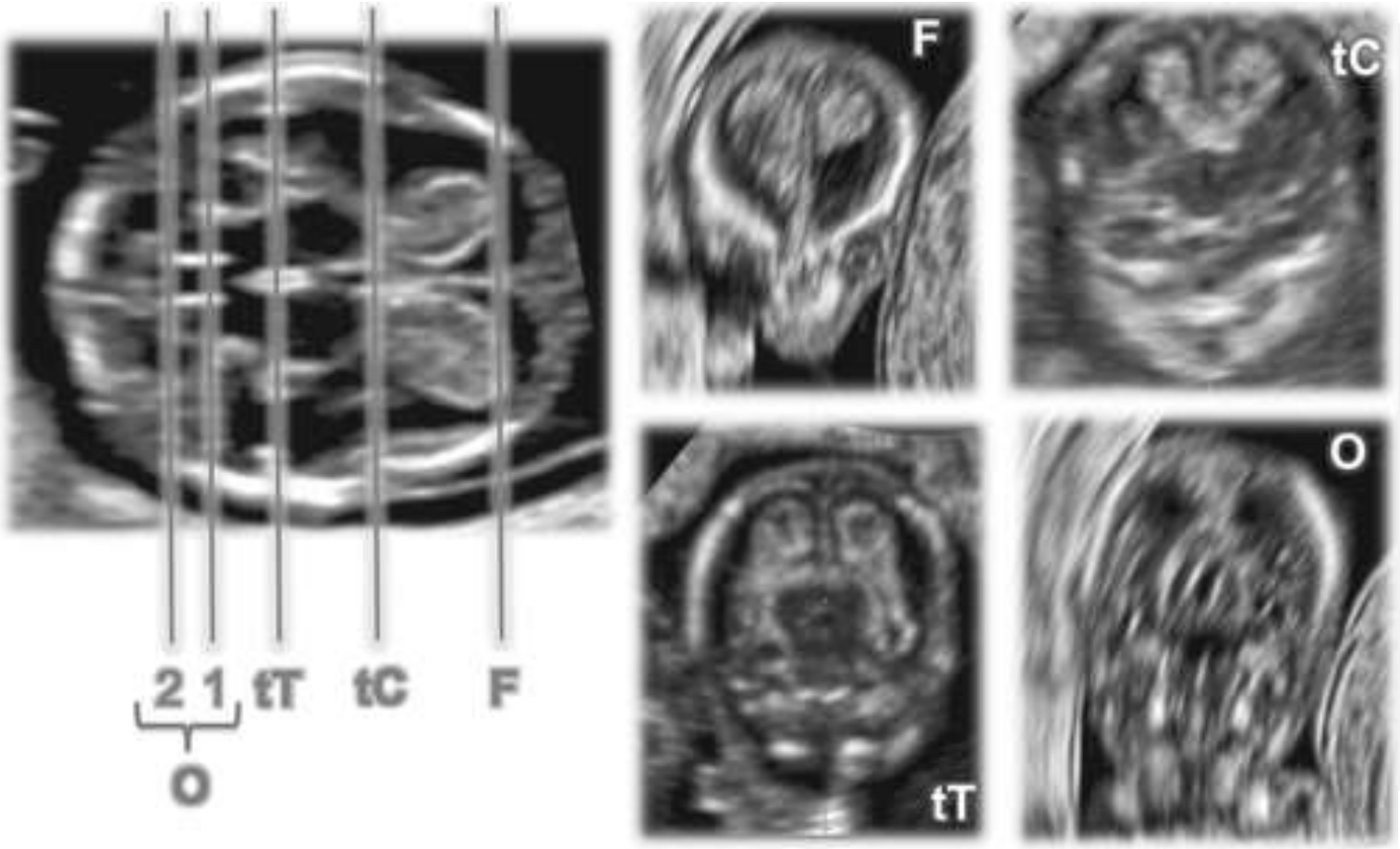
- El diencéfalo se ve como una estructura hipoecoica y redonda
- El tallo cerebral incluye el Mesencéfalo, Puente y Médula.
- En la fosa posterior se ve el 4° V y la cisterna magna
- El tallo, el 4°V y la CM forman 3 líneas paralelas anecoicas entre el esfenoides y el hueso occipital
- Relación BS/BSOB 0,5-1

PLANO SAGITAL



- Plano medio sagital visto desde la fontanela posterior
- El acueducto y el 4°V separados por el itsmo a nivel del ángulo mesencefálico
- El techo del acueducto es formado por el Tectum
- Sobre el plexo coroídeo del 4°v es posible identificar el vermis
- Bajo el plexo coroideo el velum del 4°v protruye en la CM formando la bolsa de blake

CORTES CORONALES



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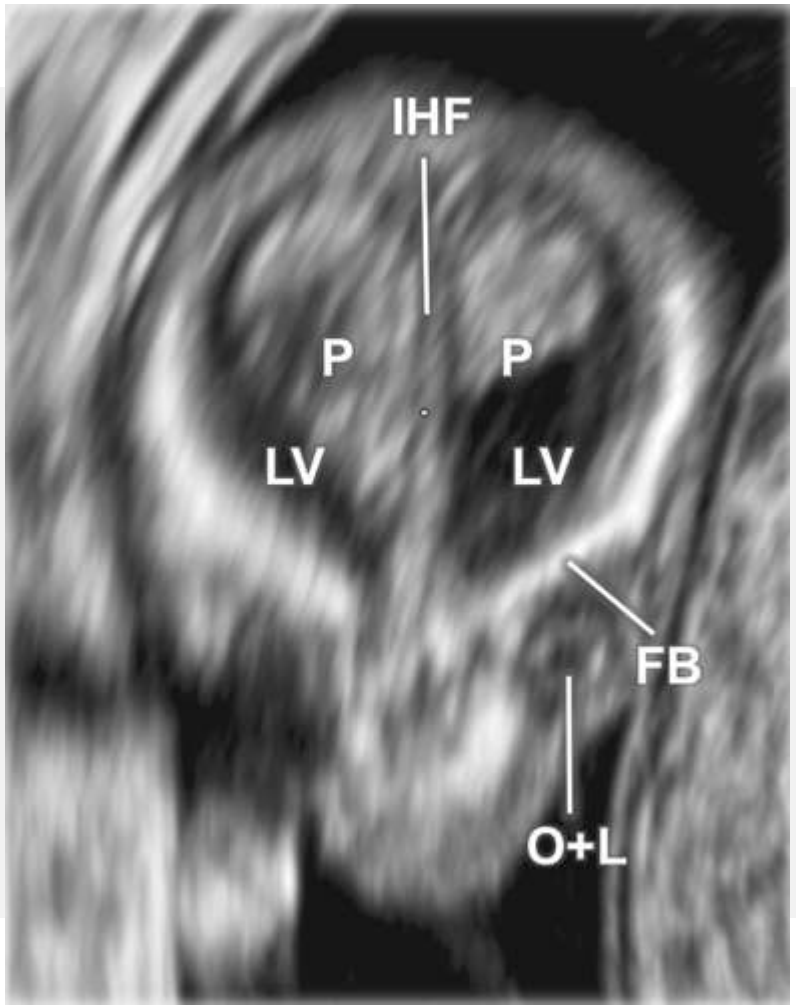

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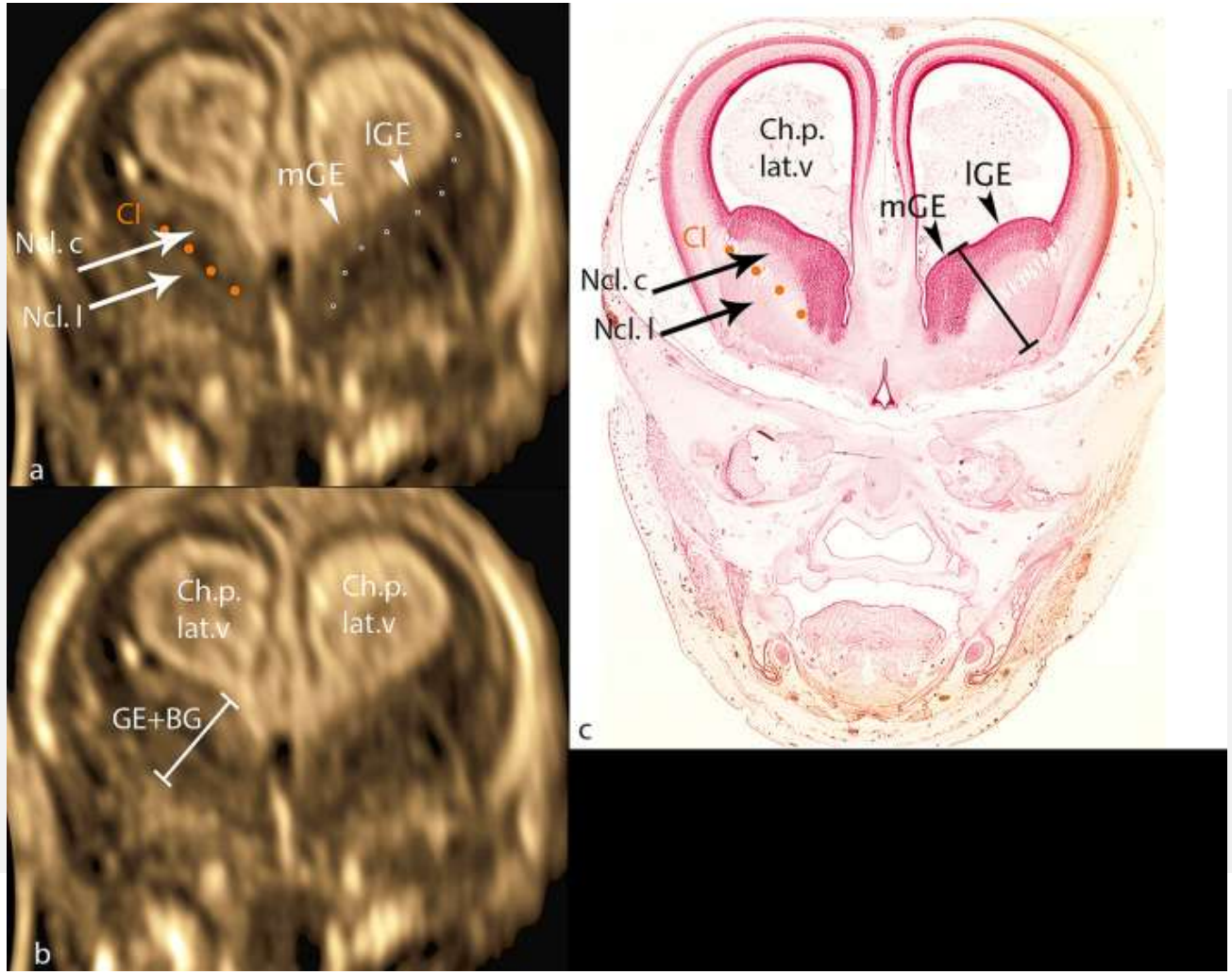
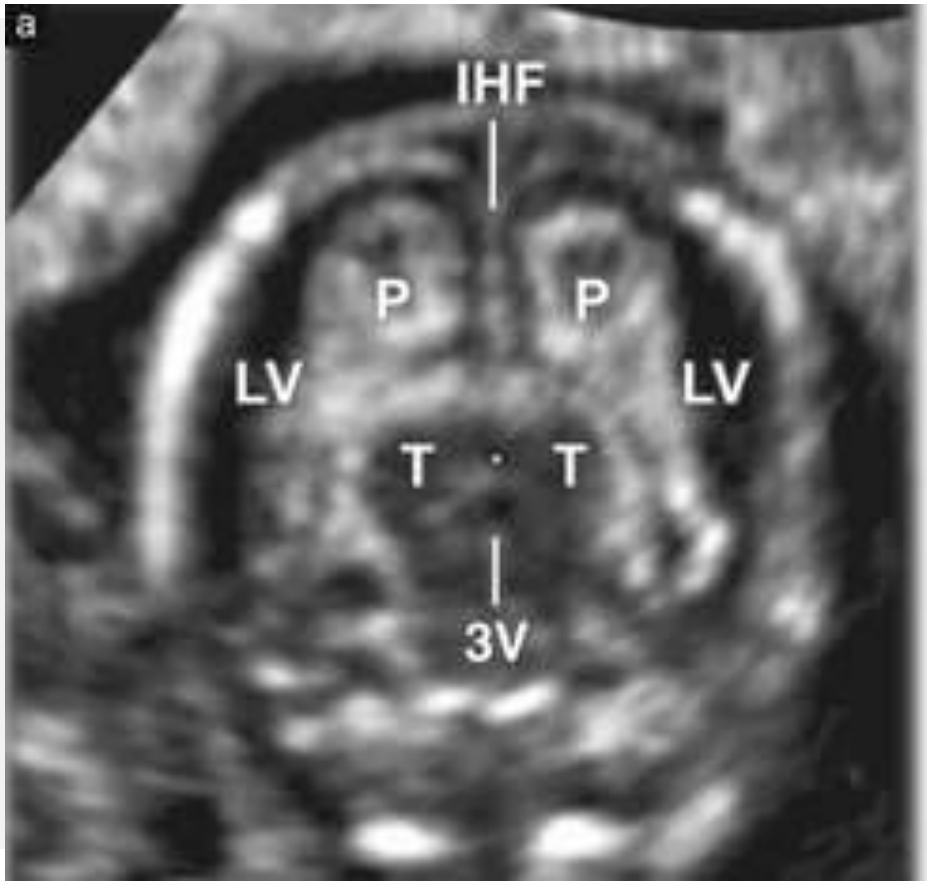
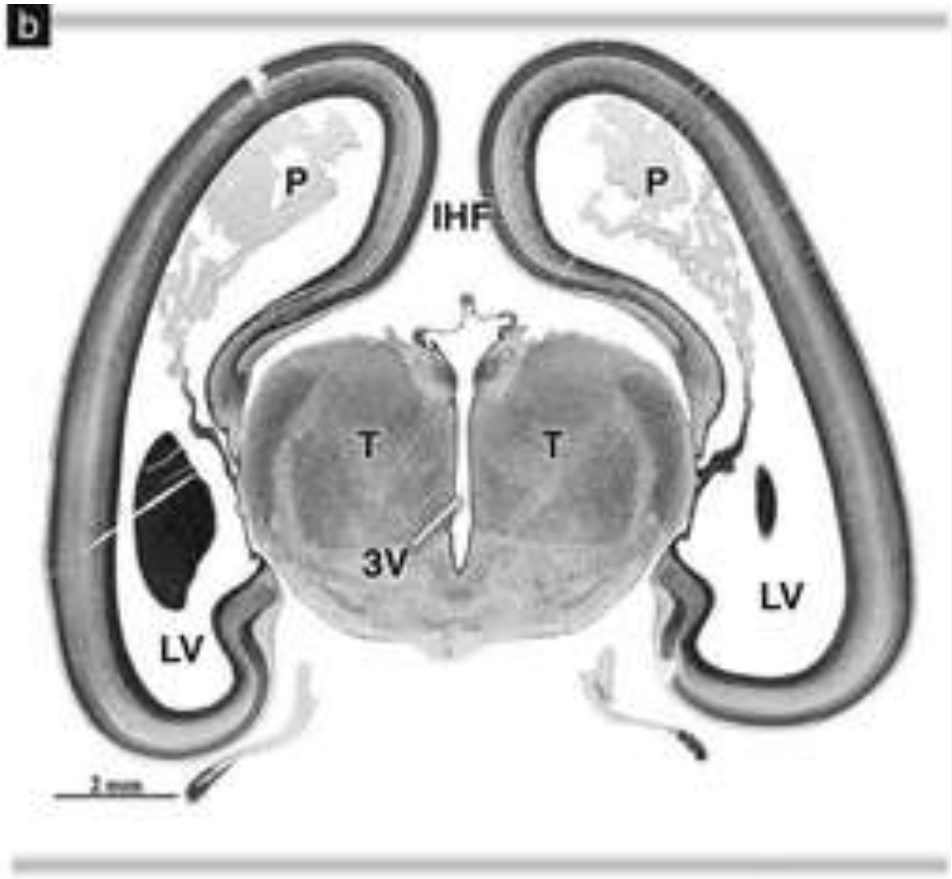


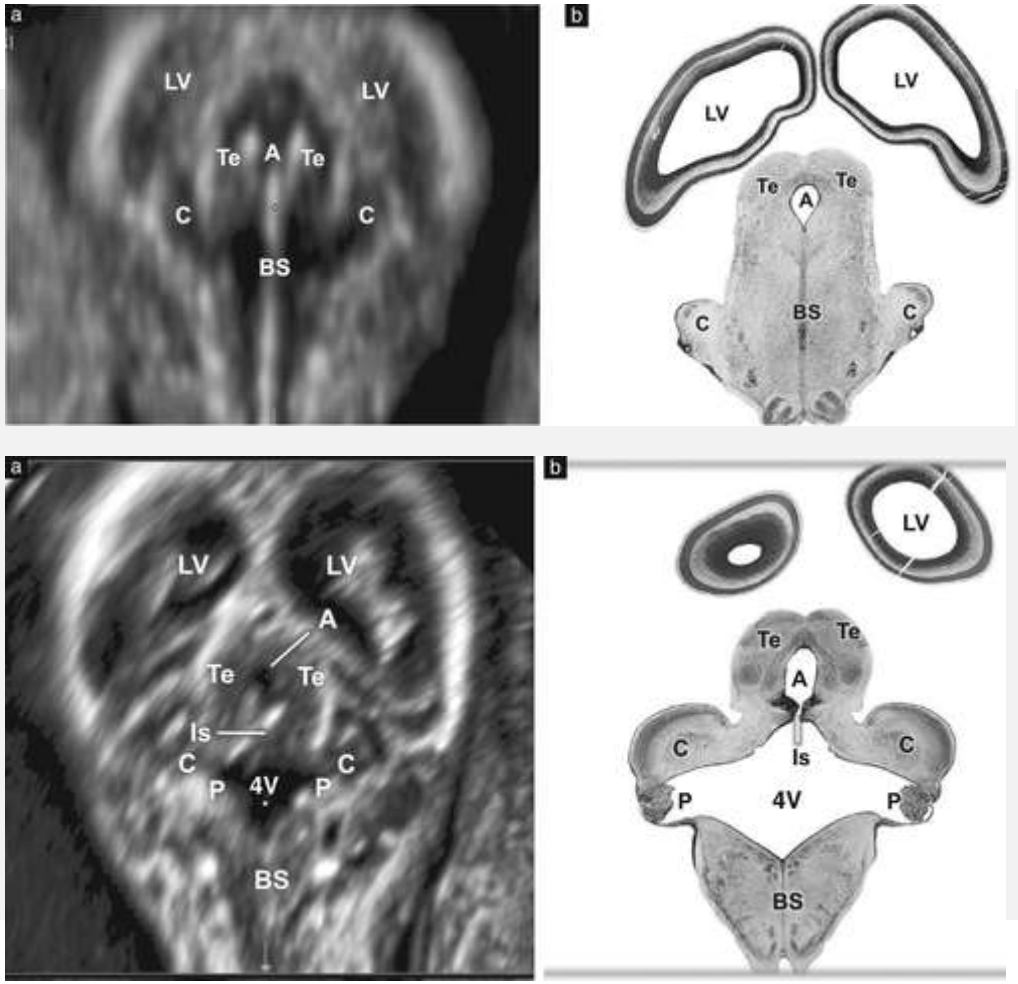
CORTES CORONALES





CORTES CORONALES

- Astas posteriores
- Acueducto
- La porción superior del acueducto rodeada de Tectum
- Es posible distinguir hemisferios cerebelosos rudimentarios



ANATOMÍA VASCULAR

- Arteria pericallosa
- A. Carótida interna
- Seno sagital superior
- Seno recto
- Vena de Galeno

DIAGNOSTICO PRENATAL DE ANOMALÍAS DEL SNC EN EL 1° TRIMESTRE

Fetal abnormality	Total	NT>95th percentile	Diagnosis 11–13 weeks	20–24 weeks	>24 week
Neural tube					
Acrania/iniencephaly	29	8 (27.6%)[a]	29 (100%)	—	—
Encephalocele	—	—	—	—	—
Open *spina bifida*	21	1 (4.7%)	3 (14.3%)	18	—
Hemivertabrae	1	—	—	1	—
Sacrococcygeal teratoma	—	—	—	—	—
Brain					
Microcephaly	1	—	—	—	1[a]
Craniosynostosis	1	—	—	1	—
Corpus callosum agenesis	10	1 (10.0%)	—	10	—
Ventriculomegaly	11	—	1 (9.1%)	9	1[b]
Holoprosencephaly	—	—	—	—	—
Alobar	2	—	2 (100%)	—	—
Semilobar	1	—	—	1	—
Cerebellar hypoplasia	3	—	—	3	—
Vermian agenesis	4	—	—	4	—

Defect	*Total*	*NT > 95th percentile*	*First trimester*
Central nervous system			
Acrania	48	0 (0)	48 (100)
Alobar holoprosencephaly	10	2 (20.0)	10 (100)
Encephalocele	15	5 (33.3)[c]	15 (100)
Open spina bifida	59	6 (10.2)[c]	35 (59.3)
Hypoplastic cerebellum/vermis	15	0 (0)	2 (13.3)
Agenesis of corpus callosum	26	2 (7.7)	0 (0)
Schizencephaly	3	0 (0)	0 (0)
Septo-optic dysplasia	1	0 (0)	0 (0)
Microcephaly	9	0 (0)	0 (0)
Severe ventriculomegaly	18	0 (0)	0 (0)
Arachnoid cyst	14	1 (7.1)	0 (0)
Brain hemorrhage	2	0 (0)	0 (0)
Dural venous sinus thrombosis	2	0 (0)	0 (0)
Craniosynostosis	2	1 (50.0)	0 (0)
Occipital dermoid cyst	1	1 (100)[c]	0 (0)
Blake's pouch cyst	4	0 (0)	0 (0)
Brain tumor	2	0 (0)	0 (0)

Syngelaki, A., Chelemen, T., Dagklis, T., Allan, L., & Nicolaides, K. H. (2011). Challenges in the diagnosis of fetal non-chromosomal abnormalities at 11–13 weeks. *Prenatal Diagnosis*, *31*(1), 90–102. https://doi.org/10.1002/PD.2642

Syngelaki, A., Hammami, A., Bower, S., Zidere, V., Akolekar, R., & Nicolaides, K. H. (2019). Diagnosis of fetal non-chromosomal abnormalities on routine ultrasound examination at 11–13 weeks' gestation. *Ultrasound in Obstetrics & Gynecology*, *54*(4), 468–476. https://doi.org/10.1002/UOG.20844

DIAGNOSTICO EN 1° TRIMESTRE

Baseline characteristics of the 38,586 pregnancies and 141 cases with CNS abnormalities

Characteristic		Fetuses with normal CNS	Cases with CNS abnormalities	P
Maternal age, years	Range	17–51	24–44	0.242
	Average (SD)	31.5 (4.0)	31.9 (3.9)	
Maternal height, cm	Range	140–186	150–178	0.918
	Average (SD)	162.8(5.0)	162.8(5.3)	
Maternal weight, kg	Range	35.0-99.5	43.5–97.0	0.498
	Average (SD)	59.0 (9.3)	58.4 (9.9)	
Body-mass index, kg/m^2	Range	13.0–40.0	16.9–37.5	0.443
	Average (SD)	22.2 (3.3)	22.0 (3.5)	
Fetal CRL, mm		63.8 (7.2)	61.3 (8.4)	< 0.001
Fetal NT, mm		1.4 (0.5)	1.7 (1.4)	0.035

Table 2 Fetal CNS abnormalities diagnosed during first-trimester scans

Fetal CNS abnormality	Total	Increased NT	Diagnosis						Pregnancy outcome		
			11-14w	rate, %	22-24w	28-34w	≥35w	Post-natal/MRI	TOP	Misc/IUD	LB
Anencephaly	5		5	100	0	0	0		5	0	0
Exencephaly	22	1	22	100	0	0	0		22	0	0
Meningoencephalocele	4	1	4	100	0	0	0		3	1	0
Holoprosencephaly	14		11	79	2	1	0		14	0	0
Alobar	5		5	100	0	0	0		5	0	0
Semi lobar	8		6	75	2	0	0		8	0	0
Lobar	1		0	0	0	1	0		1	0	0
Spina bifida	9		2	22	3	3	0	1	5	0	4
Open	3		2	67	1	0	0		3	0	0
Closed	6		0	0	2	3	0	1	2	0	4
PFA	10	1	2	20	5	1	1	1	5	0	5
Dandy-Walker	1		1	100	0	0	0		1	0	0
Vermis hypoplasia	1		0	0	1	0	0		1	0	0
Joubert Syndrome	1		1	100	0	0	0		1	0	0
Cerebellar hypoplasia	1		0	0	1	0	0		1	0	0
Blake's cyst	5	1	0	0	3	1	1		1	0	4
Mega Cisterna Magna	1		0	0	0	0	0	1	0	0	1
Severe ventriculomegaly	12	1	1	8	5	5	1		10	1	1
Vein of Galen aneurysmal malformation	1		0	0	0	0	1		0	0	1
Arachnoid cyst	32	2	0	0	8	9	15		6	0	26
Microcephaly	1		0	0	1	0	0		1	0	0
Agenesis of the Corpus Callosum	12	2	0	0	10	1	0	1	10	0	2
Intracranial infection	1		0	0	0	1	0		0	0	1
Subependymal cyst	19		0	0	0	13	6		1	0	18
Septum pellucidum cyst	4		0	0	0	3	1		0	0	4
Isolated absence of septum pellucidum	1		0	0	0	1	0		0	0	1
cerebral infarction	2		0	0	0	0	0	2	1	1	0
cortex dysplasia	4		0	0	0	0	0	4	3	1	0

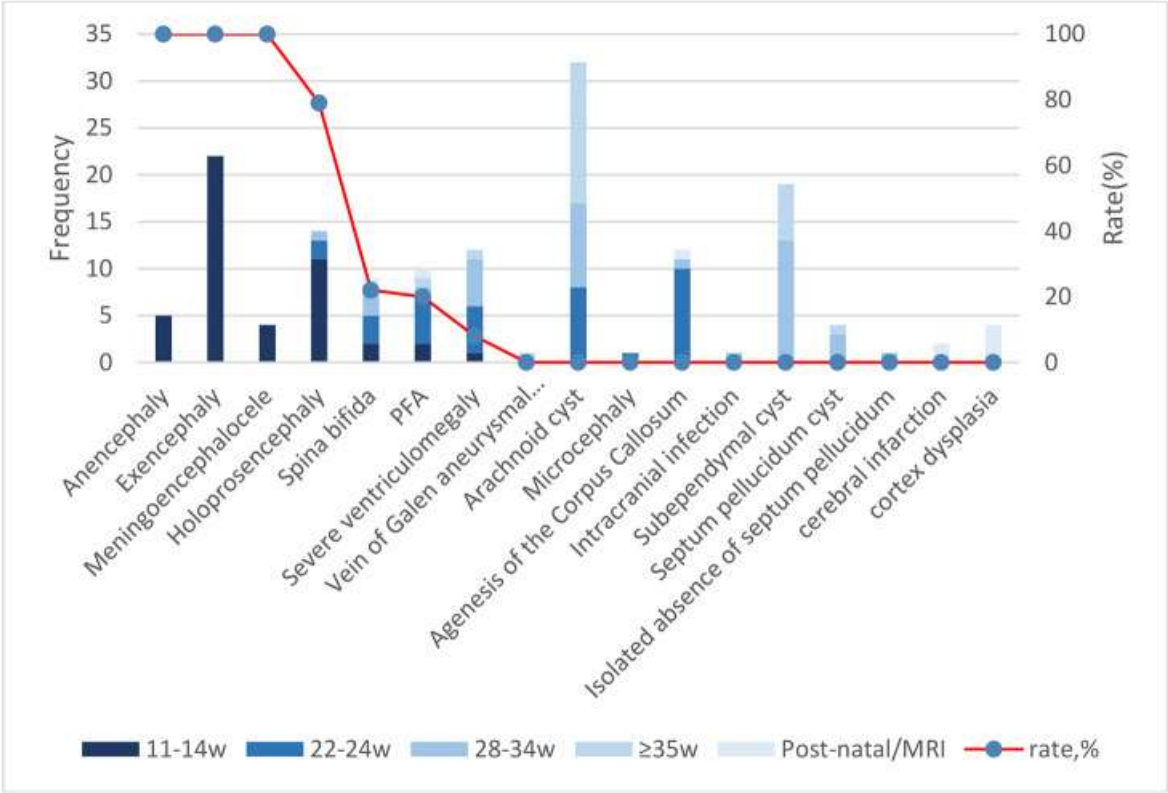
NT: nuchal translucency; PFA: posterior cranial fossa anomalies; TOP: Termination Of Pregnancy; Misc: Miscarriage; IUD: Intrauterine Death; LB: Live Birth

Hu, Y., Sun, L., Feng, L., Wang, J., Zhu, Y., & Wu, Q. (2023). The role of routine first-trimester ultrasound screening for central nervous system abnormalities: a longitudinal single-center study using an unselected cohort with 3-year experience. *BMC Pregnancy and Childbirth*, *23*(1). https://doi.org/10.1186/S12884-023-05644-Z

DIAGNOSTICO EN 1° TRIMESTRE

The pregnancy outcomes of the cases with CNS anomalies

Gestational age of diagnosed or suspected abnormality	Number of cases with CNS anomalies (detection rate)	Pregnancy outcome		
		TOP	Misc/IUD	LB
1st trimester scan	45(32%)	43(96%)	2(4%)	0
2nd trimester scan	31(22%)	26(84%)	0	5(16%)
3rd trimester scan	35(25%)	5(14%)	0	30(86%)
Late 3rd trimester scan	23(16%)	0	0	23(100%)



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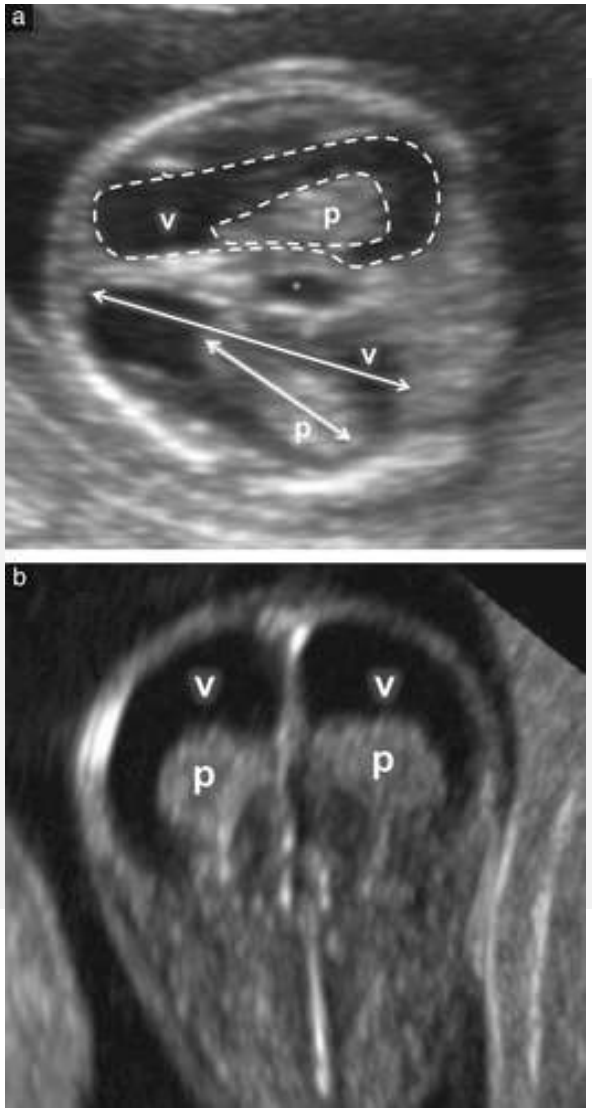
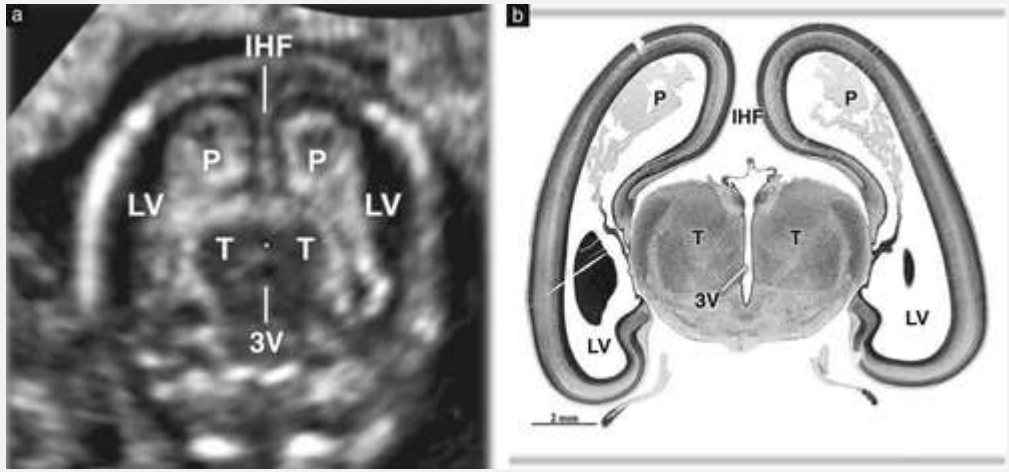
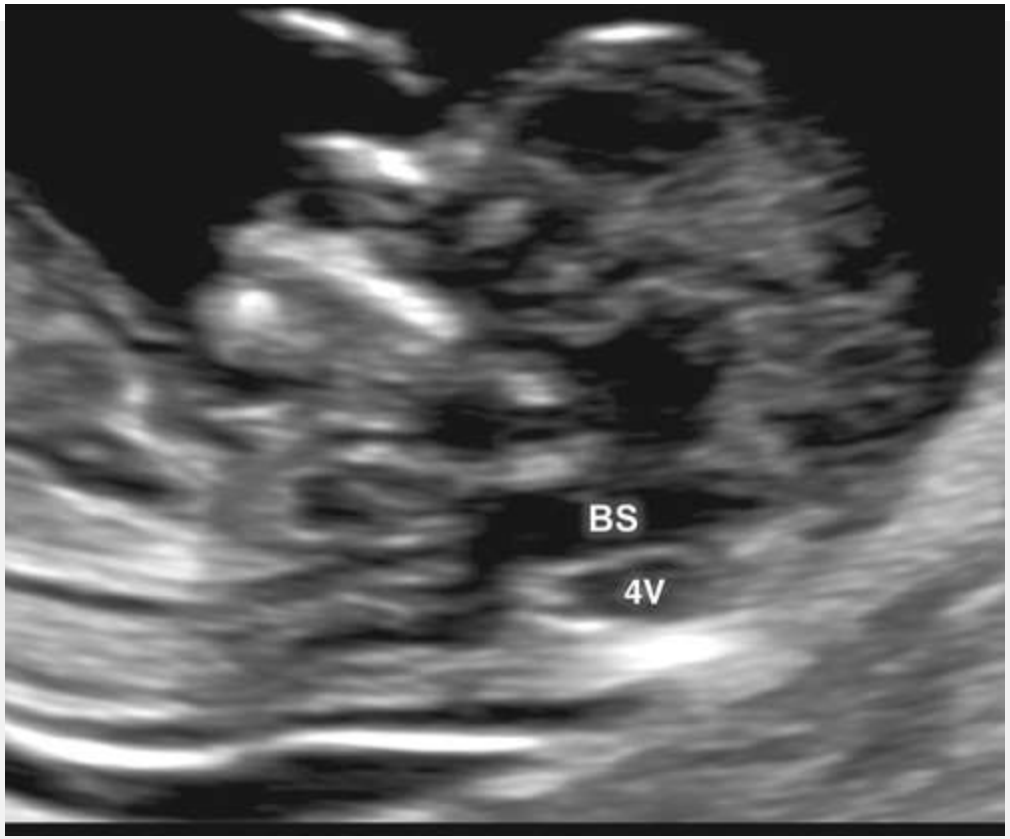
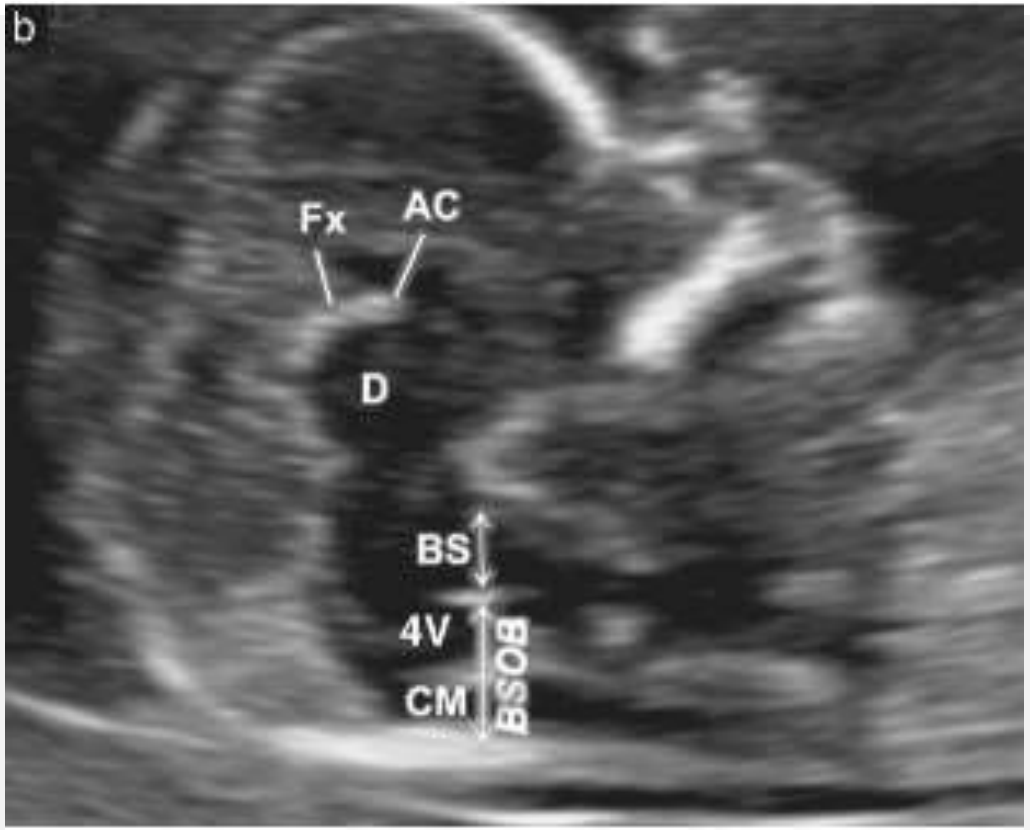
Ventriculomegaliaen 1° trimestre

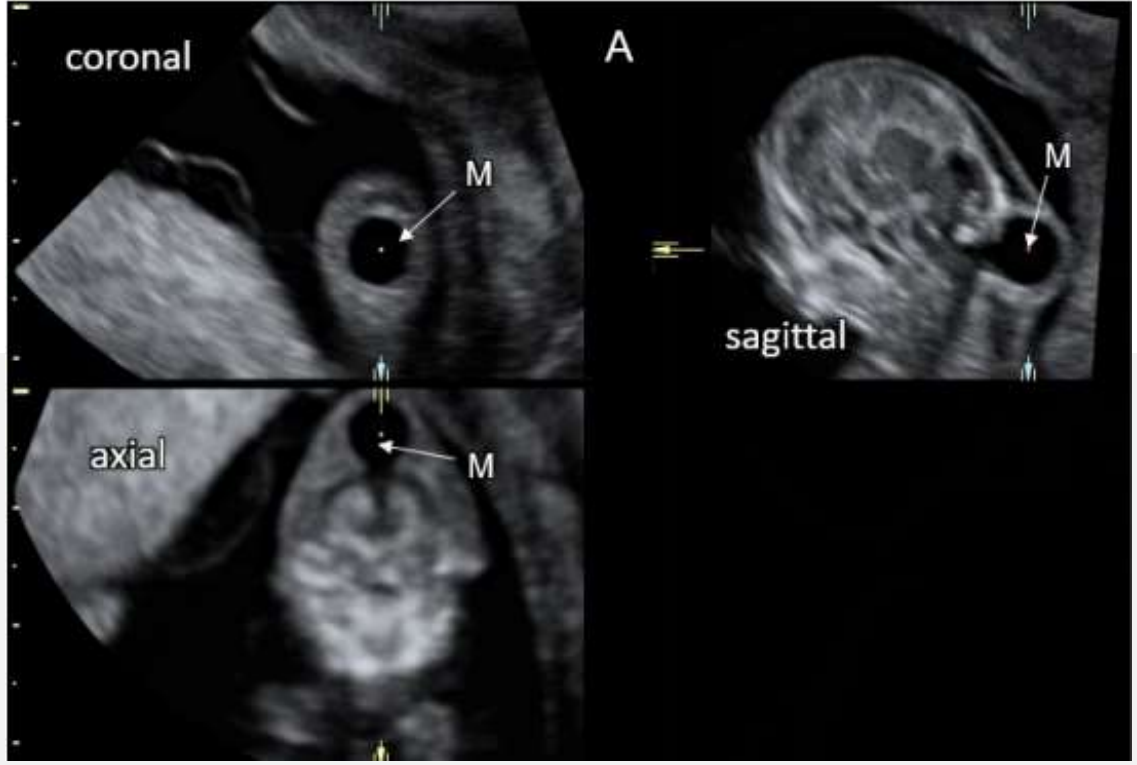




Espina bífida



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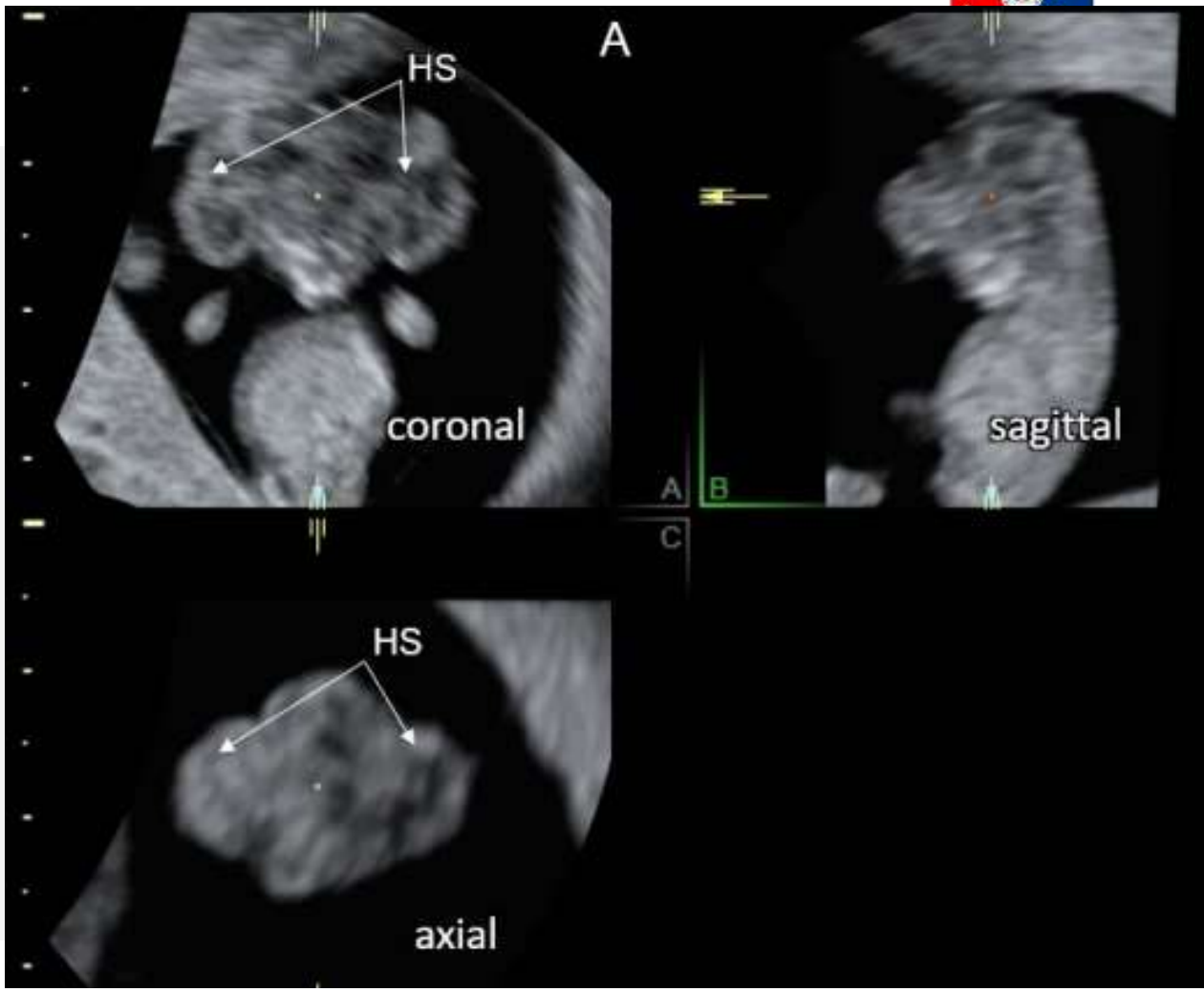
M
M
sagittal
axial
M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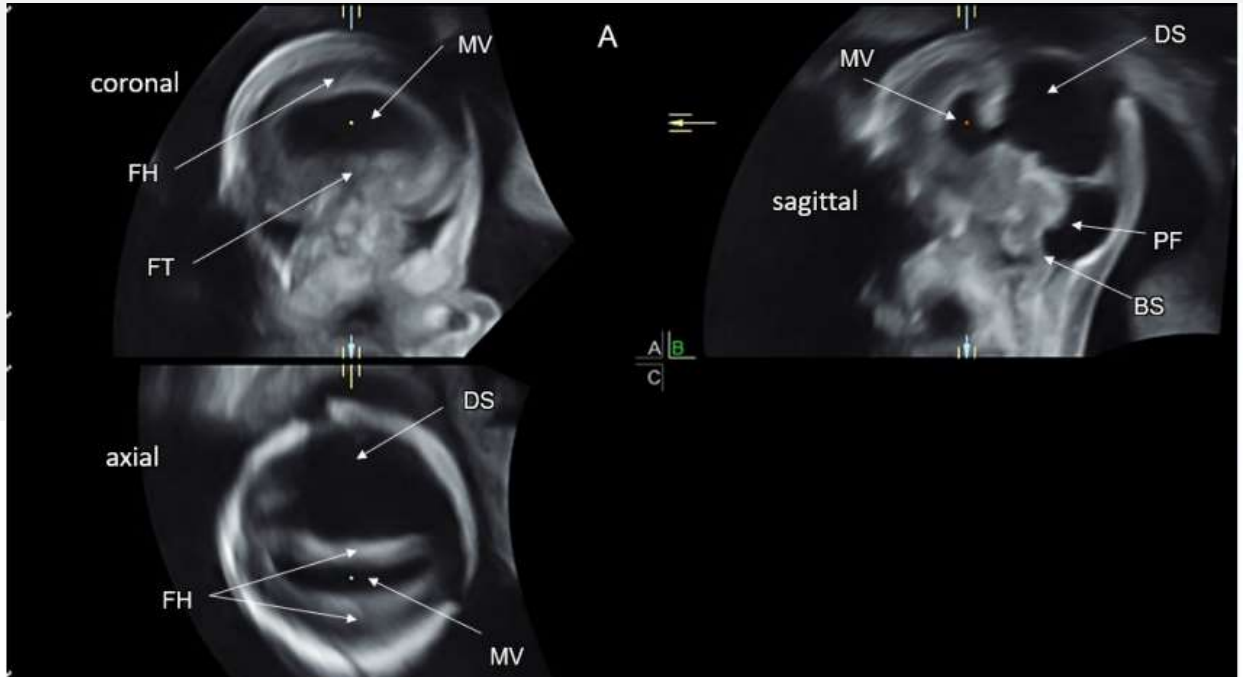
HS
coronal
sagittal
A
B
C
HS
axial


coronal
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FH
FT
A
MV
DS
sagittal
PF
BS
A
B
C
axial
DS
FH
MV

CONCLUSIONES

- El diagnóstico prenatal de las malformaciones del SNC continúa siendo un desafío
- Durante el primer trimestre es posible identificar precozmente la mayoría de las condiciones letales
- Una evaluación detallada permitiría sospechar otras malformaciones del SNC y guiar el manejo

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Examen del SNC en el 1° trimestre

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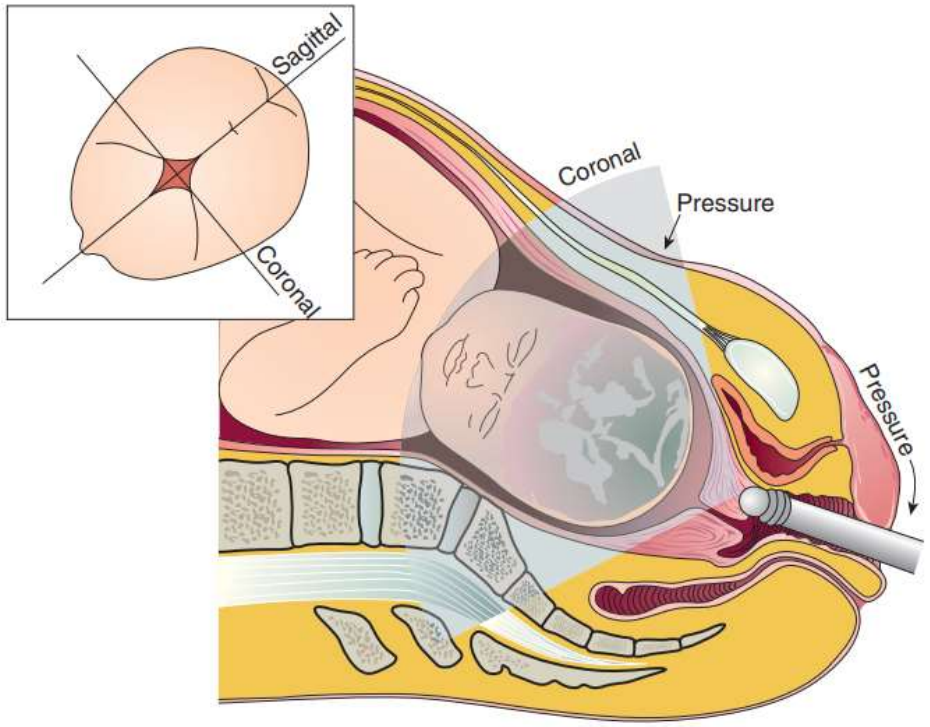
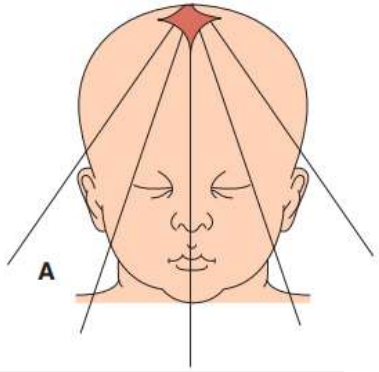
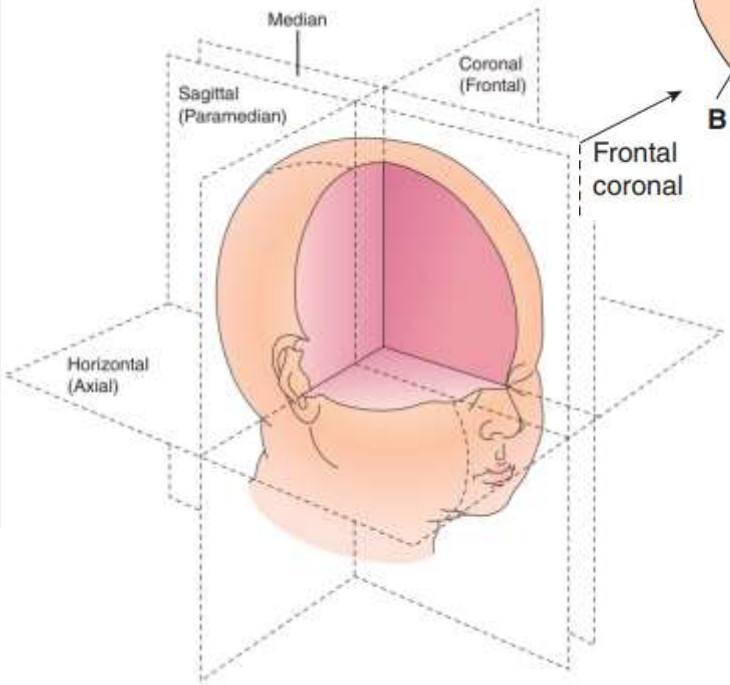
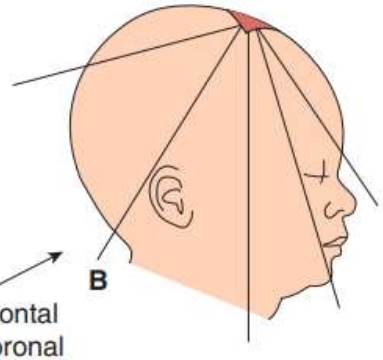
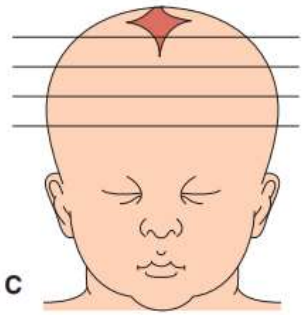
Planos



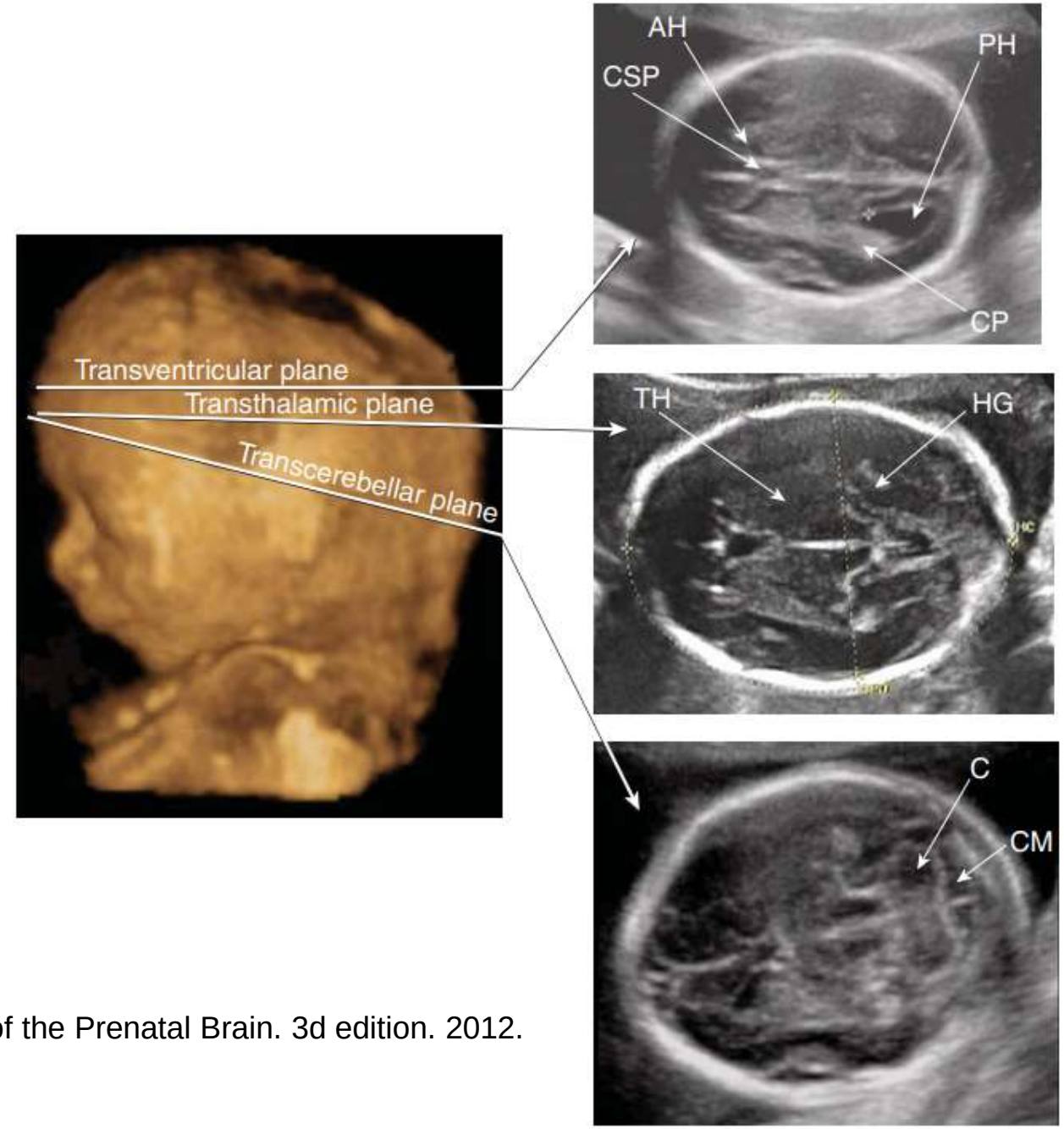








Timor-Tritsch. Ultrasonography of the Prenatal Brain. 3d edition. 2012.



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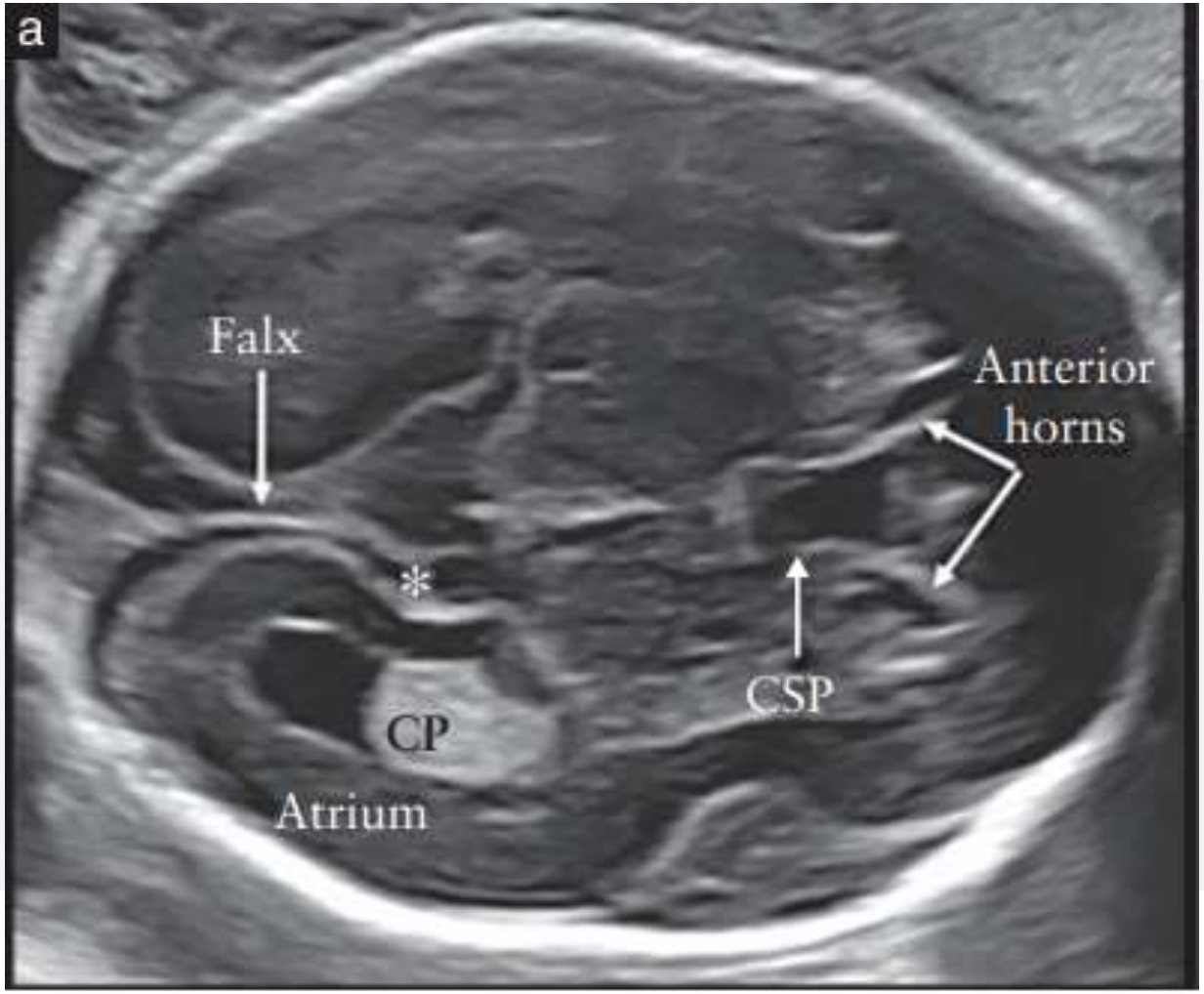

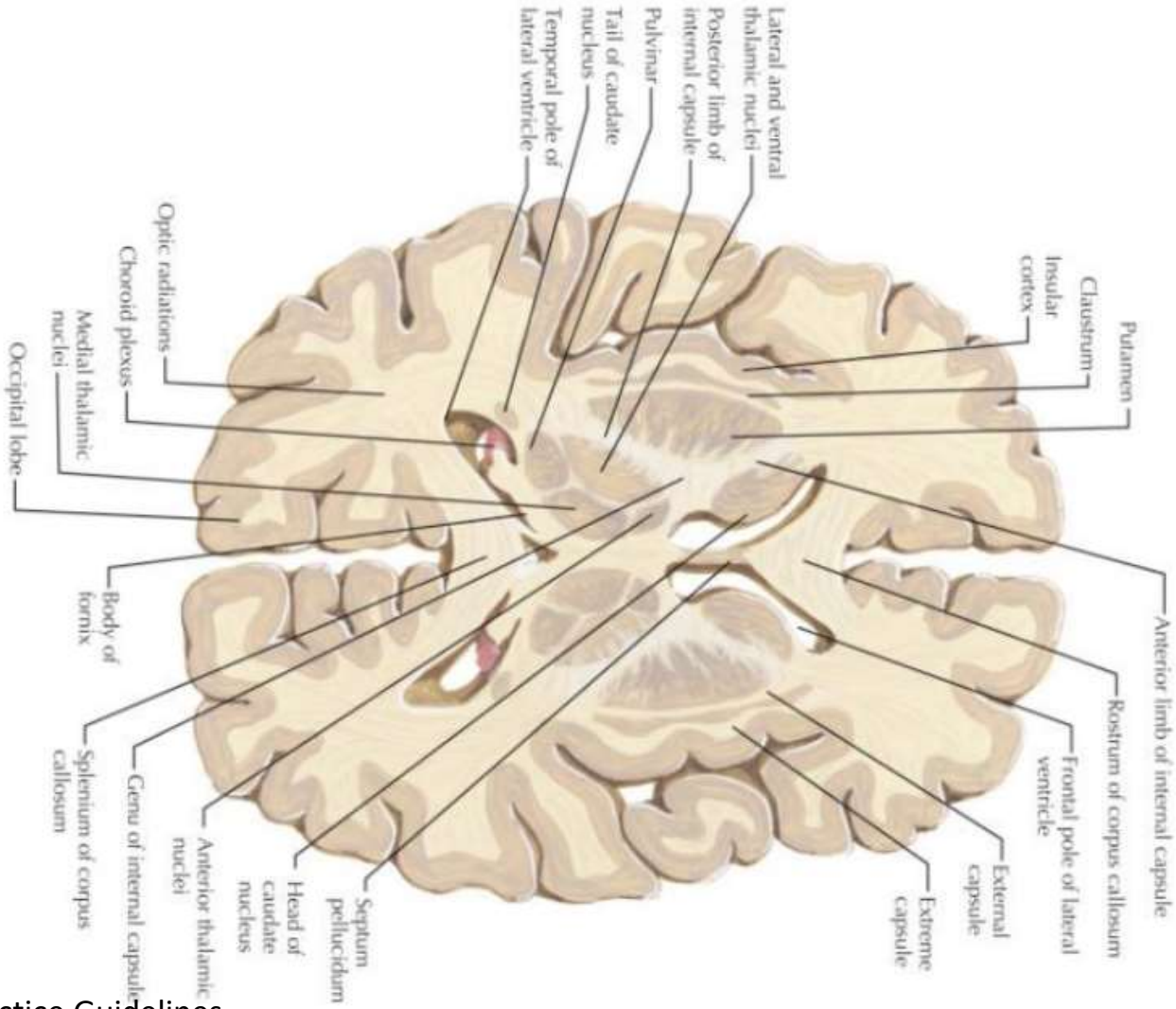

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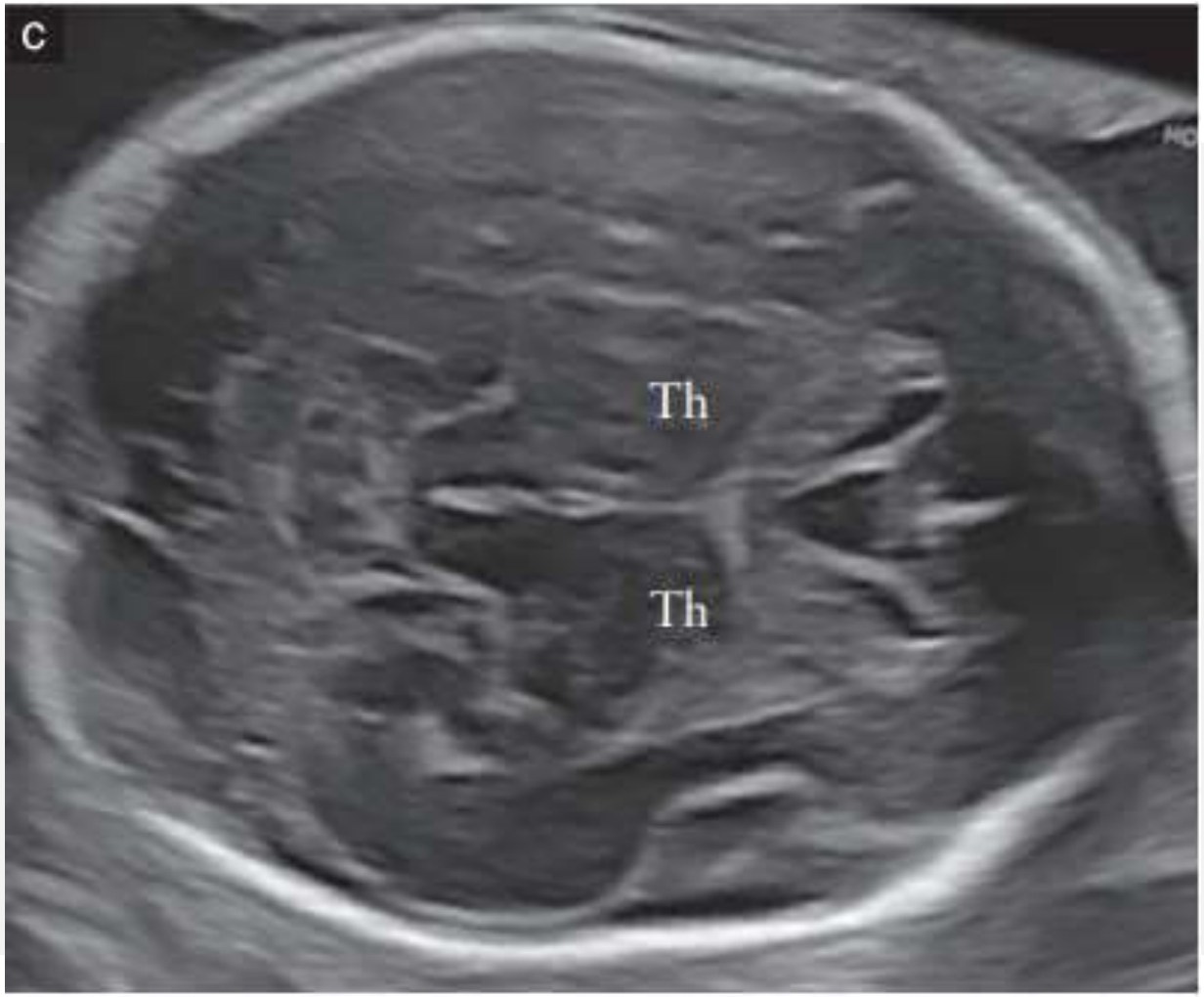

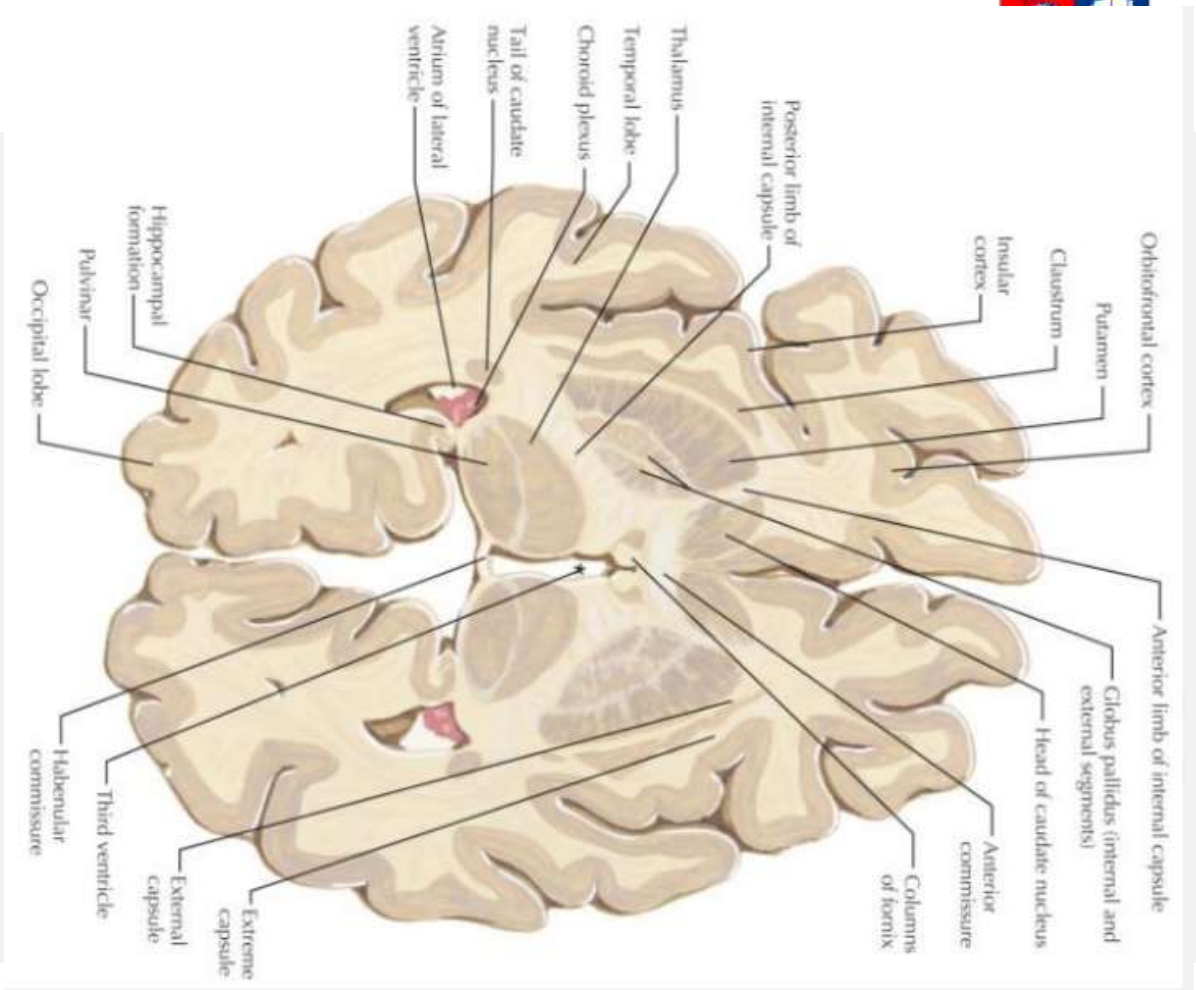

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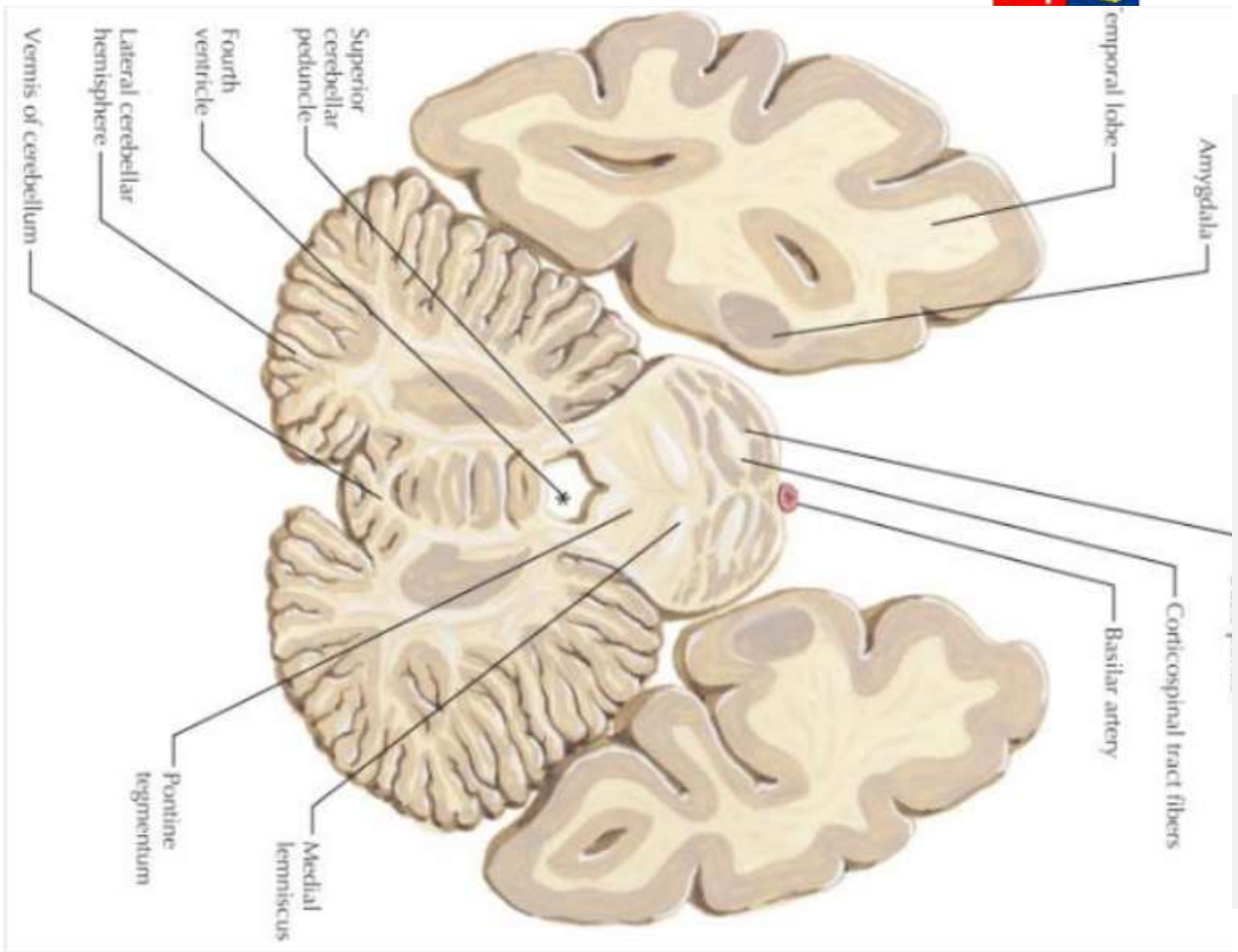

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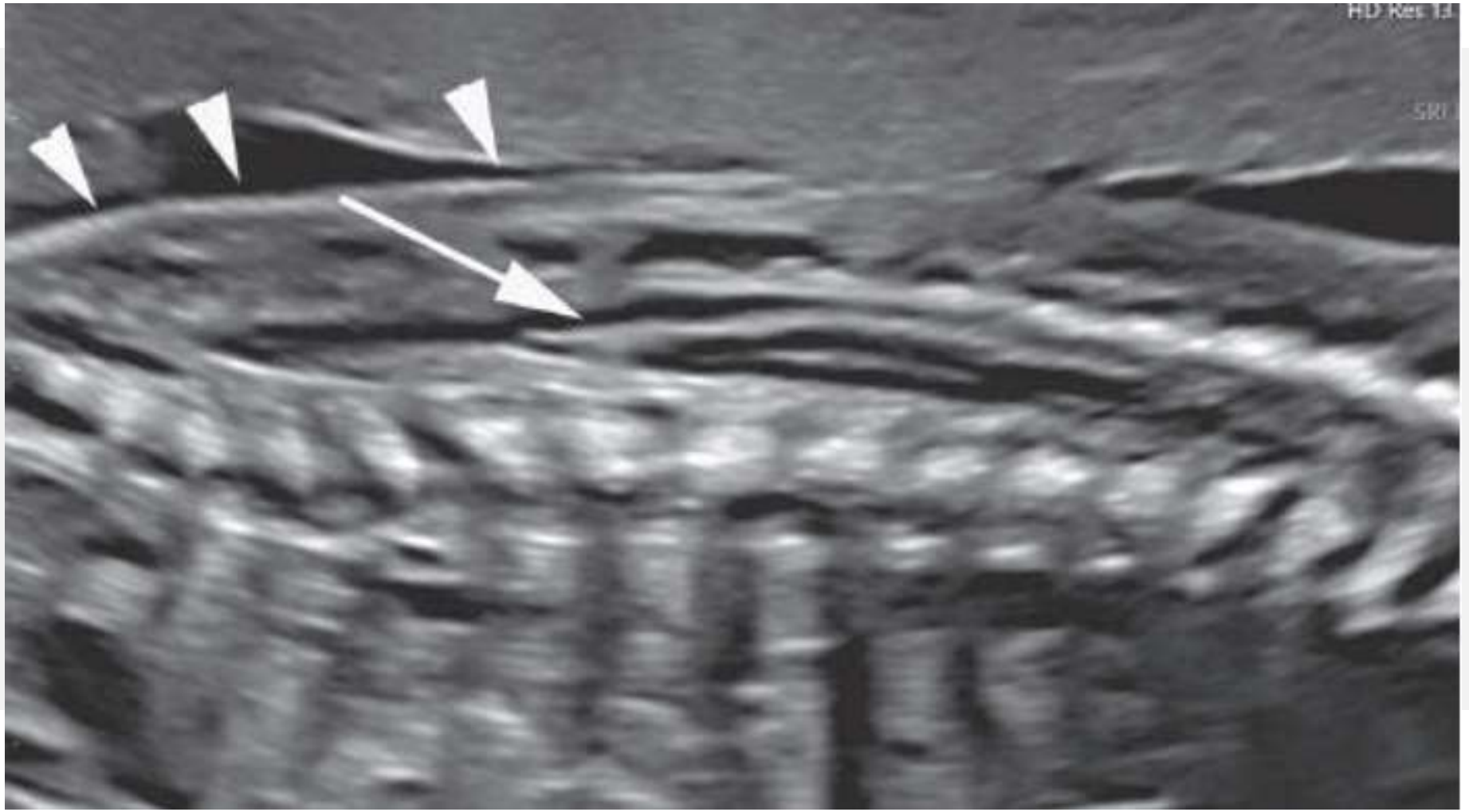

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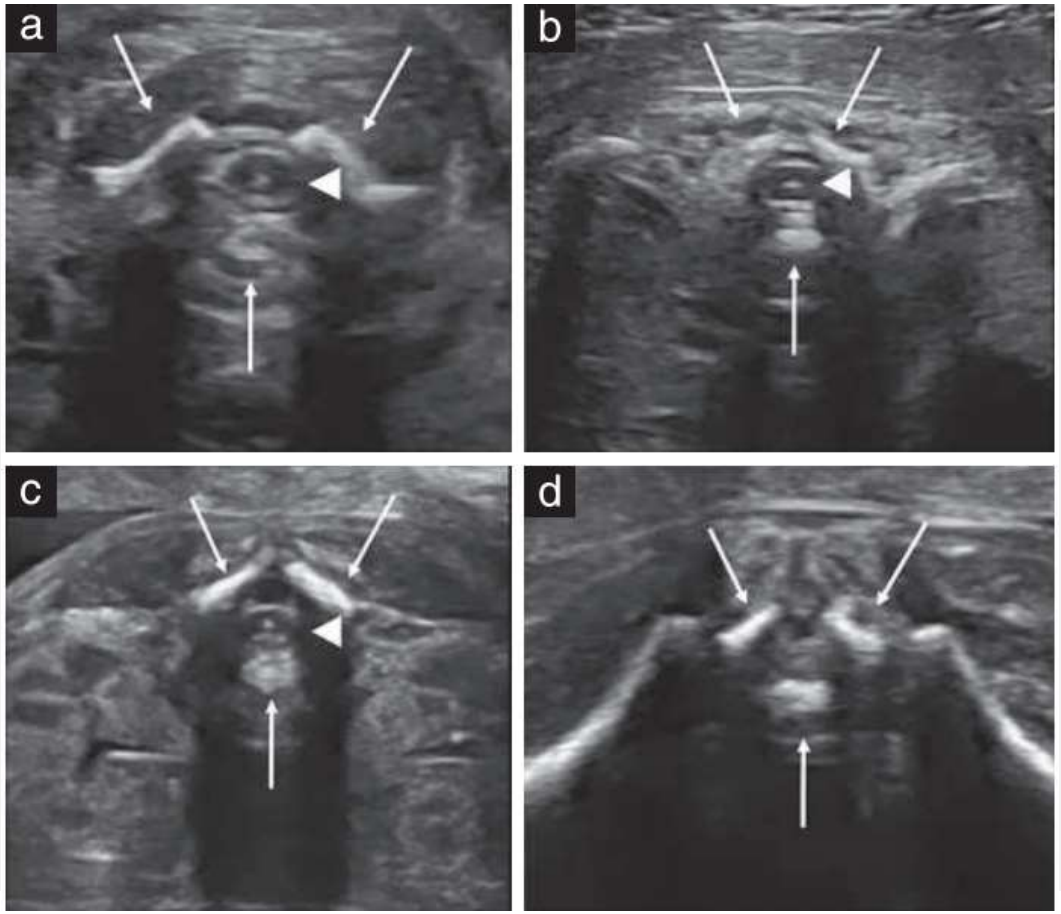

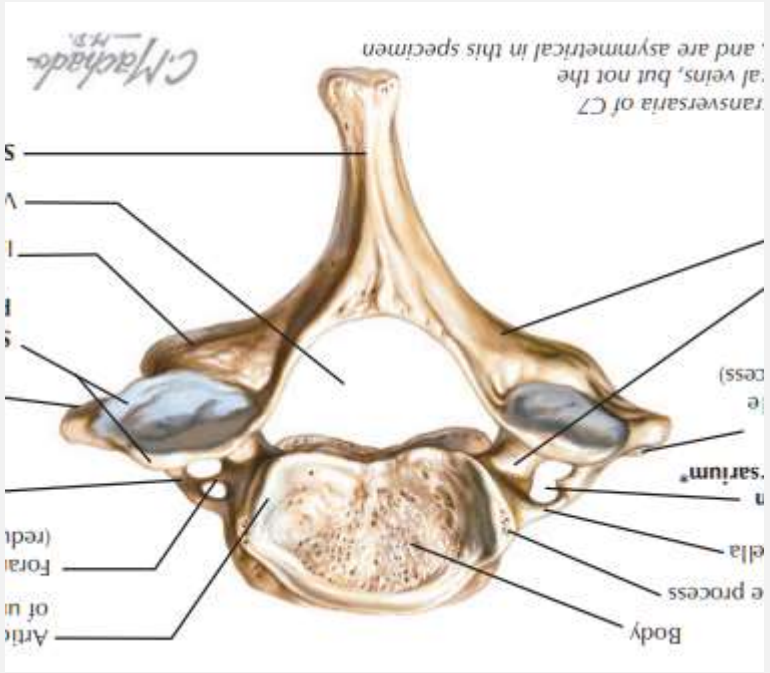

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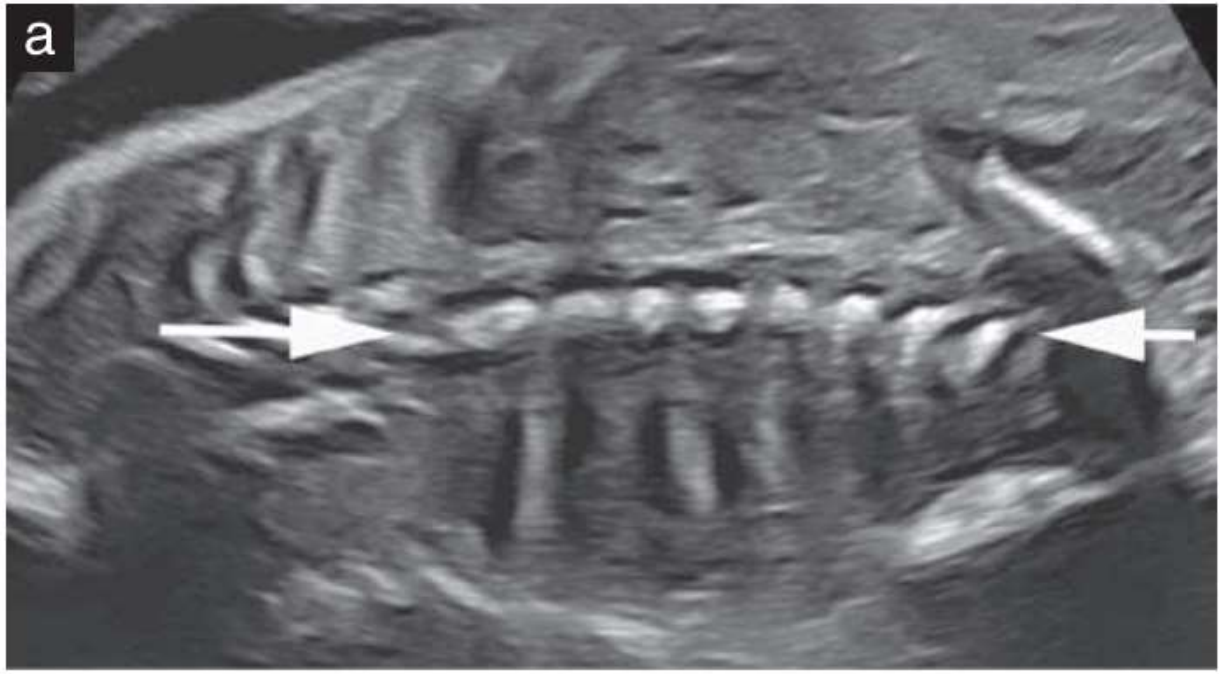

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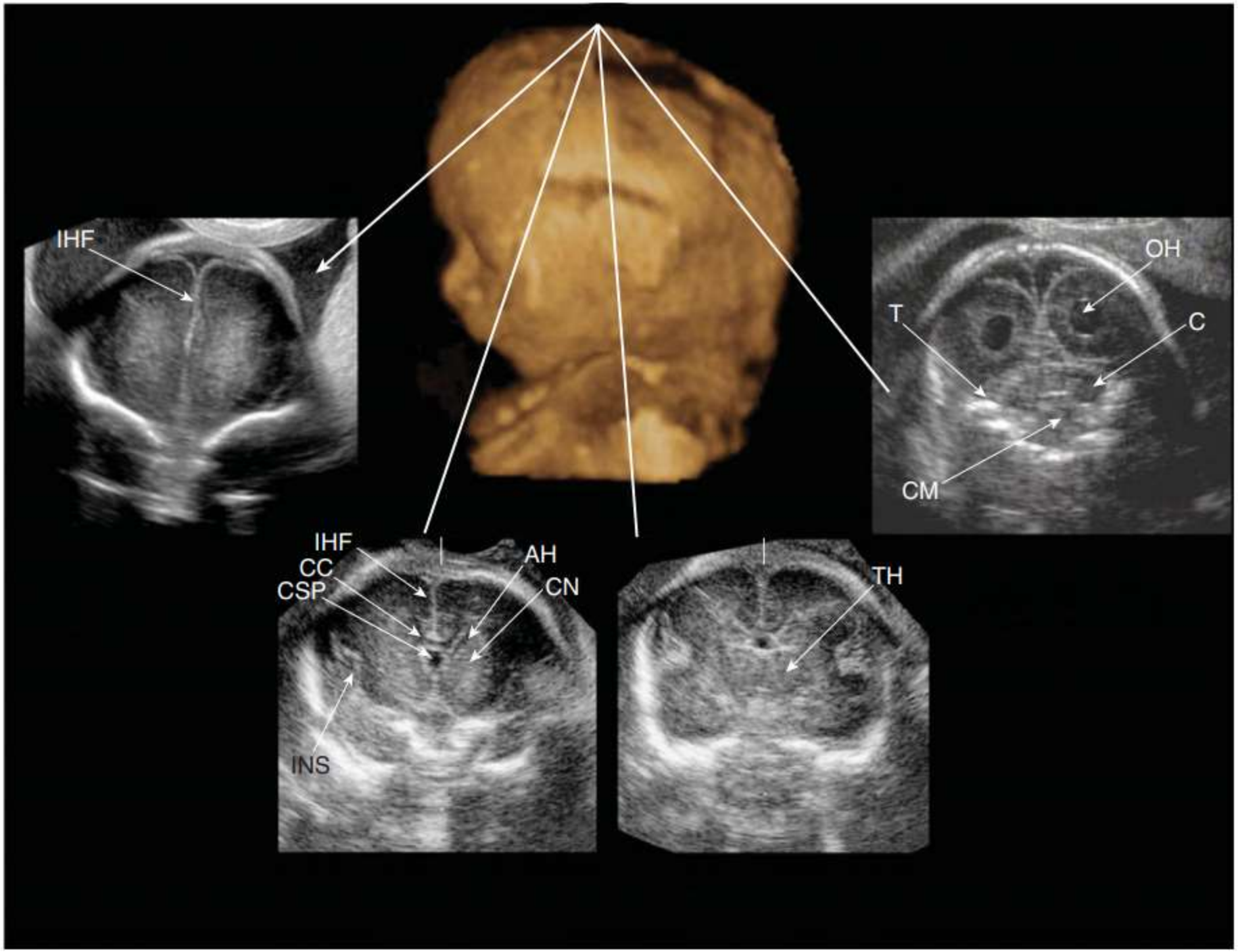
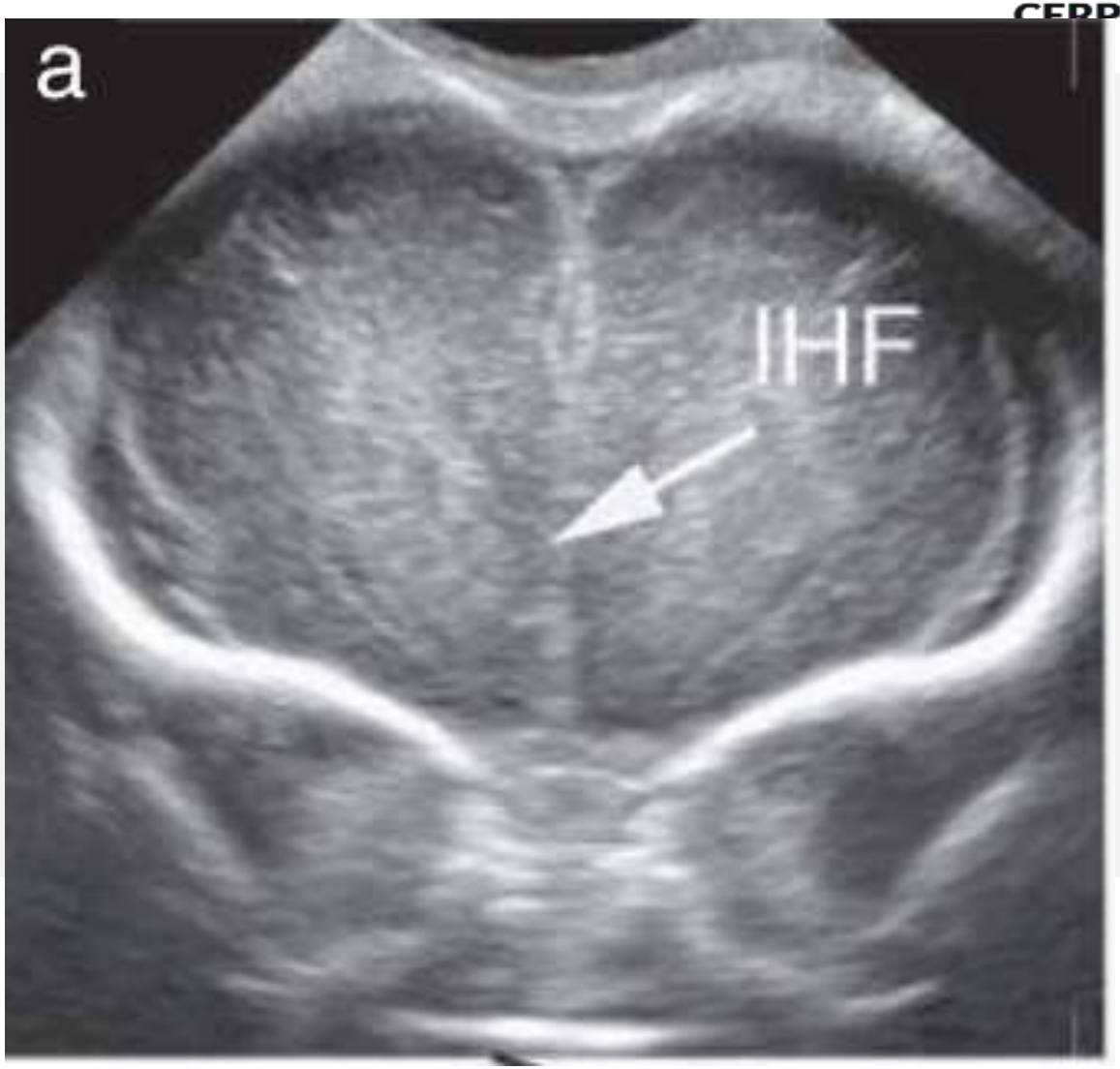
Table 2 Indications for targeted fetal neurosonography

- Suspicion of CNS or spinal malformation at routine screening ultrasound
- Suspicion of CNS or spinal malformation at nuchal translucency scan
- Family history of inheritable CNS or spinal malformations
- Previous pregnancy complicated by fetal brain or spinal malformation
- Fetus with congenital heart disease
- Monochorionic twins
- Suspected congenital intrauterine infection
- Exposure to teratogens known to affect neurogenesis
- Chromosomal microarray findings of unknown significance

CNS, central nervous system.


IHF
OH
T
C
CM
IHF
CC
CSP
AH
CN
INS
TH

Transfrontal



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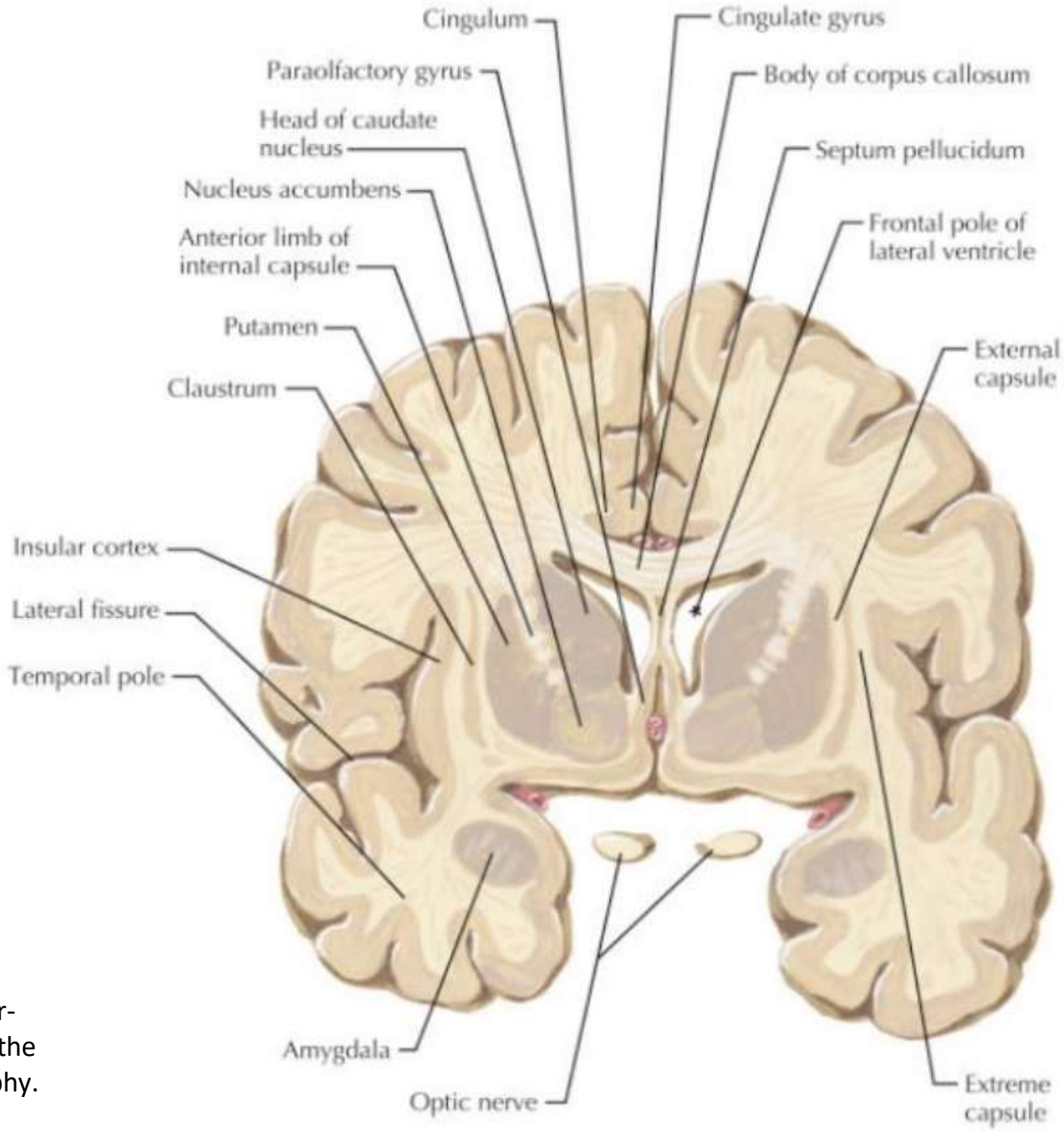

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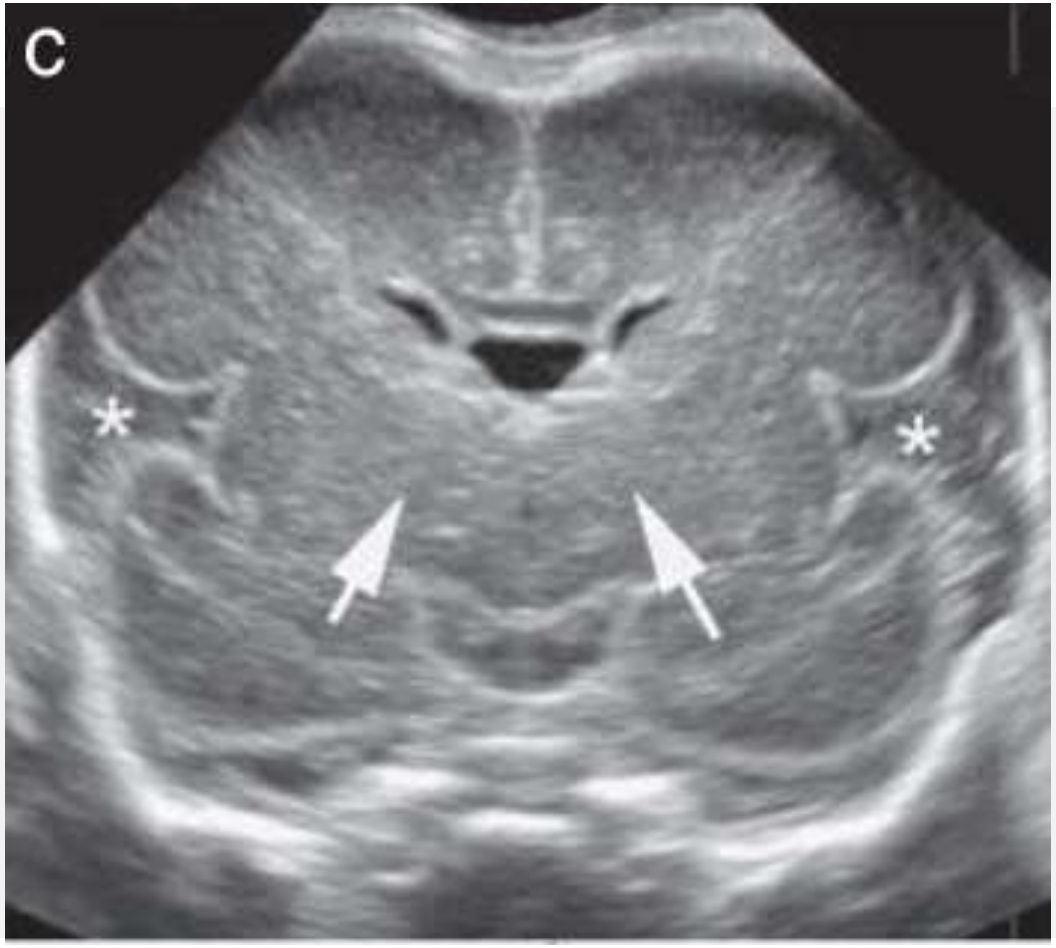

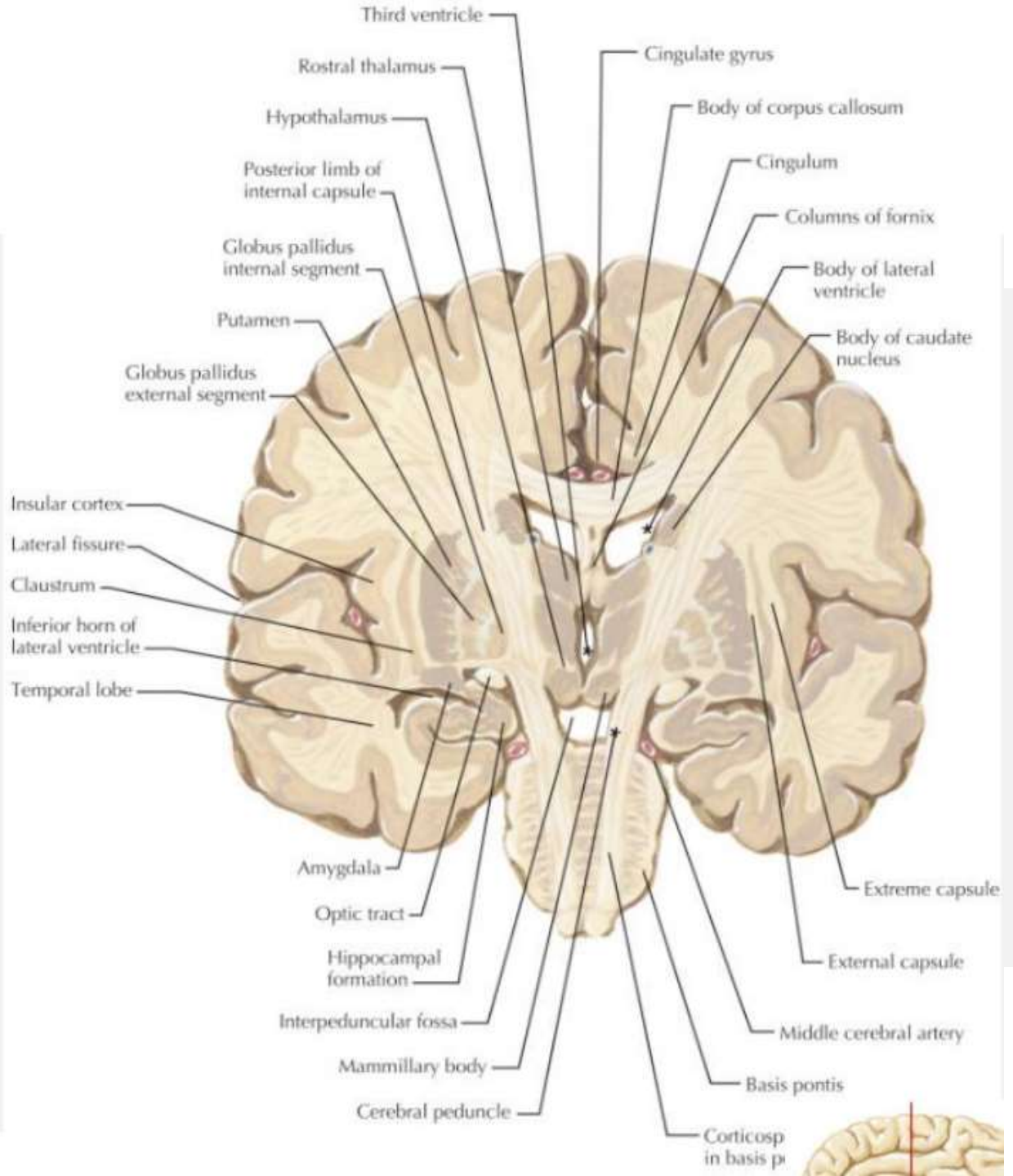

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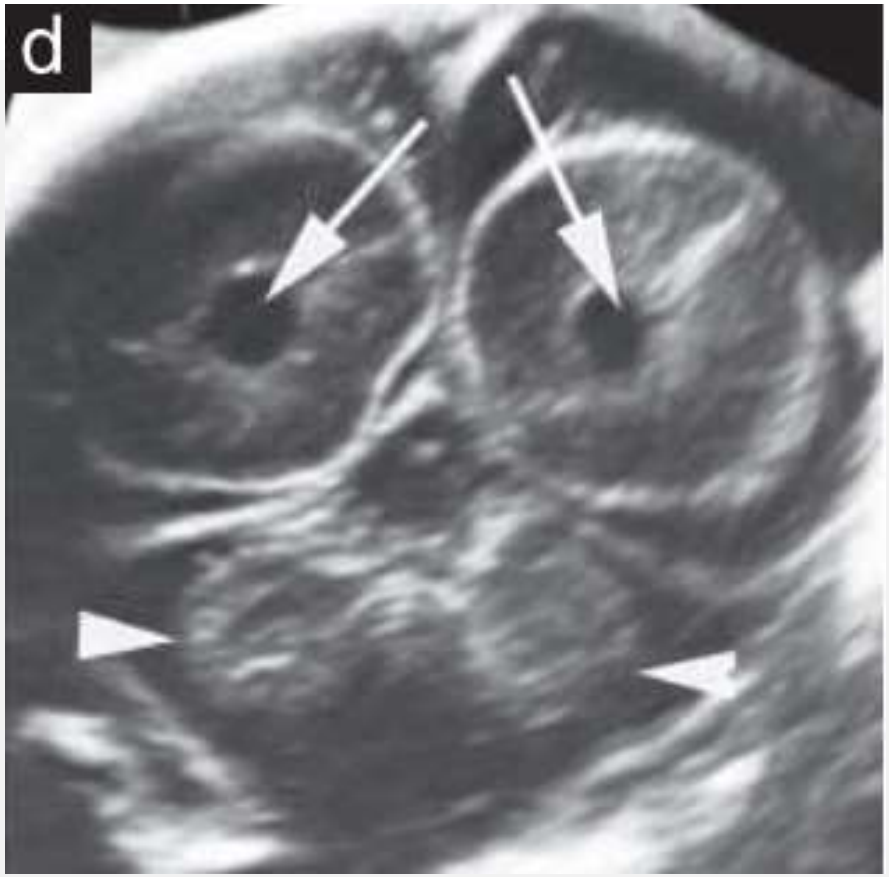

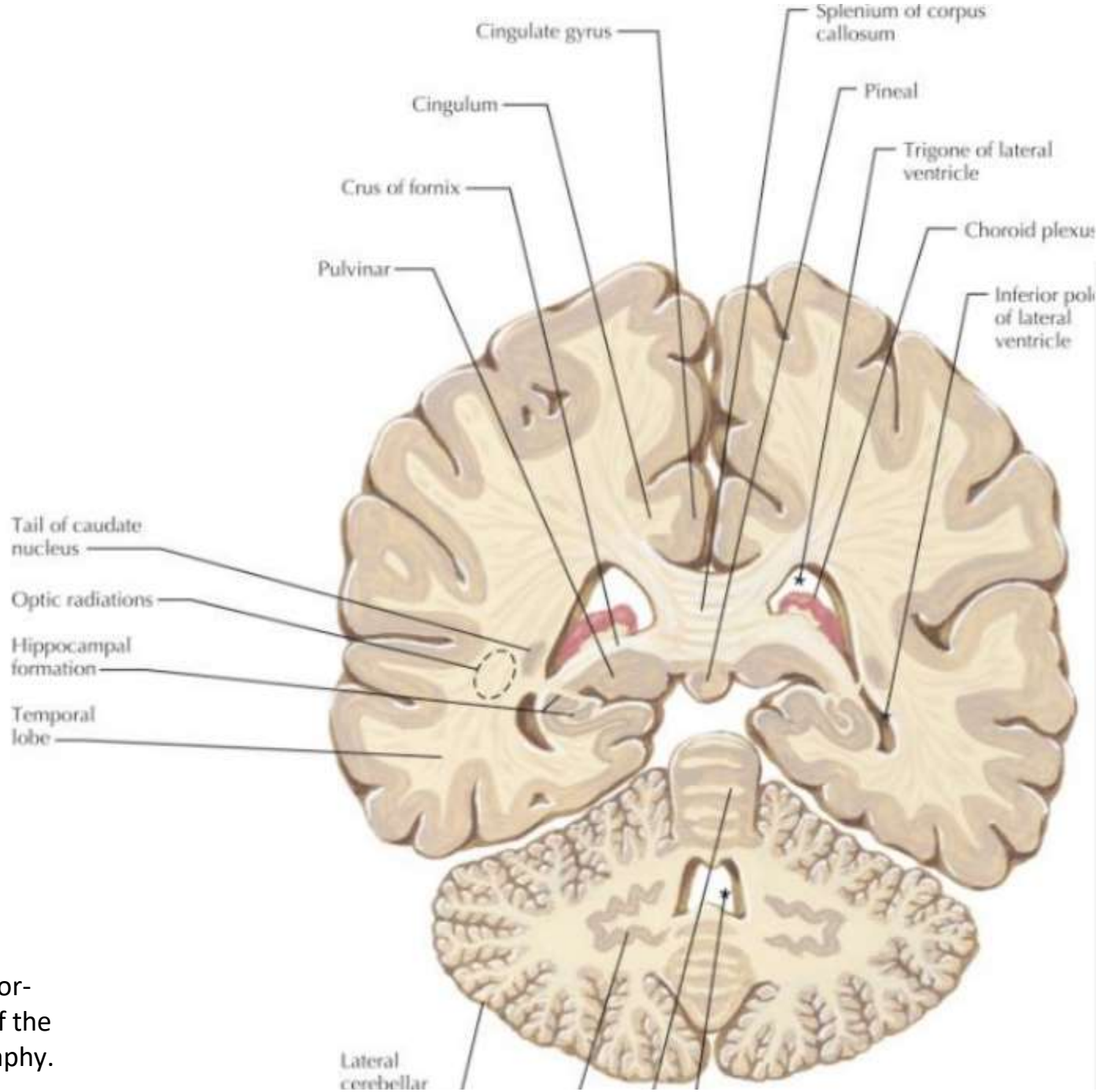

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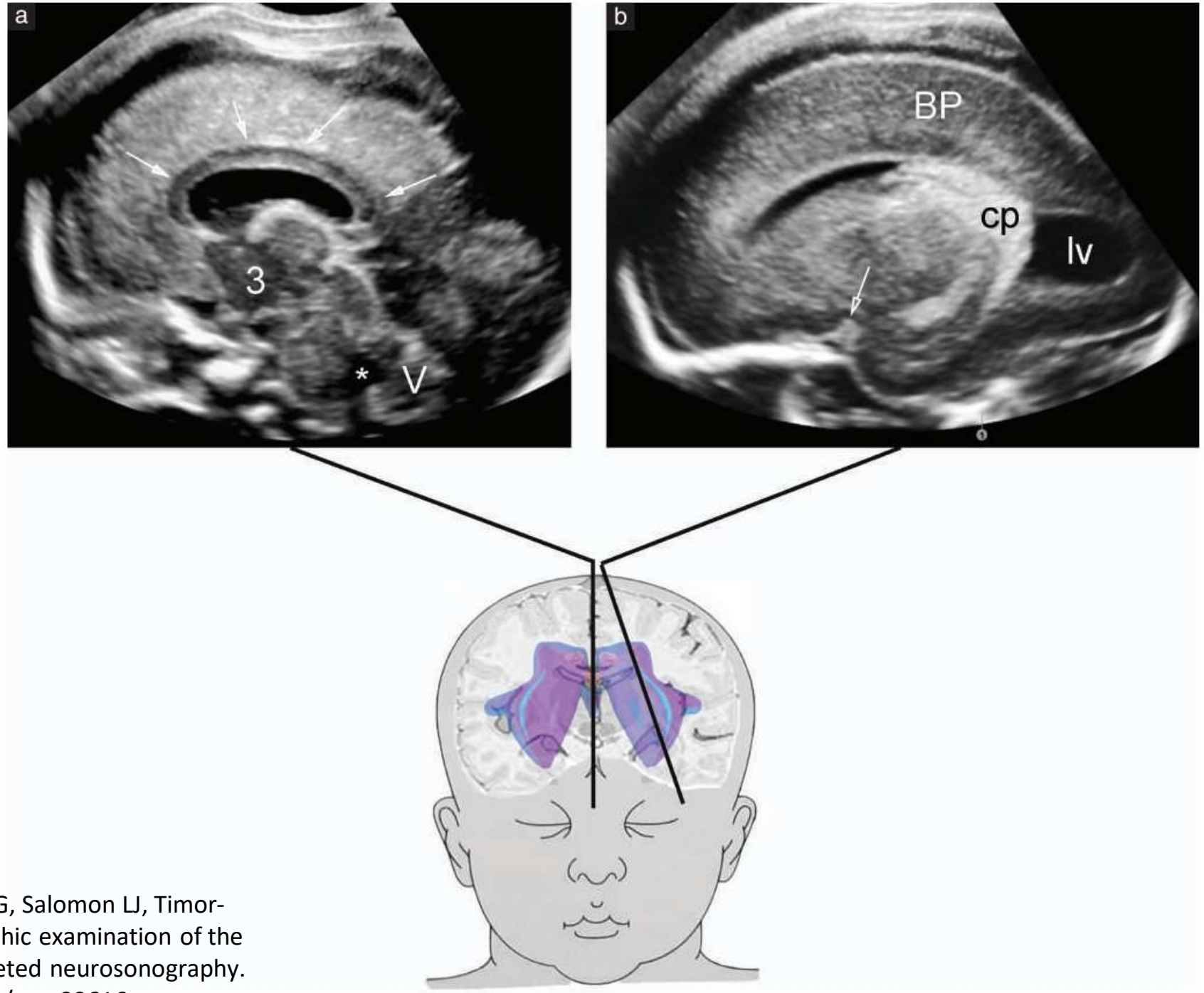

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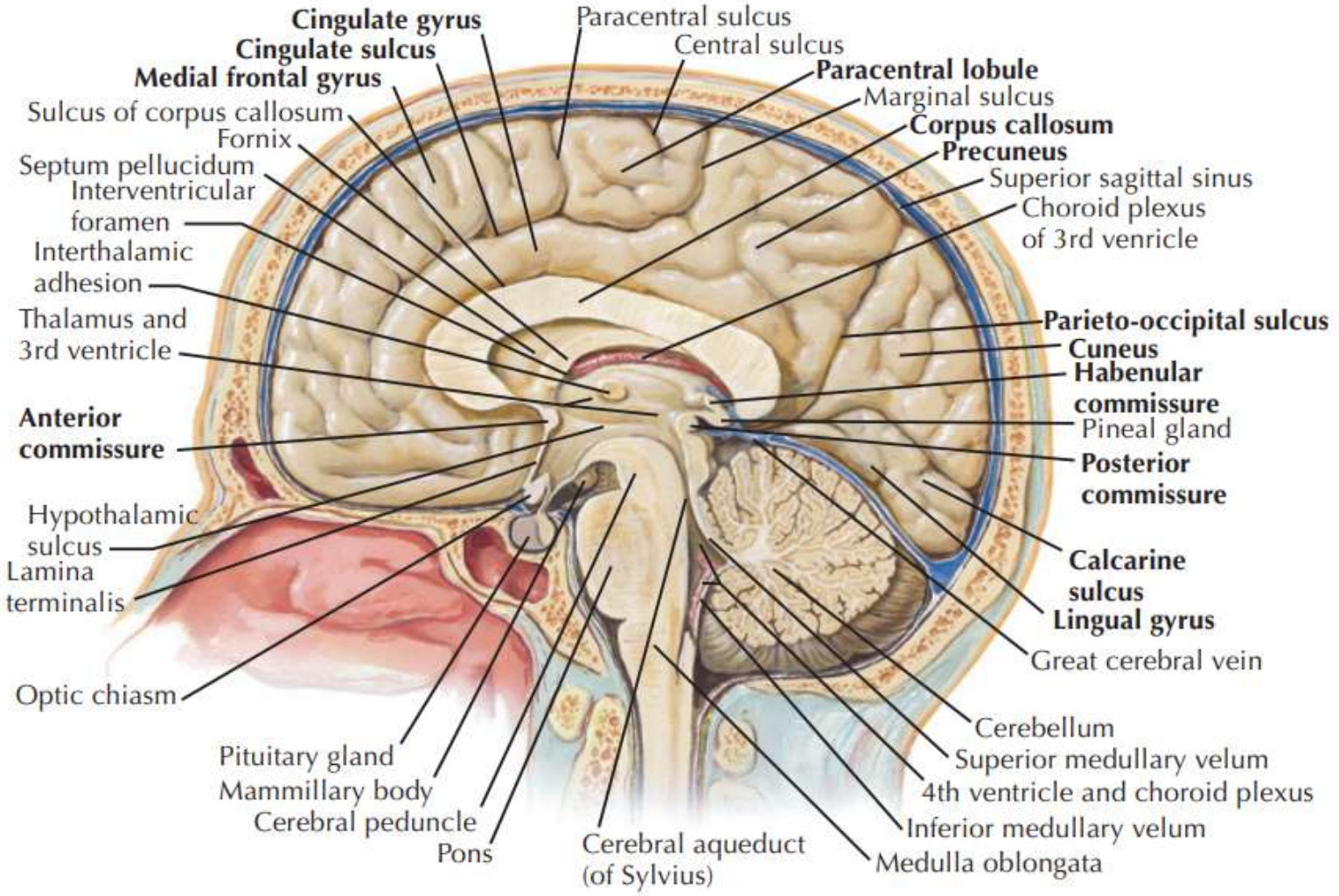

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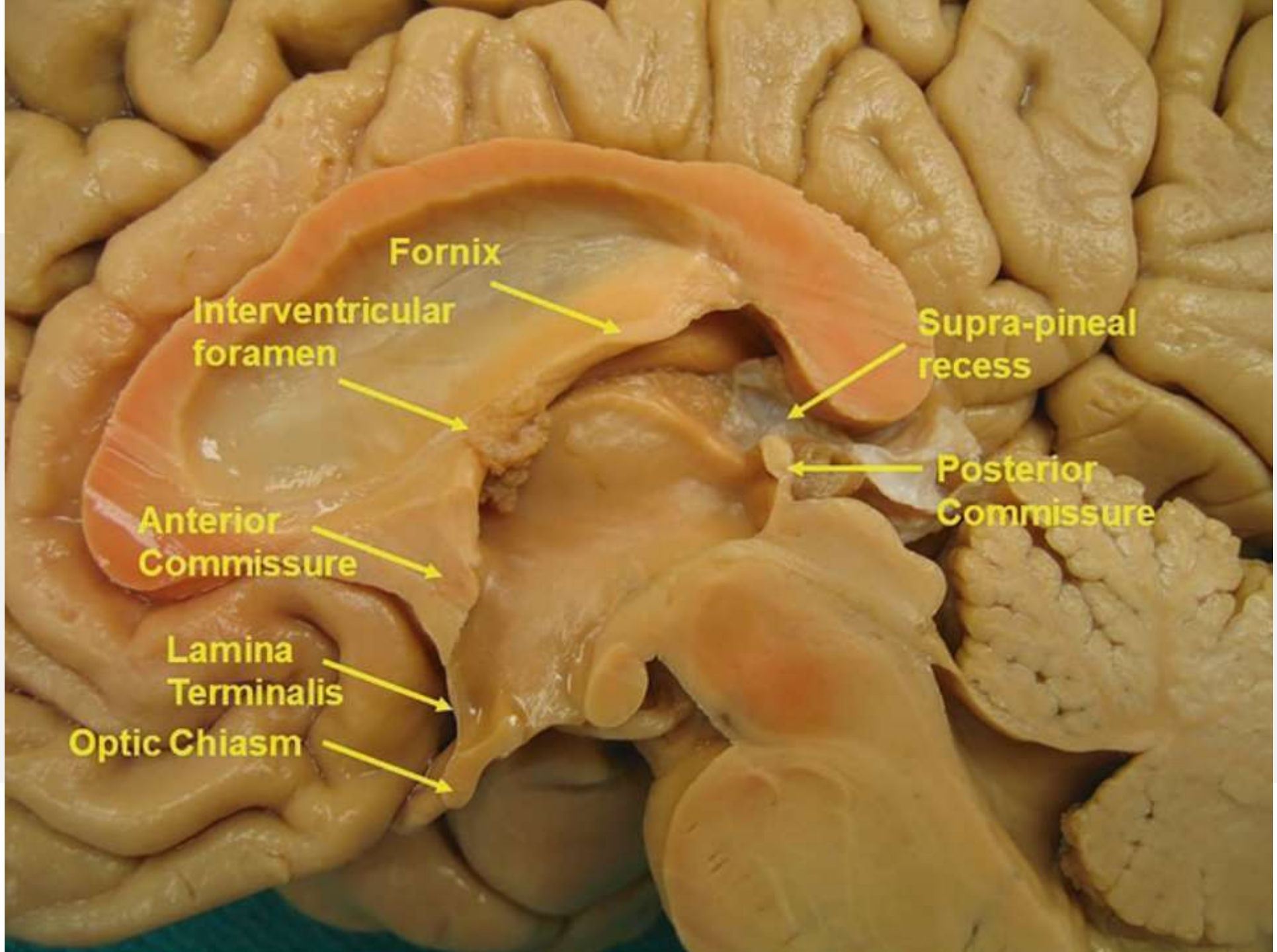
Fornix
Interventricular foramen
Supra-pineal recess
Posterior Commissure
Anterior Commissure
Lamina Terminalis
Optic Chiasm

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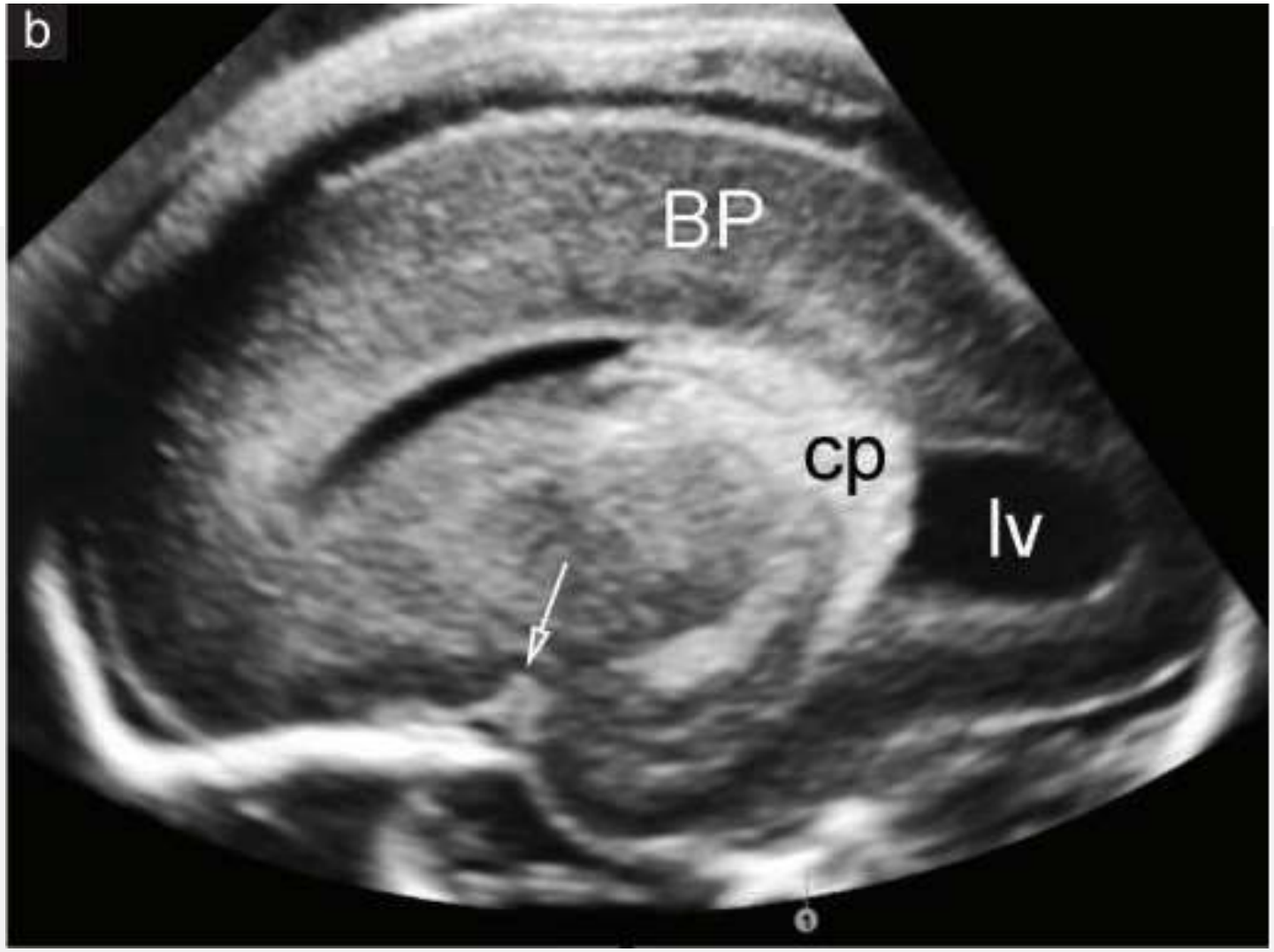

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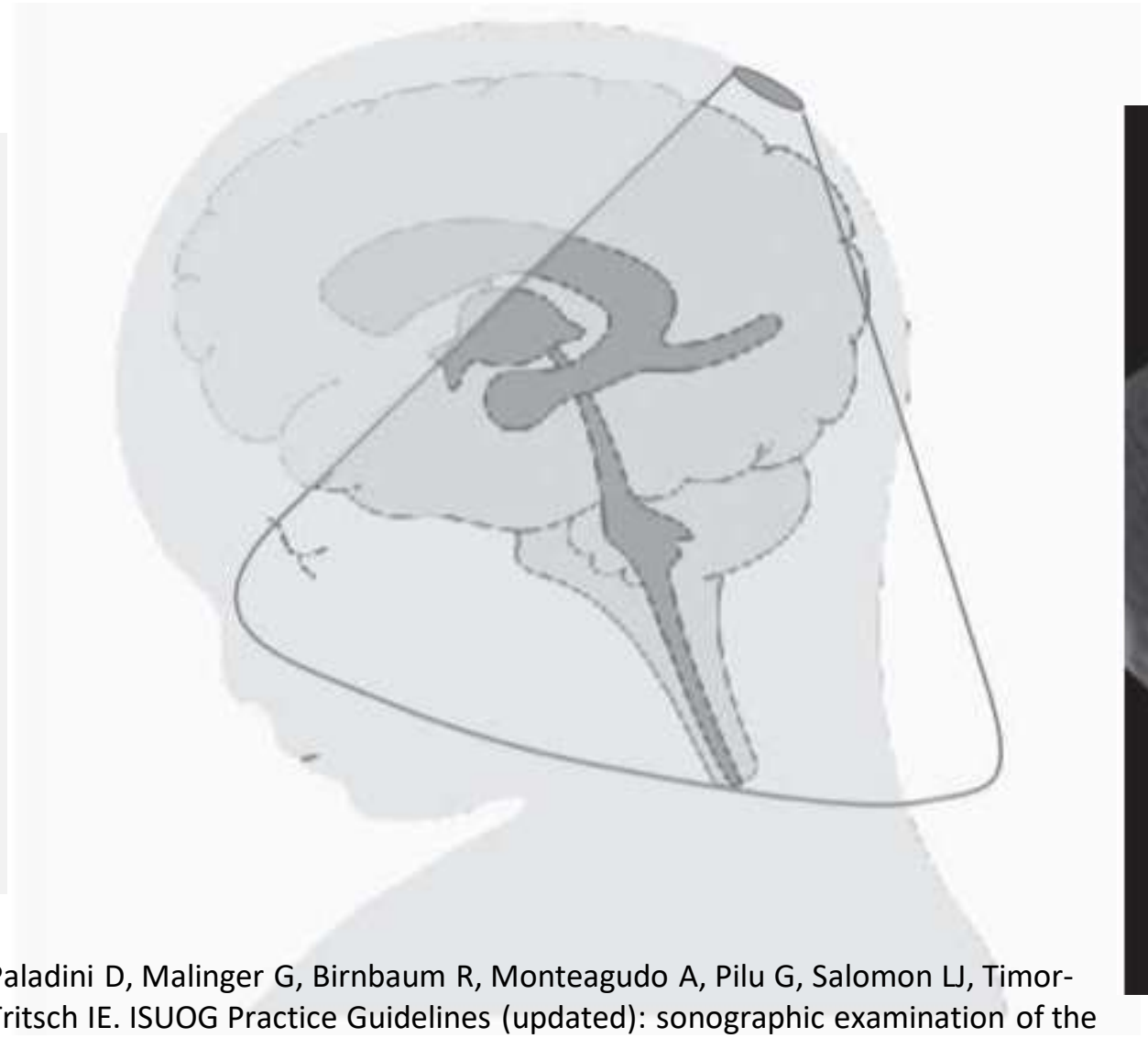

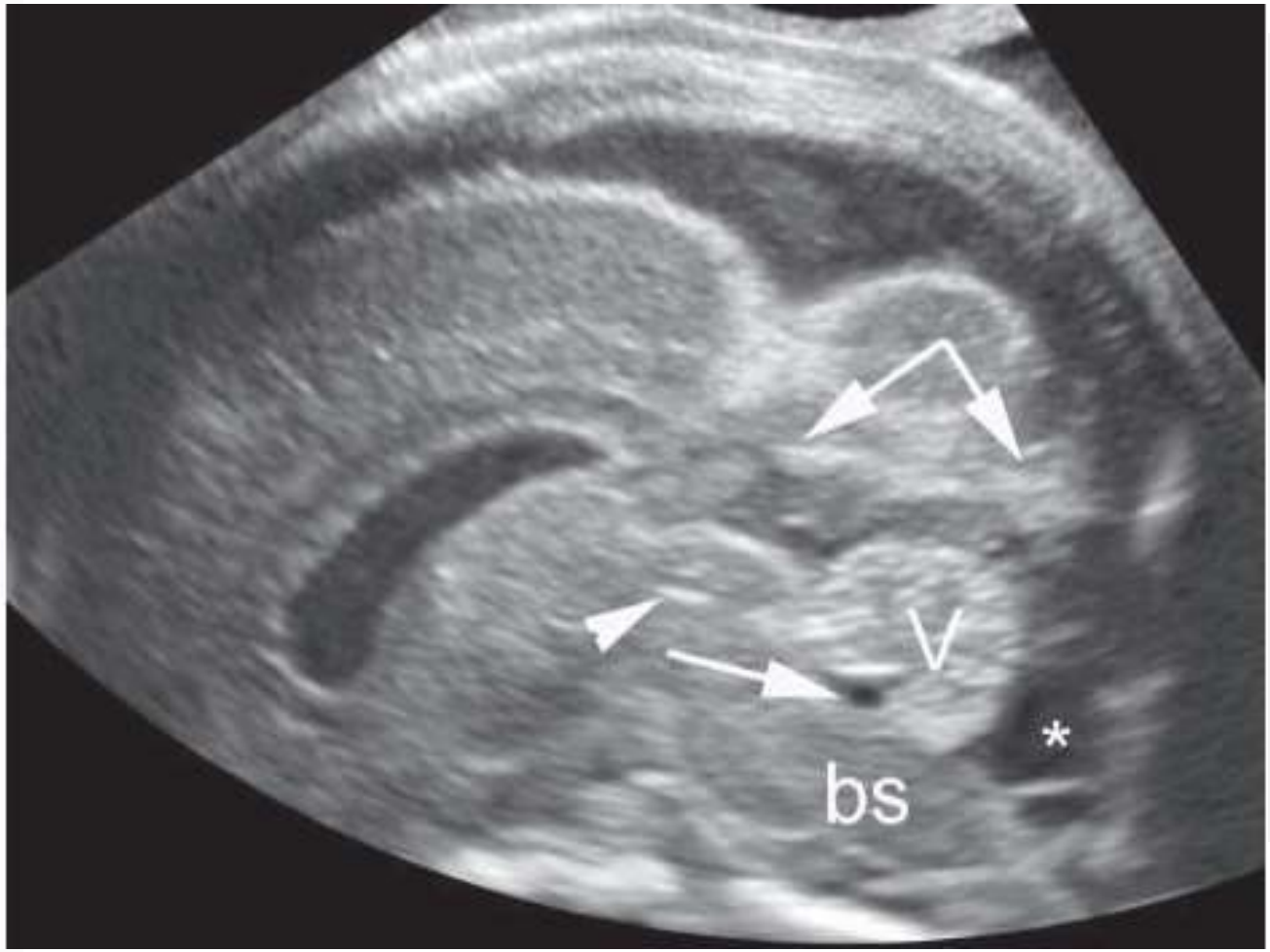

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