

Anomalías Callosas

diagnóstico y asociaciones

XII Jornada de Investigación de SADIPT



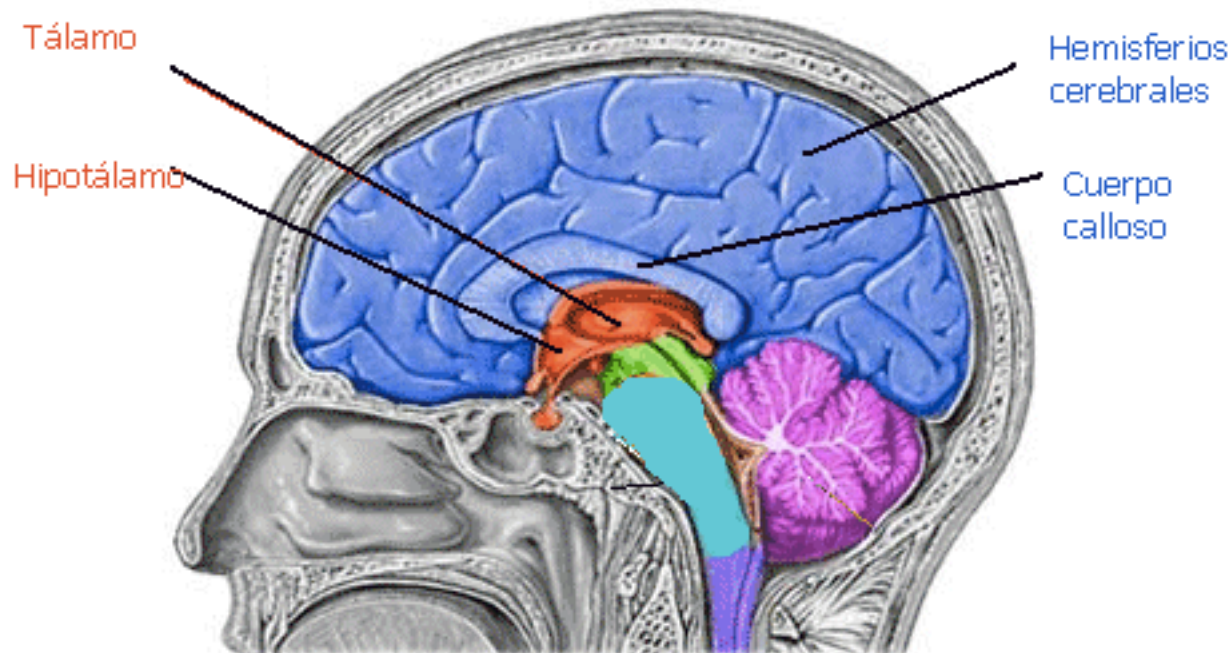
UNIDAD DE
MEDICINA
FETAL

HOSPITAL UNIVERSITARIO AUSTRAL

Josefina Maquieira
maquieirajosefina@gmail.com

Telencéfalo

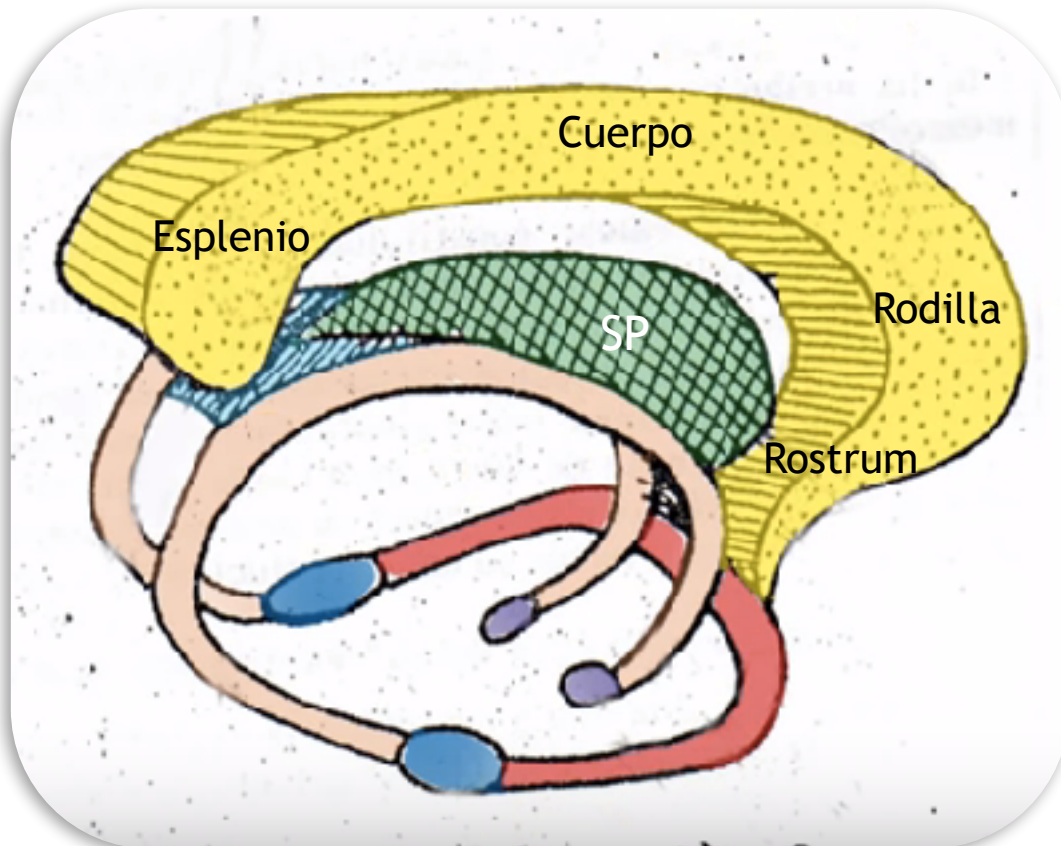
- Hemisferios cerebrales + comisuras interhemisféricas



■ Médula espinal	■ Metencéfalo o cerebelo	■ Diencefalo
■ Mielencéfalo o Bulbo raquídeo	■ Mesencéfalo	■ Telencéfalo

El Cuerpo Calloso

- Comisuras interhemisféricas de sustancia blanca
- Pico/Rostrum - Rodilla/Genu - Cuerpo - Rodete/Esplenio



Cuerpo: anterior, medio (anterior y posterior), istmo

CC

Trígono

Septum pellucidum

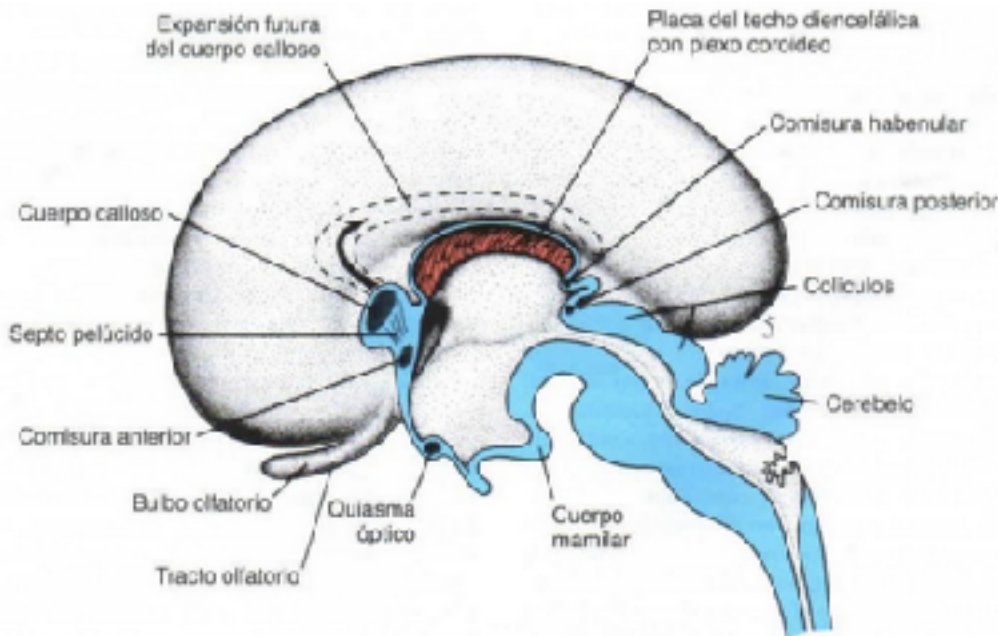
A Pilares anteriores (tuberculos mamilares)

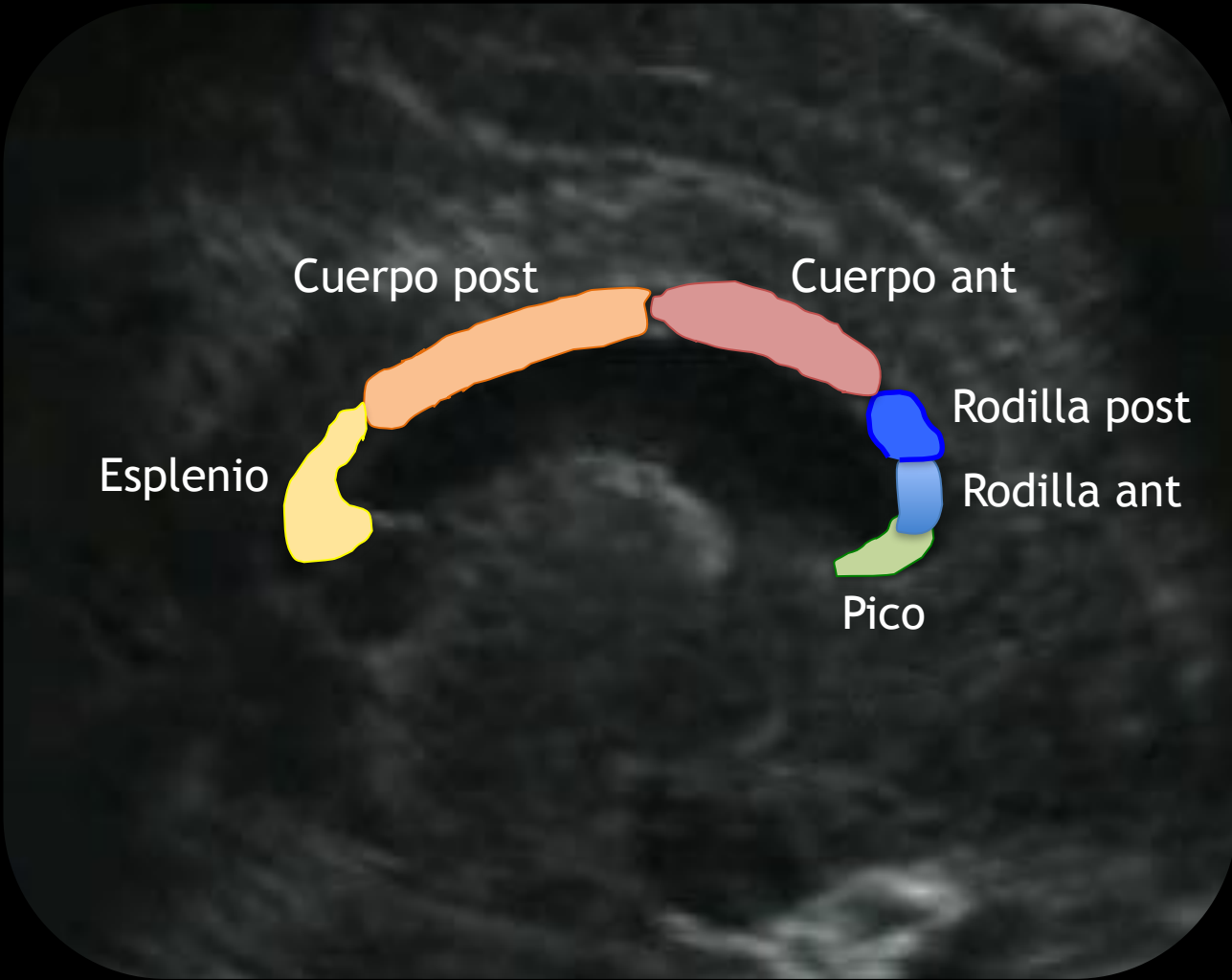
Pilares posteriores (Núcleos amigdalinos)

Comisura blanca anterior

Embriología

- Engrosamiento *lámina terminalis* del telencéfalo: sentido AP
- Comienza: 8s y completa en su extensión cráneo-caudal a las 18-19s.





Esplenio

Cuerpo post

Cuerpo ant

Rodilla post

Rodilla ant

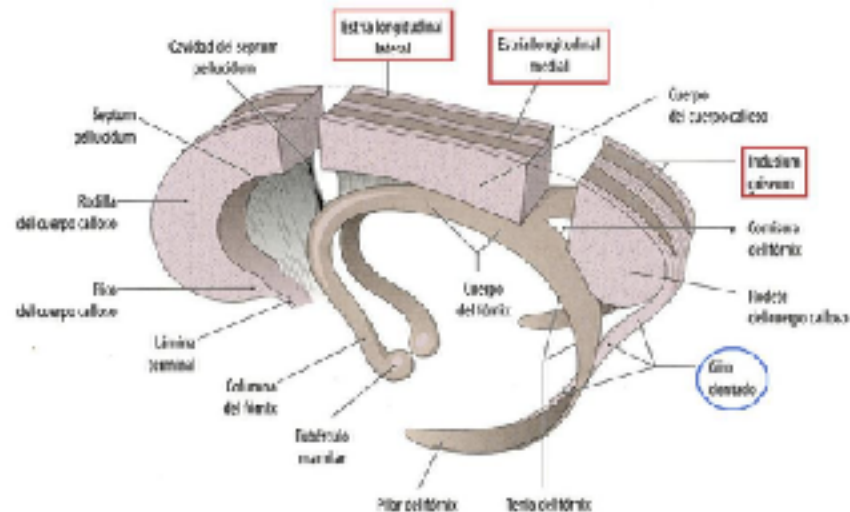
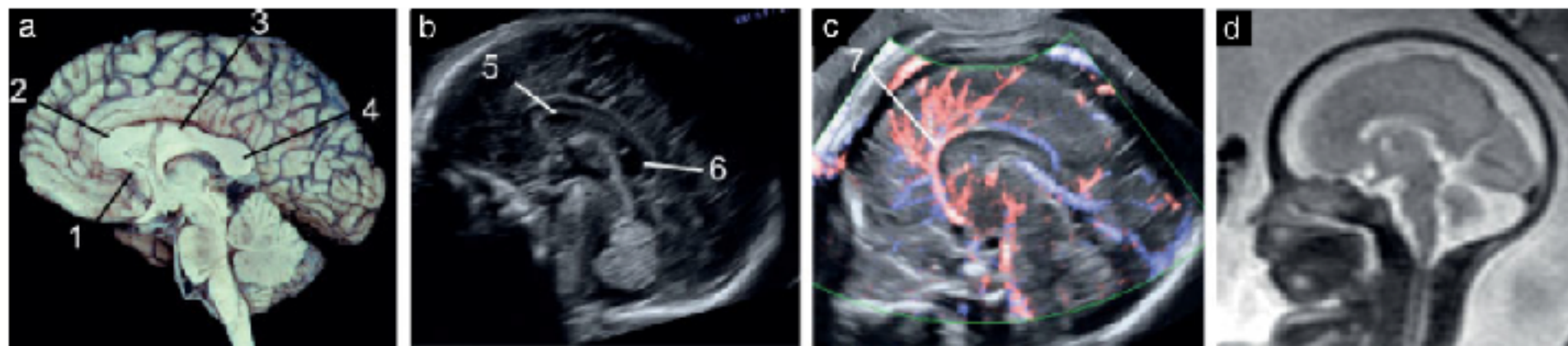
Pico

Cuerpo Calloso: *hallazgos radiológicos normales*

AP

ECOGRAFIA

RMf



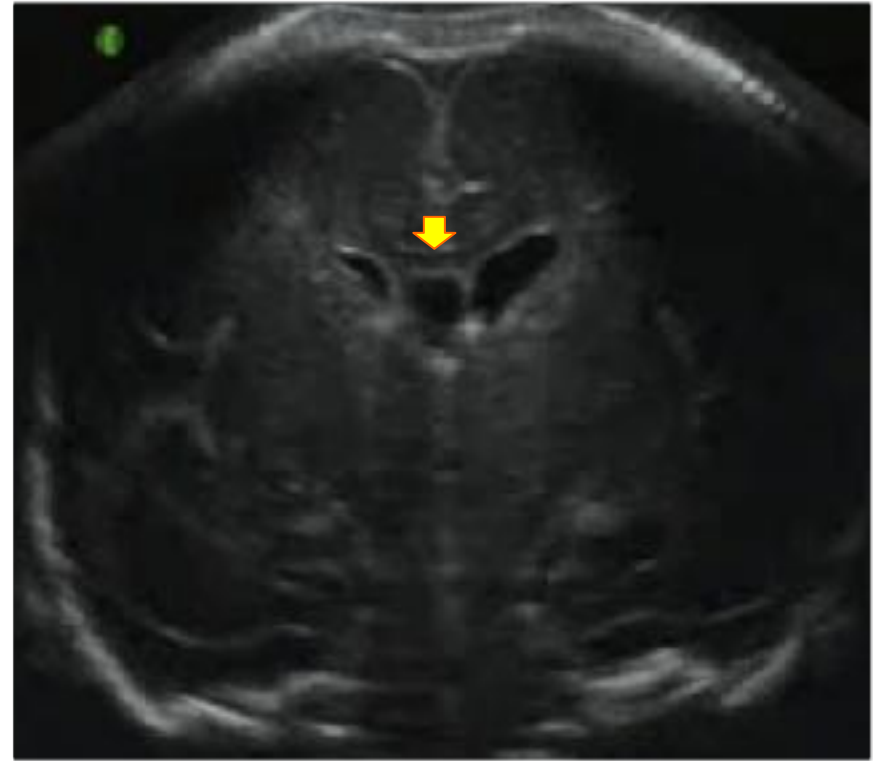
Hallazgos radiológicos NORMALES del CC

Ecografía en el planos sagital: el CC se observa como una imagen lineal hipocogénica, rodeado por imágenes lineales hiperecogénicas

SAGITAL



CORONAL

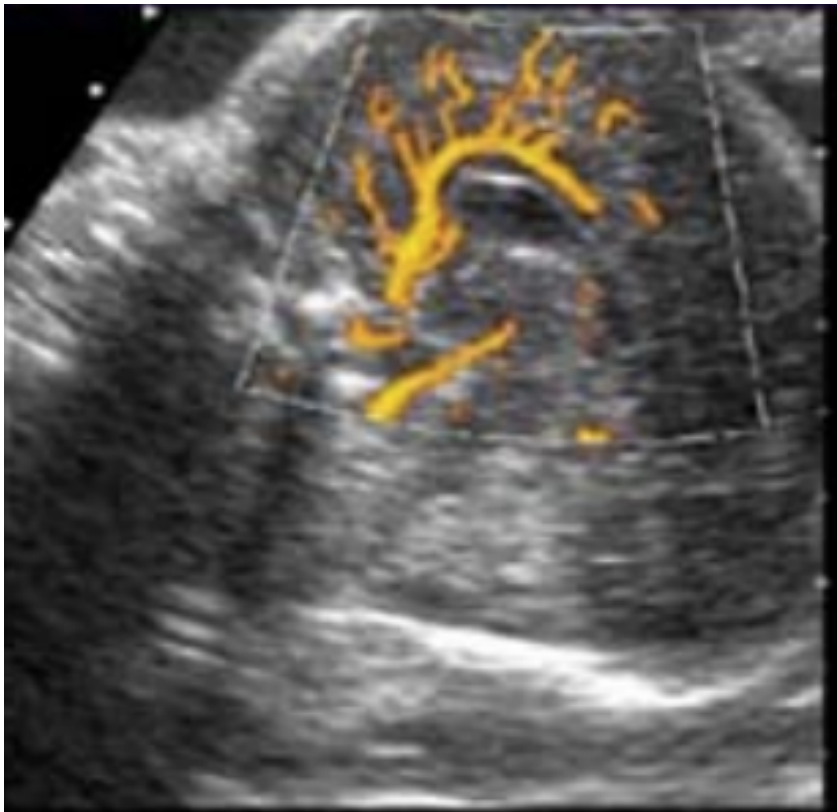


Corte transcaudal: rodilla interrumpe la fisura interhemisferica

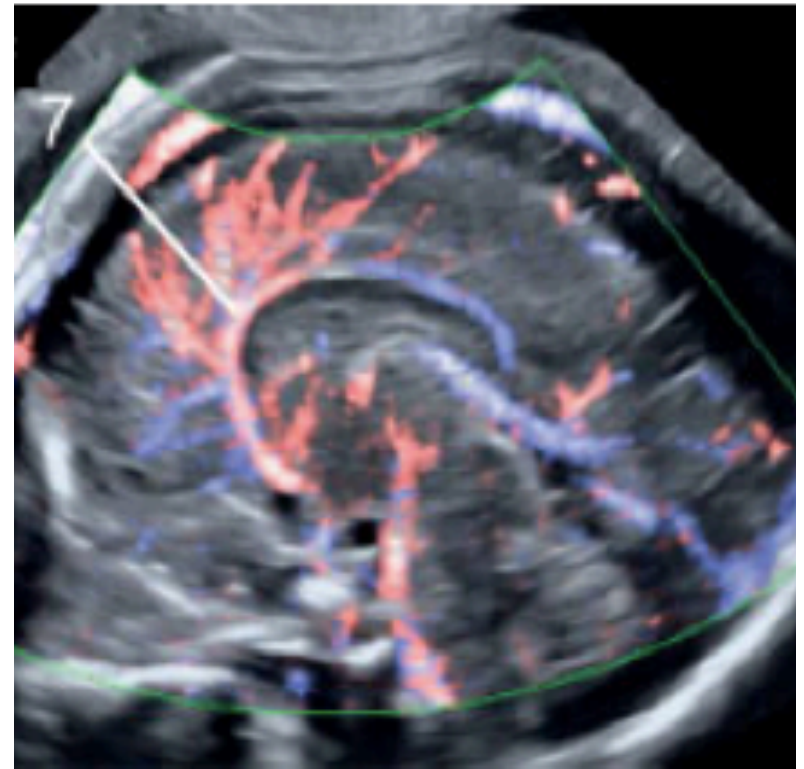
Hallazgos radiológicos NORMALES del CC

Arteria pericallosa: la evaluación vascular mediante Doppler es útil para demostrar la presencia del CC.

15s visible porción anterior



19s visible CC completo

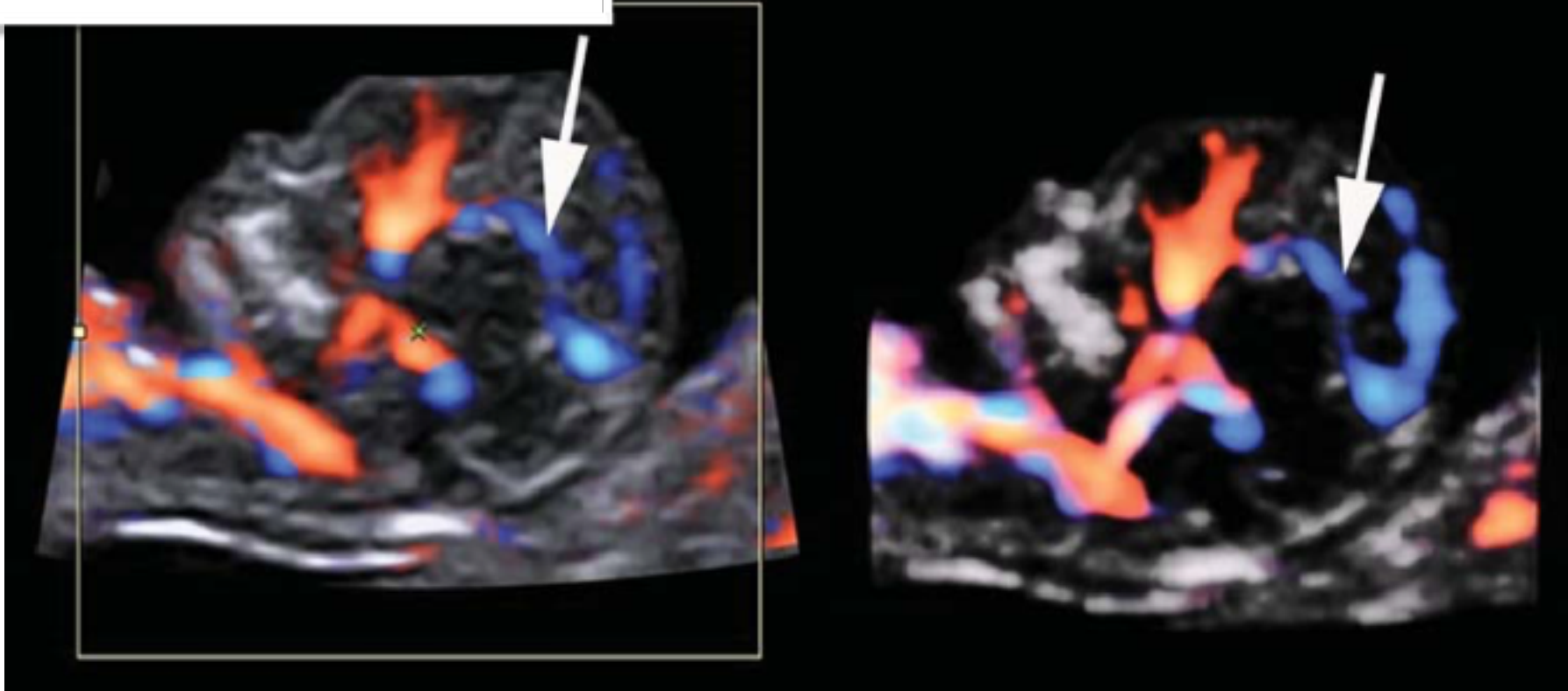


Arteria pericallosa en T1

Demonstration of the Pericallosal Artery at 11–13 Weeks of Gestation Using 3D Ultrasound

Roberto Conturso^a, Elena Contro², Federica Bellussi^b, Aly Youssef²,
Giuseppina Pasella^b, Federika Martelli^{b,c}, Nicola Rizzo^b, Gianluigi Filu^b,
Tullio Ghi^b

^aDepartment of Obstetrics and Gynecology, Widened Hospital, Como; ^bDepartment of Obstetrics and Gynecology, S. Orsola-Malpighi University Hospital, University of Bologna, Bologna; and ^cDepartment of Obstetrics and Gynecology, University of Tor Vergata, Rome, Italy



Secuencia T2, plano medio sagital

25s

33s

38s

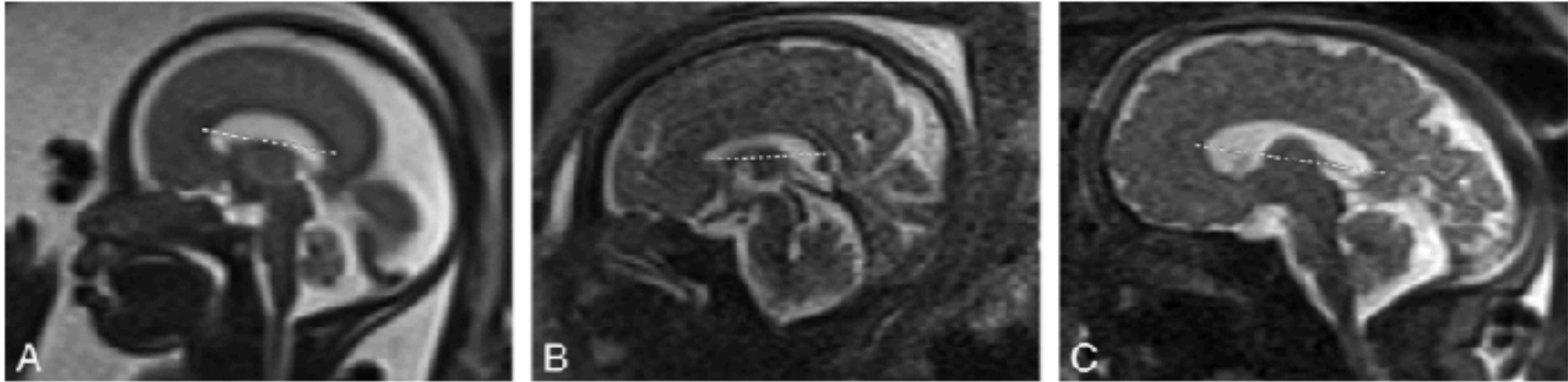


Fig 1. Sagittal MR image of the fetal brain (TE_{eff} , 50–100 ms; NEX, 0.5). Measurement of the corpus callosum from anterior to posterior on fetuses of 25 (A), 33 (B), and 38 (C) weeks' gestational age.

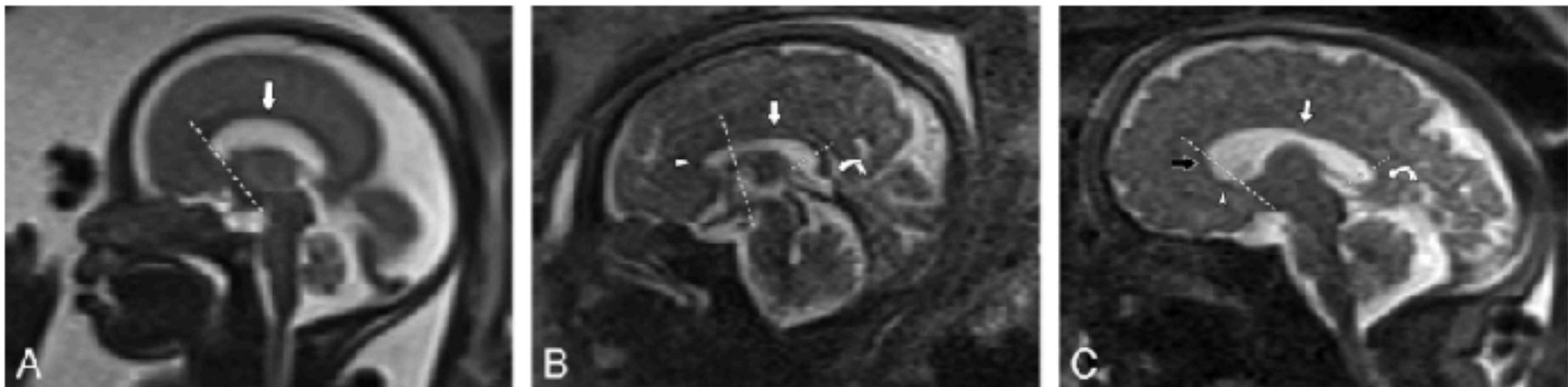
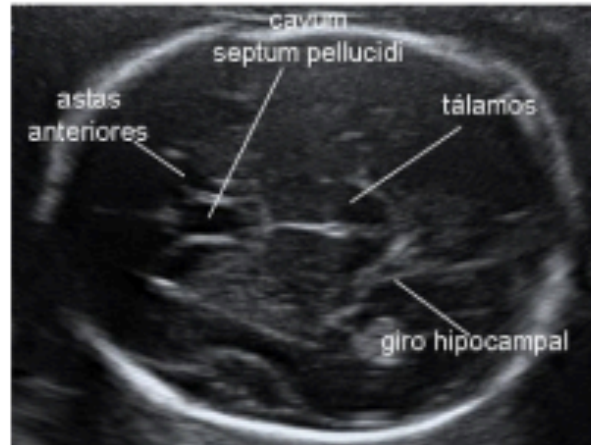
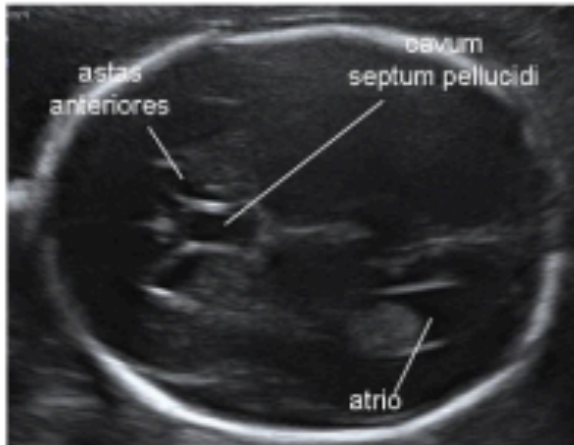


Fig 2. Sagittal MR image of the fetal brain (TE_{eff} , 50–100 ms; NEX, 0.5). Identification of the body (straight arrow), genu (open arrow), splenium (curved arrow), and rostrum (arrowhead) of the fetal corpus callosum at 25 (A), 33 (B), and 38 (C) weeks' gestational age. The genu lies anterior to an approximate line connecting the mammillary bodies, anterior commissure, and corpus callosum.¹¹ The short dotted line separates the splenium from the body.

Examen Ecográfico Básico SNC

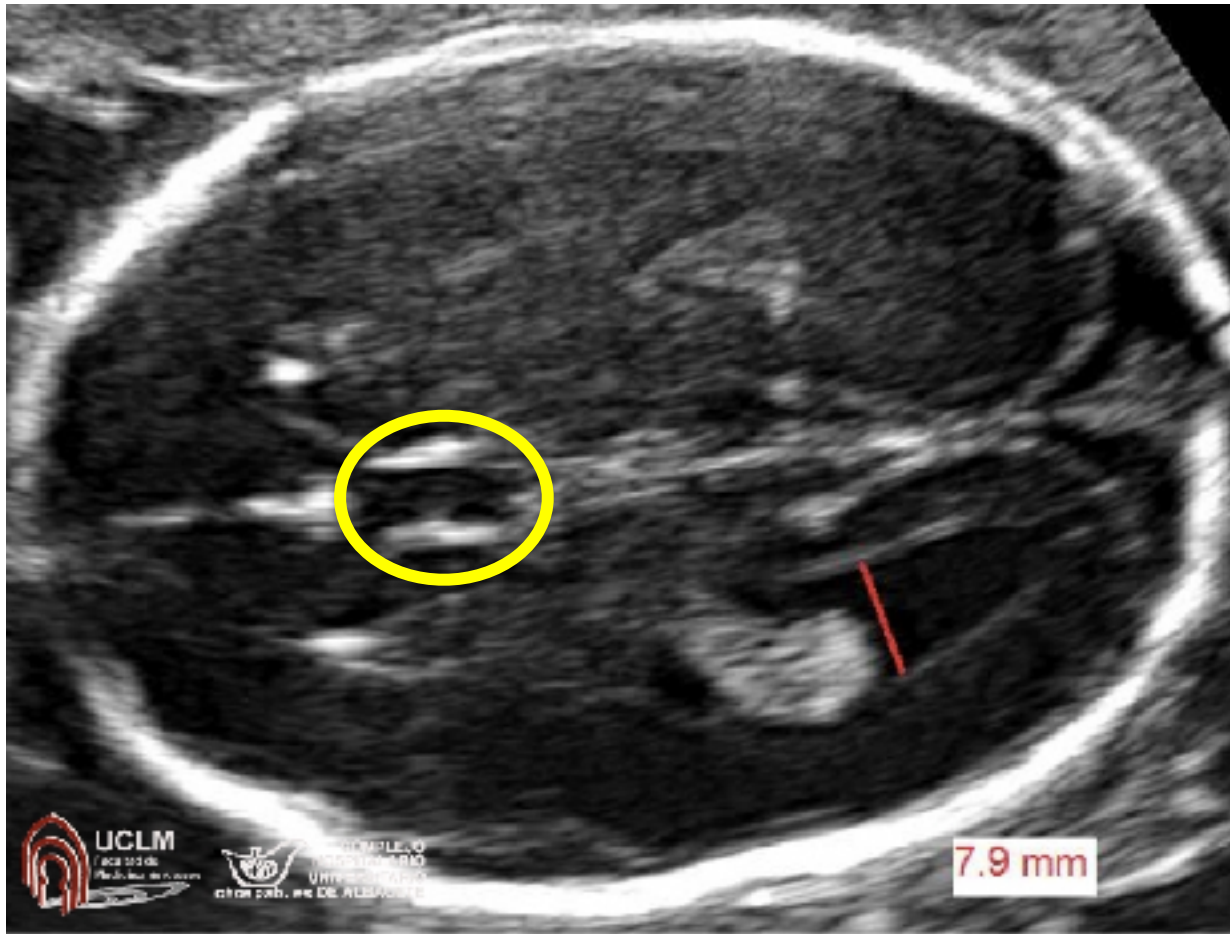
- Incluye: 3 planos
 - Transventricular
 - Transtalámico
 - Transcerebelar

CAVUM SEPTUM PELLUCIDUM

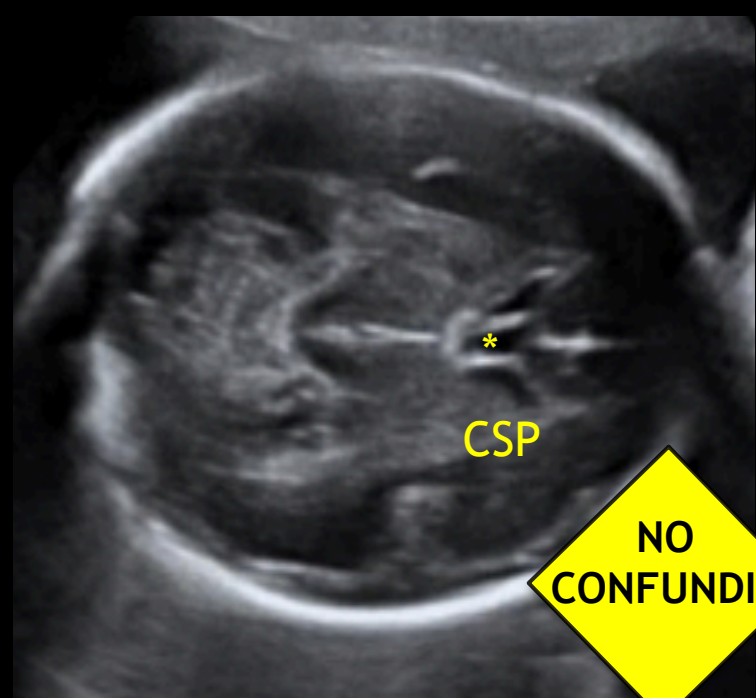
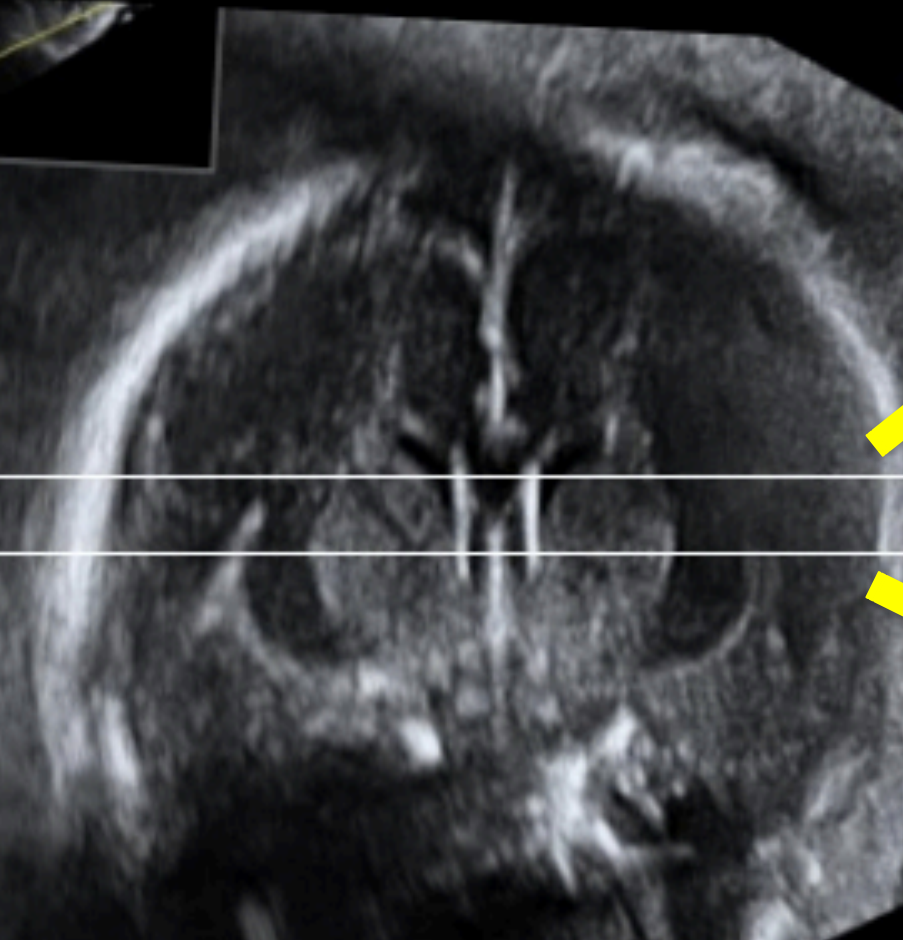


Cavum del Septum Pellucidum (CSP)

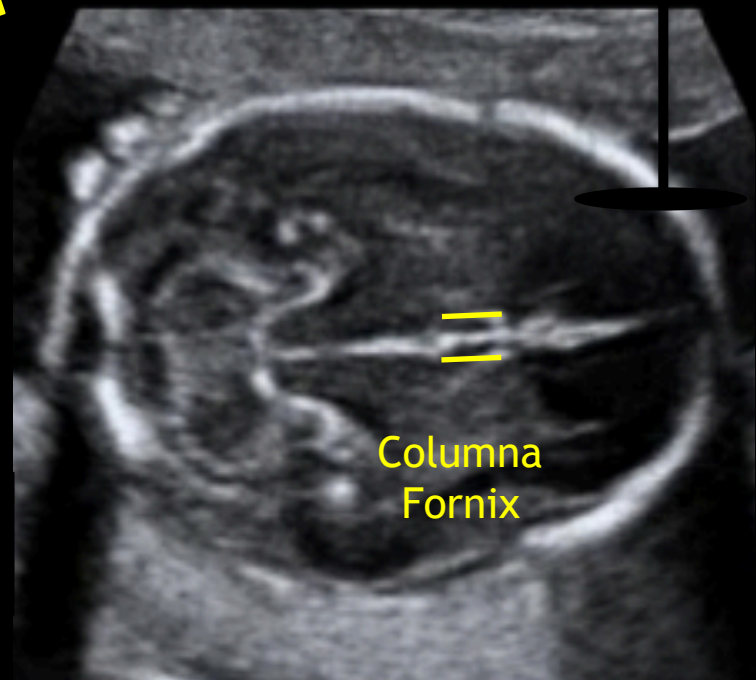
- Paredes laterales: *astas anteriores VL* y Techo: *CC*
- Puede ser visible a partir 16s (VER SIEMPRE 18-37s)



CSP y TRIGONO



**NO
CONFUNDIR**

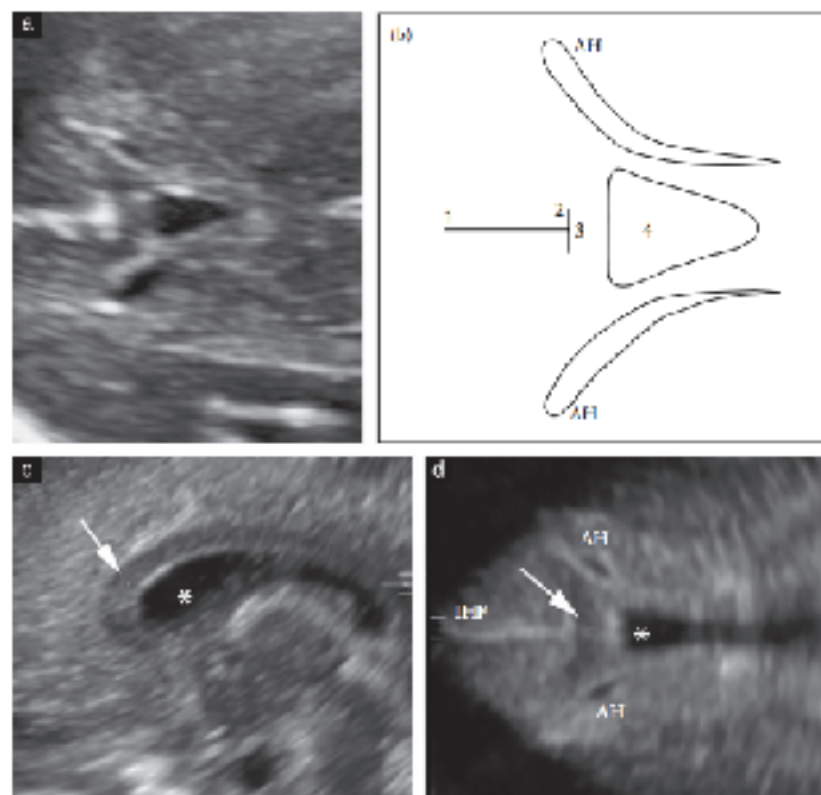


Anterior and posterior complexes: a step towards improving neurosonographic screening of midline and cortical anomalies

F. VIÑALS*, F. CORREA† and P. M. GONÇALVES-PEREIRA‡

*Centro AGB Ultrasonografía, Clínica Sanatório Alameda, Facultad de Medicina, Universidad de Concepción, Concepción, Chile; †Hospital Fernandó Fonseca, Lisboa, Portugal; ‡Neuroanatomy Department, Hospital Lusitano, Lisboa, Portugal

AC



PC

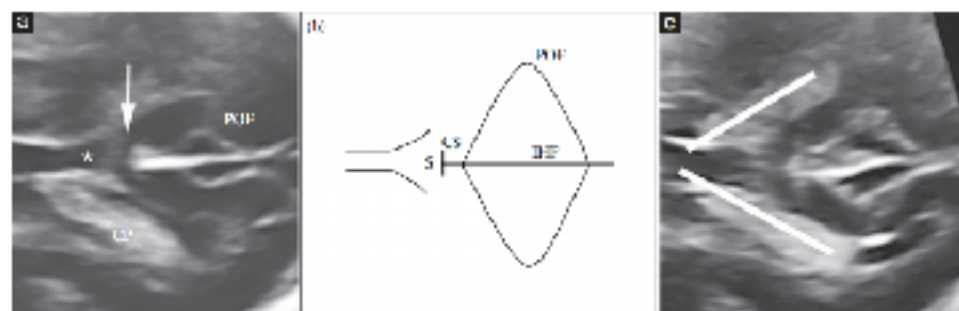
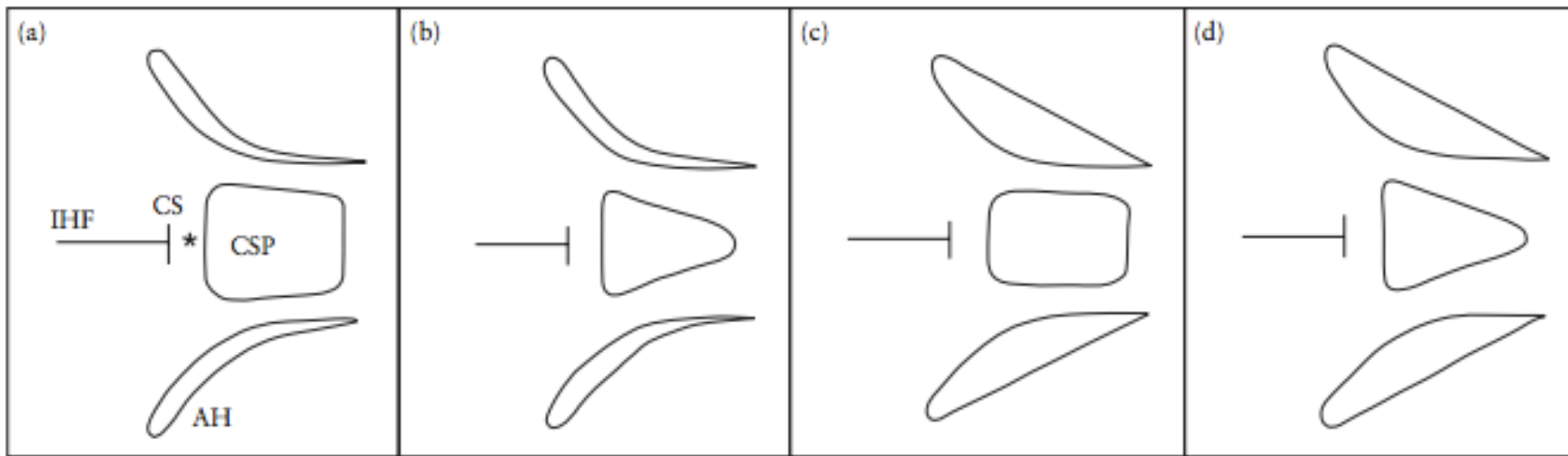


Figure 2 Ultrasound images (a,c) and diagram (b) of normal posterior complex in a 24-week fetus. Part (c) shows the oblique to midline disposition of the lateral ventricle and their choroid plexus (arrow). CP, choroid plexus; PCP, parieto-occipital fissure; S and arrow, splenium of the corpus callosum; CC, corpus callosum; PH, neurohypophyseal fissure.

73%

CSP

27%



92%

AH

18%

Neurosonografía



Coronal transcavado



Rodilla



Coronal transtálamico



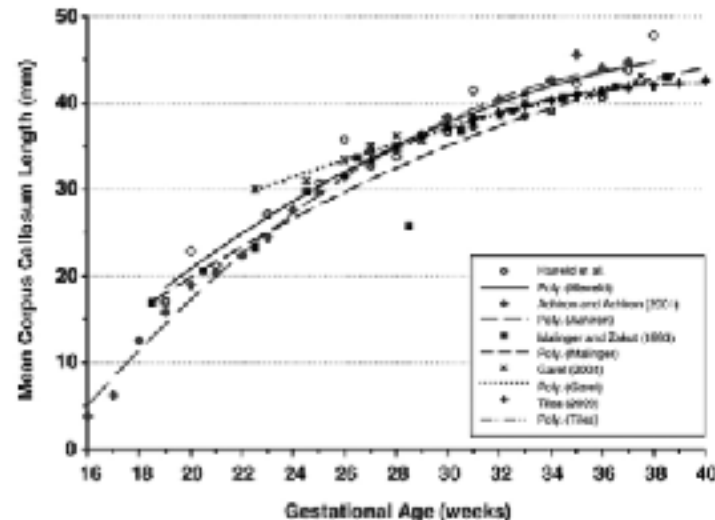
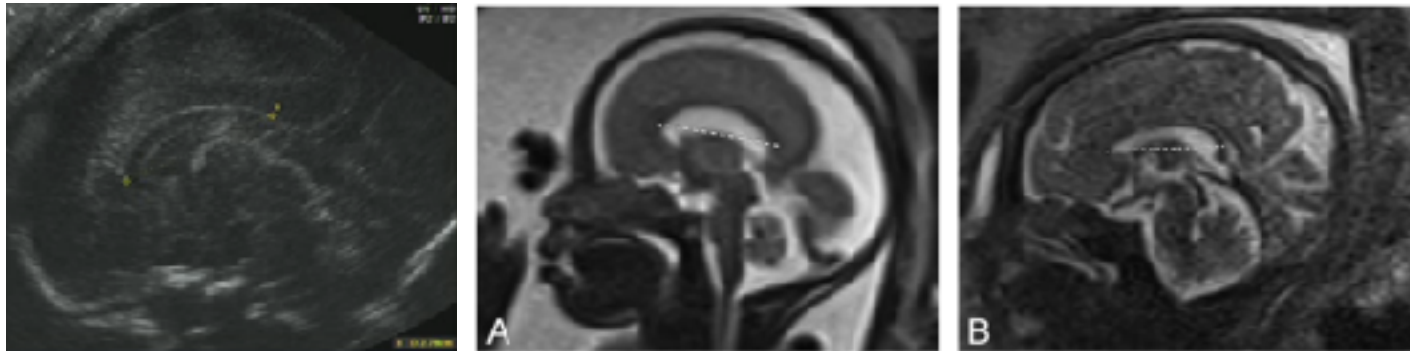
Cuerpo



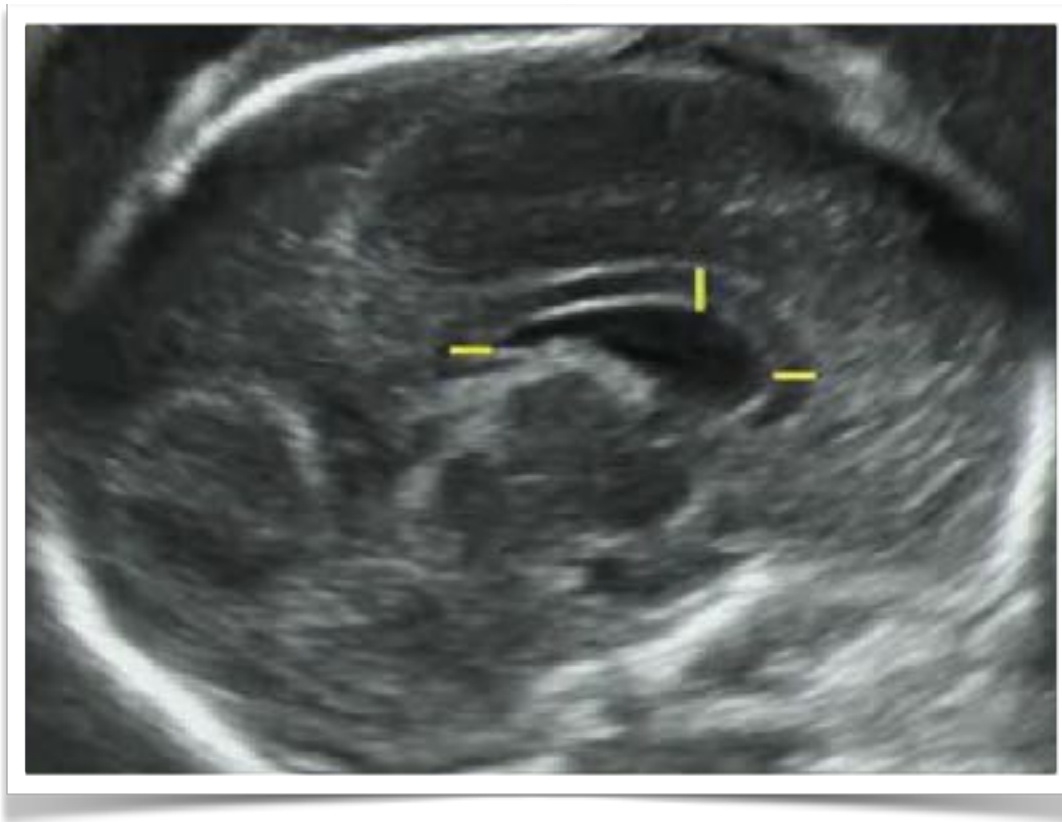
Mediosagital: CC y CSP

Crecimiento y Medicion

Corpus Callosum Length by Gestational Age as Evaluated by Fetal MR Imaging



Crecimiento y Medición



GROSOR

Anomalías del CC

PREVALENCIA

- 0.3-0.7% población general
- 2-3% población con retraso mental
 - Agenesia Parcial: 0,4/10000 RNV
 - Agenesia Completa: 1,4/10000 RNV
- >M:F

Anomalías del CC

CLASIFICACION

- **Hipoplasia:** adelgazamiento con diámetro AP normal
- Hiperplasia
- **Agenesia**
 - Completa: CACC
 - Parcial: PACC (35%)
- **Disgenesia / hipogenesia:** ACC parcial por desarrollo incompleto

Anomalías del CC

- AISLADAS
- ASOCIADAS
 - Síndromes génicos
 - Cromosomopatías (trisomías)
 - Anomalías cerebrales y extracerebrales
 - otras: infecciones - teratógenos

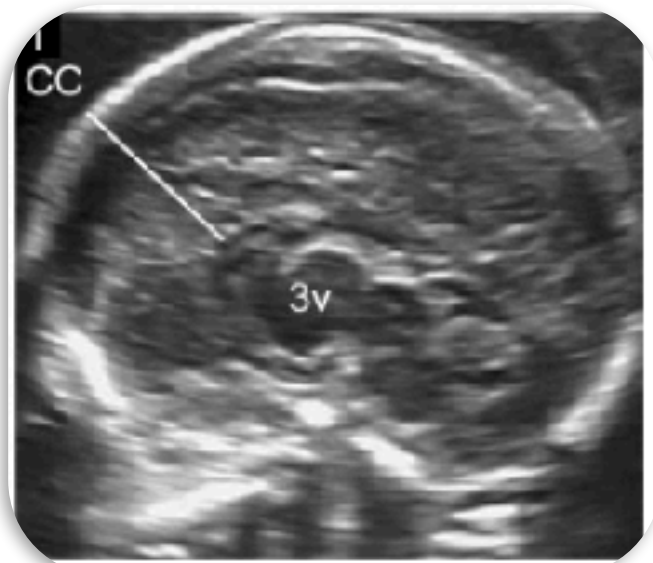
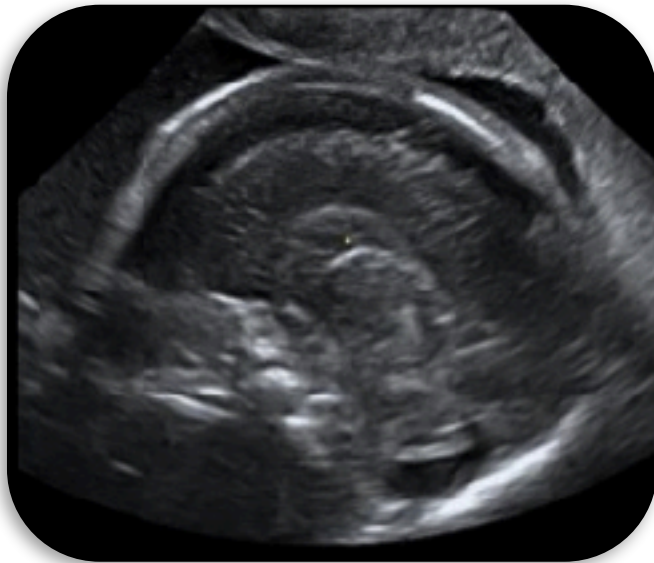
Etiología

Un evento malformativo o agudo (isquémico/
infeccioso)

Temprano | Tardío

CACC

PACC: pico / esplenio

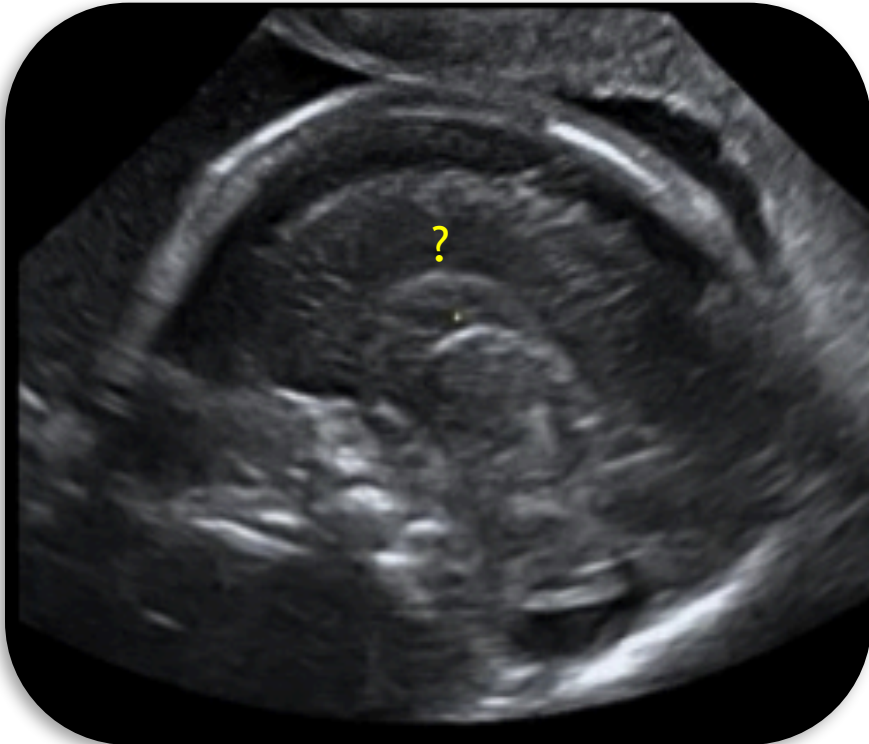


DIAGNOSTICO

ACC: signos directos

- No visualización del CC

SAGITAL 20s



SAGITAL 22s



ACC: signos directos



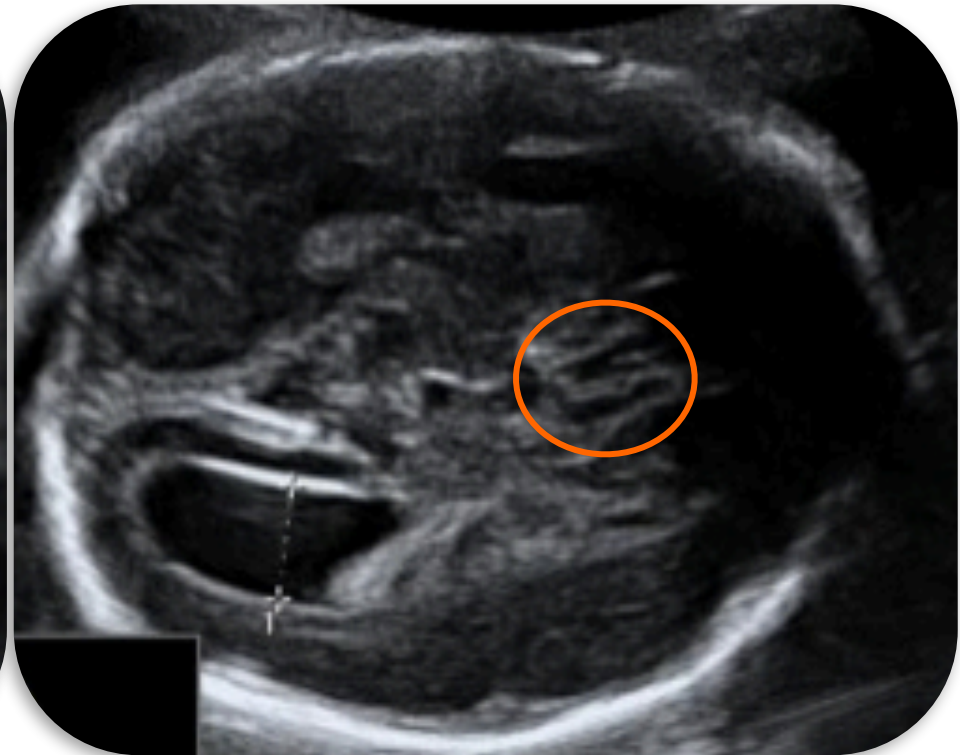
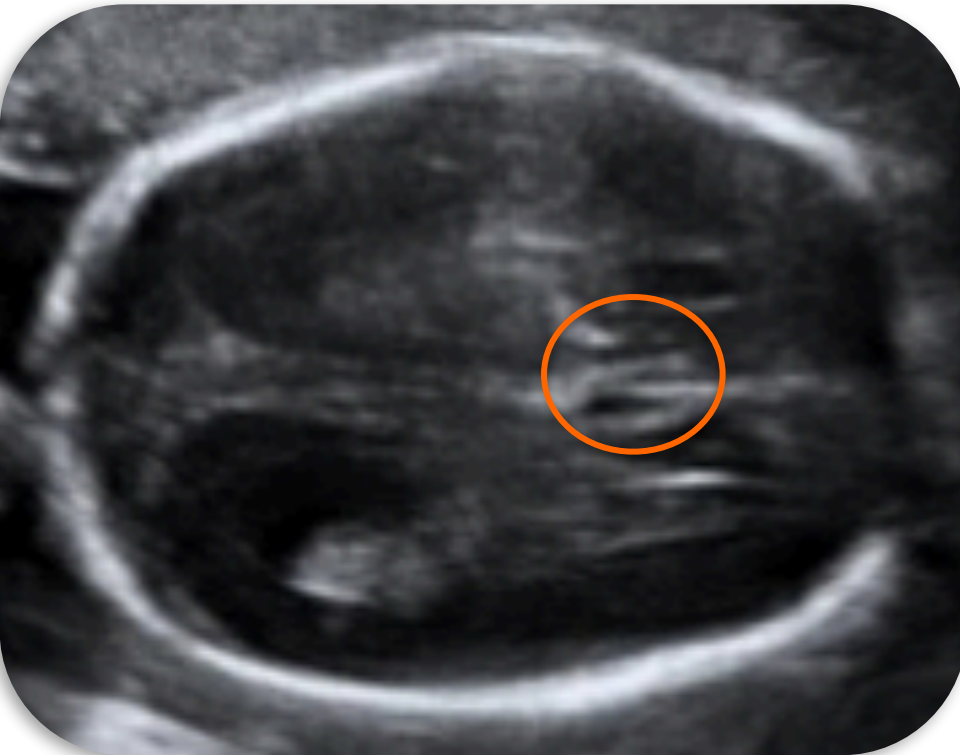
Signos indirectos ACC

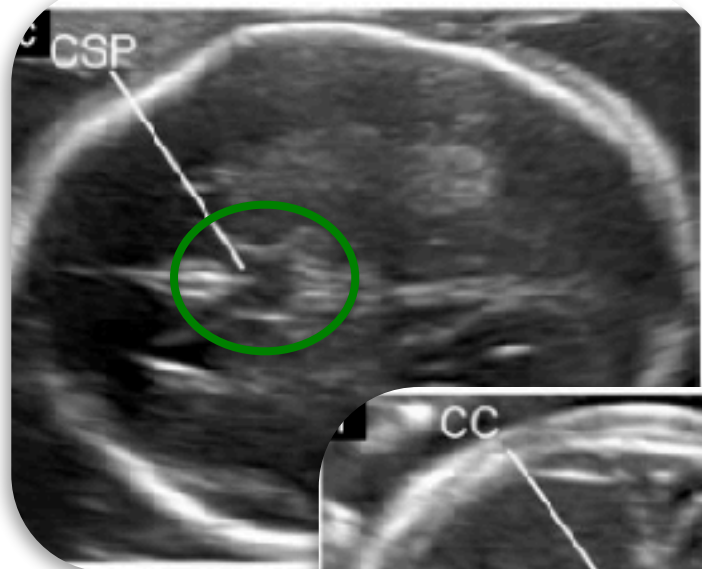
- Ausencia del CSP
- Anomalías del SV
- Distribución anormal Arteria Pericallosa
- Engrosamiento de la fisura interhemisférica
- Disposición radiada de los surcos de la convexidad

ACC: signos indirectos

Ausencia del CSP

- No específico de ACC (HPE - hidrocefalia - DSO - esquinencefalia - encefalocele - porencefalia - hidranencefalia)
- En agenesia parcial mayormente visible

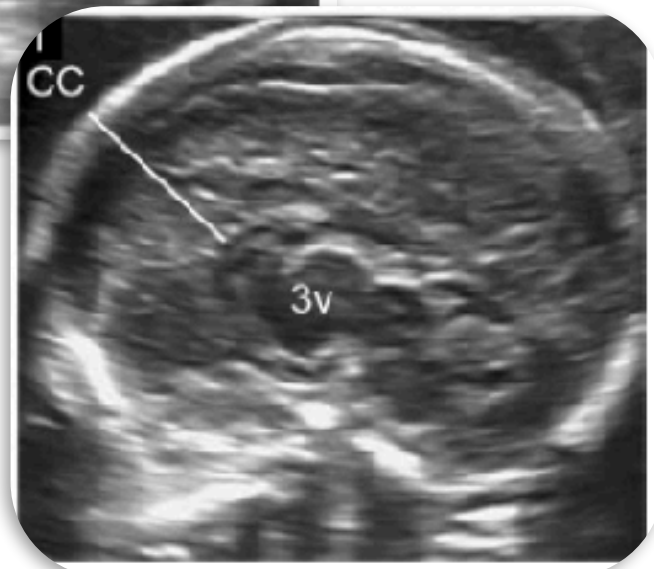
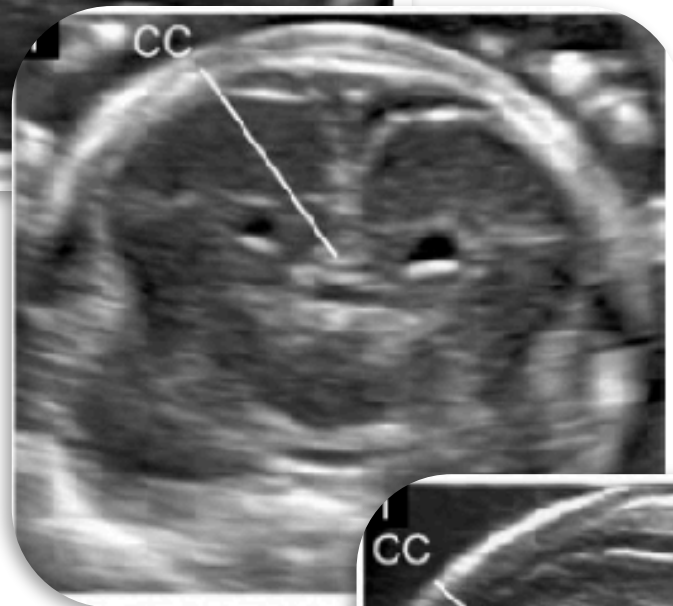




PACC: CSP presente habitualmente



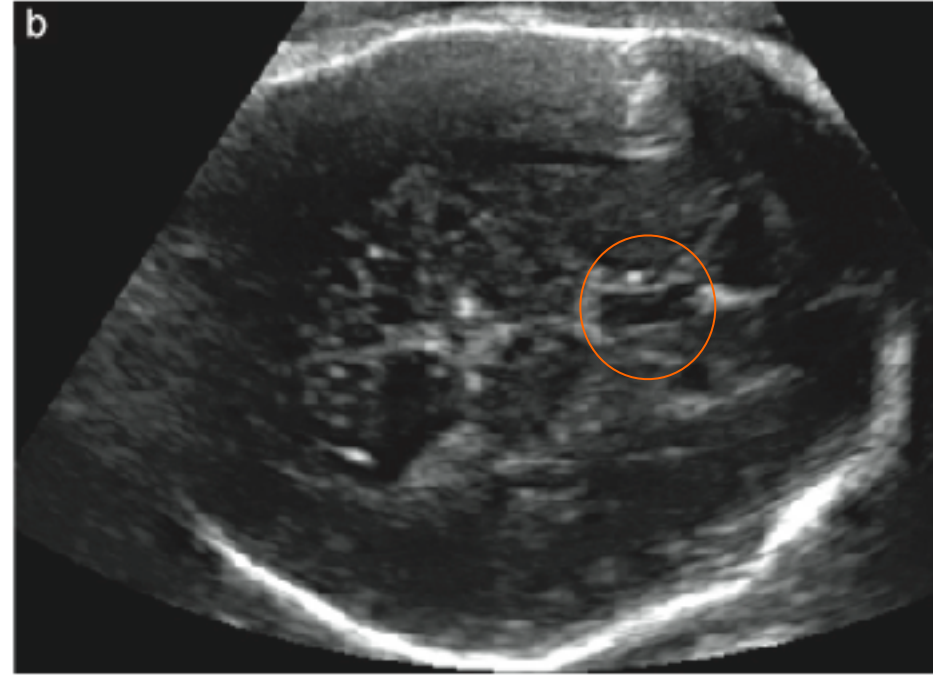
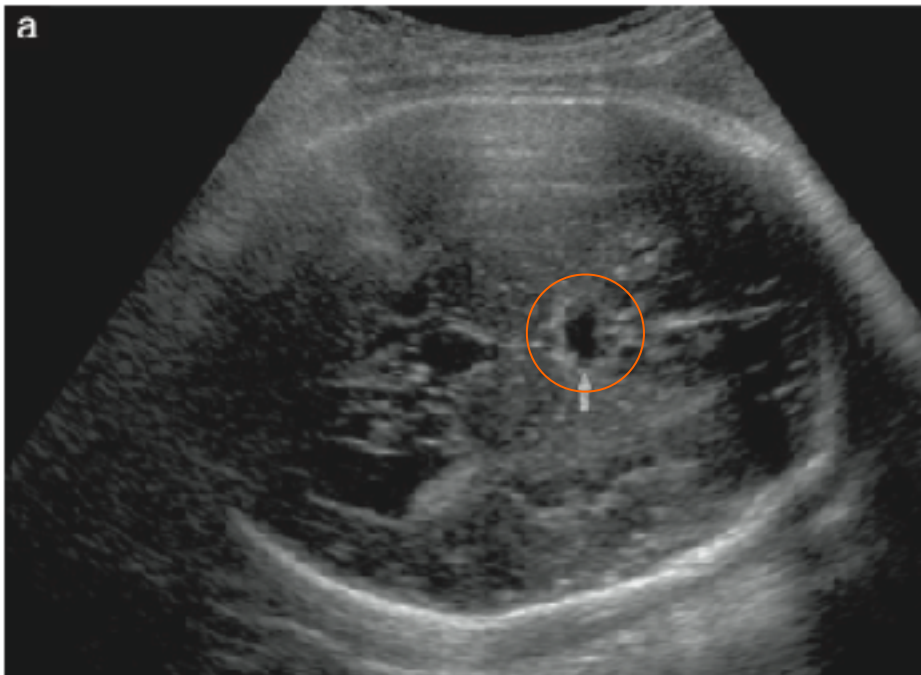
DISMORFICO???



Abnormal shape of the cavum septi pellucidi: an indirect sign of partial agenesis of the corpus callosum

O. SHEN^{*}, A. B. GELOT[†], M. L. MOUTARD[†], J. M. JOUANNIC[§], H. Y. SELA^{*} and C. GAREL[¶]

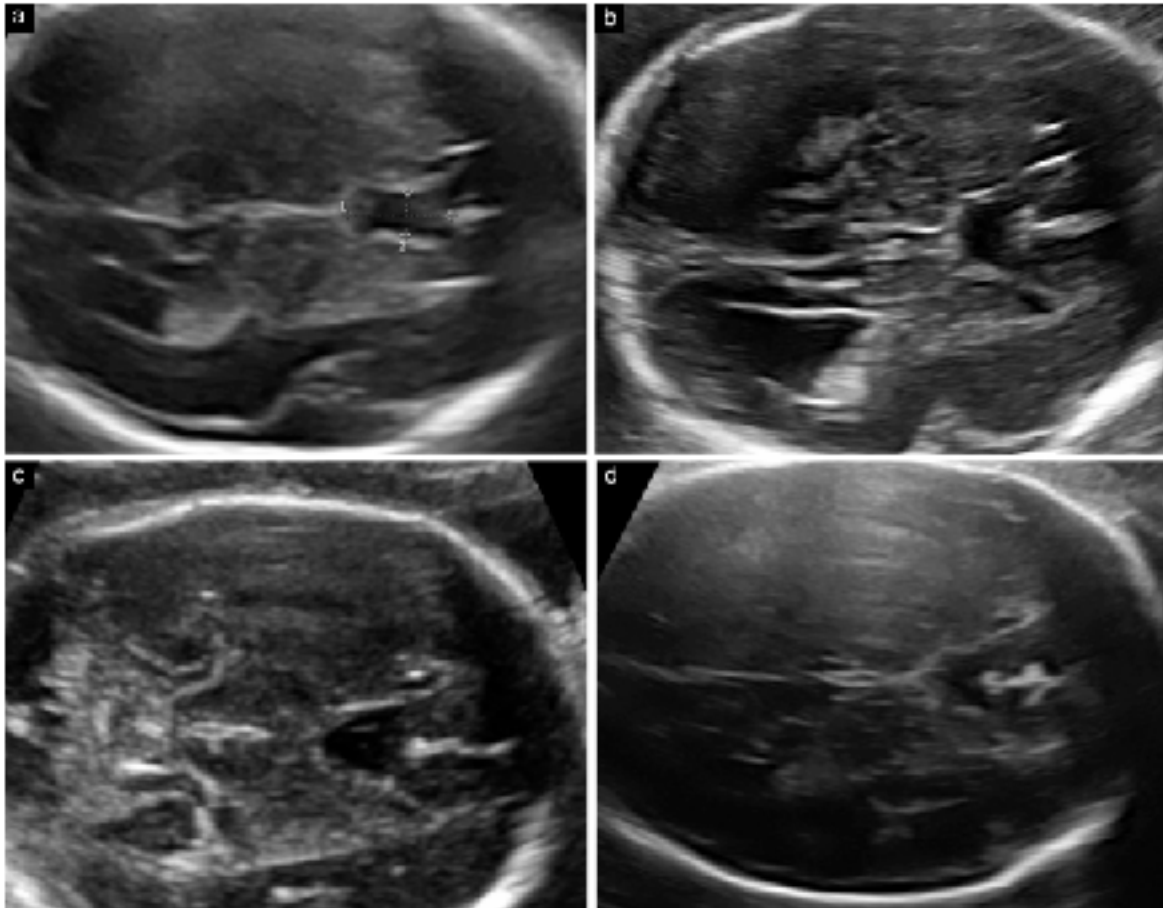
Serie de casos PACC: 52% CSP anormal
(diámetro T > diámetro AP)



Cavum septi pellucidi (CSP) ratio: a marker for partial agenesis of the fetal corpus callosum

K. KARL^{1,2}, T. ESSER¹, K. S. HELING³ and R. CHAOUT³

¹Center for Prenatal Diagnosis Munich, Munich, Germany; ²Department of Obstetrics and Gynecology, Ludwig-Maximilians-University, Munich, Germany; ³Center for Prenatal Diagnosis and Human Genetics, Berlin, Germany



- Retrospectivo de casos y controles
- Midieron la longitud y el ancho del CSP en el plano axial y calcularon el RADIO, compararon casos de PACC (n20) y controles sanos (n323)
- Correlación con DBP

Cavum septi pellucidi (CSP) ratio: a marker for partial agenesis of the fetal corpus callosum

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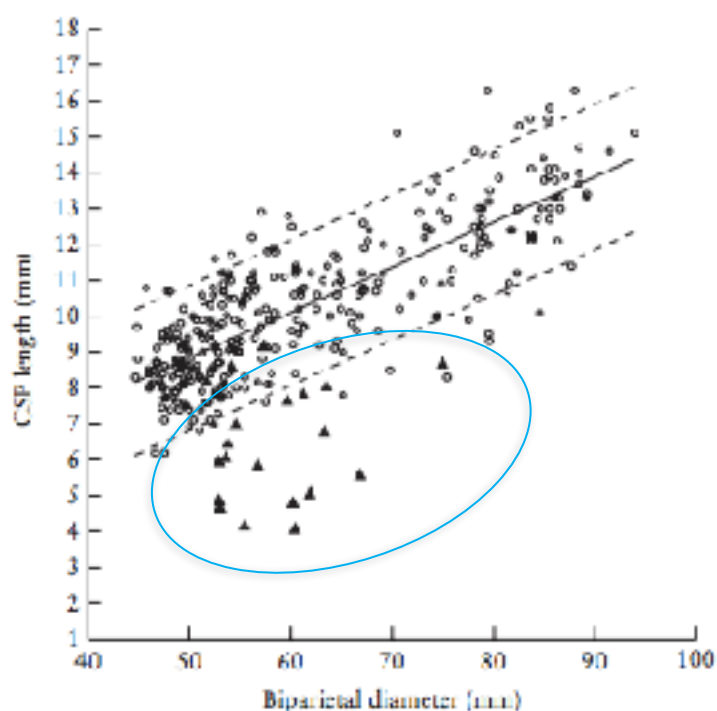


Figure 3 Individual measurements of cavum septi pellucidi (CSP) length in normal fetuses (○), with reference range (median and 5th and 95th centiles), and in 20 fetuses with partial agenesis of the corpus callosum (▲), in relation to biparietal diameter.

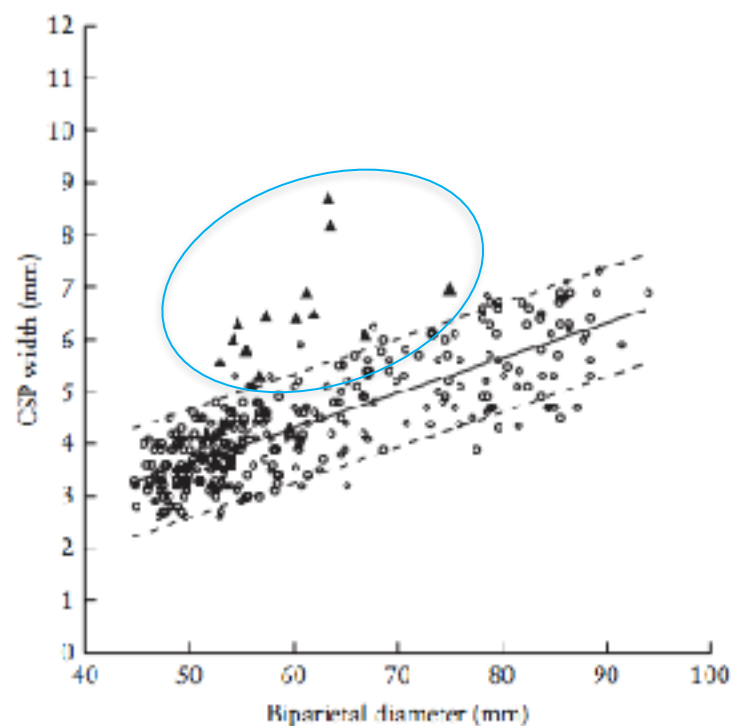


Figure 4 Individual measurements of cavum septi pellucidi (CSP) width in normal fetuses (○), with reference range (median and 5th and 95th centiles), and in 20 fetuses with partial agenesis of the corpus callosum (▲), in relation to biparietal diameter.

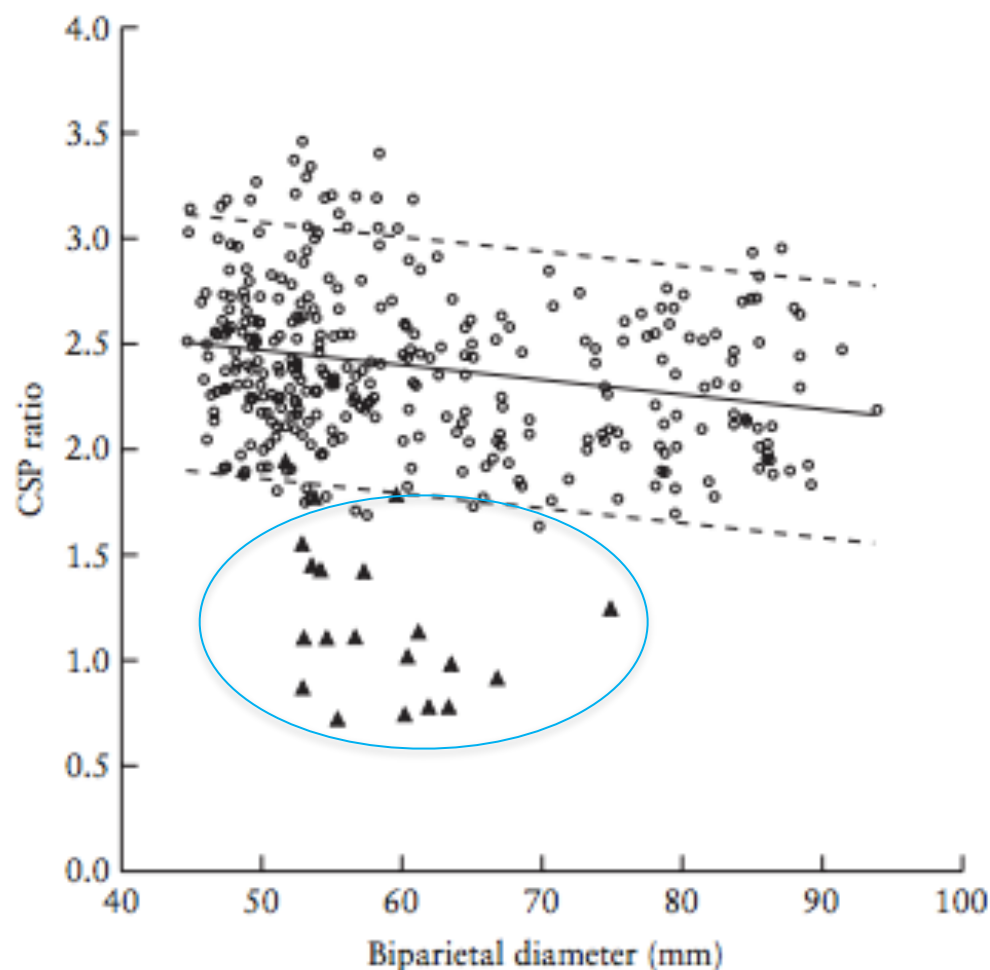


Figure 5 Individual measurements of length-to-width ratio of cavum septi pellucidi (CSP) in normal fetuses (O), with reference range (median and 5th and 95th centiles), and in 20 fetuses with partial agenesis of the corpus callosum (▲), in relation to biparietal diameter.

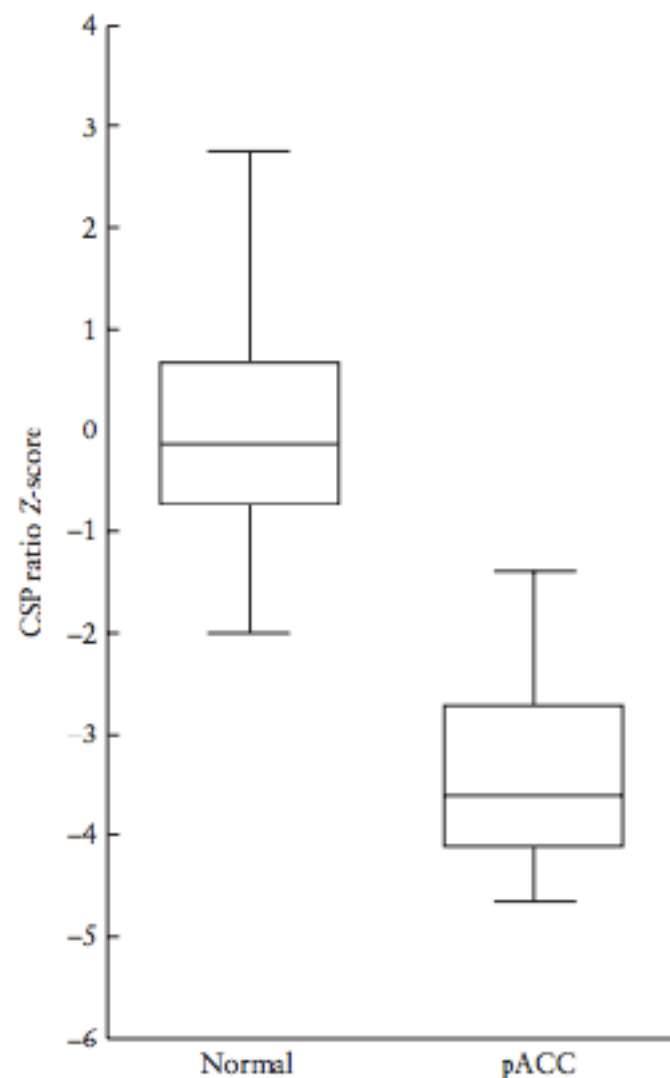


Figure 6 Box-and-whisker plot of cavum septi pellucidi (CSP) ratio (length to width) expressed as Z-score in normal fetuses and in study group of 20 fetuses with partial agenesis of the corpus callosum: (pACC). Boxes and internal lines show median and interquartile range and whiskers represent range. Fetuses with pACC had a highly significantly smaller CSP ratio ($P < 0.0001$) compared with normal population.

Cavum septi pellucidi (CSP) ratio: a marker for partial agenesis of the fetal corpus callosum

K. KARL^{1,2}, T. ESSER¹, K. S. HELING³ and R. CHAOUT³

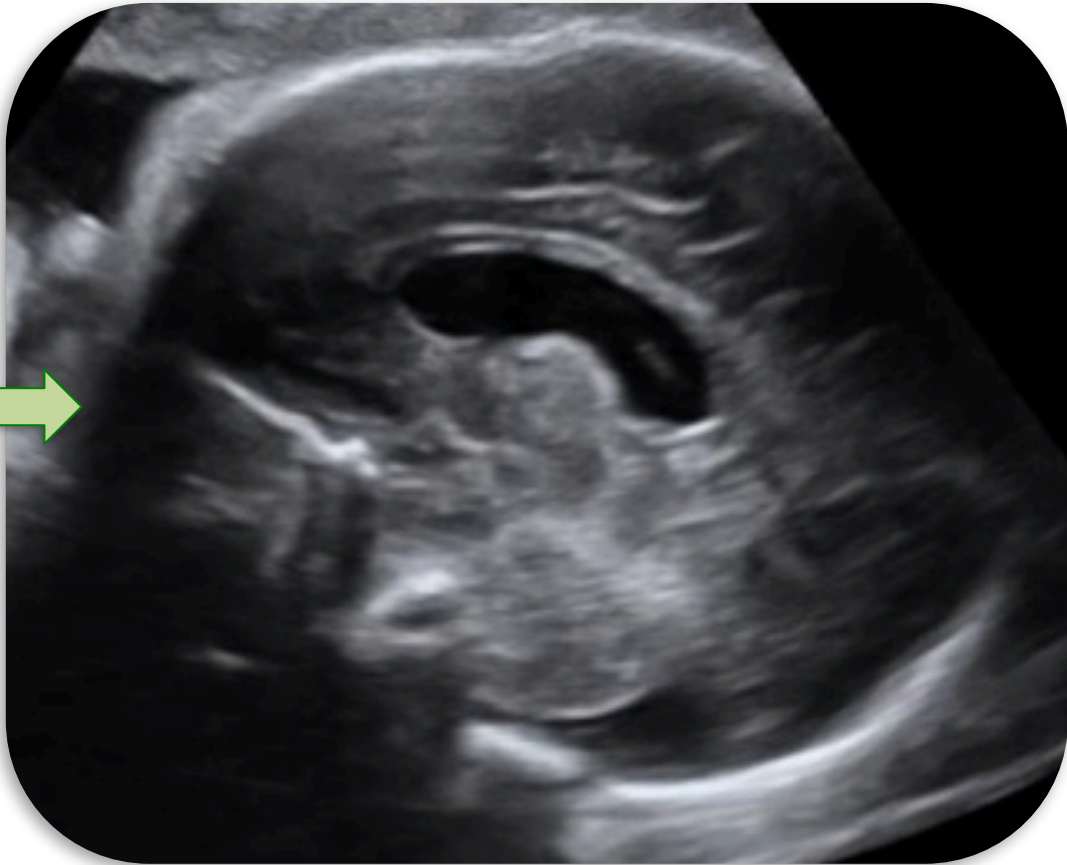
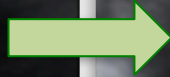
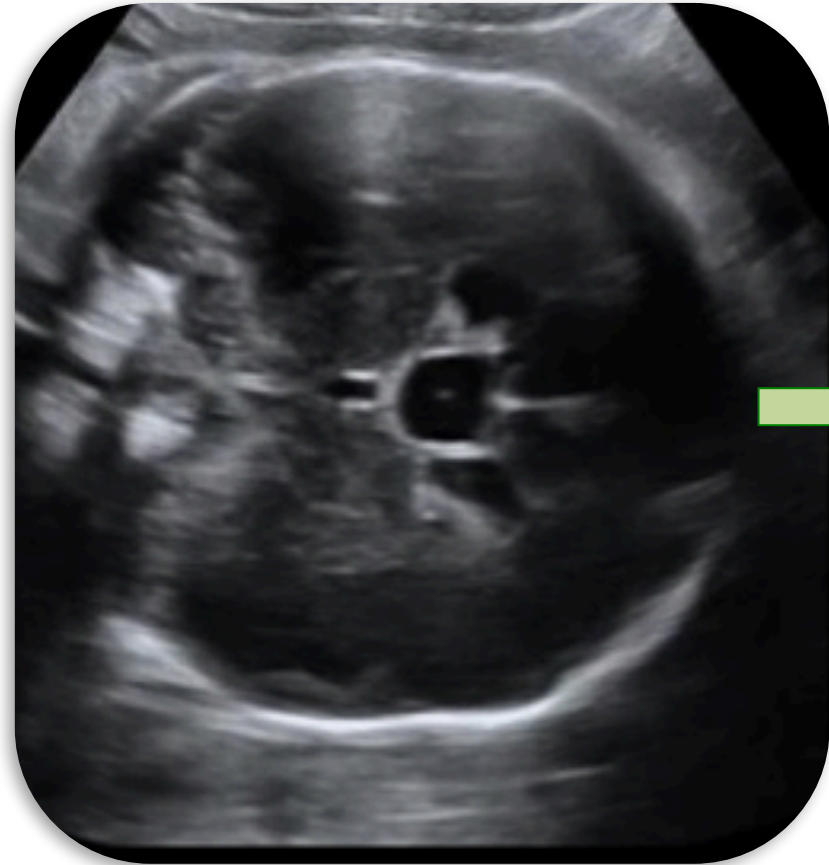
¹Center for Prenatal Diagnosis Munich, Munich, Germany; ²Department of Obstetrics and Gynecology, Ludwig-Maximilians-University, Munich, Germany; ³Center for Prenatal Diagnosis and Human Genetics, Berlin, Germany

Resultados:

- En la población normal el largo y el ancho aumentan a medida que aumenta el DBP, mientras que el RADIO disminuye.
- CSP fue corto (<p5) en 85% de PACC y ancho (>p95) en 65% de PACC
- *RADIO fue pequeño (<p5) en el 95% de PACC*

CSP anómalo

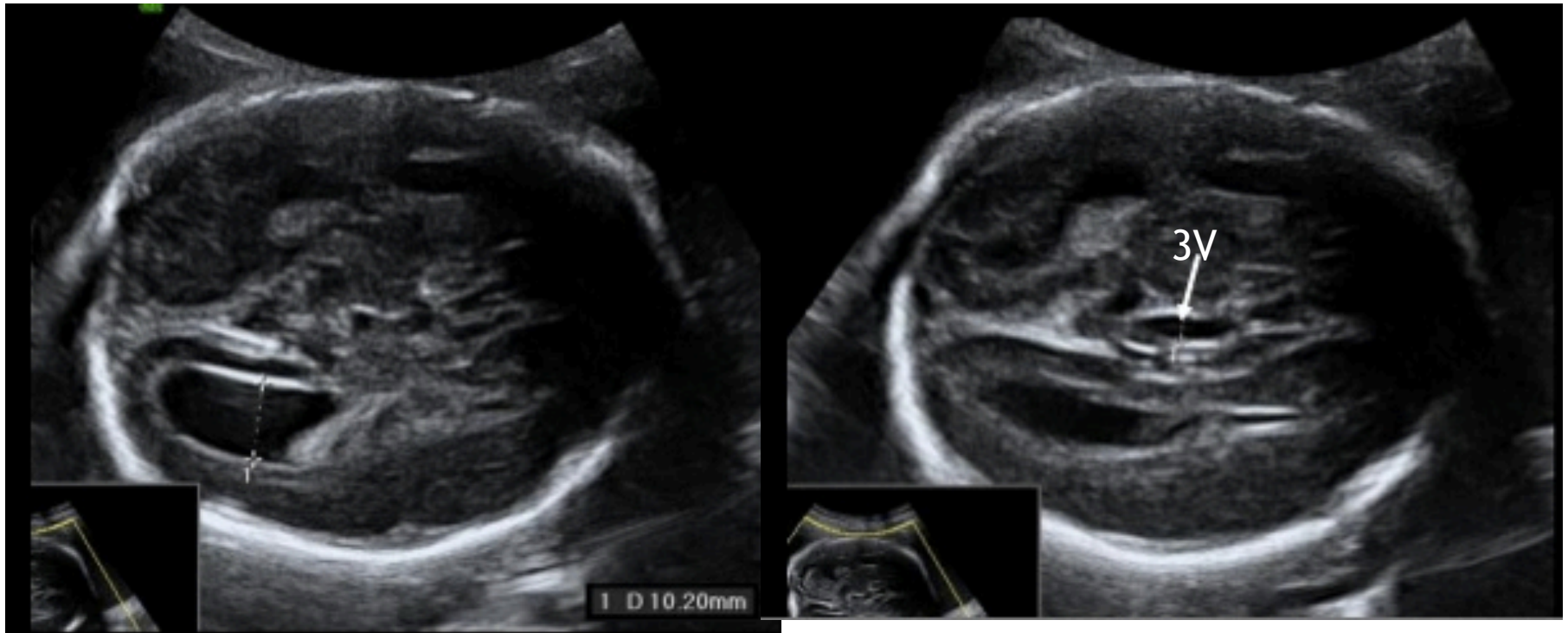
CC hipoplásico



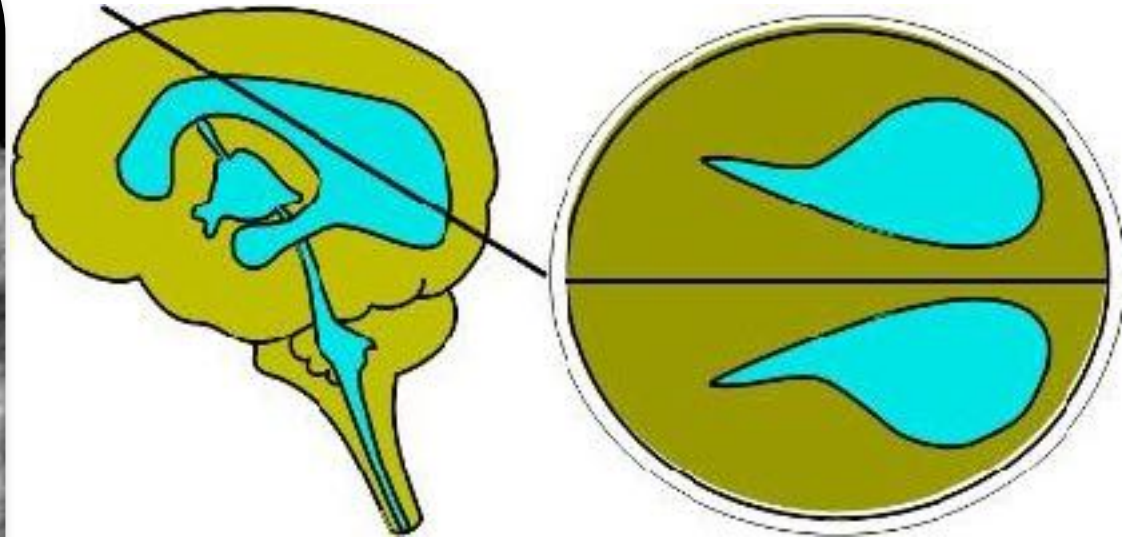
ACC: signos indirectos

Anormalidades SV

- Colpocefalia: debido a ausencia de la porción posterior del CC.
- CORONAL: desplazamiento lateral de los VL (haz Probst), “cabeza de alce”
- Tercer ventriculo elevado al nivel de VL



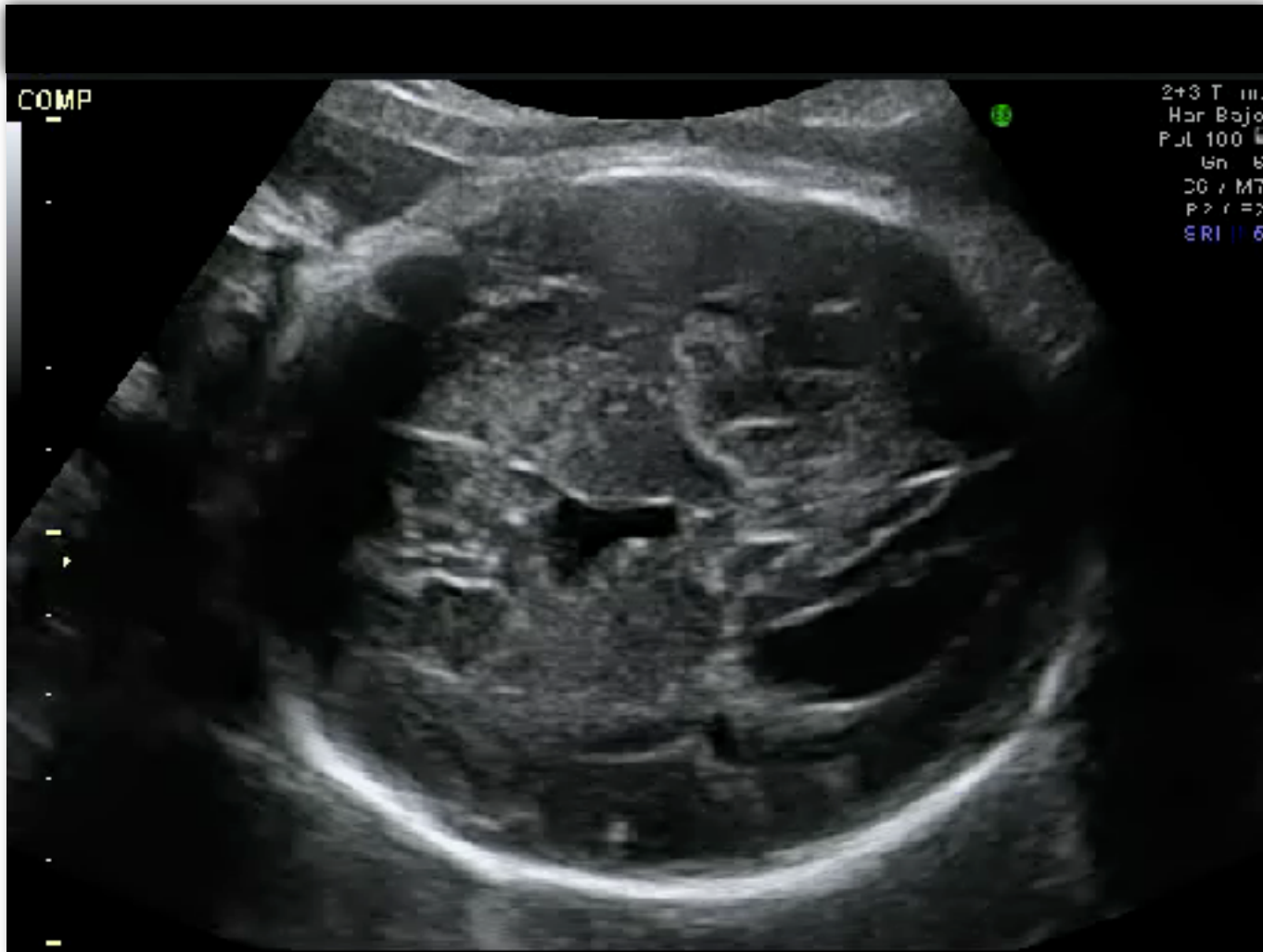
Colpocefalia



Dilatación de la prolongación occipital VL en el plano axial transventricular:

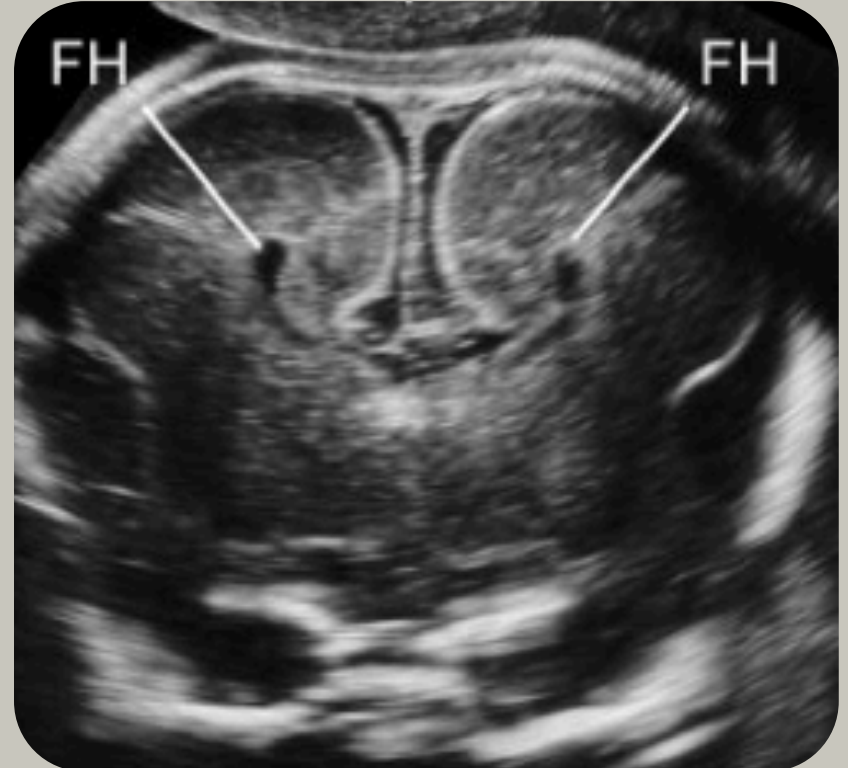
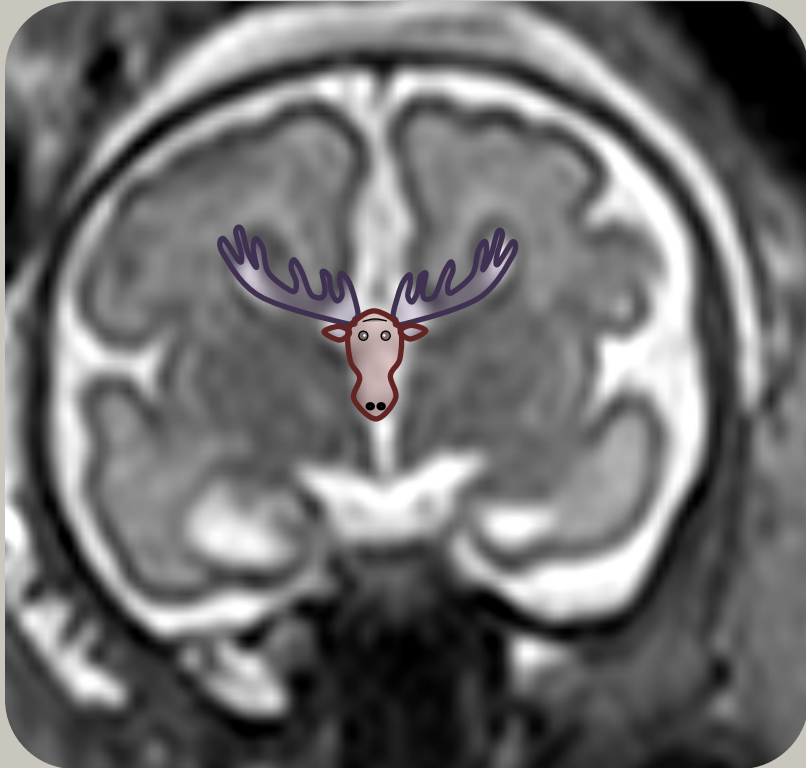
Forma de lagrima

ACC: desplazamiento superior del 3V



3er ventrículo anormal: 60% de los casos

Signos de la cabeza de Alce



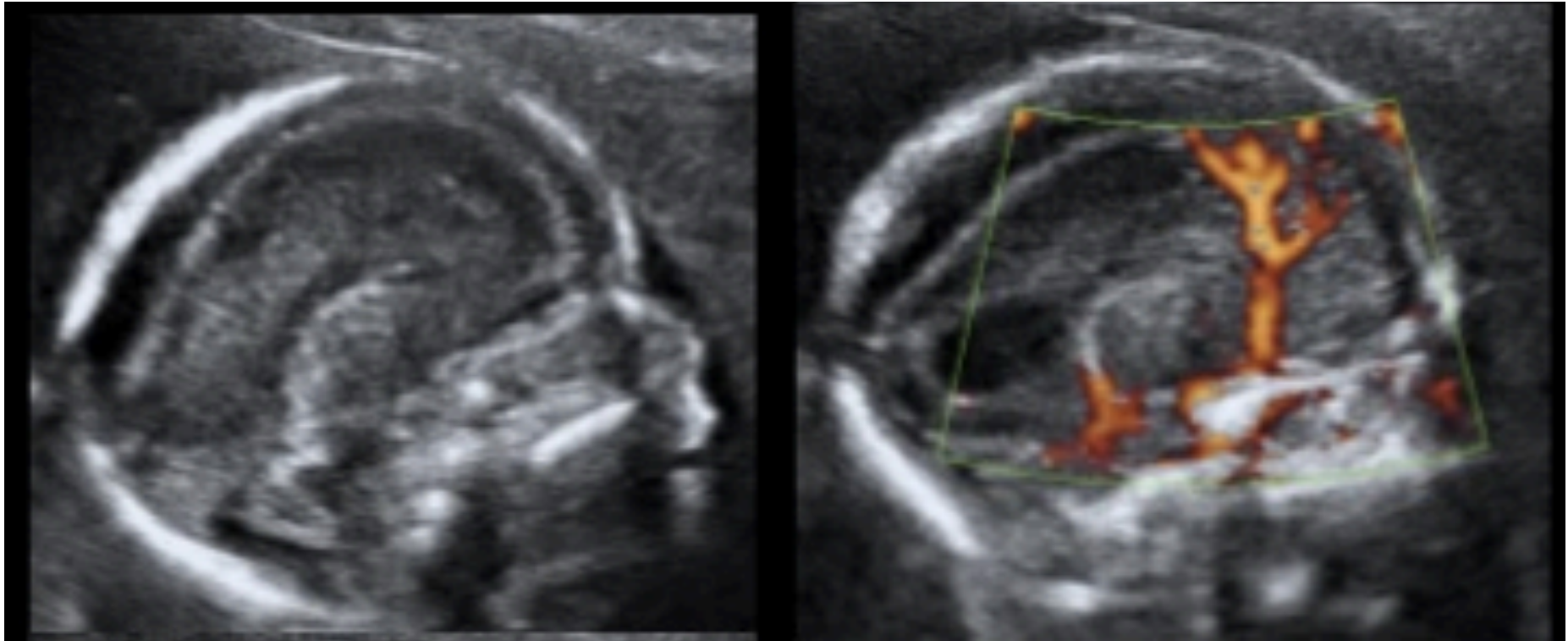
ACC: signos indirectos

Distribución anormal de la arteria pericallosa

ACC completa: ausencia loop semicircular de arteria pericallosa

- ACC parcial: arteria pericallosa recorre la porción anterior del CC y luego asciende posterior oblicua.

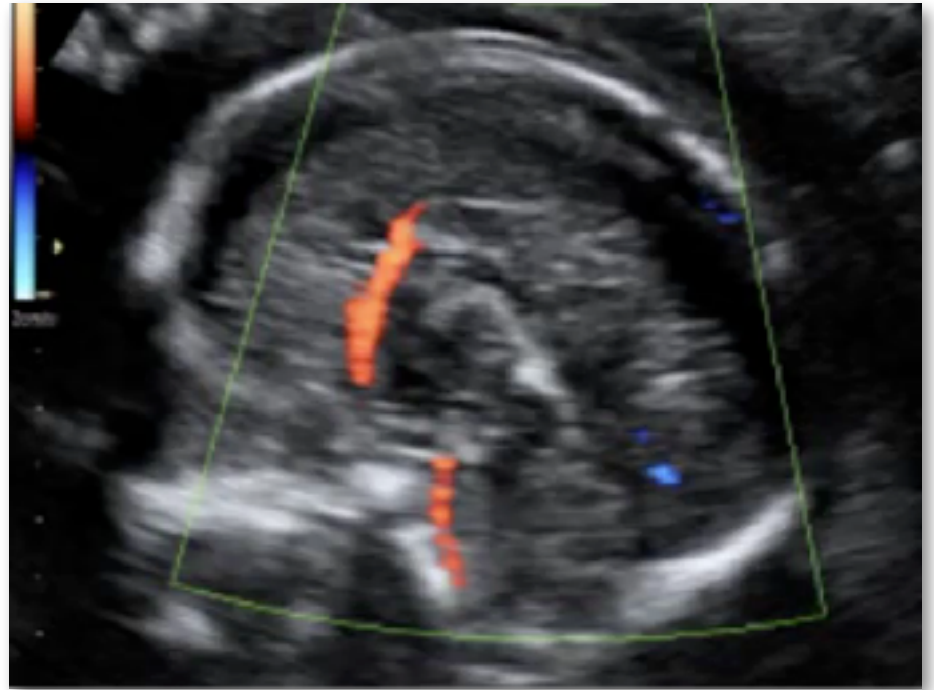
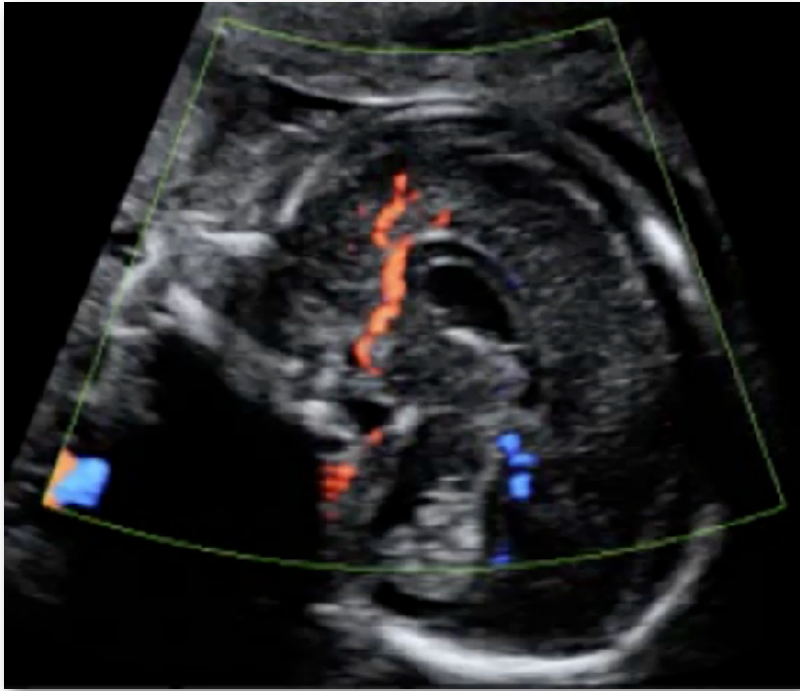
19s



Arteria Pericallosa NORMAL



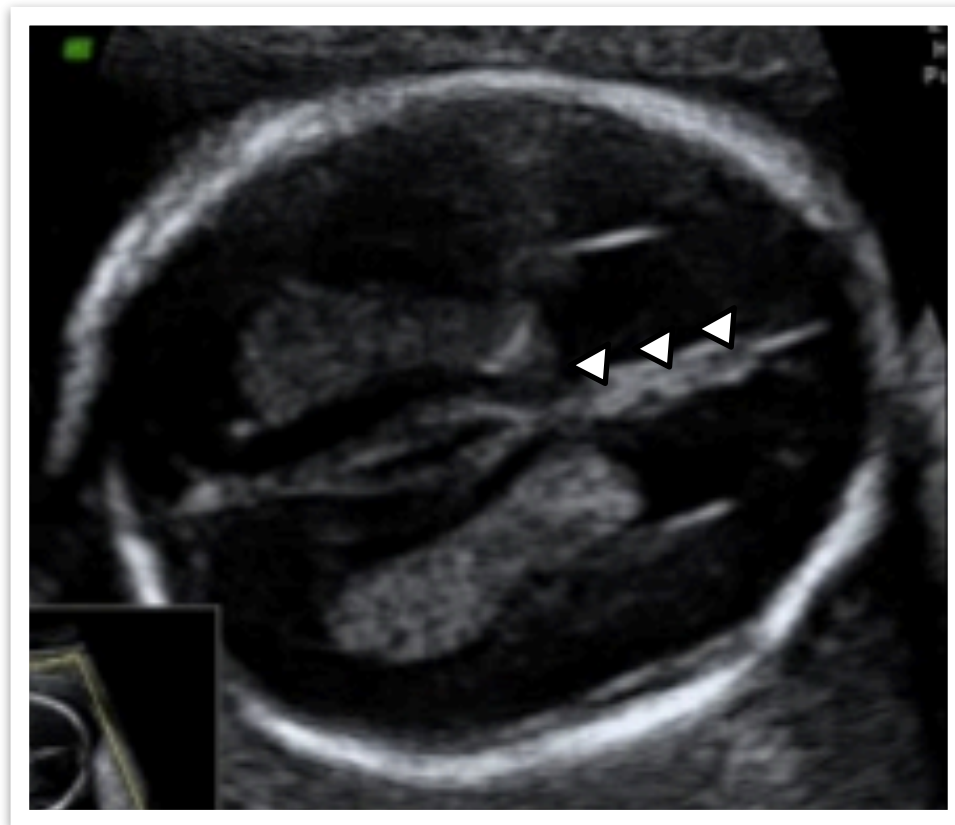
Arteria Pericallosa



ACC: signos indirectos

Engrosamiento de la fisura interhemisférica

- 3 líneas hiperecogénicas paralelas: hoz del cerebro y lateralmente el margen cerebral



ACC: signos indirectos

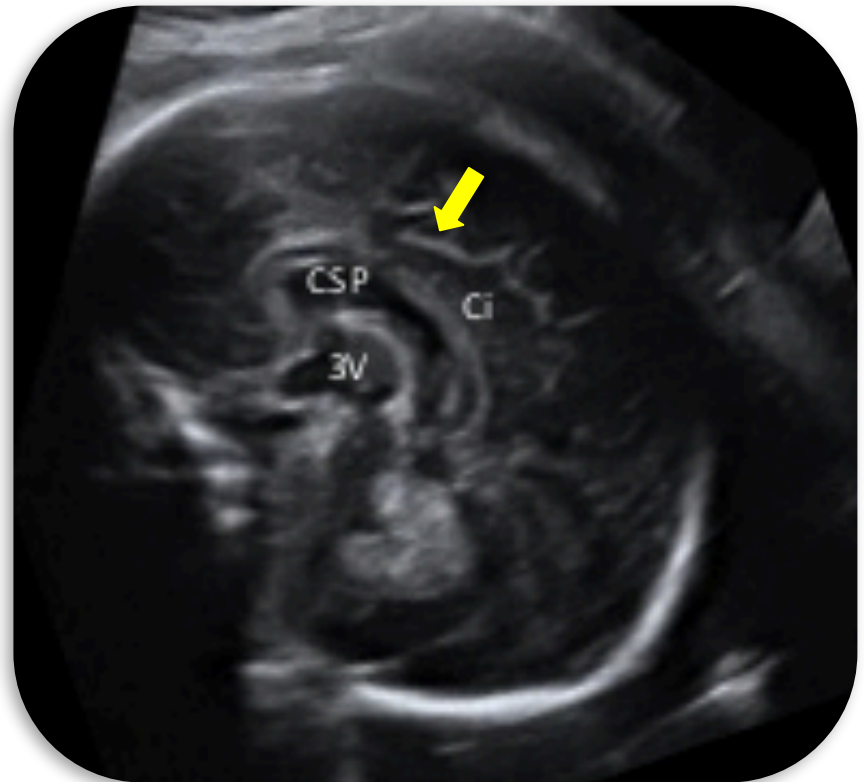
Disposición radial surcos convexidad hacia el techo 3V



Circunvolución pericallosa

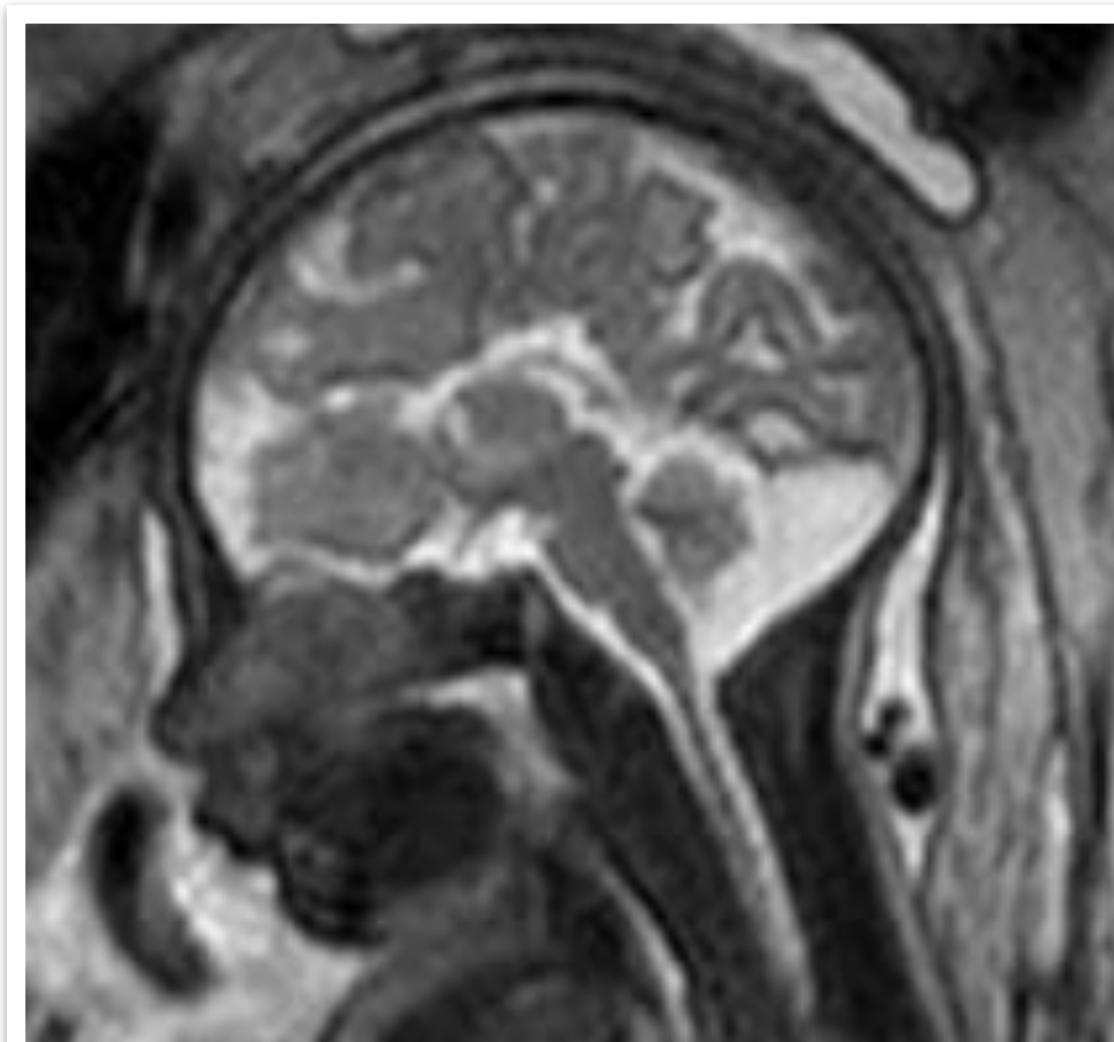


34s



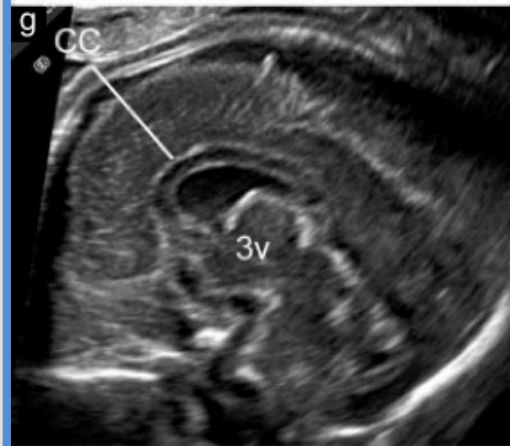
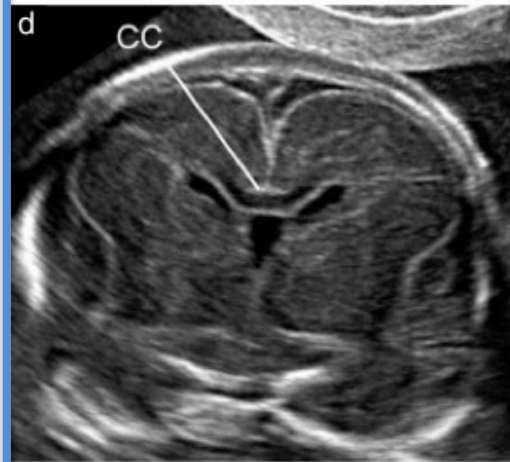
36s

ACC: signos indirectos

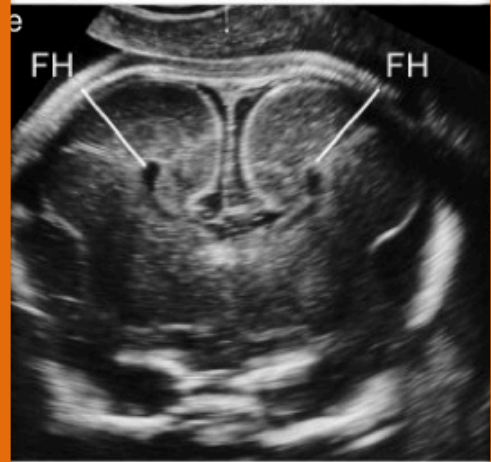
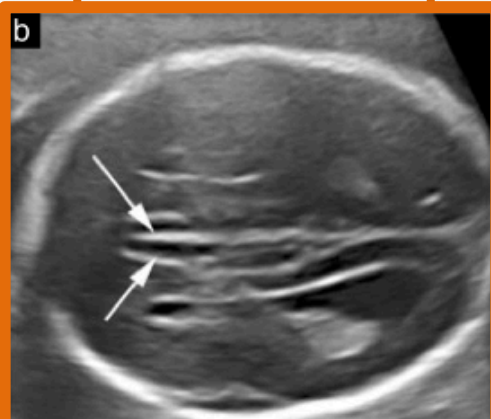


RMf
34s

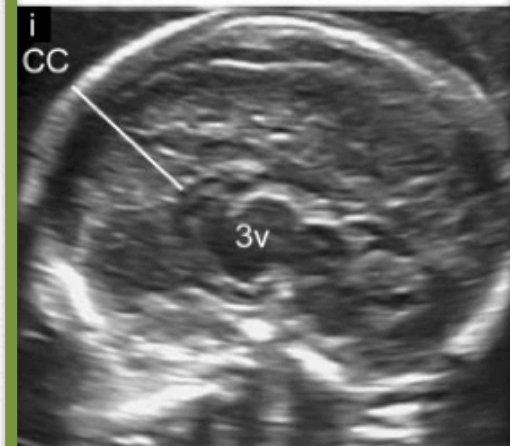
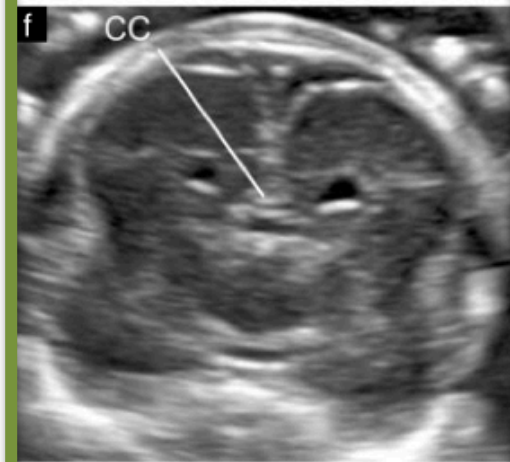
CEREBRO NORMAL



ACC COMPLETA



ACC PARCIAL



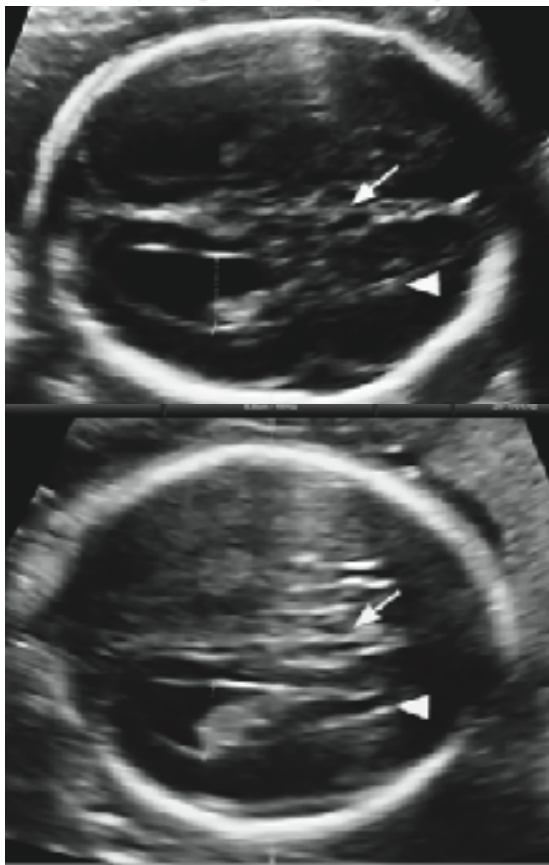
Dx ecografico: plano axial



Aggenesis of the fetal corpus callosum: sonographic signs change with advancing gestational age

D. PALADINI*, G. PASTORE†, A. CAVALLARO†, M. MASSARO† and C. NAPPI†

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Retrospectivo
54 casos de ACC
31 CACC
23 PACC

Table 1 Sonographic signs of fetal partial/total agenesis of the corpus callosum (ACC) according to gestational age

Sonographic sign	< 24 weeks (n = 34)	≥ 24 weeks (n = 35)
Atrial width > 9.9 mm	9/34 (26.5)	26/35 (74.3)
Colpocephaly	7/34 (20.6)	24/35 (68.6)
Spear-shaped ventricles	7/34 (20.6)	3/35 (8.6)
Non-visualization of CSP*	6/17 (35.3)	2/10 (20.0)

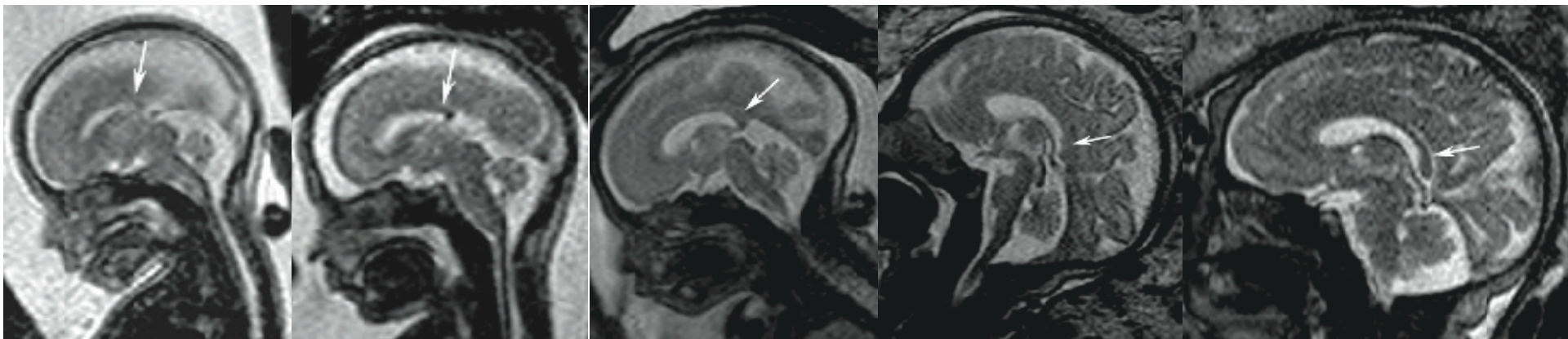
Data are presented as n(%). *Non-visualization of cavum septi pellucidum (CSP) evaluated only in fetuses with partial ACC.

Resultados:

- Luego de las 24 semanas los signos indirectos mas frecuentes: alteraciones del SV.
- Por lo tanto en la ecografía morfológica puede no ser DX
- 1/3 pACC sin signos US <24 s

RMf

- Visualización directa del CC
- Confirma ACC (completa/parcial)
- Anormalidades coexistentes



21

24

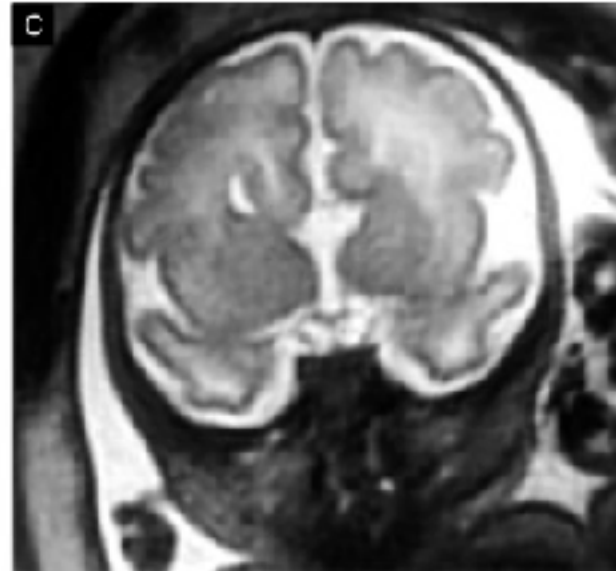
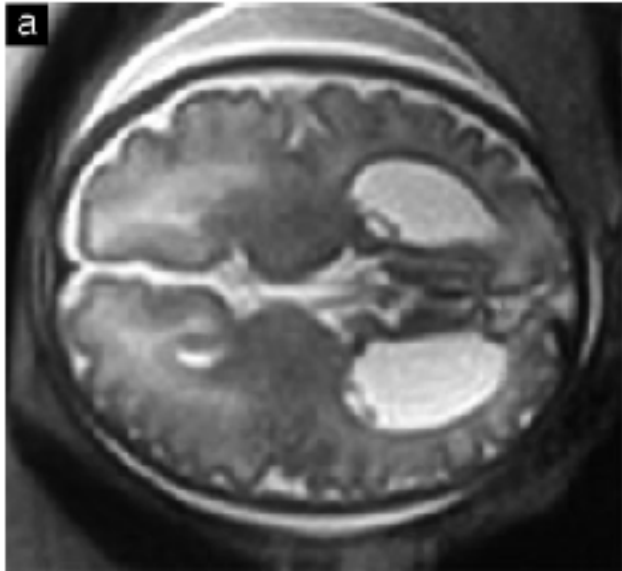
28

32

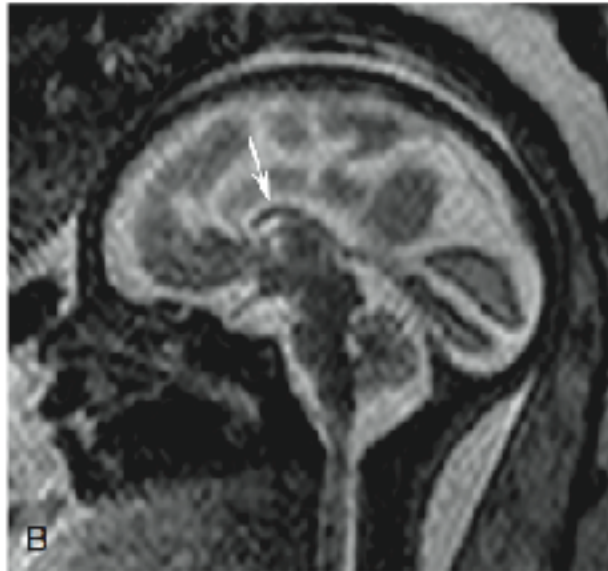
36

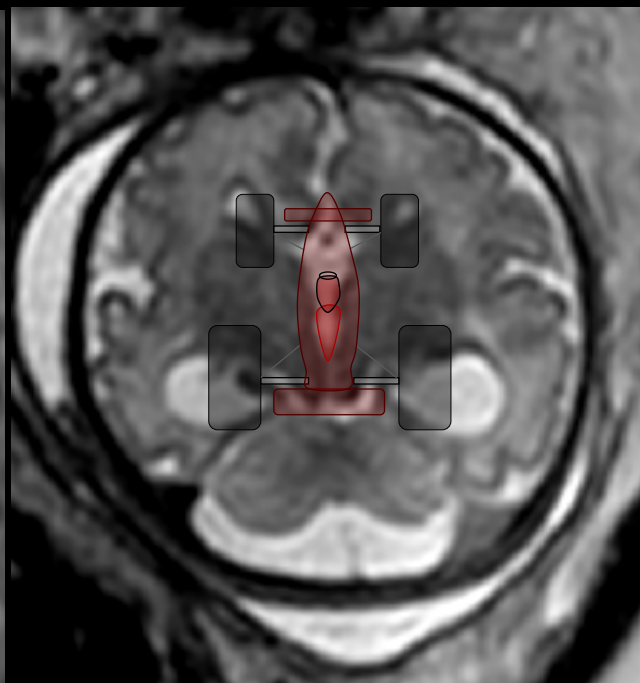
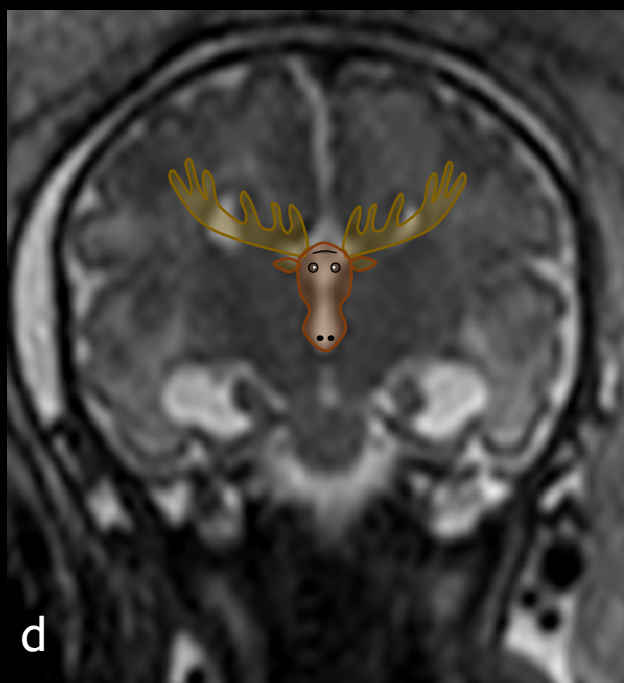
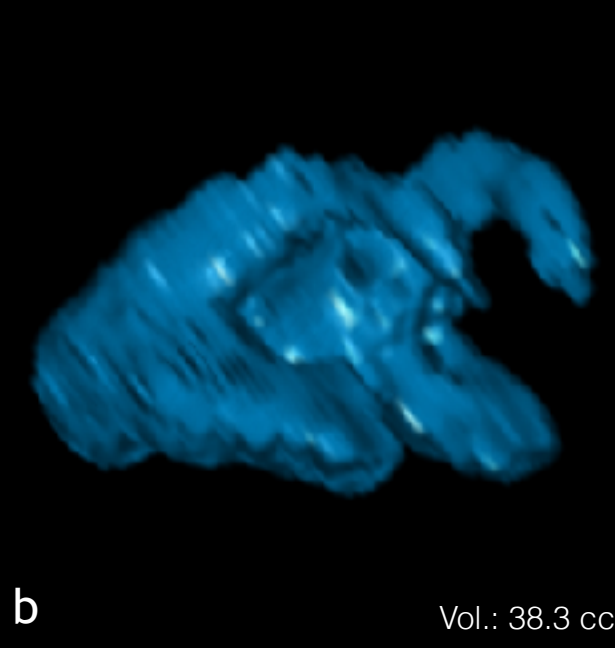
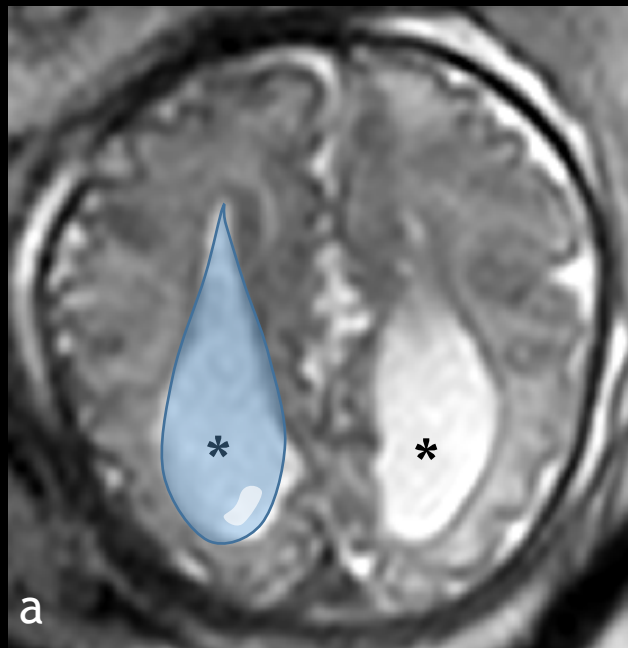
El esplenio es el mejor visualizado por este método

CACC

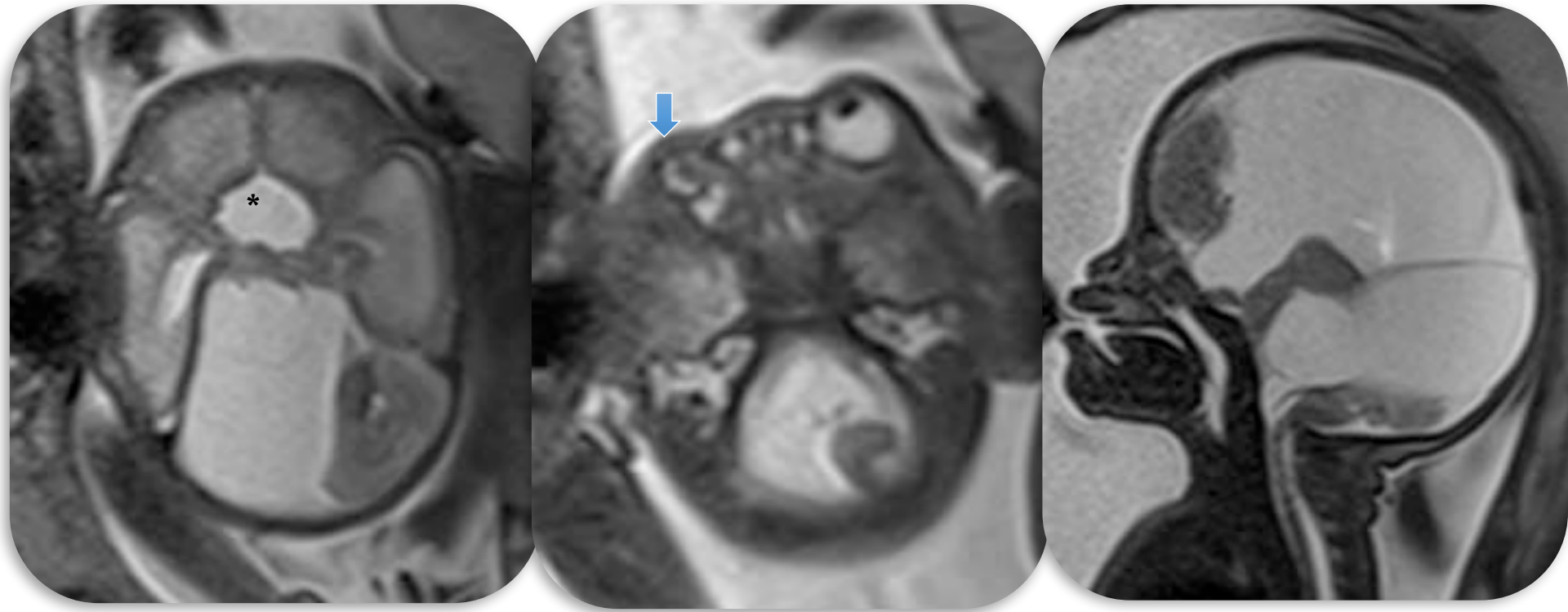


PACC



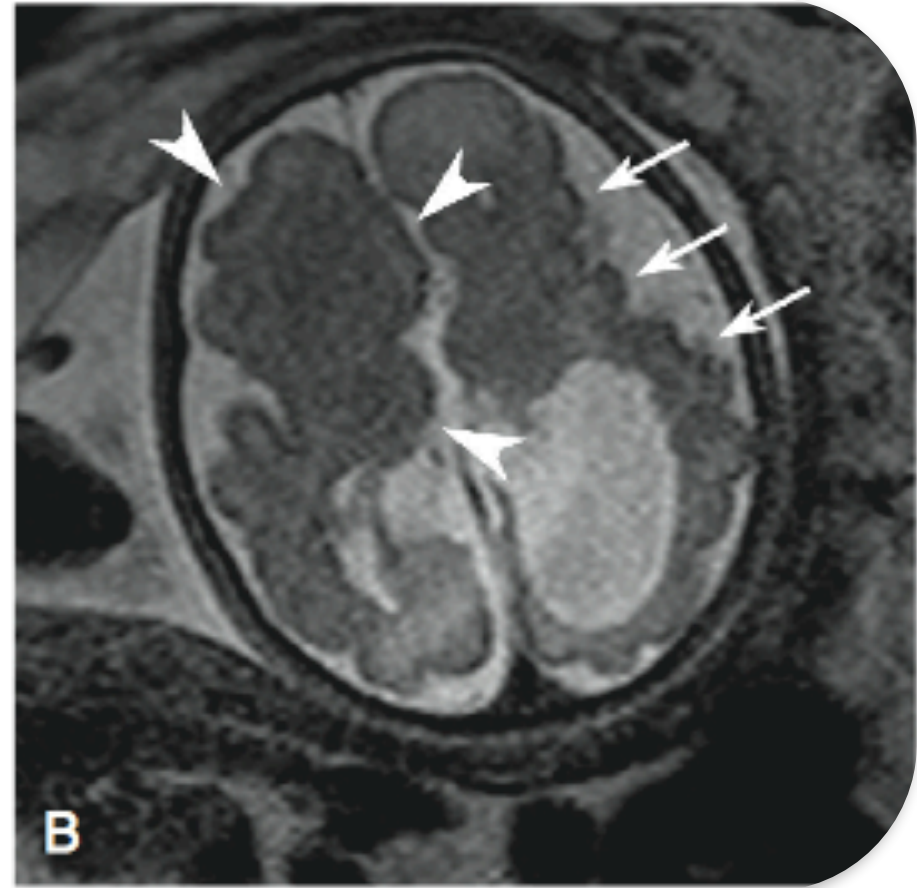
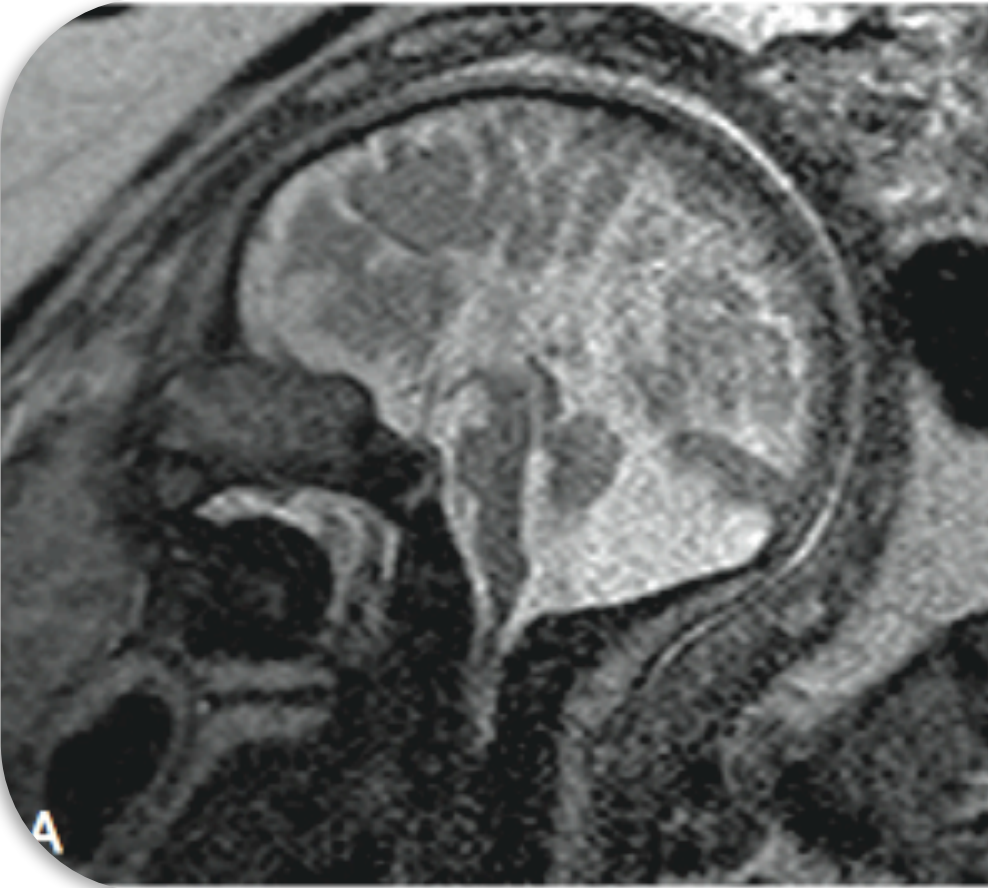


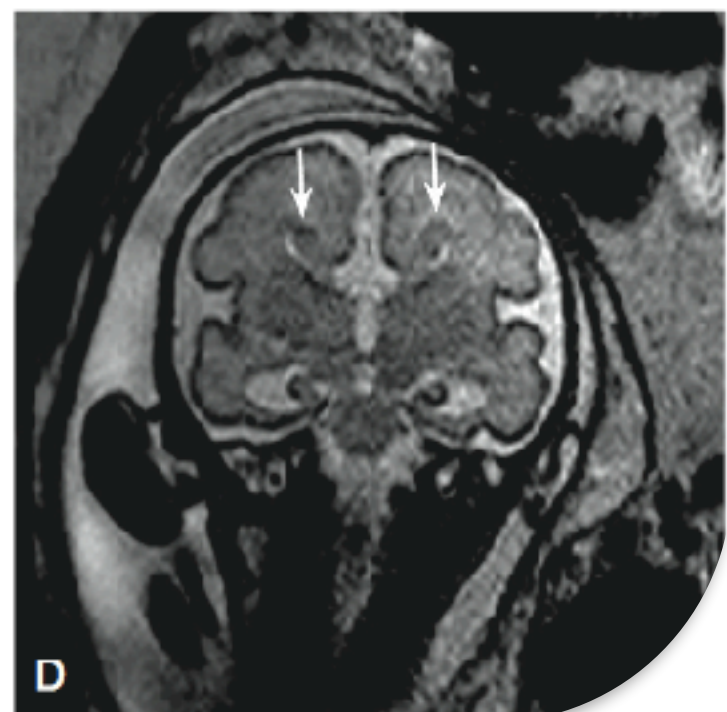
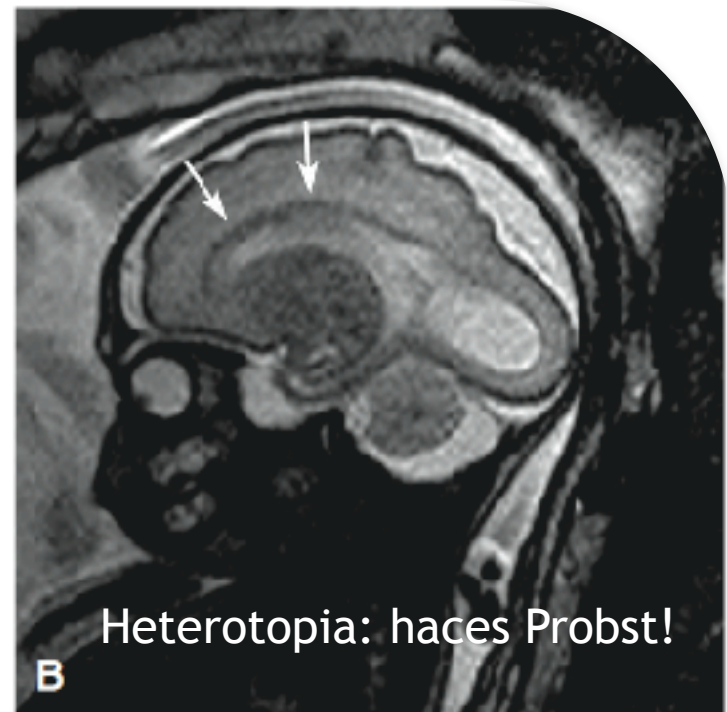
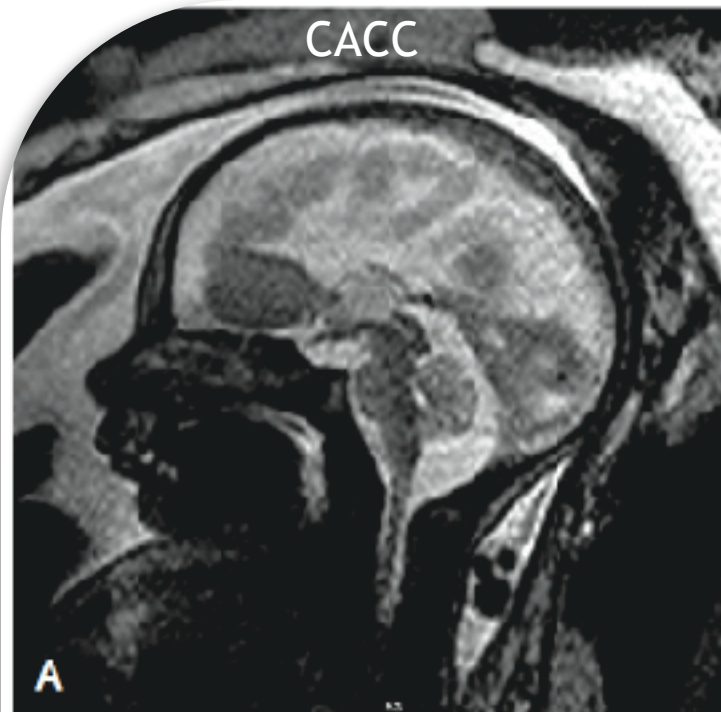
AVID: VM asimétrica + quistes interhemisféricos + disgenesia callosa



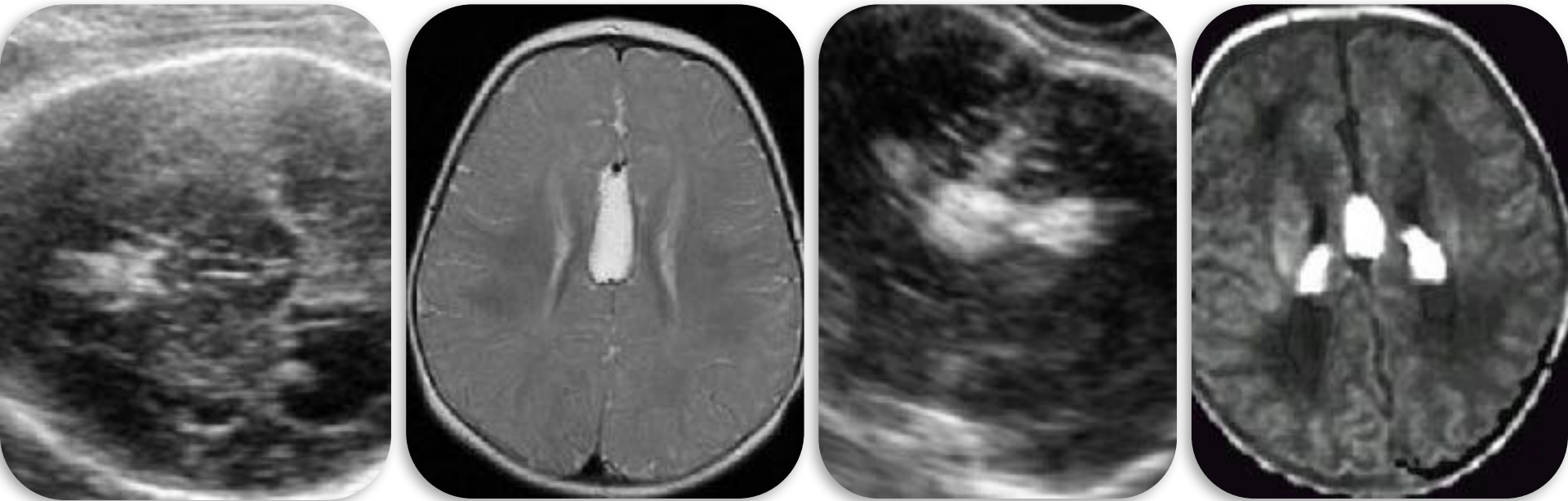
CACC

Polimicrogiria y quistes interhemisferico



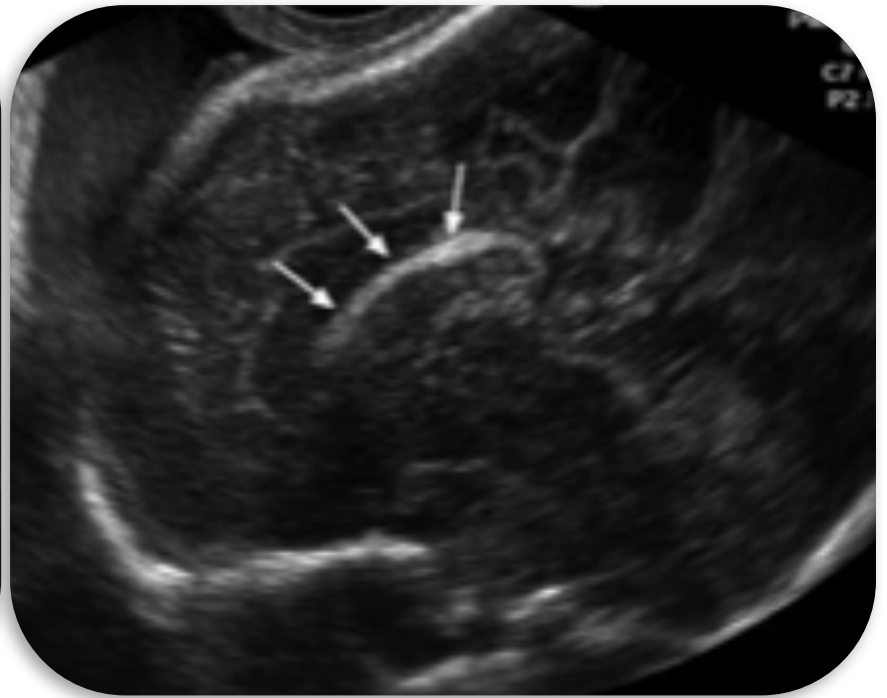
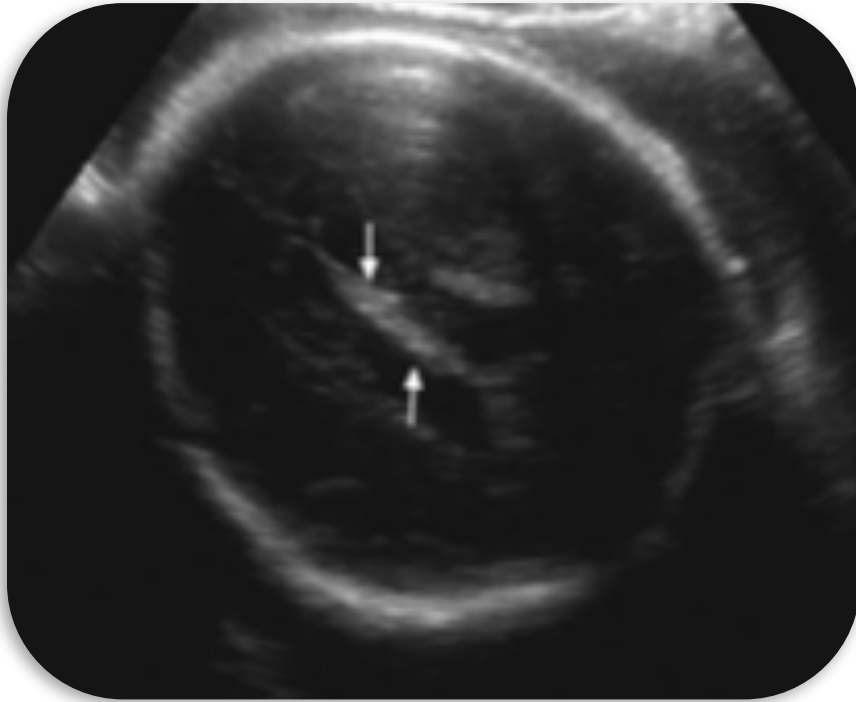


Lipoma



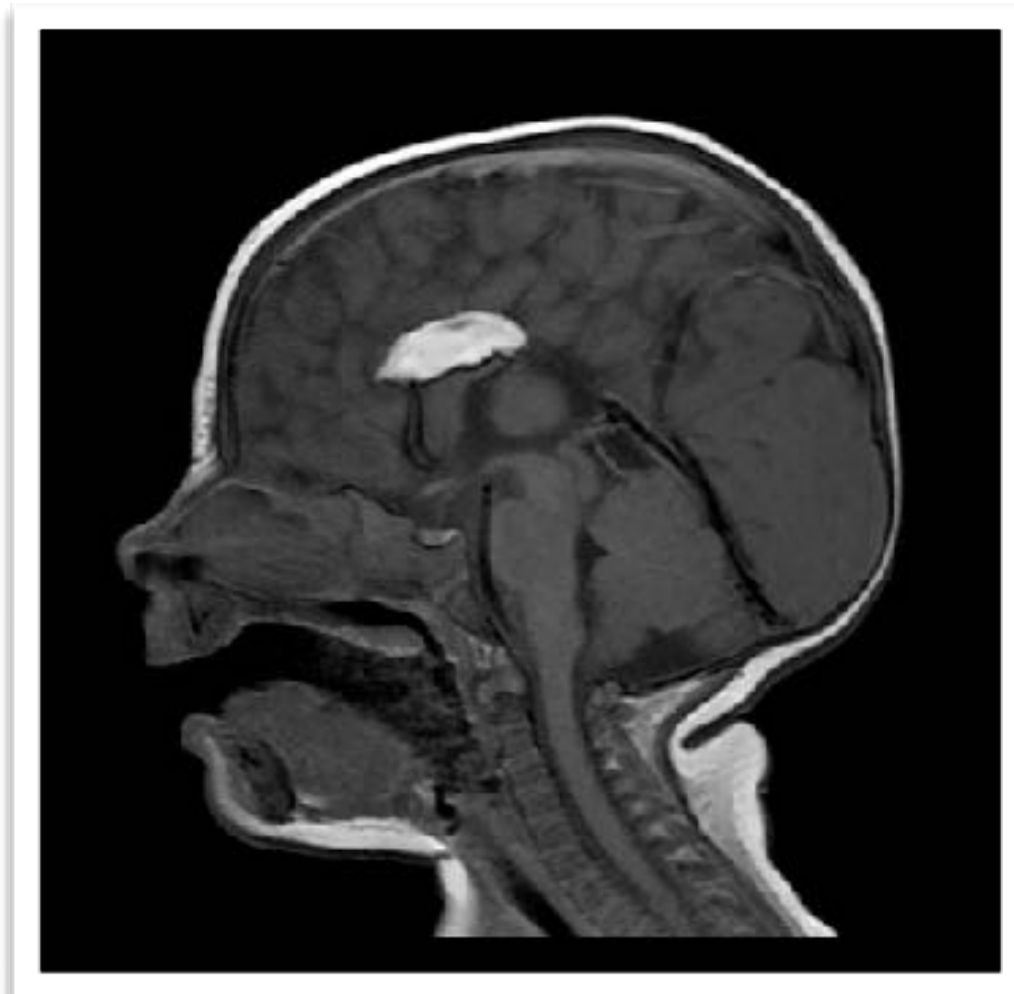
50% de los lipomas tienen ACC

Lipoma pericalloso



3er Trimestre

Lipoma pericalloso



ASOCIACIONES

- Síndromes Genéticos: >50
- Cromosomopatías: 20%
- Malformaciones cerebrales adicionales 46-80% (malformación de la fosa posterior, quistes interhemisféricos - alteración de la migración neuronal)
- Malformaciones extra-SNC adicionales: 60%
- Otras: alteraciones metabólicas, alcoholismo e infecciones

Table 1 — Causes and conditions associated with ACC

Cause/Condition	Diagnostic aids
<u>Chromosomal abnormalities</u>	
Trisomy 18	
Trisomy 8	
Trisomy 21	<i>Fetal</i>
Trisomy 22	<i>karyotyping</i>
Others: other trisomies, deletions, translocations, duplications	
Cryptic unbalanced aberrations	<i>FISH, CGH array, parental karyotyping</i>
<u>Genetic syndromes</u>	
Autosomal dominant	<i>Associated US features—family history (see Tables 2–4)</i>
Autosomal recessive	
X-linked	
<u>Environmental factors</u>	
Alcoholism	<i>Prenatal history</i>
Maternal rubella	
<u>Metabolic disorders</u>	
Adenylocuccinase deficiency	
Adipsic hypernatremia	
B-hydroxyisobutyryl coA deacylase deficiency	
Maternal diabetes	
Glutaric aciduria type II	
Histidinemia	<i>Family history and physical examination at birth</i>
Harler syndrome	
Leigh syndrome	
Menkes syndrome	
Neonatal adrenoleukodystrophy	
Nonketonic hyperglycinemia	
Pyruvate dehydrogenase deficiency	
Zellweger syndrome	

Aneuploidias: T18 / T13 / T8

- Trisomía 18: síndrome de Edwards:

- ACC
- Quistes plexo coroideo
- Anomalías de la fosa posterior
- Micrognatia
- Implantación baja de las orejas
- Microftalmia
- Hipertelorismo
- Mano cerrada con dedos superpuestos
- Talipes
- Pie en mecedora
- Onfalocele
- HDC
- Anomalías renales
- Hidronefrosis
- Defectos cardiacos
- AUU
- RCIU
- Polihidramnios
- Aumento TN
- Criptorquidia
- MMC
- VM
- Fisura labio-palatina

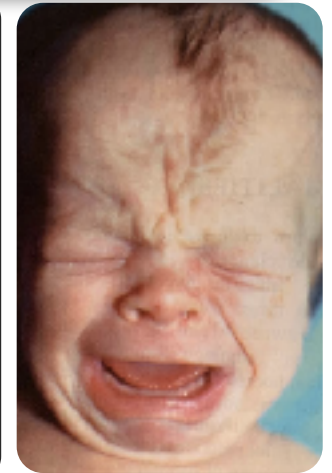
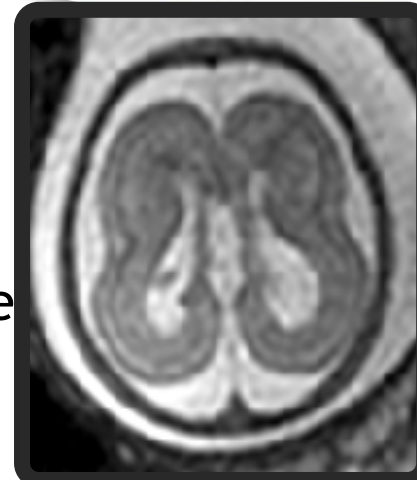
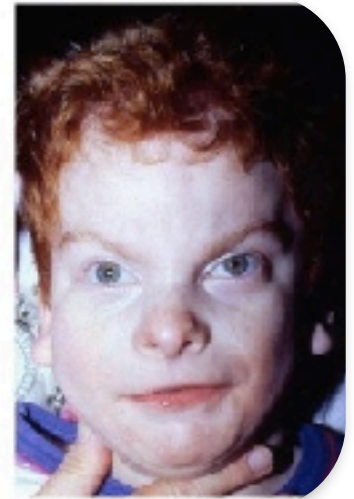
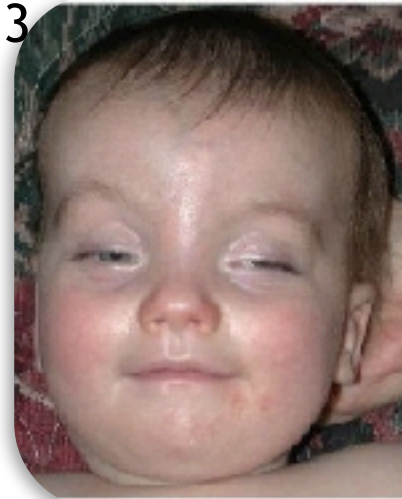


Delección 17p

- Síndrome de Miller-Dieker:

Lisencefalia tipo I delección 17p13.3

- Características: cerebro sin pliegues
- Hallazgos:
 - VM
 - PACC
 - Microcefalia
 - RCIU
 - Defectos cardiacos
 - Displasia renal
 - Atresia duodenal
 - Criptorquidia
 - Polihidramnios
- Retraso mental muy grave y convulsiones



Complex Genetic Syndromes With Agenesis of the Corpus Callosum (ACC) as a Possible Feature

	Syndrome	Chromosomal region	Gen	OMIM#	
	Autosomal-dominant				
AD	* Apert syndrome	10q26	FGFR3	#101200	
	Basal cell nevus syndrome	9q22.3	PTCH	#109400	
	Greig cephalopolysyndactyly syndrome	7p13	GLI3	#175700	
	* Miller–Dieker syndrome	17p13.3	LIS1	#247200	
	Mowat–Wilson syndrome	2q22	ZFX1B	#235730	
	Opitz GBBB syndrome	22q11.2		#145410	
	Rubinstein–Taybi syndrome	16p13.3 22q13	CREBBP EP300	#180849	
	Autosomal-recessive				
AR	* Acanthosis syndrome	7p13	GLI3	#200990	
	Andermann syndrome	15q13–q14	SLC12A6	#218000	
	* Coffin–Sims syndrome			135900	
	Dinesoy syndrome			601016	
	* Fryns syndrome			#229850	
	Fukuyama congenital muscular dystrophy	9q31	FCMD	#253800	
	Hydrocephalus syndrome	11q23–q25 9c34.3		#236680 #213300	
	* Joubert syndrome	6q23.2–q23.3	ARH1	#608620	
	Lowry–Wood syndrome			#226960	
	Lyon syndrome			225740	
	* Marden–Walker syndrome			#248700	
	* Meckel–Gruber syndrome	17q22–q23		#249000	
	Microcephalic osteodysplastic primordial dwarfism, type 1			#210710	
	Microcephalic osteodysplastic primordial dwarfism, type 5			#210730	
	Muscle-eye-brain disease	1p34–p33	POMGNT1	#243280	
	Neu–L axons syndrome			#256520	
	Ocular motor apraxia (Cogan-syndrome)	2q13		#257550	
	Peters-Plus syndrome			#261540	
	Septo-optic dysplasia	3p21.2–p21.1	HPSX1	#182230	
	Toriello–Carey syndrome			#217980	
	Vici syndrome	9q34.1 14q24.3	POMT1 POMT2	#242840	
	* Walker–Warburg syndrome	9q31	FUMD	#256670	
	Warburg–Mikro syndrome	2q21.3	RAB3GAP	#601138	
	LXR	X-linked ACC in combination with ectodermal dysplasia (hypohidrotic)			225040
		* Akcaadi syndrome	Xp22		#304050
		ATR-X syndrome	Xc13	ATR X	#01040
		FG syndrome	Xc12–q21.31		#305450
X-linked aqueductal stenosis or hydrocephalus/MASA syndrome		Xq28	L1CAM	#307000	
Craniofrontonasal syndrome		Xc12	EFNB1	#304110	
Lujan–Fryns syndrome				#309520	
MLS syndrome		Xp22.31		#309801	
Opitz GBBB syndrome		Xp22	MID1	#500000	
Oro-facial digital syndrome type 1		Xp22.3–p22.2	CXORF5	#311200	
Proud syndrome		Xp22.13	ARX	#500004	
X-linked lissencephaly		Xc22.3–q23	DCX	#300067	

Asociaciones GENICAS

- Síndrome de AICARDI:

Poco frecuente, ligado al X dominante

TRIADA diagnóstica:

- Convulsiones lactancia
- ACC
- Coriorretinitis lacunar

- US:

- Hallazgos frecuentes: VM, ACC, quistes interhemisfericos, orbitas asimétricas
- Hallazgos ocasionales: MDW, heterotopia cortical, hemivertebr

- Pronostico muy malo: retraso psicomotor y convulsiones graves



Asociaciones GENICAS

- Síndrome acrocalloso:

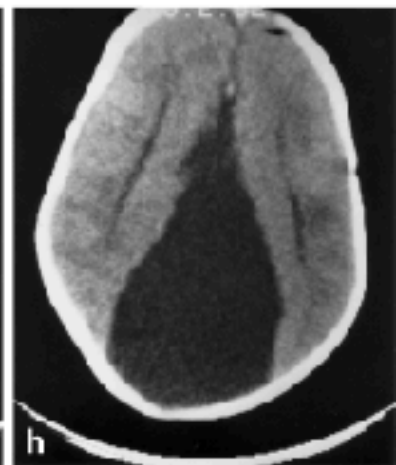
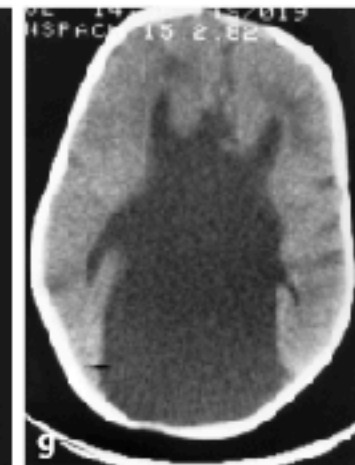
Poco frecuente, AR

- Características:

- ACC
- Anomalías distales de las extremidades
- Anomalías cráneo-faciales menores
- Retraso mental



DISGENESIA CALLOSA
EXTREMIDADES: polidactilia/
polisindactilia
Macrocefalia
Frente prominente
Hipertelorismo
Narinas antevertidas
Puente nasal amplio



Asociaciones GENICAS

- Síndrome de Apert:
Acrocefalosindactilia tipo I,
- Características:
 - Acrocefalia
 - Craneosinostosis
 - Sindactilia



Hallazgos mas frecuentes:

- Aumento TN 1T
- Braquicefalia y acrocefalia
- Craneosinostosis (sutura coronal)
- Cara plana
- Hipertelorismo
- ACC
- VM leve
- Fusión C5-6
- Sindactilia ósea y cutanea 2-3-4 dedos
- Pulgar ancho
- OTRAS: Tetralogía de Fallot - GU

Asociaciones GENICAS

- Síndrome de Meckel-Gruber
Disencefalia esplacnoquistica, AR

- Características:

- Encefalocele posterior (80%)
- Polidactilia (75%)
- Displasia renal quística (95%)
- Otras:
 - Microcefalia
 - Hipoplasia cerebelosa
 - MDW
 - ACC
 - Microftalmia
 - Micrognatia
 - Paladar hendido
 - Defectos cardiacos
 - Criptorquidea

- Los niños afectados fallecen en los primeros días de vida



Asociaciones GENICAS

- Síndrome de Walker-Warburg:
Lisencefalia tipo II, AR

- Características:

- HARD:
- Hidrocefalia (VM 100%)
- Agiria o lisencefalia generalizada
- Displasia retiniana, microftalmia, cataratas congénitas, desprendimiento retina
- Encefalocele (25%)
- MDW (50%)
- Microcefalia (16%)
- ACC

- Pronostico desfavorable, suele ser fatal dentro del primer año de vida



Asociaciones GENICAS

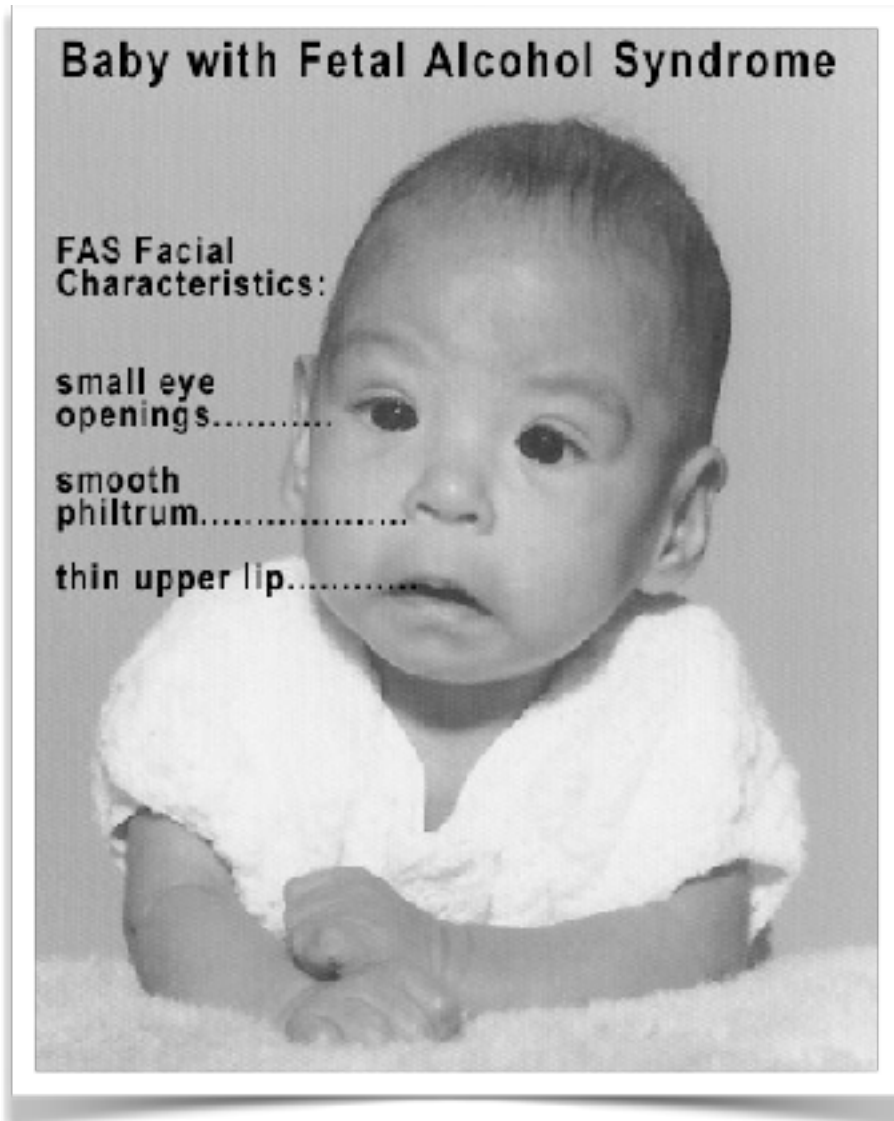
- Síndrome de Fryns:

AR



- Anomalías corneas
 - HDC
 - anomalías digitales: hipoplasia ungueal
 - Fascies tosca
 - Micrognatia
 - Orejas de implantación baja
- Trastorno fatal, fallecen intrauterino o en la vida neonatal. Si sobreviven a la reparación diafragmática: retraso mental.

Síndrome de Alcoholismo Fetal



- *RCIU*
- *Anomalías oftálmicas*
- *Cardiopatía y malformaciones viscerales*
- *Microcefalia*
- *Anomalías faciales:*
 - Puente nasal deprimido
 - Filtrum largo y liso
 - Labio superior fino
 - Hendidura palpebral corta
 - Fisura labiopalatina
 - Malformación de las orejas
 - Micrognatia

Recordar...

- Cromosomopatías

- T18
- T13
- T8
- Triploidias
- Miller Dieker
- Otras

- Síndromes no cromosómicos

- Aicardi
- Acrocalloso
- Apert
- Meckel Gruber
- Walker Warburg
- Fryns
- Alcoholismo fetal

- Malformaciones SNC

- Quistes interhemisfericos
- Malformación fosa posterior
- Alteración migración neuronal

- Chiari II : 7%
- Espectro Dandy-Walker: 11%
- HPE
- Hidrocefalia : 30%
- Lipoma : 10%
- Porencefalalia

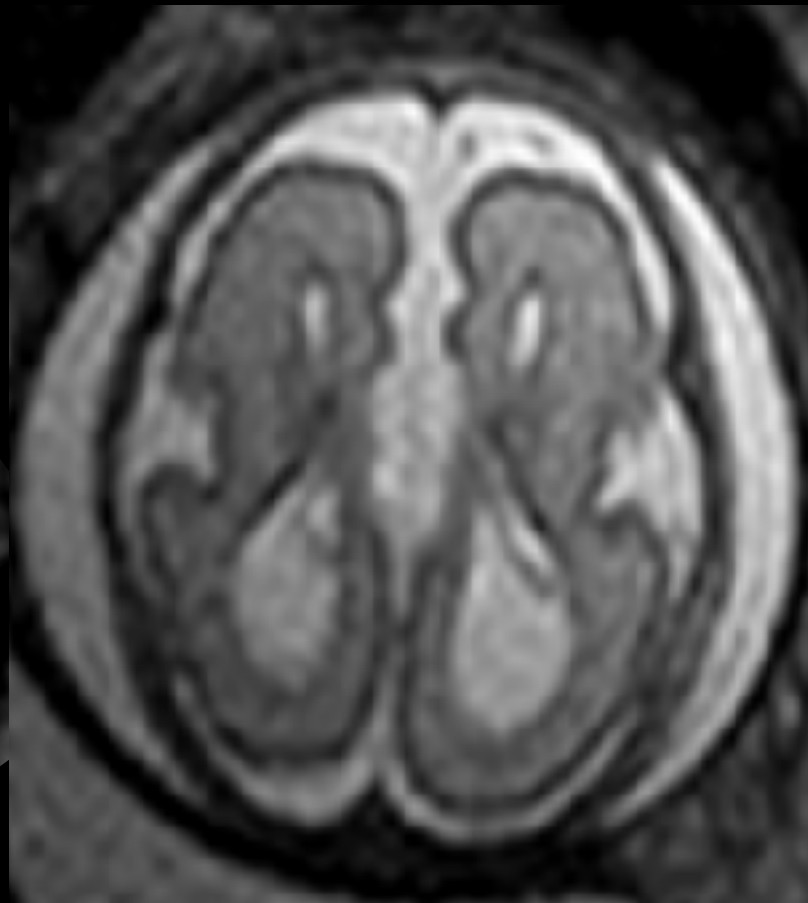
Caso n° 1

- G1, medica dermatóloga pediatra
- RA T21 1:4832 (otro centro)
- Eco Morfológica:
 - Pliegue nugal aumentado
 - VM limítrofe bilateral
 - Cervicometria 20mm

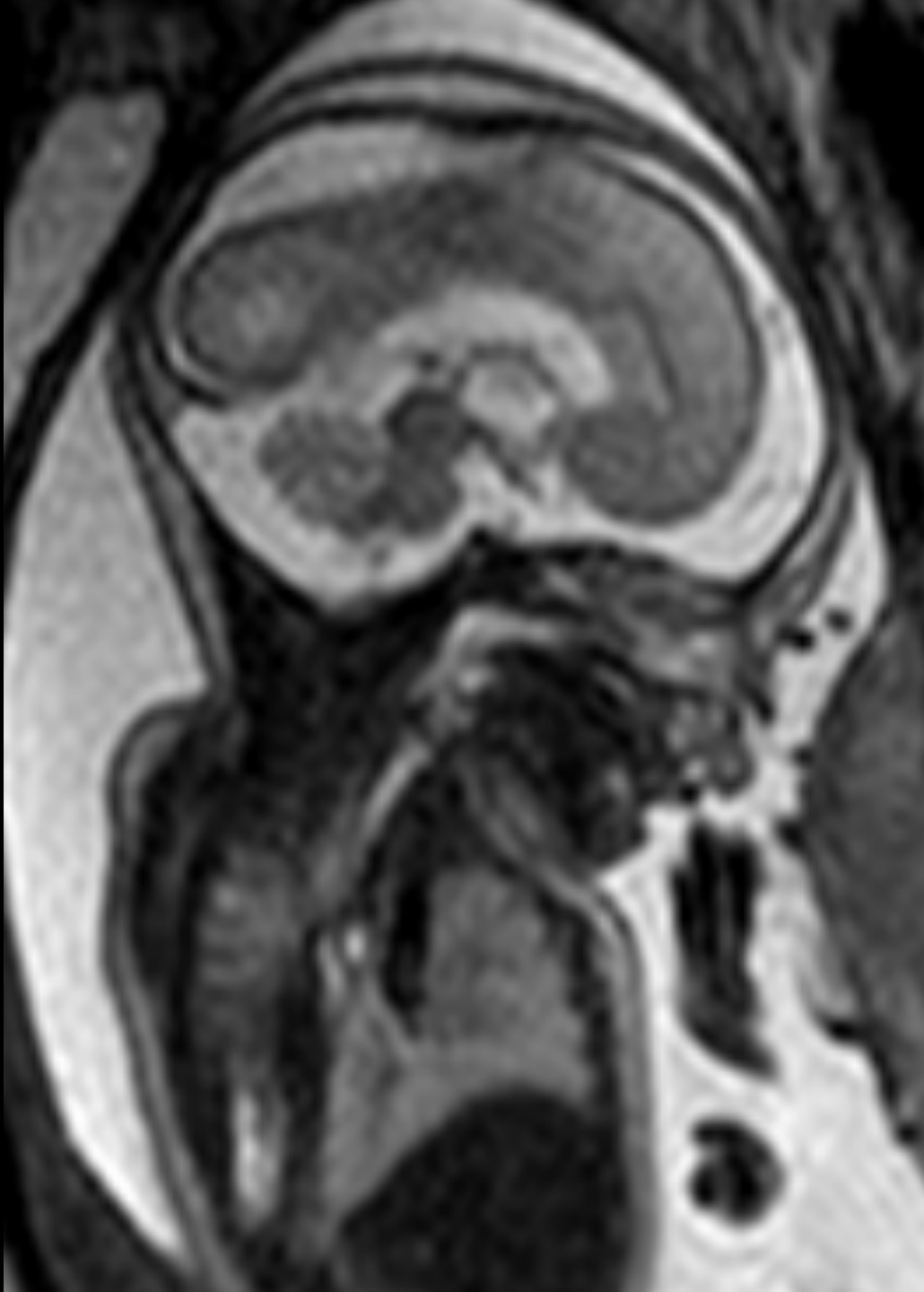
Coronal

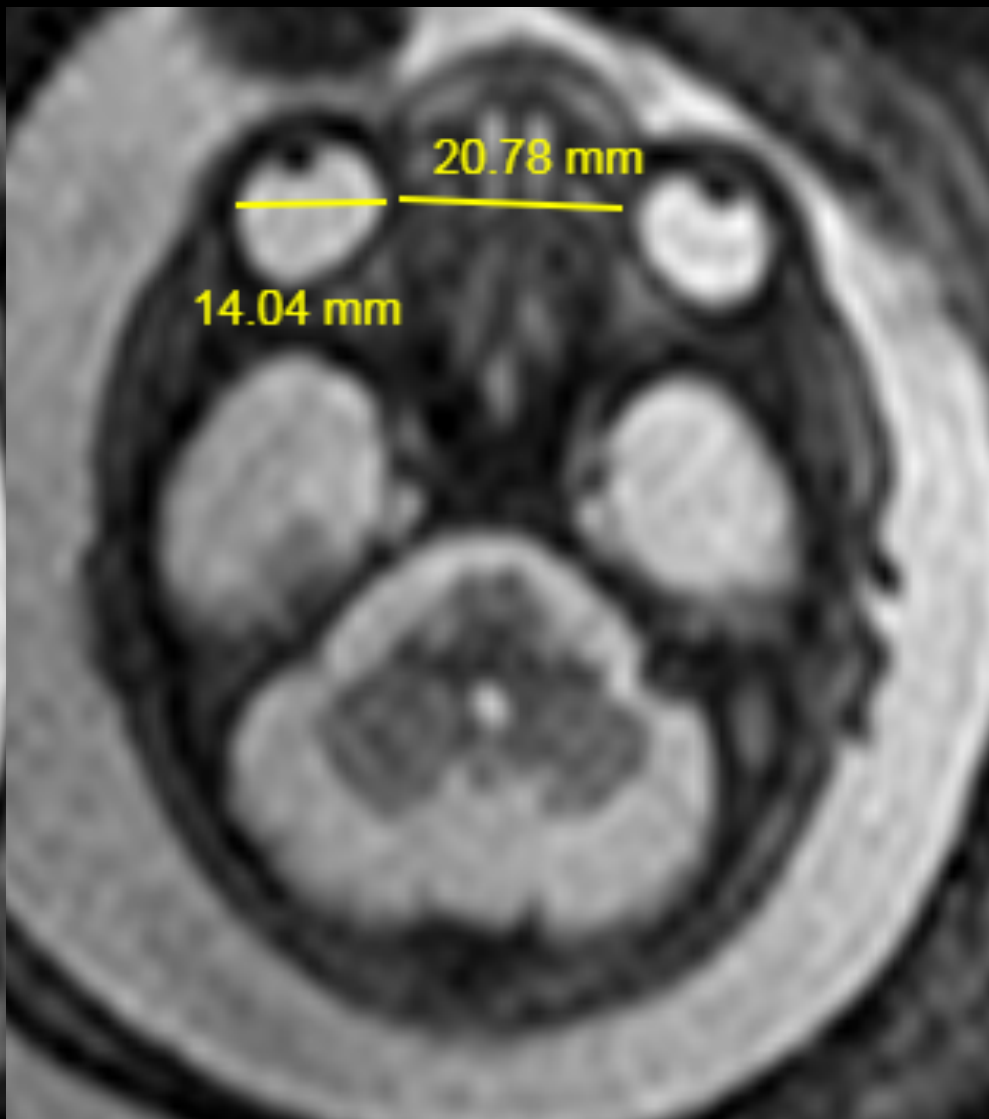
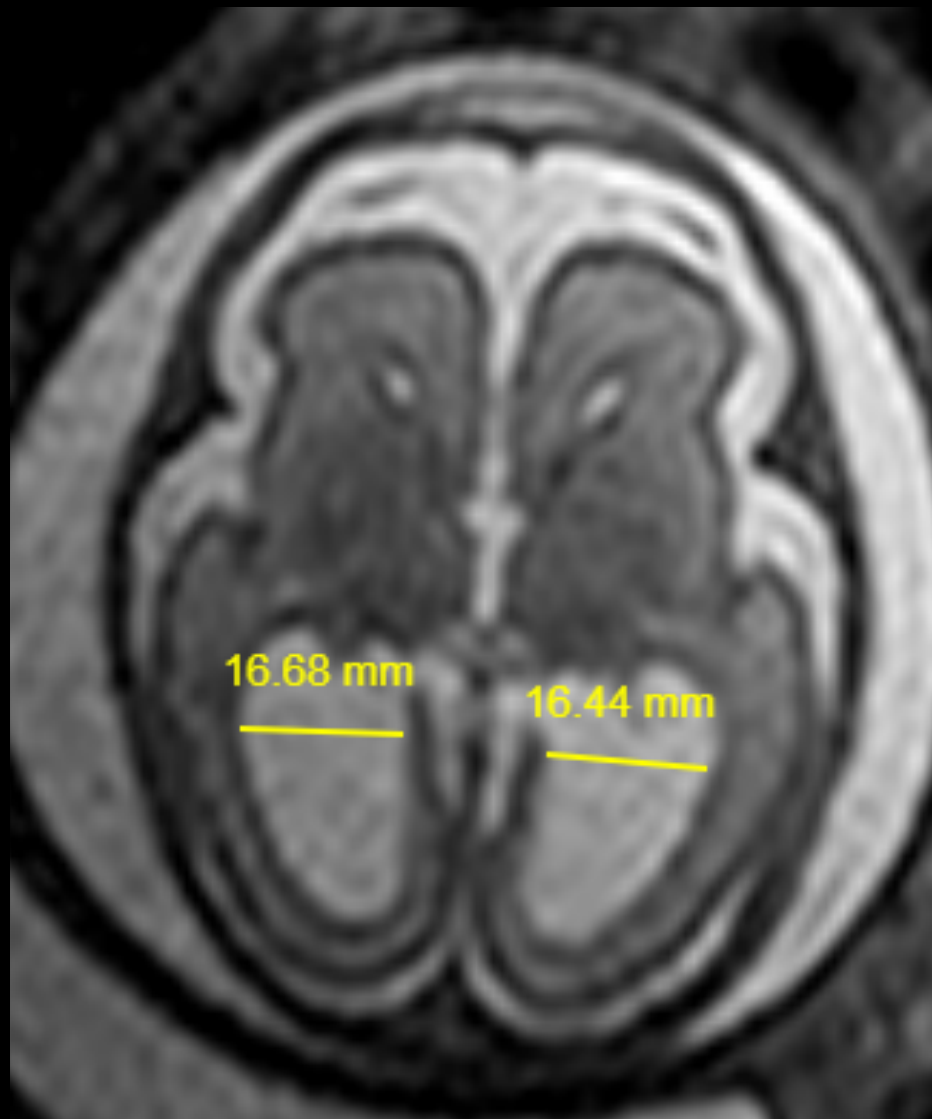


Axial



Sagittal





Caso n° 1

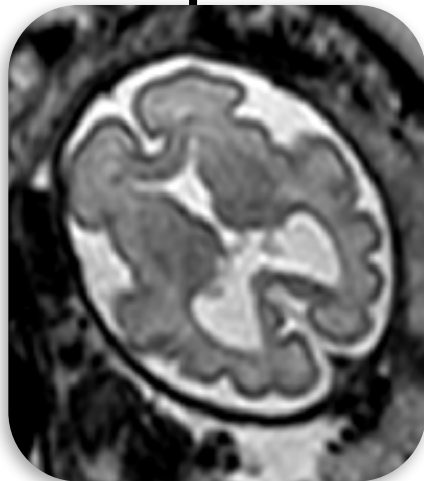
- NIPT: alto riesgo XO
- Confirma Cariotipo postnatal: Síndrome Turner

Diagnósticos diferenciales



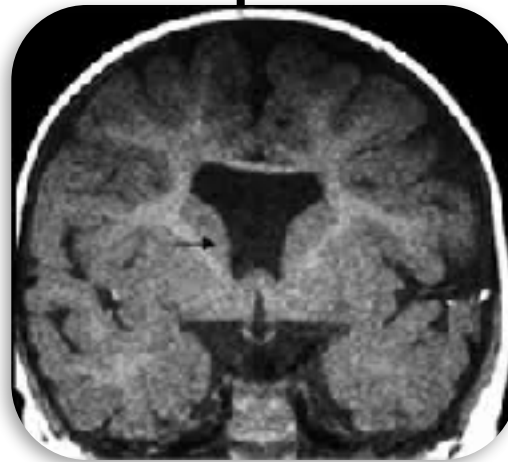
CSP ausente

monoVL ant + talamos no fusionados



Holoprosencefalia
lobar

Astas frontales fusionadas + CC presente



DSO

Astas frontales separadas + CC presente



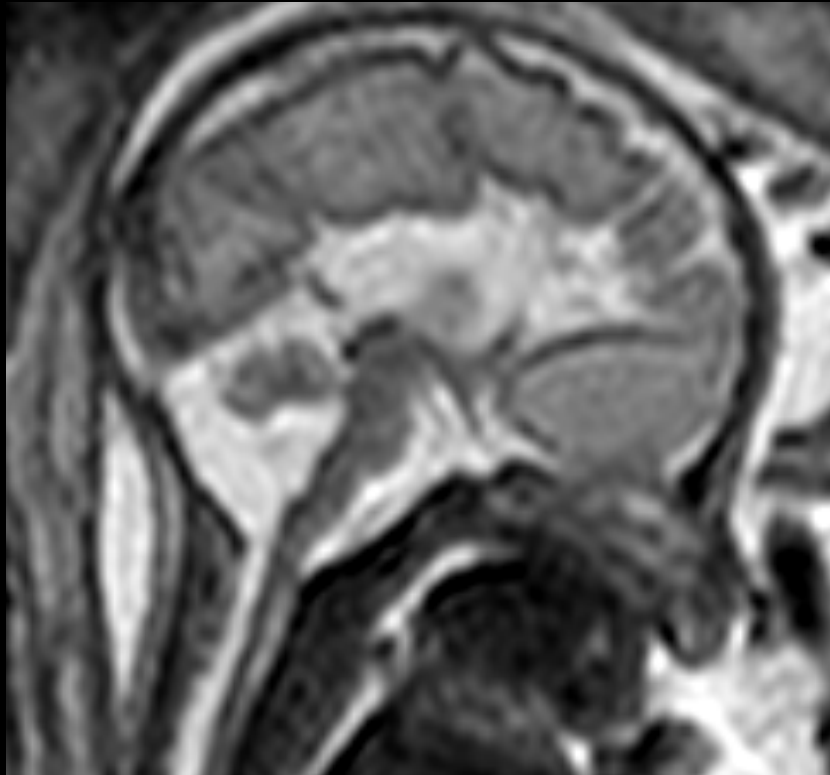
Agenesia
septal

Metodología de estudio

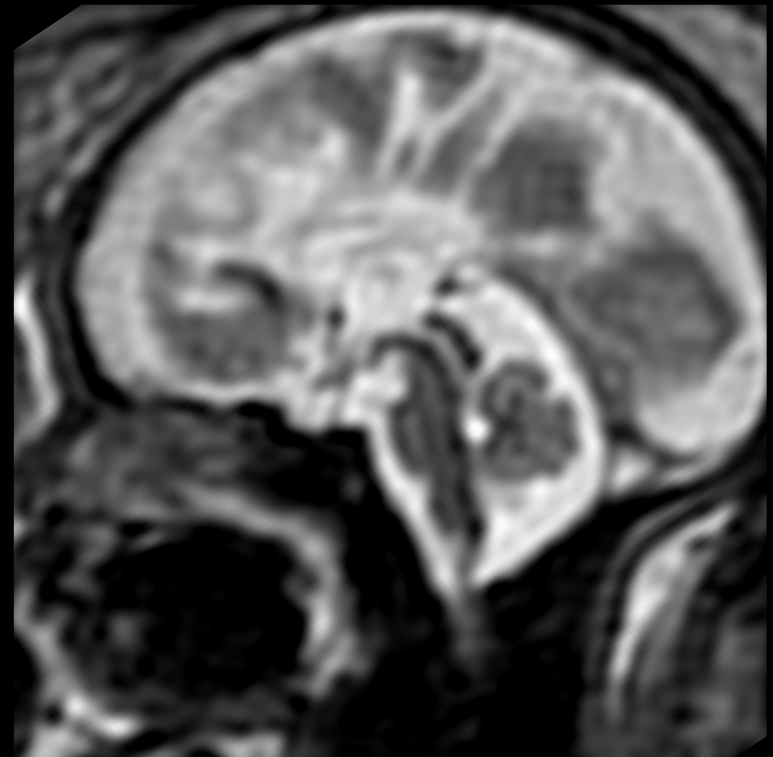
- Ecografía morfológica detallada + neurosonografía (abd/TV)
- Ecocardiograma fetal
- Cariotipo
- TORCH
- RMf a partir 30 semanas

PRONOSTICO

Asociada



Aislada



Outcome of prenatally diagnosed agenesis of the corpus callosum

N. Fratelli¹*, A. T. Papageorgiou¹, E. Prefumo¹, S. Bakalis², T. Homfray³ and B. Thilaganathan¹

¹Fetal Medicine Unit, Academic Department of Obstetrics and Gynaecology, St George's Hospital Medical School, London, UK

²Harris Birthright Research Centre for Fetal Medicine, Department of Obstetrics and Gynaecology, Kings College Hospital, London, UK

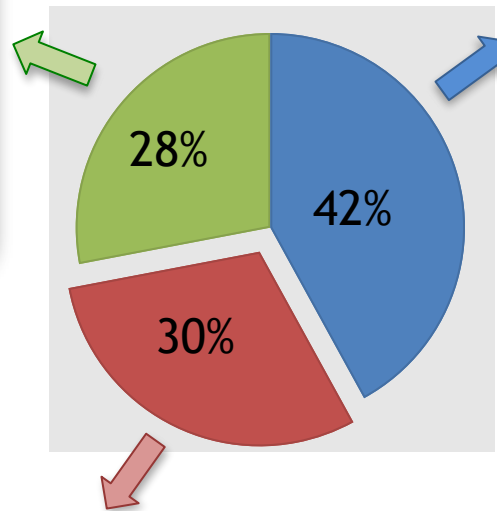
³Department of Genetics, St George's Hospital, London, UK

Table 1—Details of karyotype abnormalities occurring in 33 of the 117 cases of ACC

Karyotype abnormality	n	%
Aneuploidy	28	85
Trisomy 18	23	
Trisomy 13	1	
Trisomy 15	1	
Trisomy 21	1	
Triploidy	2	
Other abnormalities	5	15
46 XY var. (18p)	1	
46 XXdup (8 p)	1	
46 XYadd (1) (q47)	1	
45 XY -14, -20 + der(20)	1	
t (14:20) (q13.3 : q23)	1	
47 XY +der(9) t (9;9) (p11.2; q 13)	1	

T18

N: 117 casos



Defectos craneales: 60%
Defectos extracraniales: 71%

● cario normal
● aislados
● cario anormal

36% Retraso mental

Characteristics, associations and outcome of partial agenesis of the corpus callosum in the fetus

P. VOLPE*, D. PALADINI†, M. RESTA‡, A. STANZIANO*, M. SALVATORE§,
M. QUARANTELLI§, V. DE ROBERTIS*, A. L. BUONADONNA¶, G. CARUSO** and
M. GENTILE¶

Departments of *Obstetrics and Gynecology and †Medical Genetics, Hospital Di Venere, A.S.L. IASL and **Department of Pathological Anatomy and Genetics, University of Bari, Bari, ‡Fetal Cardiology Unit, Department of Gynecology and Obstetrics, University Federico II of Naples and §Biotechnology and Bio-Imaging Institute, National Research Council, Naples and ¶Department of Neuroradiology, Hospital SS Annunziata, Taranto, Italy



- Retrospectivo en pacientes con PACC, en 2 centros durante 10 años.
- N: 54 ACC → 19 PACC (35%)
- PACC es posible de diagnosticar prenatal: signos mas frecuente: Colpocefalia

47%: AISLADOS



25% retraso mental

Neurodevelopmental outcome following prenatal diagnosis of an isolated anomaly of the corpus callosum

R. MANGIONE*, N. FRIES*, P. GODARD*, C. CAPRON†‡, V. MIRLESSE*§, D. LACOMBE¶
and M. DUYME†‡

* Collège Français d'Échographie Fœtale (CFEF), France; † University of Montpellier 3, Montpellier, France; ‡ CNRS, University of Montpellier 1-Nîmes, Montpellier, France; § Maternité Pichat, Paris, France; ¶ Department of Medical Genetics, University of Bordeaux 2, CHU Bordeaux, Bordeaux, France

- Prospectivo: casos y controles
- ACC aislados con diagnóstico US + RMf
- Los niños fueron evaluados mediante un neuropediatra para responder un cuestionario (CDI, Ireton`s Child Developmental Inventory)
- N: 27 ACC. Casos control: 44.

15% (4 casos)
Hallazgos adicionales

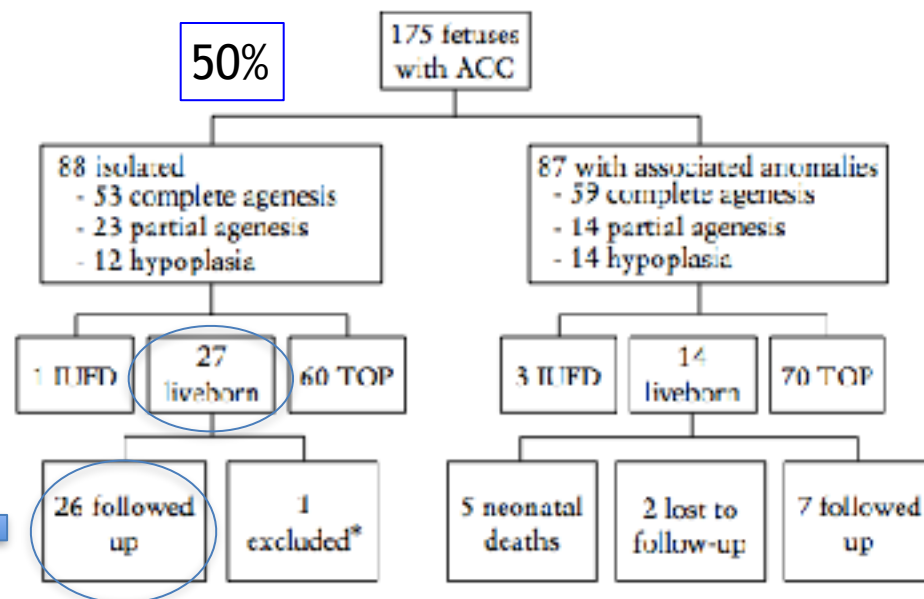


Figure 1 Outcome of pregnancies with fetal anomalies of the corpus callosum (ACC). IUFD, intrauterine fetal death; TOP, termination of pregnancy. *Excluded because age 14 months at assessment.

Neurodevelopmental outcome following prenatal diagnosis of an isolated anomaly of the corpus callosum

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Resultados:

- Mejores resultados para el grupo control ($p < 0.05$) con respecto a motor grueso, fino, comprensión del lenguaje, números y desarrollo general.
- 7 niños de los 26 (27%) tuvieron retraso del neurodesarrollo, en contraste solo 1 niño de los controles tuvo desarrollo borderline.

Conclusiones:

- DX prenatal de ACC debe realizarse mediante eco + RMf (S85%) → 15% hallazgos adicionales postnatales.
- Meta-análisis 9 estudios publicados sugieren que el desarrollo de los niños con dx prenatal de ACC aislada es NORMAL en 70%. → significa que el riesgo de retraso mental en un feto con dx de ACC aislada es de 27%.

Conclusiones

- Disgenesias callosas son poco frecuente: 0,5%
- No tan sencillo diagnóstico, *pero posible!!*
- Aisladas (30%) o Asociadas
- Aisladas: el pronóstico similar en CACC y PACC → retraso mental en 30%
- 70% buen pronóstico, pueden aparecer defectos neuropsicológicos, motores o sensoriales sutiles en etapas mas tardías