idcsalud
Hospital General de Catalunya

Perfil Pla Fetal

XXII JORNADA CATALANA DE DIAGNÒSTIC PRENATAL

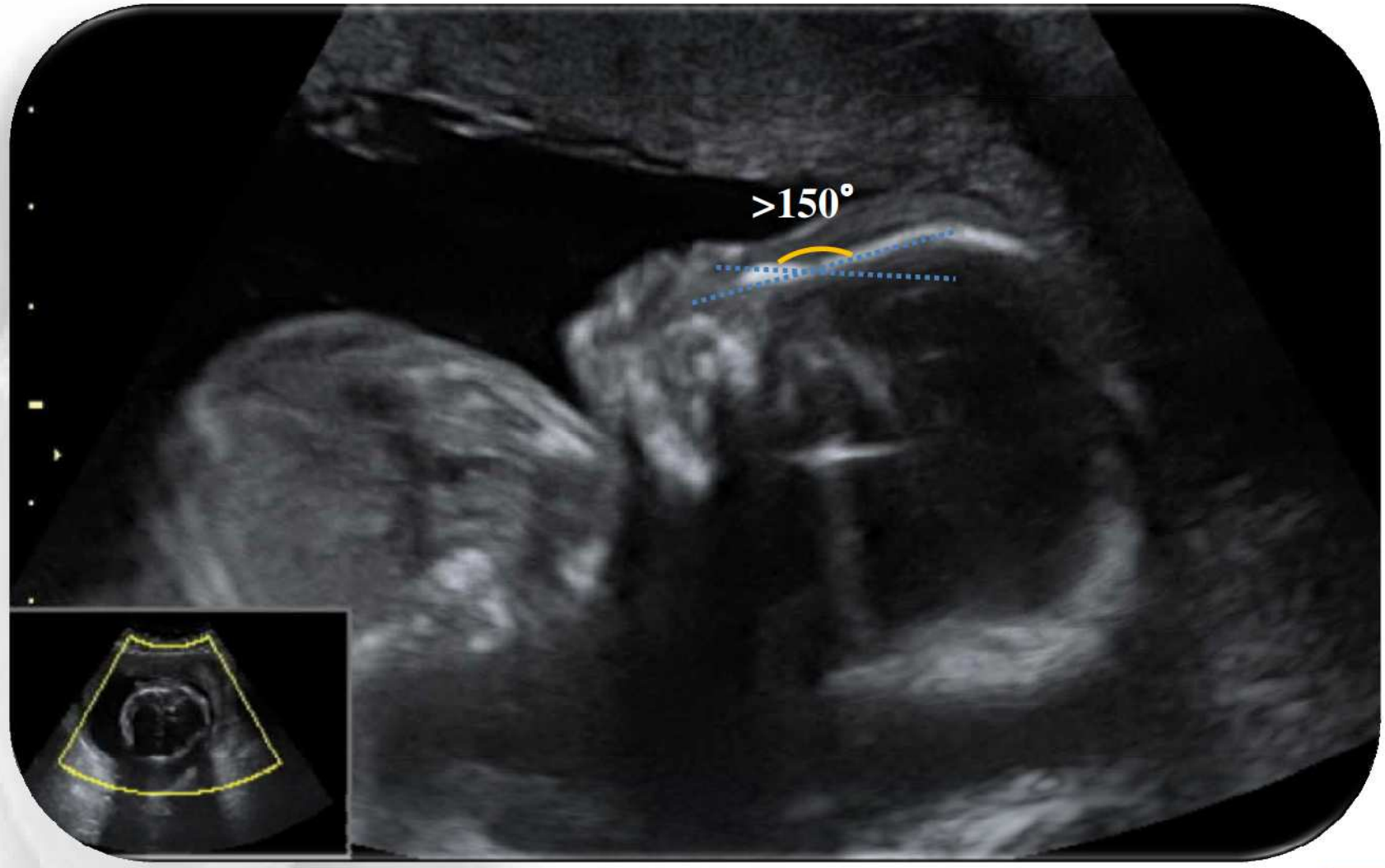
Dra N. Souliman, Dra E. Gil, Dra H. Bagan, Dra M. Gómez, Dra L. Rodellar, Dr M.A. Jiménez

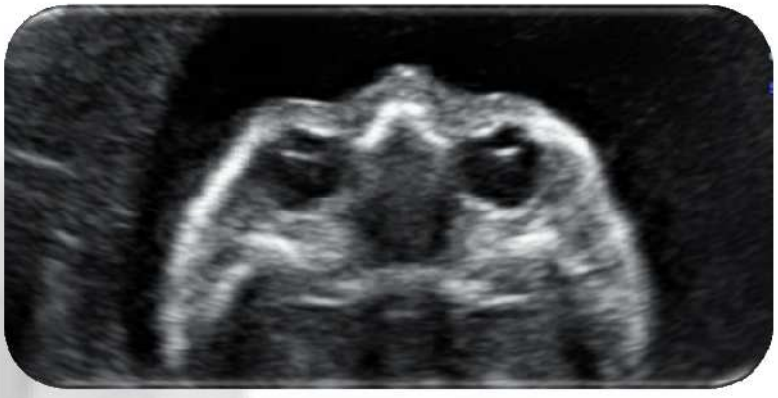
Març 2014

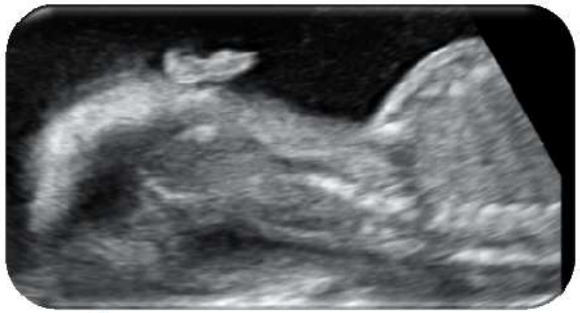
Cas Clínic

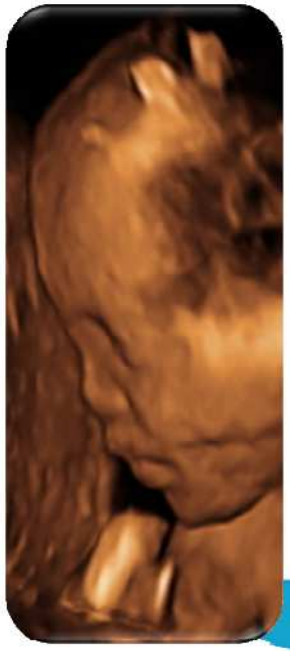
- **34 anys, CAUCÀSICA**
- **TPAL 0000**
- **No antecedents familiars.**
- **No consanguinitat.**
- **Tabaquisme**
- **Sense exposicions prenatals conegudes . No consum d'alcohol ni fàrmacs anticoagulants.**
- **Sense antecedents d'hiperemesi ni dèficits vitamínics.**
- **Sense antecedents patològics d'interès.**
- **Risc T21: 1/341**

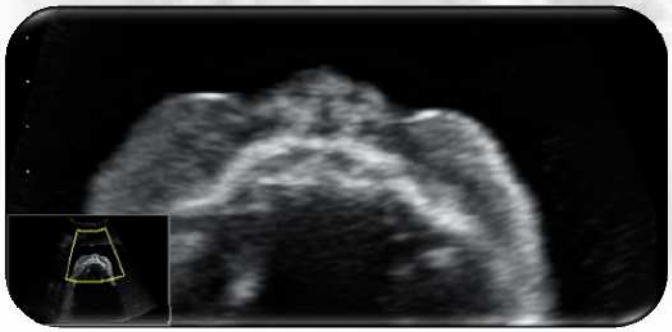

96
Gn -3
WMF 60 Hz
Ángulo SV 0
Tamaño 2.0mm
Frq baja
PRF 4.4kHz
11cm/s
-11cm/s
Voluson
E6
1 Trim.
Har-alta
82
Gn 1
C7 / M7
P3 / E3
SRI II 3
100
Gn -9.2
Frq baja
Calidad norm
WMF baja2
PRF 0.9kHz
-45
-30
-15
cm/s
15
30
45


>150°

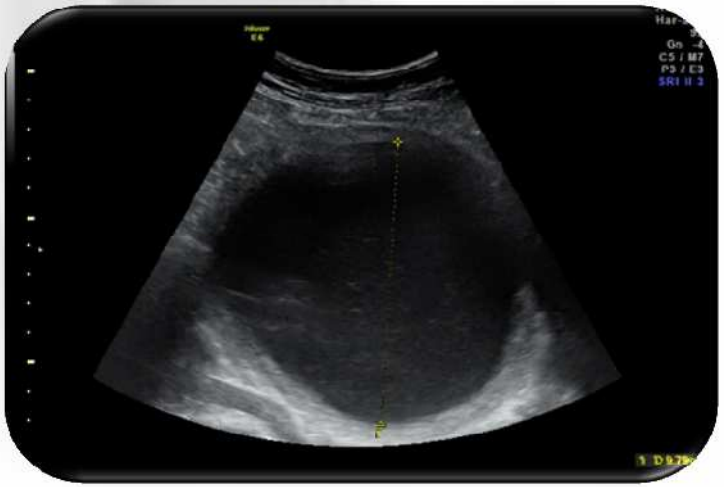


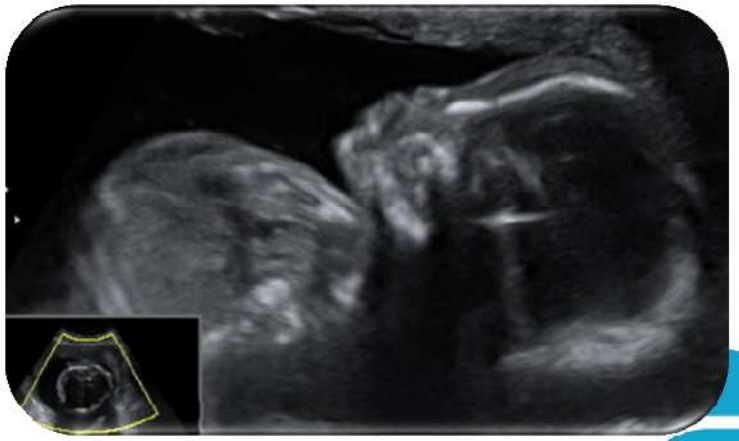


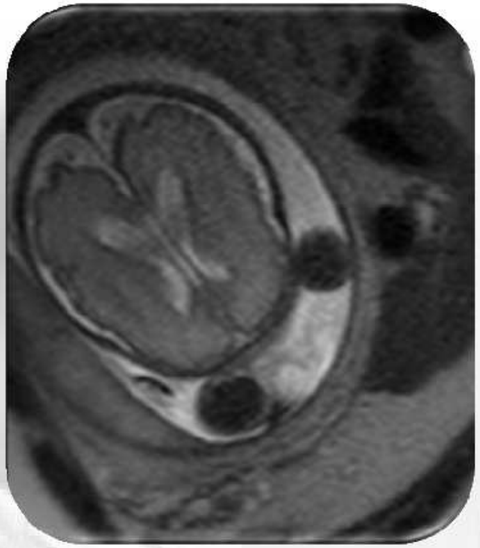


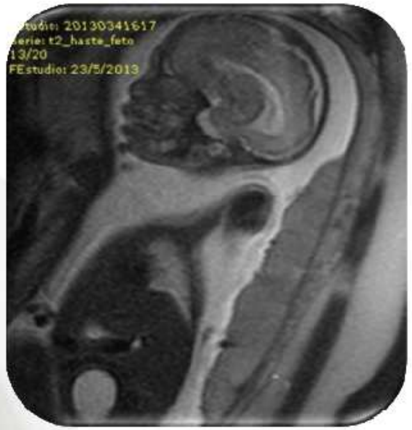



Ecografía
Morfològica





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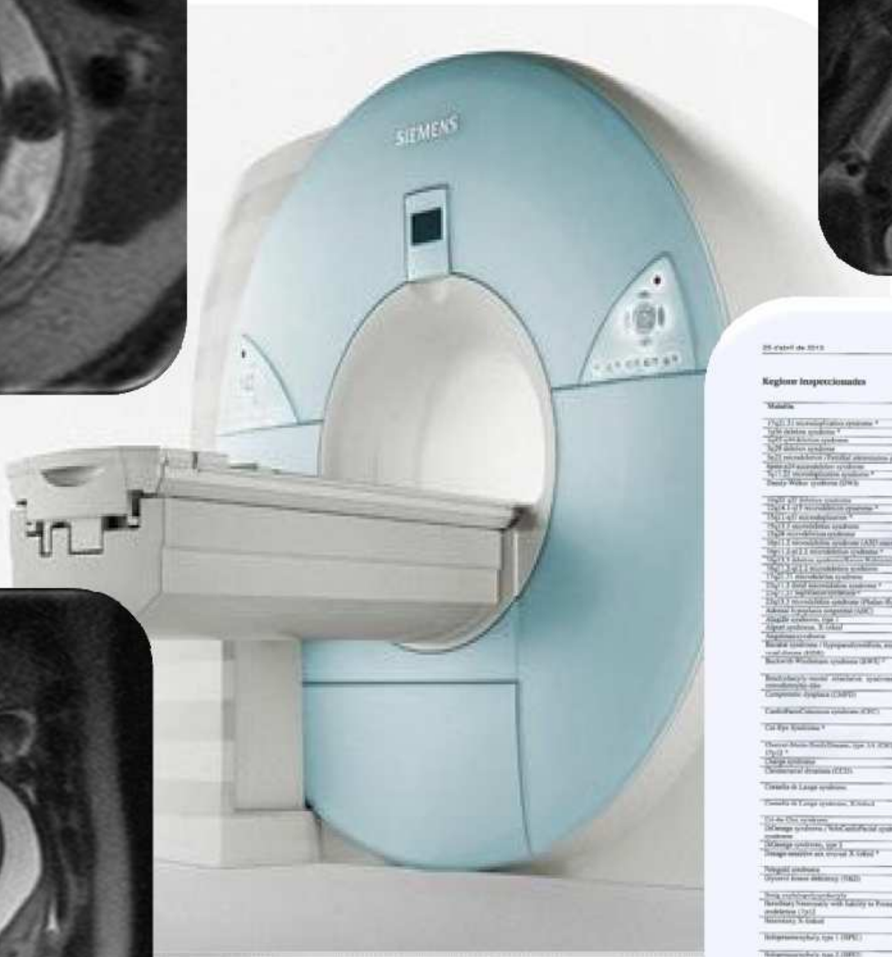
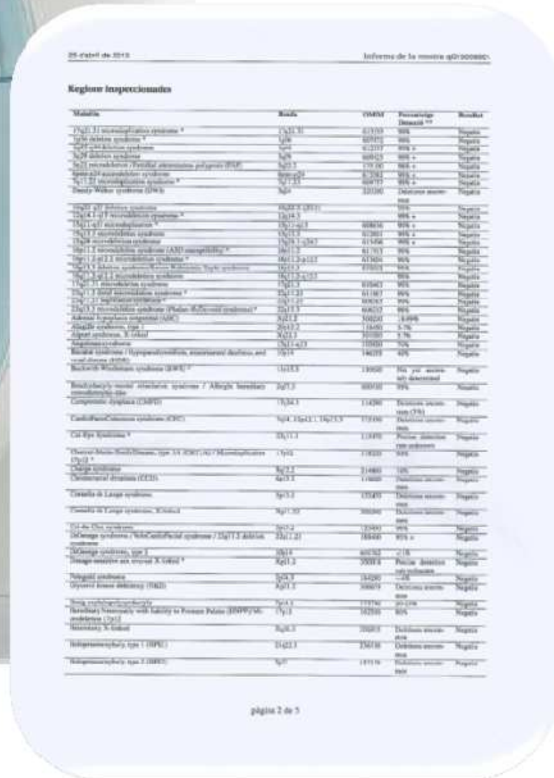
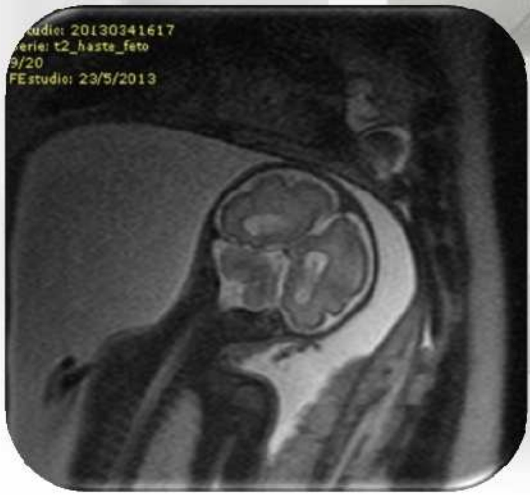
erie: t2_haste_feto
13/20
FEstudio: 23/5/2013


SIEMENS

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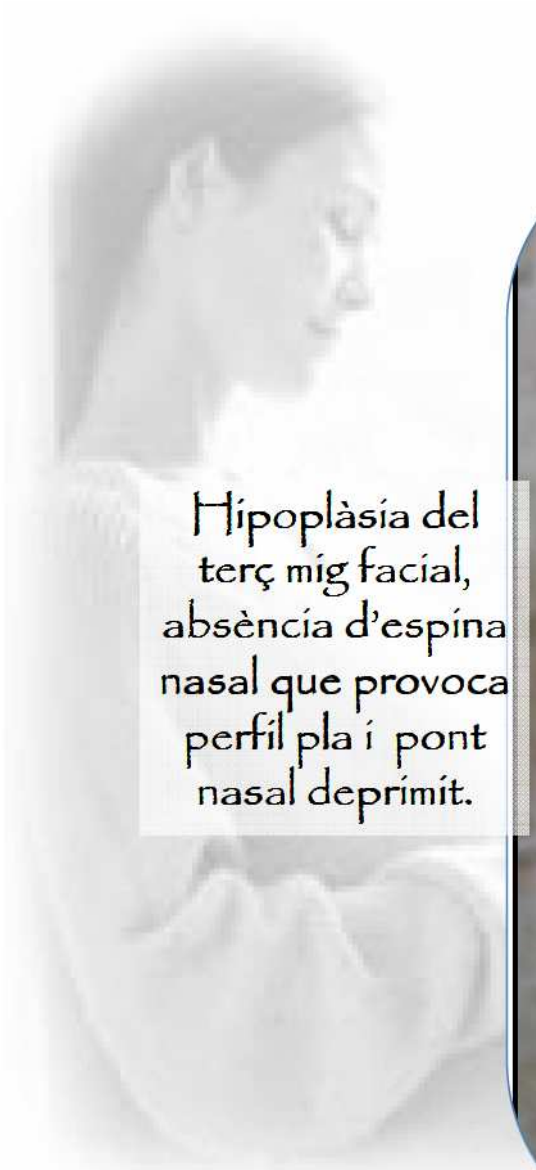
FEstudio: 23/5/2013

Fenotip Binder



Hipoplàsia del terç mig facial, absència d'espina nasal que provoca perfil pla i pont nasal deprimit.

- ✓ Incidència < 1/10.000. (infradiagnòstic)
- ✓ Fenotip heterogeni associat a diferents etiologies. Pot ser un signe important de condrodisplasia punctata.
- ✓ Anomalia cromosòmica (Sd Down)
- ✓ Metabolopaties (Sd de Zellweger)
- ✓ Alteracions del metabolisme de la vitamina K
  - ✧ factors intrínsecs (autoinmunes LES, litiasi biliar)
  - ✧ factors extrínsecs (warfarina, alcohol...)
- ✓ Microdeleció 4p Sd Wolf-Hirschhorn, Sd Apert, Sd Crouzon, Sd Robinow, Sd Keutel...
- ✓ Braquitelefalangia

Ultrasound Obstet Gynecol 2000; 16: 578–581.

The prenatal diagnosis of Binder syndrome before 24 weeks of gestation: case report

K. COOK, F. PREFUMO, F. PRESTI, T. HOMFRAY* and S. CAMPBELL
*Department of Obstetrics and Gynaecology and *Department of Genetics, St. George's Hospital Medical School, London, UK*

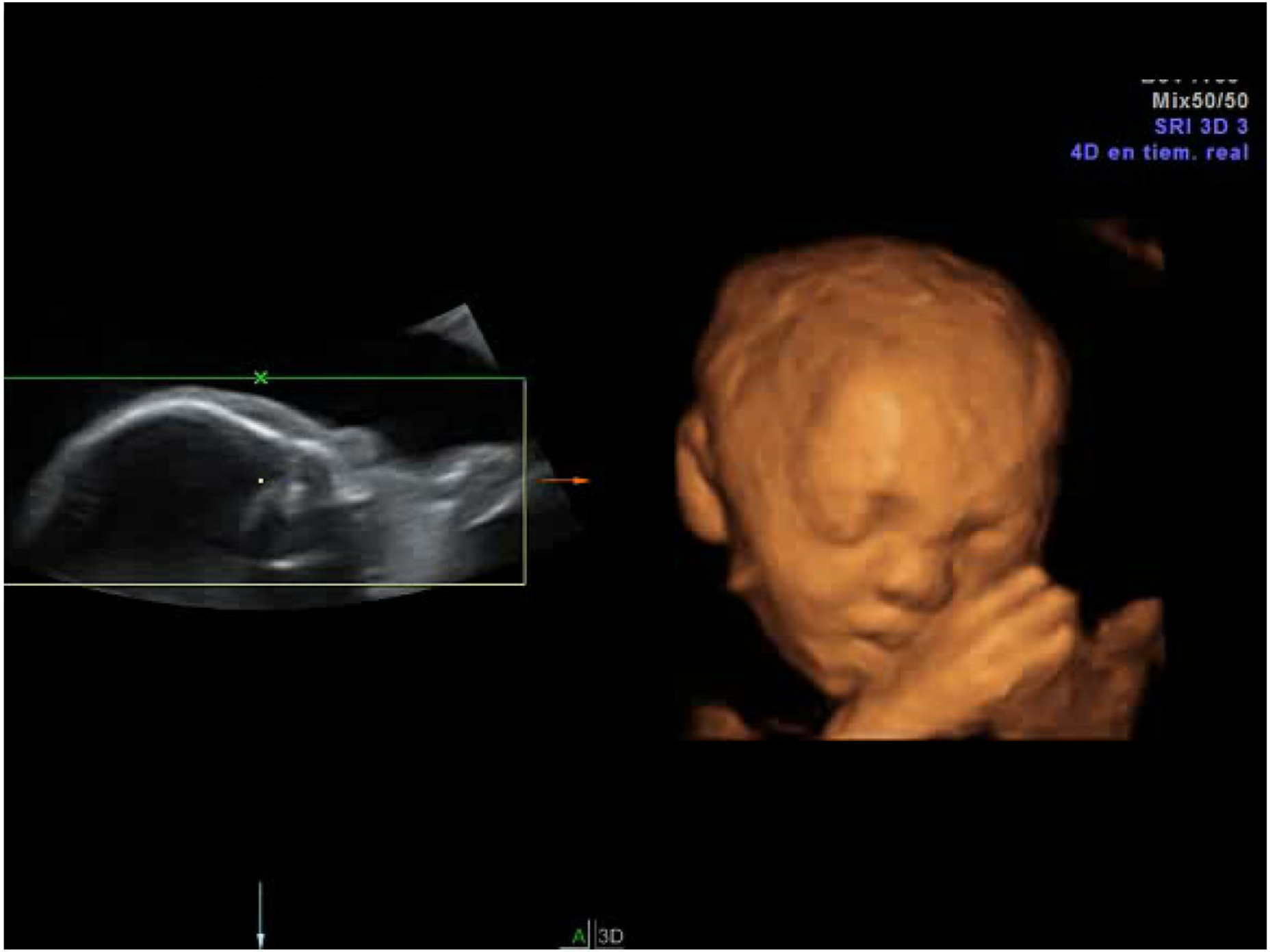
J Matern Fetal Neonatal Med. 2012 Aug;25(8):1413-8. doi: 10.3109/14767058.2011.636105. Epub 2011 Dec 7.

Binder phenotype in mothers affected with autoimmune disorders.

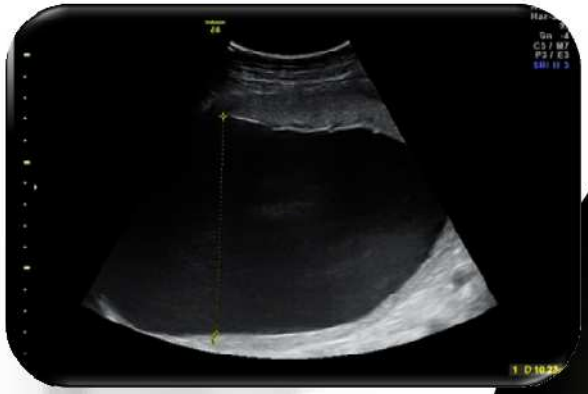
Colin E[1], Touraine R, Levaillant JM, Pasquier L, Boussion F, Ferry M, Guichet A, Barth M, Mercier A, Gérard-Blanluet M, Odent S, Bonneau D.

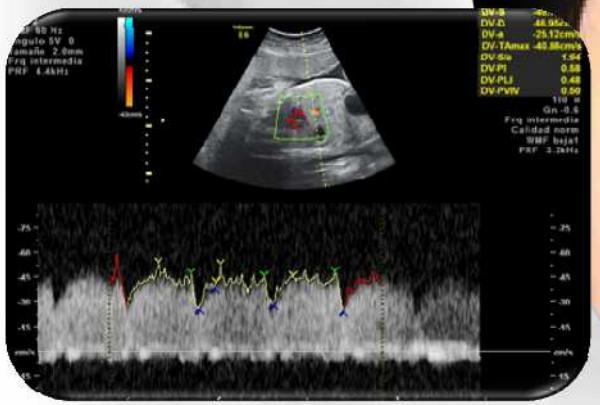
Table 1 Common facial and other ultrasound findings in relation to their most common syndrome[3–4,7,8–9]

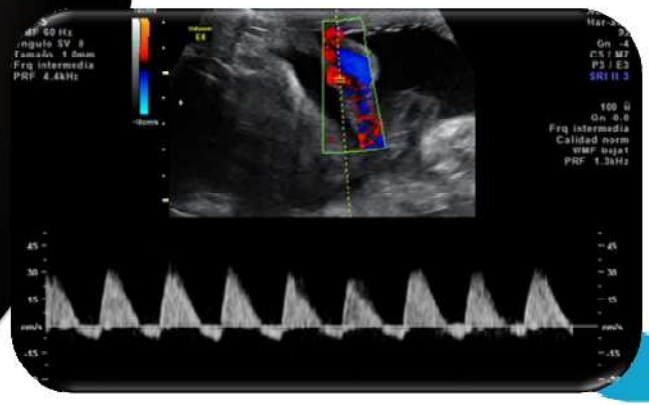
	Common facial ultrasound features	*Other ultrasound features*	*Inheritance*	*Karyotype*
Binder syndrome	Flat mid-face, nasal hypoplasia, hypertelorism		Autosomal recessive Autosomal dominant	Normal
Chondrodysplasia punctata (CDP), X linked	Nasal hypoplasia	Scoliosis, asymmentrical shortening of the limbs	X-linked dominant	Deletion/translocation of chromosome Xp22.3
CDP, rhizomelic type	Flat face	Epiphyseal stippling, talipes, short limbs	Autosomal recessive	Normal
Robinow syndrome	Flat face, hypertelorism	Short forearms, clinodactyly, macrocephaly	Autosomal dominant	Normal
Aarksog syndrome	Flat nose, hypertelorism	Brachydactyly, clinodactyly of the 5th fingers	X-linked recessive	Normal
Crouzon syndrome	Maxillary hypoplasia, hypertelorism	Craniosynostosis, short occipital-frontal diameter	Autosomal dominant	Normal
Apert syndrome	Flat nose, maxillary hypoplasia, hypertelorism	Irregular craniosynostosis, short occipital-frontal diameter, flat occiput, ventriculomegaly, syndactyly	Autosomal dominant	Normal
Achondroplasia	Mid-face hypoplasia low nasal bridge	Short tubular bones, megalocephaly	Autosomal dominant	Normal
Fetal warfarin syndrome	Flat face	Stippling (mild)	(Teratogen)	Normal
Rudiger syndrome	Flat nasal bridge	Short digits	Autosomal recessive	Normal
Stickler syndrome	Facial cleft, micrognathia, flat face	Osteo-chondrodysplasia, talipes	Autosomal dominant	Normal


Mix50/50
SRI 3D 3
4D en tiem. real
A 3D

Perfil Pla Fetal







Conclusions

- Descartar:
  - Cromosomopaties
  - Metabolopaties
  - Malalties autoinmunes
  - Dèficits de vitamina K

- Informar a la família de la possibilitat de que es tracti d'una variant de la normalitat (Herència autosòmica dominant)

Gràcies per la seva atenció