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

MARCADORES BLANDOS Y SU PODER DIAGNÓSTICO EN ANEUPLOIDÍAS

Dr. Nicolás Zapata Lara
Becado Obstetricia y Ginecología
Universidad de Concepción
CERPO

Semifinal

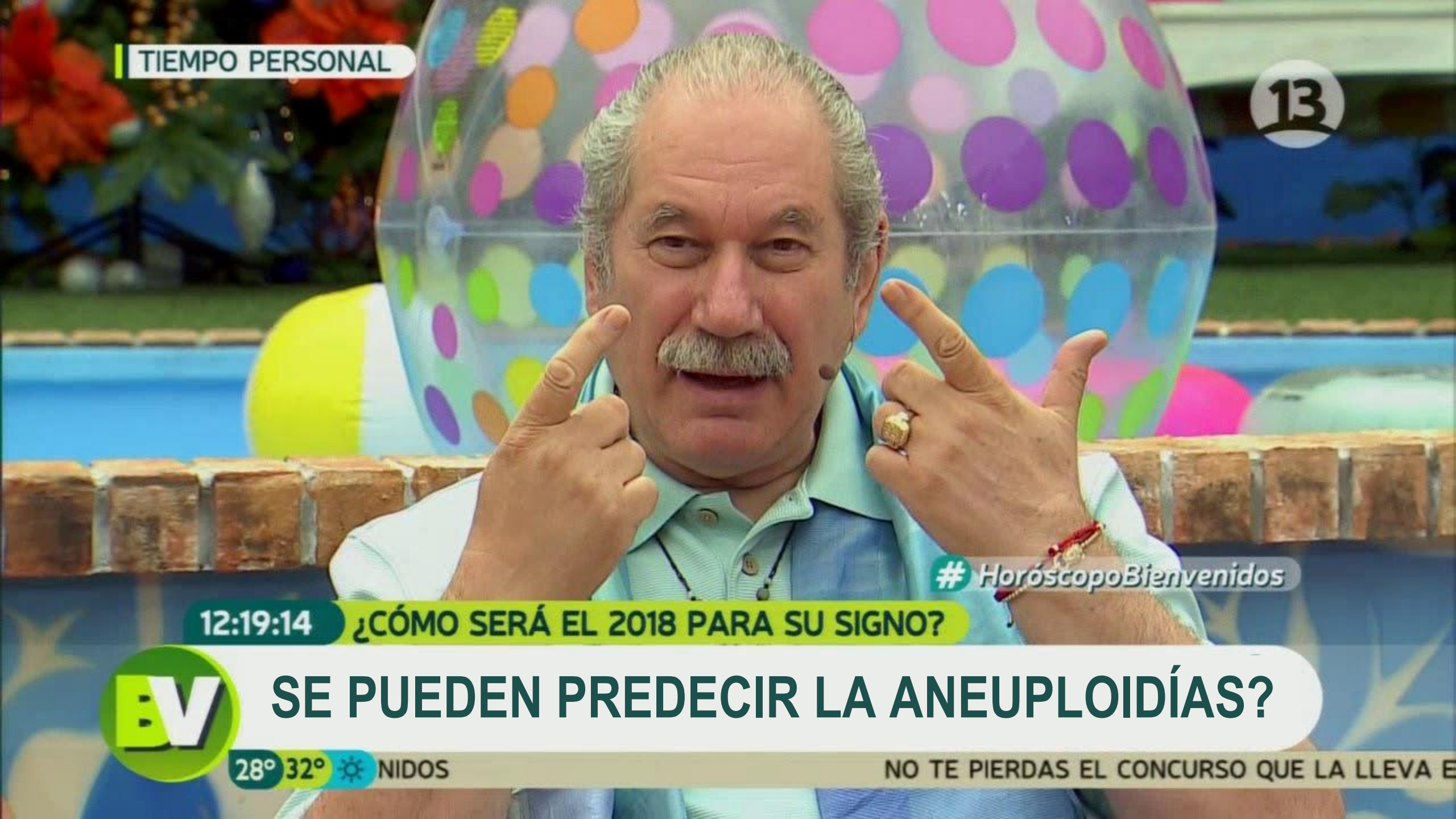
Brasil	Argentina	Mañana 20:30	Chile	Perú	Mié., 03-07 20:30

Partido por el tercer lugar

Por definirse	Por definirse	Sáb., 06-07 15:00			

Final

Por definirse	Por definirse	Dom., 07-07 16:00			


TIEMPO PERSONAL
13
#HoróscopoBienvenidos
12:19:14
¿CÓMO SERÁ EL 2018 PARA SU SIGNO?
SE PUEDEN PREDECIR LA ANEUPLOIDÍAS?
28° 32°
NIDOS
NO TE PIERDAS EL CONCURSO QUE LA LLEVA E

CERPO

LH+ / LH-

HIPOPLASIA/ AGENESIA HUESO NASAL

- Corte sagital medio
- Cabeza fetal utilizando la mayor parte de la imagen
- Ángulo del HN cercano a 45°
- Se considera hipoplasia p < 2,5
- 30 - 60% T21
- 0,5 - 7% euploides

Sonek and croom. Second trimester ultrasound markers. Clinical Obstetrics and Gynecology Volume 57, number 1, 159–181. 2014

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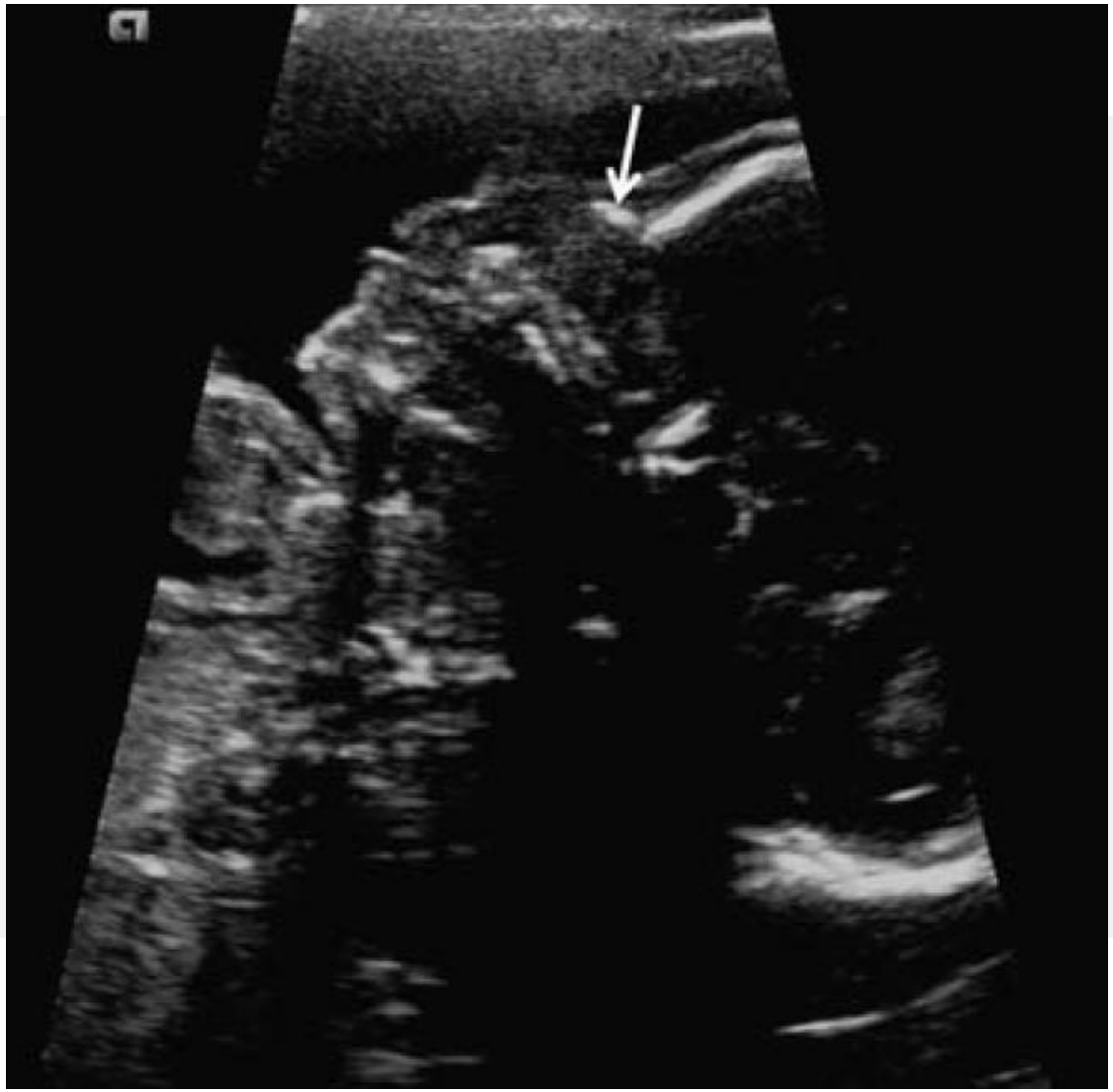


BEDON VILLANUEVA, MARIA 04.09.198
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05.09.2018 11:32:22 AM
TIs 0.2
TIb 0.2
MI 0.9
RAB6-D
OB
15.0cm / 2.5
65°/ 41Hz
3 Trim.
HL 7.00 - 4.40
Gn -12
C6 / M7
FF4 / E3

The Journal of Maternal-Fetal and Neonatal Medicine, April 2011; 24(4): 555–558

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healthcare

Isolated absent or hypoplastic nasal bone in the second trimester fetus: is amniocentesis necessary?

YUEN HA TING, TERENCE T. LAO, TZE KIN LAU, MAN KIN CHUNG, & TAK YEUNG LEUNG

Department of Obstetrics and Gynaecology, Fetal Medicine Unit, Prince of Wales Hospital, The Chinese University of Hong Kong, Hong Kong

- Retrospectivo
- 14 pacientes con ausencia/hipoplasia
- 0% aneuploidía al ser aislado
- 75% T21 al tener otros hallazgos asociados (p=0,01)

J. Fetal Med. (June 2015) 2:75–78
DOI 10.1007/s40556-015-0051-1

ORIGINAL ARTICLE

Absent/Hypoplastic Fetal Nasal Bone and Its Association with Aneuploidies

Pratima Dash[1] · Ratna Dua Puri[1] · Manisha Goyal[2] · Sunita Bijarnia[1] · Meena Lall[1] · Udhaya Kotecha[1] · Ishwar Chander Verma[1]

- Prospectivo
- 92 pacientes con agenesia/hipoplasia
- Aislado 12,9%
- Con otros hallazgos 40%

AOGS
Acta Obstetricia et Gynecologica Scandinavica

AOGS ORIGINAL RESEARCH ARTICLE

Absent fetal nasal bone in the second trimester and risk of abnormal karyotype in a prescreened population of Chinese women

YAN DU[1], YUNYUN REN[2], YINGLIU YAN[2] & LI CAO[2]

[1]Office of Clinical Epidemiology, Obstetrics and Gynecology Hospital of Fudan University, Shanghai, and [2]Ultrasound Department, Obstetrics and Gynecology Hospital of Fudan University, Shanghai, China

- Retrospectivo
- 56707 pacientes -> 71 casos (ausencia HN)
- Ausencia sola: 7,1%
- Ausencia + blando: 17,6%
- Ausencia + estructural: 83,3%

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Zhonghua Yi Xue Za Zhi. 2018 Nov 20;98(43):3532-3535. doi: 10.3760/cma.j.issn.0376-2491.2018.43.0

[Application of chromosomal analysis for 29 cases of fetuses with nasal bone absence or hypoplasia].

[Article in Chinese; Abstract available in Chinese from the publisher]

Hou L[1], Wang XX, Jiang HL, Zhang T, Li L, Zhang WY, Wang X.

- Retrospectivo
- 29 pacientes hipoplasia/agenesia
- Hipoplasia 40%
- Ausencia 54,2%

Ultrasound Obstet Gynecol 2005; 25: 19–24
Published online in Wiley InterScience (www.interscience.wiley.com). **DOI:** 10.1002/uog.1805

Three-dimensional ultrasound with maximal mode rendering: a novel technique for the diagnosis of bilateral or unilateral absence or hypoplasia of nasal bones in second-trimester screening for Down syndrome

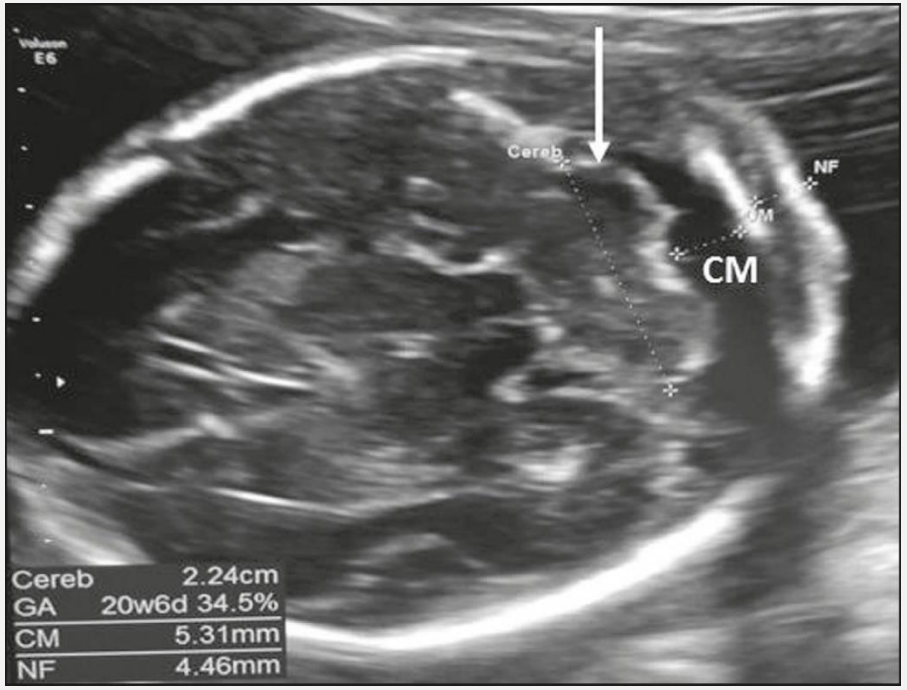
B. BENOIT* and R. CHAOUI†

**Maternité, Hôpital Princesse Grace, Monaco and †Unit of Prenatal Diagnosis and Therapy, Charité University Hospital CCM, Berlin, Germany*

- 20 pacientes con dg prenatal de T21
- 9 con ausencia HN en eco 2D
- 3 con discrepancia al 3D (33%)

PLIEGUE NUCAL AUMENTADO

- Corte transversal
- CSP – Tálamo – Cerebelo
- Borde externo occipital a borde externo de la piel
- 6 mm: p = 99
- 20-33% T21
- 0,5-2% euploides



Sonek and croom. Second trimester ultrasound markers. Clinical Obstetrics and Gynecology Volume 57, number 1, 159–181. 2014

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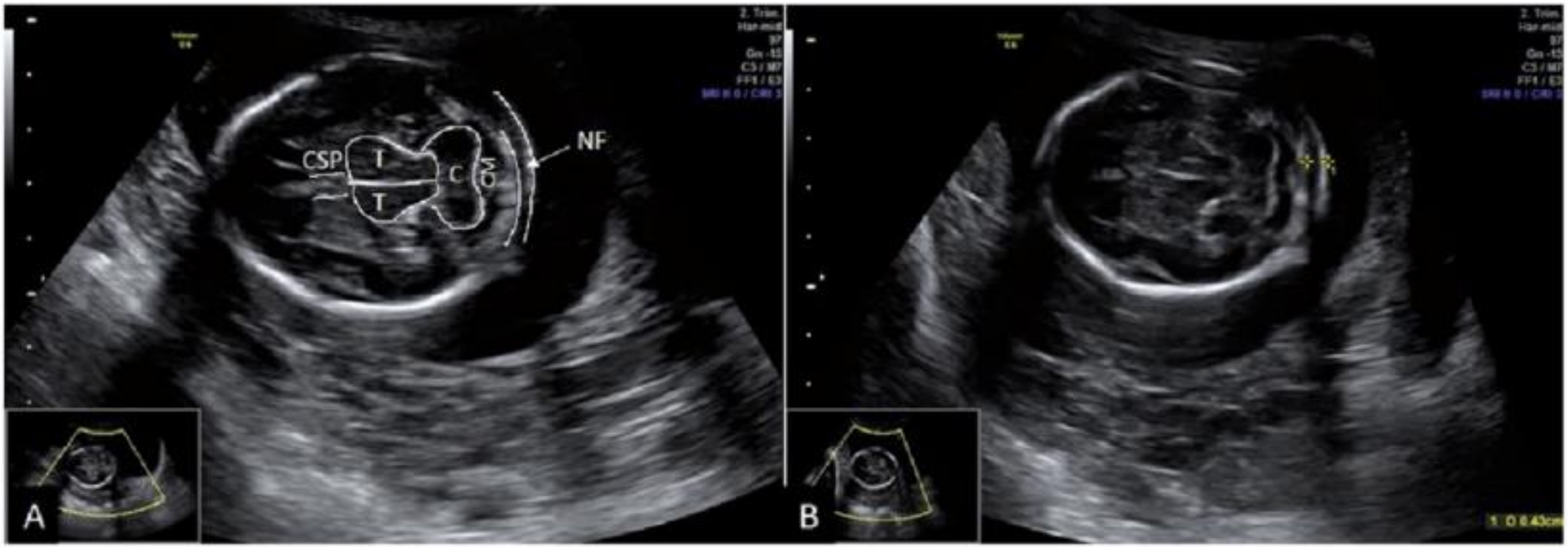

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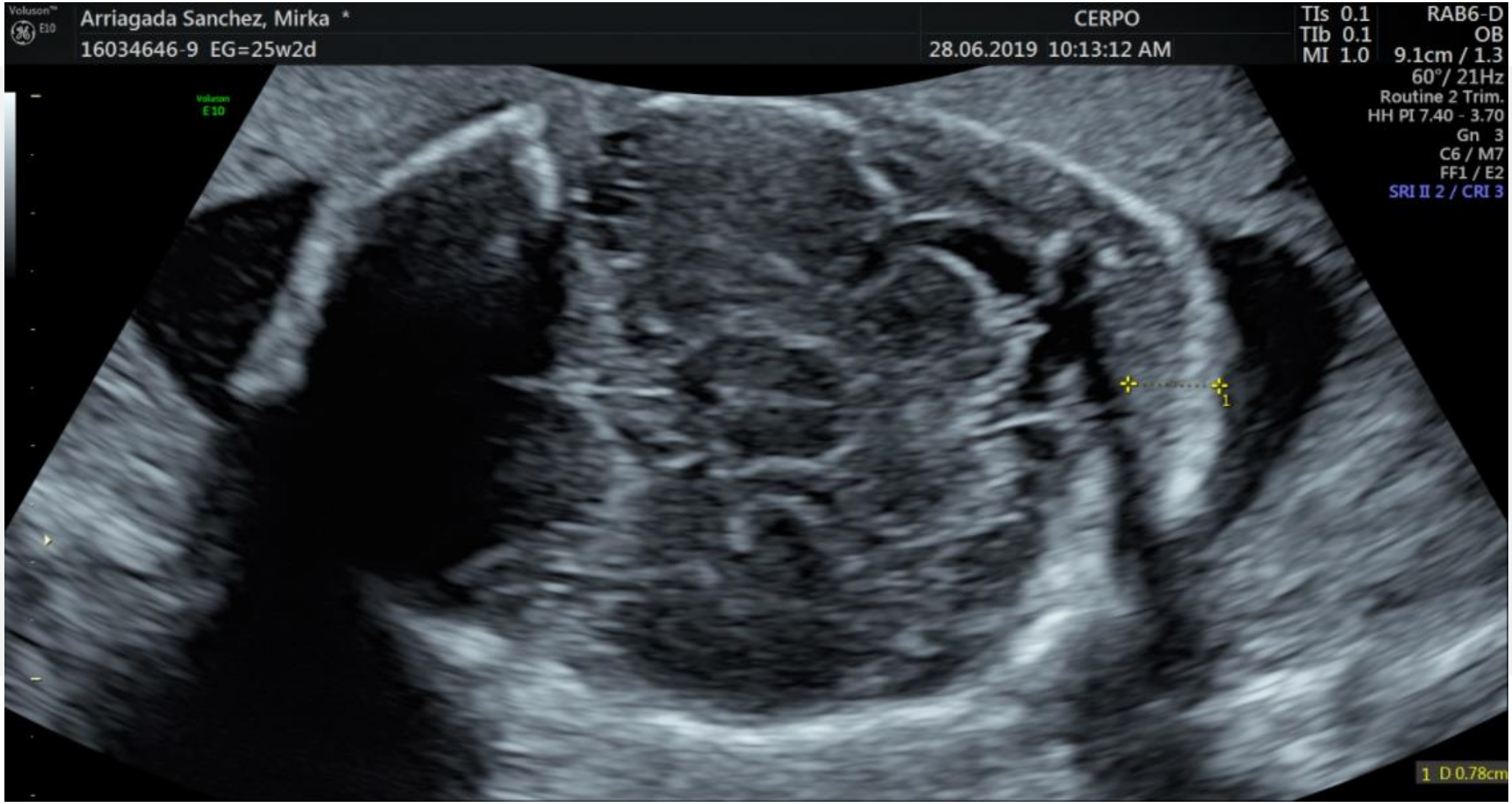
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TIb 0.1
MI 1.0
RAB6-D
OB
9.1cm / 1.3
60°/ 21Hz
Routine 2 Trim.
HH PI 7.40 - 3.70
Gn 3
C6 / M7
FF1 / E2
SRI II 2 / CRI 3
1 D 0.78cm

260 Second trimester nuchal fold thickness and pregnancy outcomes

Shelly Soni, Burton Rochelson, Matthew J. Blitz, Nidhi Vohra, Caroline Drewes Pessel

Hofstra-Northwell School of Medicine, Manhasset, NY

- Retrospectivo
- 10416 pacientes, 149 NF > 6mm
- 11% aneuploidía
- Euploides -> sin riesgo de malformación

Observational Study

Medicine®

OPEN

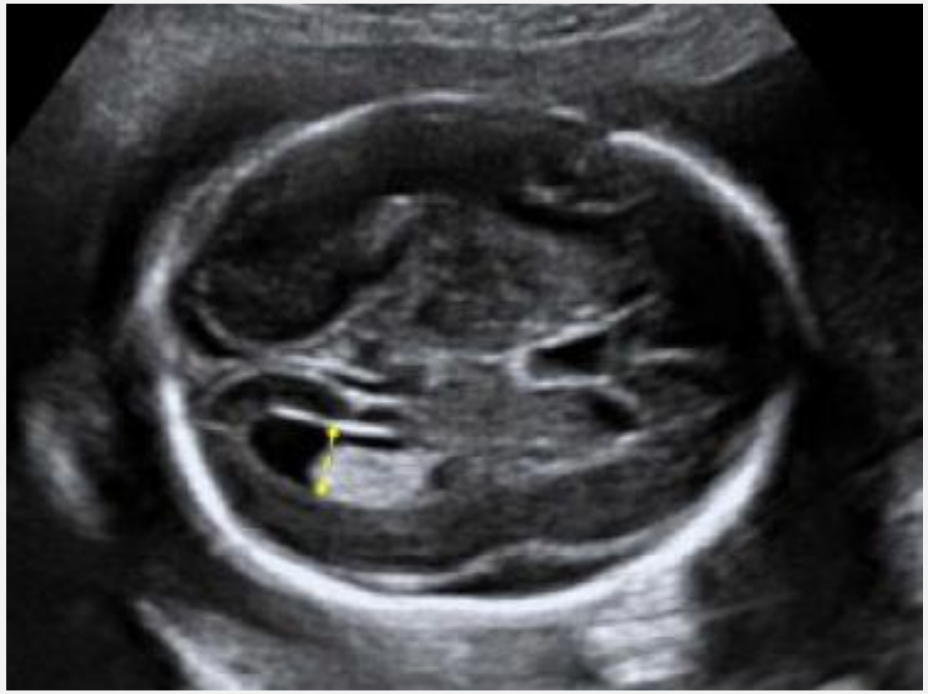
Prenatal diagnosis and pregnancy outcome analysis of thickened nuchal fold in the second trimester

Lushan Li, MD*, Fang Fu, PHD, Ru Li, PHD, Zequn Liu, MD, Can Liao, MD*

- Retrospectivo
- 72 pacientes
- Asociado a anomalía estructural 35,5%
- Aislado 3,7%

VENTRICULOMEGALIA LEVE

- Corte transversal
- CSP – Tálamo – Atrium
- Borde interno a borde interno
- Valor entre 10 y 15 mm
- 0,15% de los euploides
- 4,7% aneuploidías



Sonek and croom. Second trimester ultrasound markers. Clinical Obstetrics and Gynecology Volume 57, number 1, 159–181. 2014

Aneuploidy and Isolated Mild Ventriculomegaly

Attributable Risk for Isolated Fetal Marker

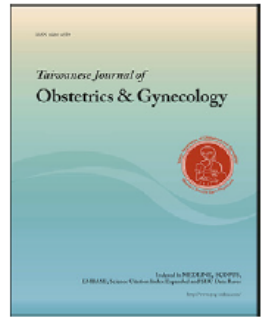
Melissa Terry Byron C. Calhoun William Walker Christina Apodaca
Laura Martin Brian Pierce Roderick F. Hume Mark I. Evans

- Retrospectivo
- 46 pacientes con VM.
- 25 con AMCT -> 12% aneuploidía

Contents lists available at ScienceDirect

Taiwanese Journal of Obstetrics & Gynecology

journal homepage: www.tjog-online.com



Original Article

The application of chromosomal microarray analysis to the prenatal diagnosis of isolated mild ventriculomegaly

Hong-Lei Duan, Xiang-Yu Zhu, Yu-Jie Zhu, Xing Wu, Guang-Feng Zhao, Wan-Jun Wang, Jie Li*

Department of Obstetrics and Gynecology, Affiliated Drum Tower Hospital, Medical School of Nanjing University, Nanjing, China

- Retrospectivo
- 101 pacientes con VM leve
- 3% de aneuploidías (2 T21 – 1 Turner)

Perinatal and long-term outcome in fetuses diagnosed with isolated unilateral ventriculomegaly: systematic review and meta-analysis

Scala C, Familiari A, Pinas A, Papageorghiou AT, Bhide A, Thilaganathan B and Khalil A

Fetal Medicine Unit, St George's Hospital, St George's University of London, London, UK

- Metanalisis
- 108 pacientes con ventriculomegalia unilateral aislada
- 0/108 aneuploidías
- 8,2% TORCH

FOCO HIPERECOGÉNICO CARDIACO

- Foco con ecogenicidad similar al hueso
- Habitualmente se visualizan en el corte 4C
- 90% en el VI
- 30% asiáticos, 11% blancos, 6% negros.

Sonek and croom. Second trimester ultrasound markers. Clinical Obstetrics and Gynecology Volume 57, number 1, 159–181. 2014

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PIZARRO, HELEN *
E64166-18-04-25-1 EG=28w2d
CERPO
31.05.2018 10:39:54 AM
TIs 0.2
TIb 0.2
MI 1.0
RAB6-D
OB
11.8cm / 2.2
30°/ 91Hz
2+3 Trim. Card.
HL 7.00 - 4.40
Gn 8
C7 / M7
FF5 / E3
COMP

American Journal of Obstetrics and Gynecology (2005) **192**, 2021–8

American Journal of OBSTETRICS and GYNECOLOGY
www.ajog.org

An isolated intracardiac echogenic focus as a marker for aneuploidy

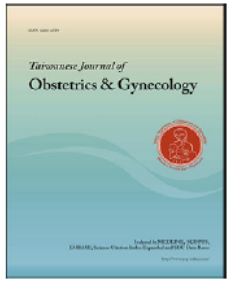
Kathleen E. Bradley, MD,* Thomas S. Santulli, MD, Kimberly D. Gregory, MD, MPH, William Herbert, MS, Dru E. Carlson, MD, Lawrence D. Platt, MD

- 10875 pacientes -> 176 con foco (1,6%)
- Aislado -> sin aneuploidías
- RR 2.55 para AMCT

Contents lists available at ScienceDirect

Taiwanese Journal of Obstetrics & Gynecology

journal homepage: www.tjog-online.com



Review Article

Meta-analysis of validity of echogenic intracardiac foci for calculating the risk of Down syndrome in the second trimester of pregnancy

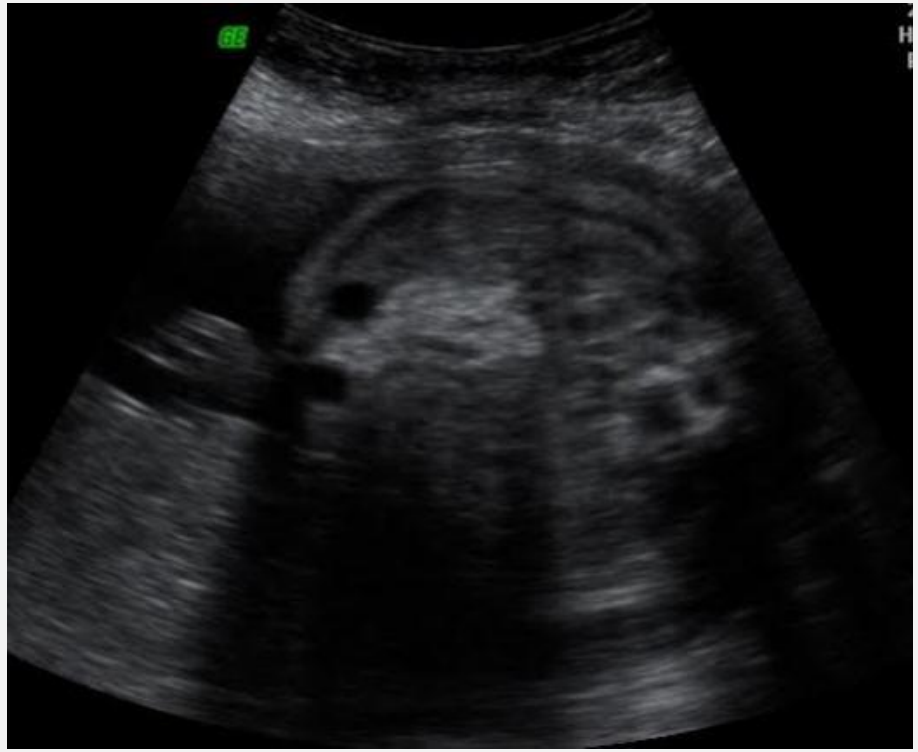
Ana María Rubio Lorente [a, *], María Moreno-Cid [a], María José Rodríguez [a], Gema Bueno [a], José María Tenías [b], Carmen Román [b], Ángel Arias [b], Ana Pascual [a]

[a] Obstetrics and Gynaecology Department, Hospital La Mancha Centro, Alcázar de San Juan, Ciudad Real, Spain
[b] Research Support Unit, Hospital La Mancha Centro, Alcázar de San Juan, Ciudad Real, Spain

- Metanalisis
- 9 trabajos: foco hiperecogénico aislado, población bajo riesgo
- LH+ 5.08

INTESTINO HIPERECOGENICO

- Aumento de la ecogenicidad intestinal
- Disminuye con la edad gestacional
- Se cree es secundario a disminución de la peristalsis
- 0,2-1% euploides
- 3-25% aneuploidías
- Se asocia a FQ, Hirschsprung, RCIU y TORCH



Sonek and croom. Second trimester ultrasound markers. Clinical Obstetrics and Gynecology Volume 57, number 1, 159–181. 2014

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BJOG. 2000 Mar;107(3):426-9.

Hyperechogenic fetal bowel: a prospective analysis of sixty consecutive cases.

Ghose I[1], Mason GC, Martinez D, Harrison KL, Evans JA, Ferriman EL, Stringer MD.

- Prospectivo
- 60 pacientes con IHE
- 5% aneuploidía
- 5% FQ, 0% TORCH, 10% RCIU

THE JOURNAL OF
MATERNAL-FETAL
& NEONATAL
MEDICINE

http://informahealthcare.com/jmf
ISSN: 1476-7058 (print), 1476-4954 (electronic)

J Matern Fetal Neonatal Med, 2016; 29(1): 41–45
 DOI: 10.3109/14767058.2014.986091

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ORIGINAL ARTICLE

The effect of associated structural malformations in the prediction of chromosomal abnormality risk of fetuses with echogenic bowel

Atalay Ekin, Cenk Gezer, Cuneyt Eftal Taner, and Mehmet Ozeren

- Retrospectivo
- 281 fetos con IHE
  - 105 aislado
  - 78 con otro marcador
  - 98 con malformación
- 6,7%; 7,7% y 17,4% respectivamente.
- Sin diferencia entre aislada y con otro marcador blando.

Title: Antenatal Prognostic factor of fetal echogenic bowel

Author: Candice Ronin Pierre Mace Fabien Stenard Anderson Loundou Marianne Capelle Isabelle Mortier Marie Christine Pellissier Sabine Sigaudy Annie Levy Claude D'ercole Pascale Hoffmann Thierry Merrot Jonathan Lopater Pascal De Lagausie Nicole Philip Florence Bretelle

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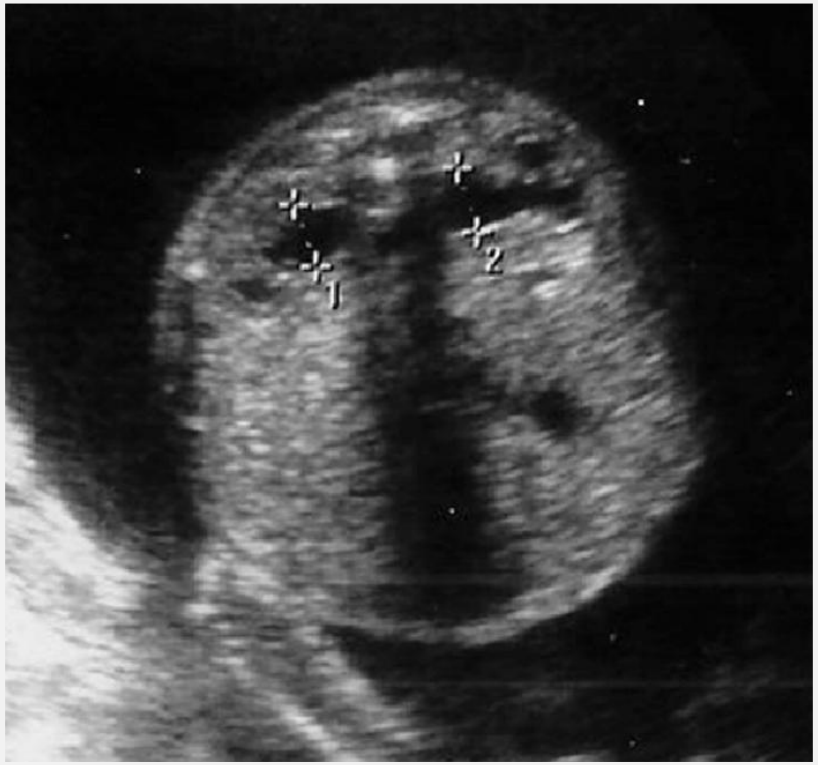
- Retrospectivo, multicéntrico
- 409 fetos
- 0,98% de aneuploidías, todas con otros marcadores.
  - 1 T13
  - 1 T18
  - 2 Triploidías

PIELECTASIA LEVE

- Se miden en un corte transverso abdominal
- Se mide el lugar más amplio
- Se considera desde 4mm, sin compromiso de los cálices
- 0-4,5% euploides
- 18% T21



Sonek and croom. Second trimester ultrasound markers. Clinical Obstetrics and Gynecology Volume 57, number 1, 159–181. 2014

Ultrasound Obstet Gynecol 2001; 17: 197–202

The association of aneuploidy and mild fetal pyelectasis in an unselected population: the results of a multicenter study

P. M. CHUDLEIGH, L. S. CHITTY*, M. PEMBREY* and S. CAMPBELL

*Department of Obstetrics and Gynaecology, St George's Hospital and *Clinical and Molecular Genetics Unit, Institute of Child Health, London, UK*

- Prospectivo, multicéntrico
- 101600 pacientes
- 737 Pielectasia (0,73%)
- 12 aneuploidías 1,7%
  - 0,46% aisladas
  - 9,2% asociada

Ultrasound Obstet Gynecol 2013; 42: 615–621
Published online 27 October 2013 in Wiley Online Library (wileyonlinelibrary.com). DOI: 10.1002/uog.12516

Isolated fetal pyelectasis and the risk of Down syndrome: a meta-analysis

K. M. ORZECHOWSKI and V. BERGHELLA

Division of Maternal Fetal Medicine, Department of Obstetrics and Gynecology, Thomas Jefferson University, Philadelphia, PA, USA

- Metanalisis
- 10 trabajos
- LH + 2.78
- LH – 0.98

Ultrasound Obstet Gynecol 2013; 41: 247–261
Published online 24 January 2013 in Wiley Online Library (wileyonlinelibrary.com). DOI: 10.1002/uog.12364

Meta-analysis of second-trimester markers for trisomy 21

M. AGATHOKLEOUS*, P. CHAVEEVA*, L. C. Y. POON*, P. KOSINSKI* and K. H. NICOLAIDES*†

**Harris Birthright Research Centre for Fetal Medicine, King's College Hospital, London, UK; †Department of Fetal Medicine, University College Hospital, London, UK*

- Metanalisis
- 38 trabajos
- Se evaluaron los marcadores combinados y aislados.

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Table 11 Pooled estimates of detection rate (DR), false positive rate (FPR) and positive and negative likelihood ratios (LR+ and LR−) of sonographic markers for trisomy 21 and estimated likelihood ratio (LR) of individual isolated markers

Marker	*DR (95% CI) (%)*	*FPR (95% CI) (%)*	*LR+ (95% CI)*	*LR− (95% CI)*	*LR isolated marker**
Intracardiac echogenic focus	24.4 (20.9–28.2)	3.9 (3.4–4.5)	5.83 (5.02–6.77)	0.80 (0.75–0.86)	0.95
Ventriculomegaly	7.5 (4.2–12.9)	0.2 (0.1–0.4)	27.52 (13.61–55.68)	0.94 (0.91–0.98)	3.81
Increased nuchal fold	26.0 (20.3–32.9)	1.0 (0.5–1.9)	23.30 (14.35–37.83)	0.80 (0.74–0.85)	3.79
Echogenic bowel	16.7 (13.4–20.7)	1.1 (0.8–1.5)	11.44 (9.05–14.47)	0.90 (0.86–0.94)	1.65
Mild hydronephrosis	13.9 (11.2–17.2)	1.7 (1.4–2.0)	7.63 (6.11–9.51)	0.92 (0.89–0.96)	1.08
Short femur	27.7 (19.3–38.1)	6.4 (4.7–8.8)	3.72 (2.79–4.97)	0.80 (0.73–0.88)	0.61
Absent or hypoplastic NB	59.8 (48.9–69.9)	2.8 (1.9–4.0)	23.27 (14.23–38.06)	0.46 (0.36–0.58)	6.58

Second trimester: echogenic intracardiac foci	Echogenic tissue in one or both ventricles of the heart seen on standard four-chamber view	LR 1.4–1.8 for Down syndrome Seen in 15–30% of Down syndrome and 4–7% euploid fetuses	1. If isolated finding, aneuploidy screening should be offered if not done previously 2. If aneuploidy screen result is negative, no further evaluation is required.
Second trimester: pyelectasis	Renal pelvis measuring ≥ 4 mm in anteroposterior diameter up to 20 weeks of gestation	LR 1.5–1.6 for Down syndrome	1. If isolated finding, aneuploidy screening should be offered if not performed previously 2. Repeat ultrasonography in third trimester for potential urinary tract obstruction
Second trimester: echogenic bowel	Fetal small bowel as echogenic as bone	LR 5.5–6.7 for Down syndrome Associated with aneuploidy, intra-amniotic bleeding, cystic fibrosis, CMV	1. Further counseling 2. Offer CMV, CF, and aneuploidy screening or diagnostic testing
Second trimester: thickened nuchal fold	≥ 6 mm from outer edge of the occipital bone to outer skin in the midline	LR 11–18.6 with 40–50% sensitivity and > 99% specificity for Down syndrome Most powerful second-trimester marker	1. Detailed anatomic survey 2. Further detailed genetic counseling and aneuploidy screening or diagnostic testing
Second trimester: mild ventriculomegaly	Lateral ventricular atrial measurement between 10–15 mm	Associated with aneuploidy LR 25 for Down syndrome	1. Genetic counseling 2. Second-trimester detailed anatomic ultrasound evaluation 3. Consider diagnostic testing for aneuploidy and CMV 4. Repeat ultrasound in third trimester

- ACOG Practice Bulletin N°163, May 2016. Screening for Fetal Aneuploidy.

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Dr. Nicolás Zapata Lara
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