NIPS

VÀ NHỮNG TRANH CÃI LÂM SÀNG

TRẦN NHẬT THĂNG

KHOA PHỤ SẢN BV ĐẠI HỌC Y DƯỢC – ĐƠN VỊ CHẨN ĐOÁN TRƯỚC SINH

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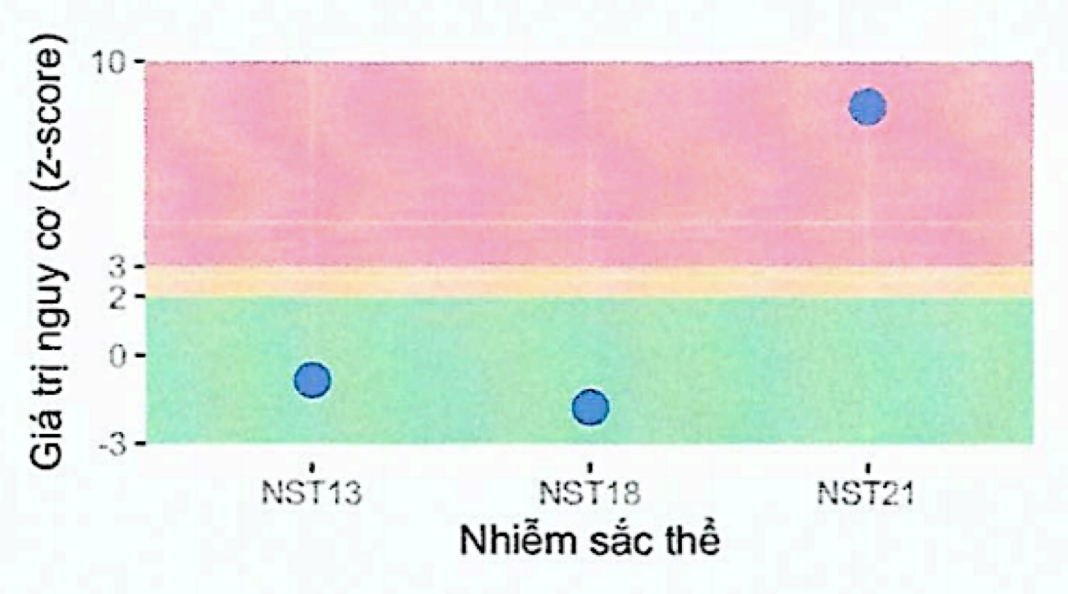
TRƯỜNG HỢP 1

- LT. Th. 1992
- G1P0: thai tự nhiên
- CRL 60mm / NT 1.0mm
- FTS (combined) 1/65 (T21): PAPP-A 0.2 / fbhCG 1.5 MoM
- NIPS: FF 9.1%



Trường hợp 1

16 tuần

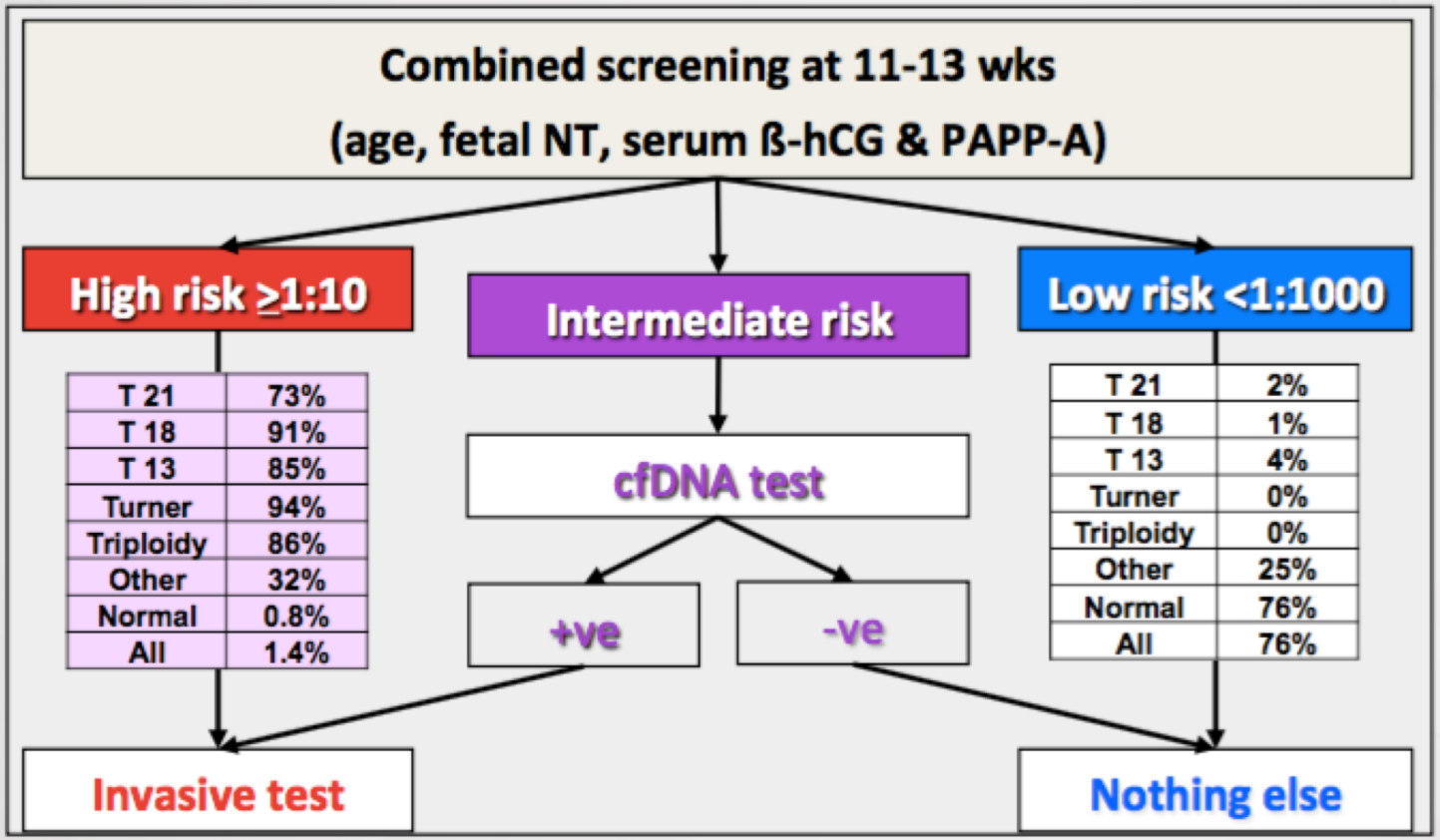
EFW 151g
Xương mũi 2.3mm
Thiểu sản đốt giữa ngón V
Dãn bể thận hai bên

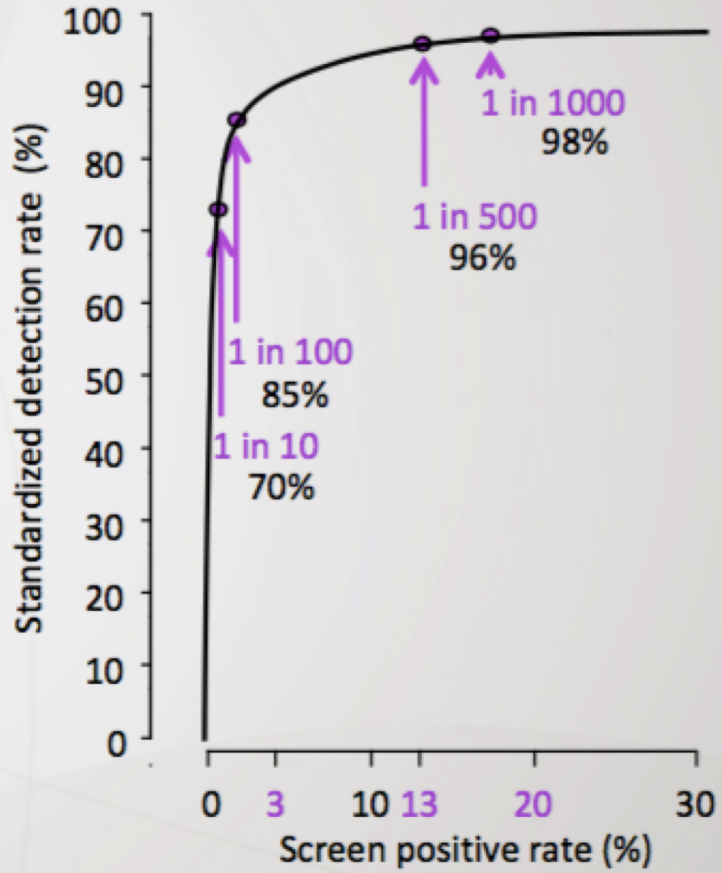
Trisomy 21

Kết thúc thai kỳ 19 tuần
Trai 255g

Cell free DNA test

Model of clinical implementation





Santorum M, Wright D, Syngelaki A, Karagioti N, Nicolaides KH. Accuracy of first trimester combined test in screening for trisomies 21, 18 and 13. Ultrasound Obstet Gynecol 2016; doi 10.1002/uog.17283

Original Article

Singapore Med J 2015; 56(1): 47-52
doi: 10.11622/smedj.2015009

The assessment of combined first trimester screening in women of advanced maternal age in an Asian cohort

Sarah Weiling Li[1,*], MBChB, BSc, *Angela Natalie Barrett*[2,*], PhD, *Leena Gole*[3], PhD, *Wei Ching Tan*[4], MBBS, FRCOG, *Arijit Biswas*[1], MD, FRCOG, *Hak Koon Tan*[4], MRACOG, FRCOG, *Mahesh Choolani*[2], FRCOG, PhD

Table I. Performance of first trimester screening in detecting trisomies 21, 18 and 13 at different selected risk cut-offs (n = 10,289).

Risk cut-off	Absolute numbers				% (95% confidence interval)			
	True positive	False positive	True negative	False negative	Sensitivity	Specificity	PPV	NPV
1:100	34	133	10,107	15	69.39 (54.58–81.74)	98.70 (98.47–98.91)	20.36 (14.53–27.27)	99.85 (99.76–99.92)
1:150	39	174	10,066	10	79.59 (65.65–89.74)	98.30 (98.03–98.54)	18.31 (13.36–24.17)	99.90 (99.82–99.95)
1:200	41	211	10,029	8	83.67 (70.34–92.66)	97.94 (97.65–98.21)	16.27 (11.94–21.42)	99.92 (99.84–99.97)
1:250*	43	245	9,995	6	87.76 (75.22–95.34)	97.61 (97.29–97.89)	14.93 (11.02–19.58)	99.94 (99.87–99.98)
1:300†	43	285	9,955	6	87.76 (75.22–95.34)	97.22 (96.88–97.53)	13.11 (9.65–17.25)	99.94 (99.87–99.98)
1:350	43	335	9,905	6	87.76 (75.22–95.34)	96.73 (96.37–97.06)	11.38 (8.36–15.02)	99.94 (99.87–99.98)
1:500	44	443	9,797	5	89.80 (77.76–96.56)	95.67 (95.26–96.06)	9.03 (6.64–11.94)	99.95 (99.88–99.98)
1:1,000	47	786	9,454	2	95.92 (85.99–99.38)	92.32 (91.79–92.83)	5.64 (4.18–7.43)	99.98 (99.92–100.00)

*Risk cut-off used to identify screen-positive women at National University Hospital. †Risk cut-off used to identify screen-positive women at Singapore General Hospital. NPV: negative predictive value; PPV: positive predictive value

Table IV. Number and cumulative number of cases, and Down syndrome cases according to different risk cut-off groups.

Risk cut-off groups	No. of cases	Cumulative no. of cases	No. of down syndrome cases
1:1–100	167	167	28
1:101–250	121	288	4
1:251–500	199	487	1
1:501–750	153	640	2
1:751–1,000	193	833	1
1:1,001–5,000	2,731	3,564	0
1:5,001–10,000	2,779	6,343	1
1:10,001–15,000	2,089	8,432	0
1:15,001–20,000	1,366	9,798	0
1:20,001 and less	491	10,289	0

TRƯỜNG HỢP 2

Srebniak *et al. Molecular Cytogenetics* (2016) 9:69
DOI 10.1186/s13039-016-0279-z

Molecular Cytogenetics

RESEARCH Open Access

Enlarged NT (≥3.5 mm) in the first trimester – not all chromosome aberrations can be detected by NIPT

Malgorzata I. Srebniak[1*], Merel C. de Wit[2], Karin E. M. Diderich[1], Lutgarde C. P. Govaerts[1], Marieke Joosten[1], Maarten F. C. M. Knapen[2,3], Marnix J. Bos[1], Gerda A. G. Looye-Bruinsma[1], Mieke Koningen[1], Attie T. J. I. Go[2], Robert Jan H. Galjaard[1] and Diane Van Opstal[1]

ULTRASOUND
in Obstetrics & Gynecology

isuog

Original Paper | Free Access

Chromosomal microarray in fetuses with increased nuchal translucency

I. C. B. Lund, R. Christensen, O. B. Petersen, I. Vogel, E. M. Vestergaard

Pregnancies with NT ≥ 3.5 mm: $n = 132$
NT: 4.2 (3.5–13.8) mm
Maternal age: 31 (18–43) years

QF-PCR

Abnormal QF-PCR results: $n = 38$ (28.8%)
NT: 4.8 (3.5–13.8) mm
Maternal age: 34 (19–43) years

Common aneuploidies detected:

Trisomy 21	$n = 28$ (21.2%)
Trisomy 18	$n = 3$ (2.3%)
Triploidy	$n = 2$ (1.5%)
45,X	$n = 4$ (3.0%)
47,XXX	$n = 1$ (0.8%)

Normal QF-PCR results: $n = 94$ (71.2%)
NT: 4.1 (3.5–8.2) mm
Maternal age: 30 (18–42) years

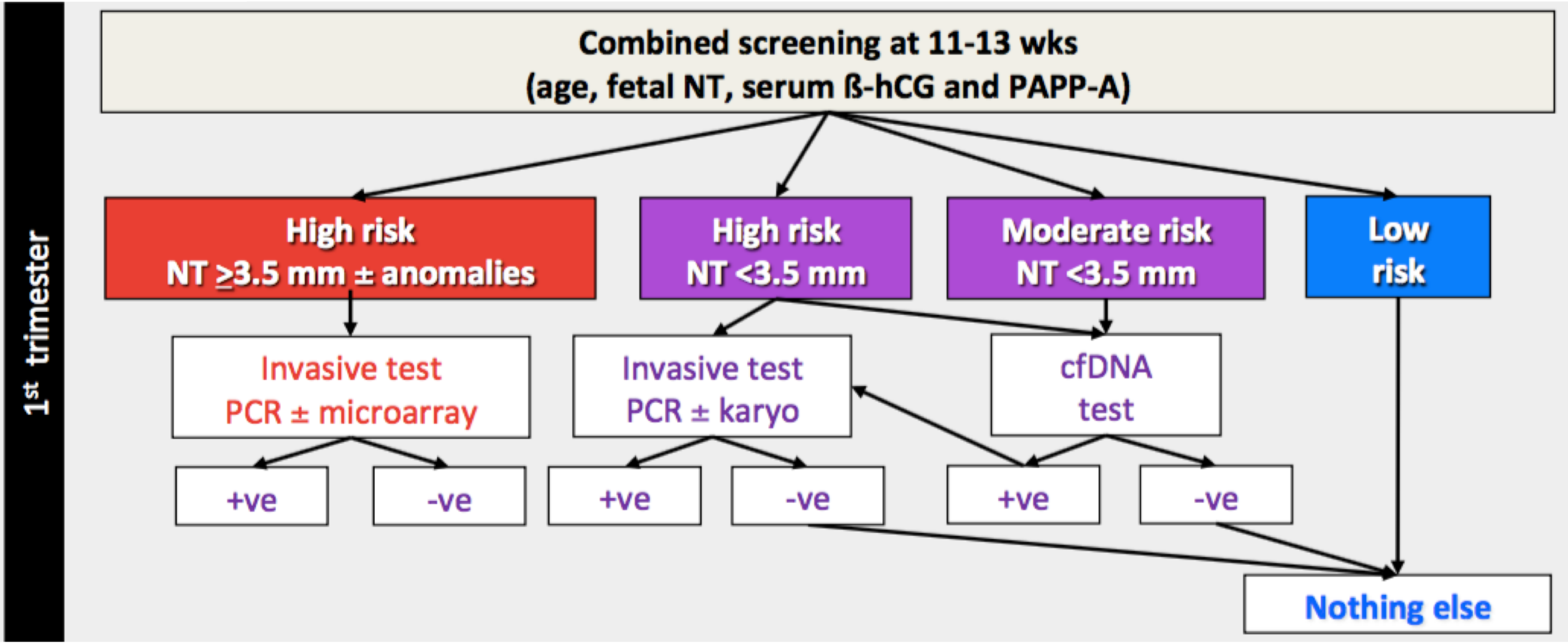
CMA

CNVs detected by CMA: $n = 15$ (16.0%)

Pathogenic	$n = 12$ (12.8%)
VOUS	$n = 3$ (3.2%)

The Fetal Medicine Foundation

Liona Poon



TRƯỜNG HỢP 3

European Journal of Obstetrics & Gynecology and Reproductive Biology 200 (2016) 72–75

Contents lists available at ScienceDirect

European Journal of Obstetrics & Gynecology and Reproductive Biology

journal homepage: www.elsevier.com/locate/ejogrb

Noninvasive prenatal testing: more caution in counseling is needed in high risk pregnancies with ultrasound abnormalities

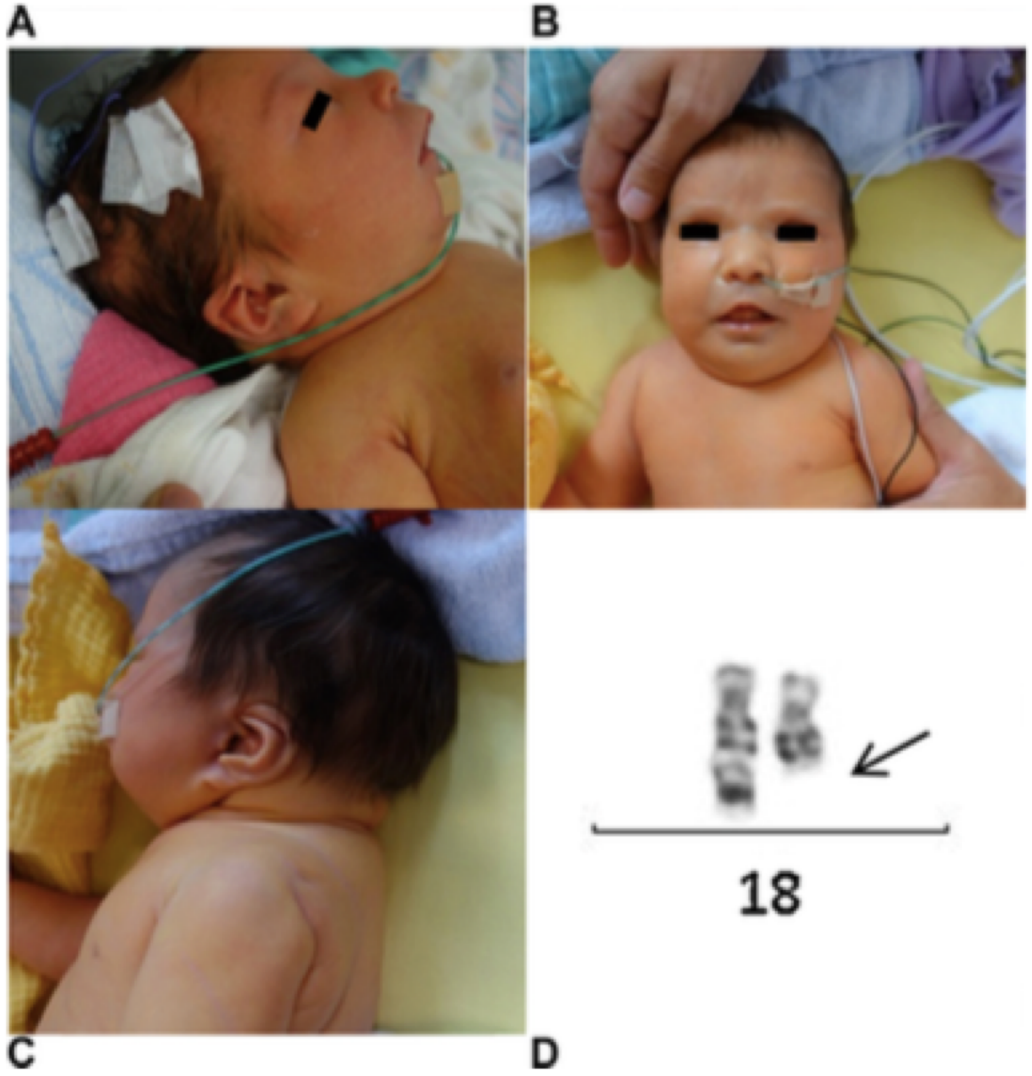
Beatrice Oneda [a,*], Katharina Steindl [a], Rahim Masood [a], Irina Reshetnikova [a], Pavel Krejci [a], Rosa Baldinger [a], Regina Reissmann [a], Malgorzata Taralczak [a], Adriano Guetg [a], Josef Wisser [b], Jean-Claude Fauchère [c], Anita Rauch [a]

[a] Institute of Medical Genetics, University of Zurich, Zurich, Switzerland
[b] Division of Obstetrics, University Hospital Zurich, Zurich 8091, Switzerland
[c] Division of Neonatology, University Hospital Zurich, Zurich, Switzerland

- NT: 3.5mm
- NIPS low risk
- Siêu âm:
  - 20-23 tuần: tư thế thai!
  - 24 tuần: bất sản thận (P)
  - 30 tuần: + bất sản thể chai, hở van 3 lá, tràn dịch đa màng (tim, bụng)
- 36 tuần sinh non

de novo 28.3 Mb deletion in 18q21.2-qter

TRƯỜNG HỢP 3



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[b] Division of Obstetrics, University Hospital Zurich, Zurich 8091, Switzerland
[c] Division of Neonatology, University Hospital Zurich, Zurich, Switzerland

cffDNA?

TRƯỜNG HỢP 4

- PTT. Th. 1977
- G P : IVF, ghi nhận 1 túi thai sau chuyển 2 phôi
- MCDA 12 tuần 6 ngày
  - CRL (A): 51mm / NT: 8.1mm
  - CRL (B): 57mm / NT: 1.3mm
- NIPS (SNP-based): FF 9.8%

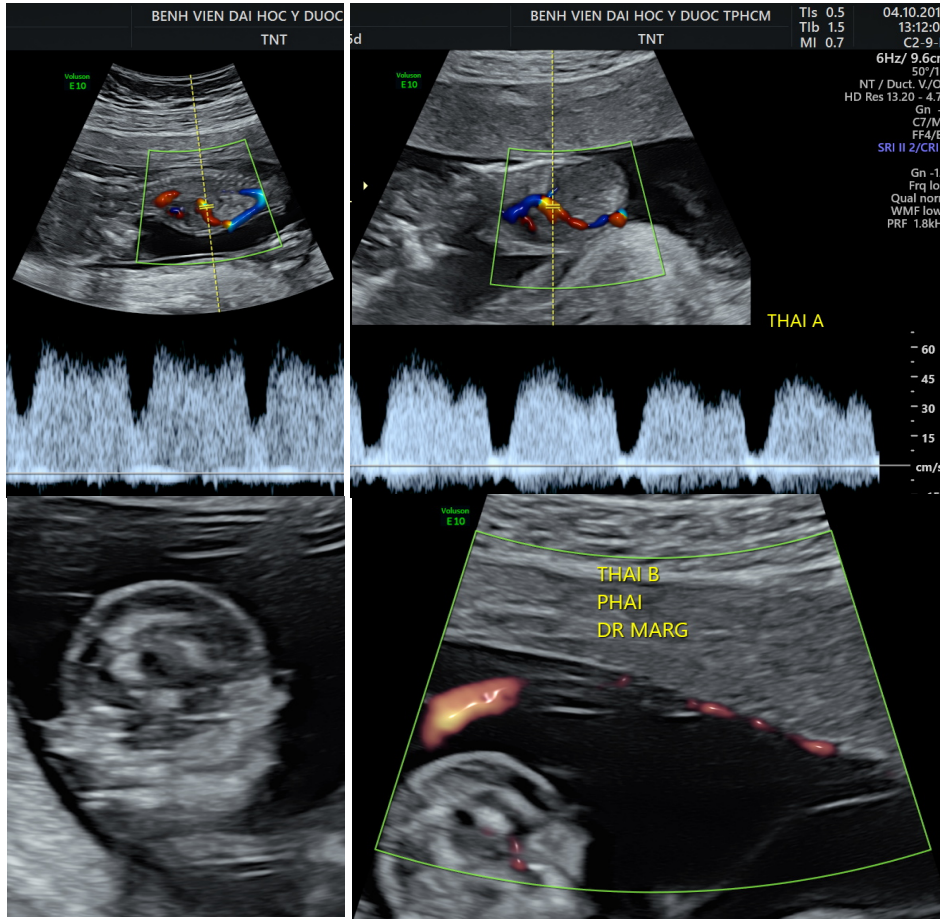
RESULT DETAILS: ANEUPLOIDIES

Condition tested[1]	*Result*	*Risk Before Test*[2]	*Risk After Test*[3]
Trisomy 21	Low Risk	1/152	<1/10,000
Trisomy 18	Low Risk	1/351	<1/10,000
Trisomy 13	Low Risk	1/1,110	<1/10,000
Monosomy X	Low Risk	1/759	<1/10,000

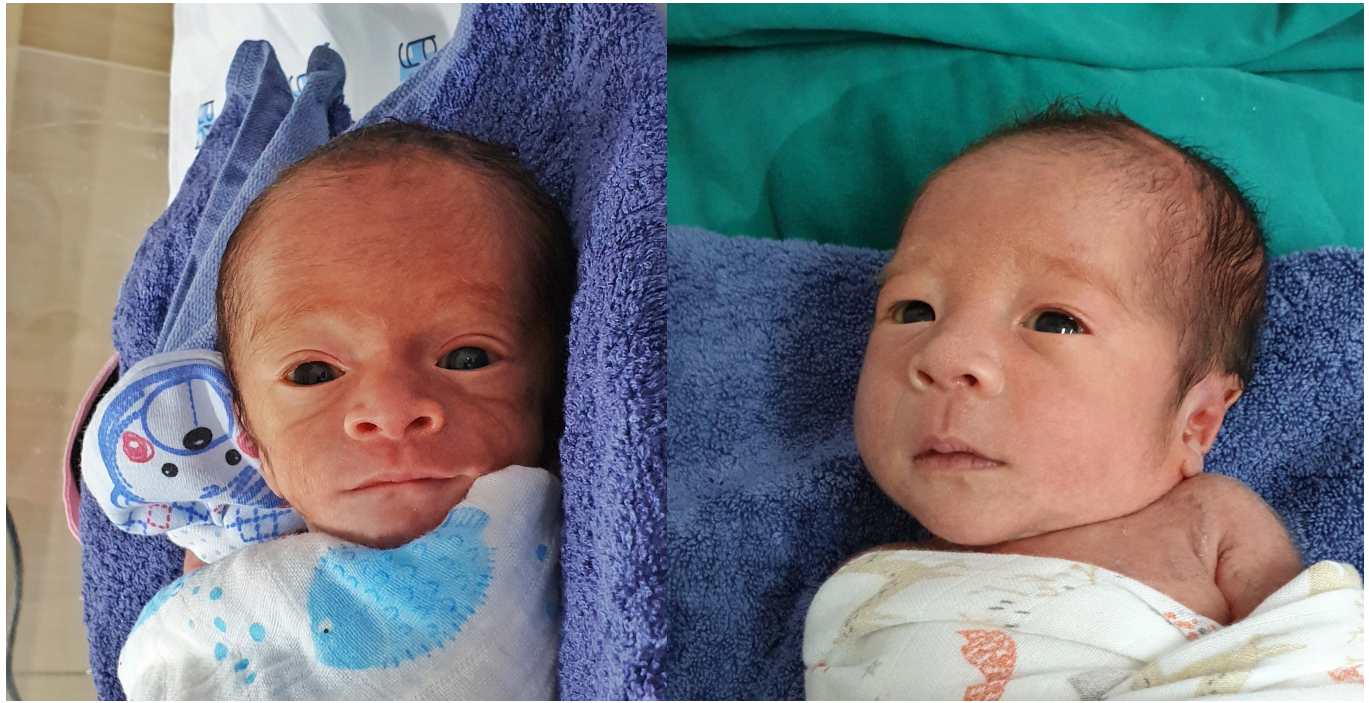
Trường hợp 4

14 tuần 5 ngày

AC 81mm vs 63mm
Dây rốn!

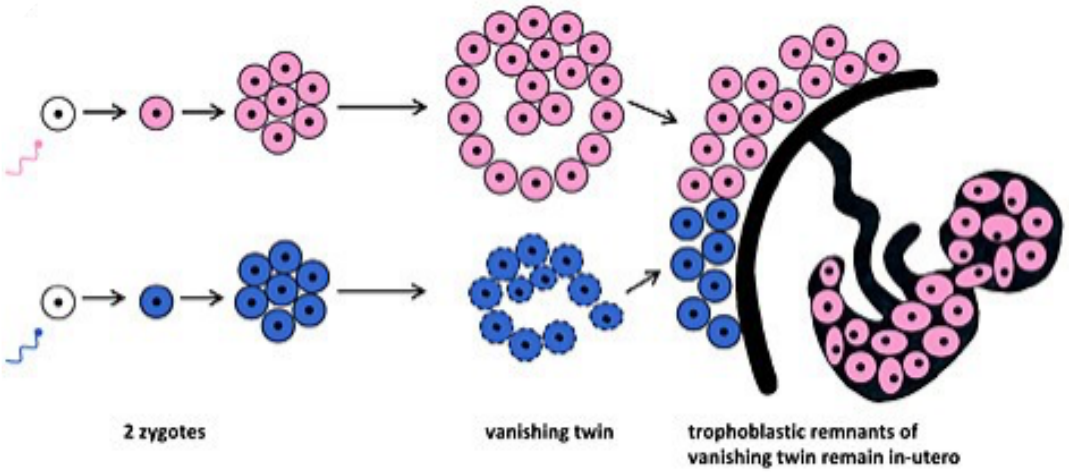


MLT 32 tuần



NIPS trong song thai

- Tranh cãi quanh việc xác định thai nào khi NIPS (+)
- Giúp khẳng định zygosity cùng với lâm sàng (siêu âm)
- Độ nhạy có thể giảm vì fetal fraction per twin thấp hơn



Song thai tiêu biến/chết:

- có thể tiếp tục còn phóng thích cfDNA
- có thể có giới tính khác gây nhầm lẫn trong kết quả thai sống làm NIPS

NIPS và NST giới tính

NIPS và sàng lọc vi mất đoạn

NIPS và sàng lọc/chẩn đoán bệnh di truyền đơn gene


THANK
YOU
GRACIAS
ARIGATO
SHUKURIA
BOLZÏN
MERCI
BİYAN
DANKSCHEEN
TASHAKKUR ATU
YAQHANYELAY
SUKSAMA
EKHMET
MEHRBANI
GRAZIE
JUSPAXAR
KOMAPSUMNIDA
GOZAIMASHITA
EFCHARISTO
PALDIES
SHUKRIA
TINGKI
SPASSIBO
SNACHALHUYA
NUHUN
CHALTU
WABEEJA
MAITEKA
YUSPAGARÀTAM
HUI
DHANYAVAAD
ANHIA
ATTO
MERSI
SPASIBO
DENKAUJA
NENACHALHIYA
UNALCHÉESH
GUI
HÄTUR
EKOJU
SIKOMO
MAKETAU
MINMONCHAR
MAAKE
SAÑCO
MERASTAWHY
GAEJTHO
LAH
TAVTAPUCH
MEDAWAGSE
AGUYJE
FAKAAUE
BAIKA