Gynea laboratorios

LABCO Quality Diagnostics

El futuro del diagnóstico prenatal.

Luis Izquierdo
Genética
Labco, Madrid
luis.izquierdo@labco.eu

Diagnóstico Prenatal de Anomalías Cromosómicas

- Análisis Citogenético necesita ≈ 2 Semanas

- Aneuploidías de X, Y, 13, 18 y 21

  ≈ 95% de Anomalías Cromosómicas

- Las Indicaciones Han Cambiado

- Cribado en el 1er trimestre

  Diagnostico en el Segundo?!

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QF-PCR en Diagnóstico Prenatal

99.9% Detección:

- **-Edad Materna**
- **-Riesgo Bioquímico**
- **-Angustia**

} **≈ 85% de Casos**

95-97% Detección:

- **TN Aumentada**
- **Marcadores Ecográficos**

} **≈ 15% de Casos**

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V.Cirigliano et.al. (2005) Reprod Biomed Online. 11(6):671-3

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Diagnóstico prenatal

Consejo genético Edad materna/historia personal o familiar)

<u>Cribado del primer trimestre</u>
1st / 2nd Suero – Ecografia (TN)

CVS / AMNIOCENTESIS

QF-PCR aCGH cariotipo

laboratorios

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Limitaciones del cribado actual

High false positive rate (5%)

Inconvenient
Multiple visits
Specialized ultrasound

Late information
Prolonged uncertainty

Safety concerns

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Limitaciones del Cribado Actual

- Falsos Positivos

  Técnicas invasivas innecesarias, angustia

- Tiempo

  Puede extenderse al segundo trimestre

- Conveniencia

  Múltiples visitas y ecografía limitan acceso/eficacia

- Seguridad

  Rechazo a técnicas invasivas por el riesgo de perdida fetal

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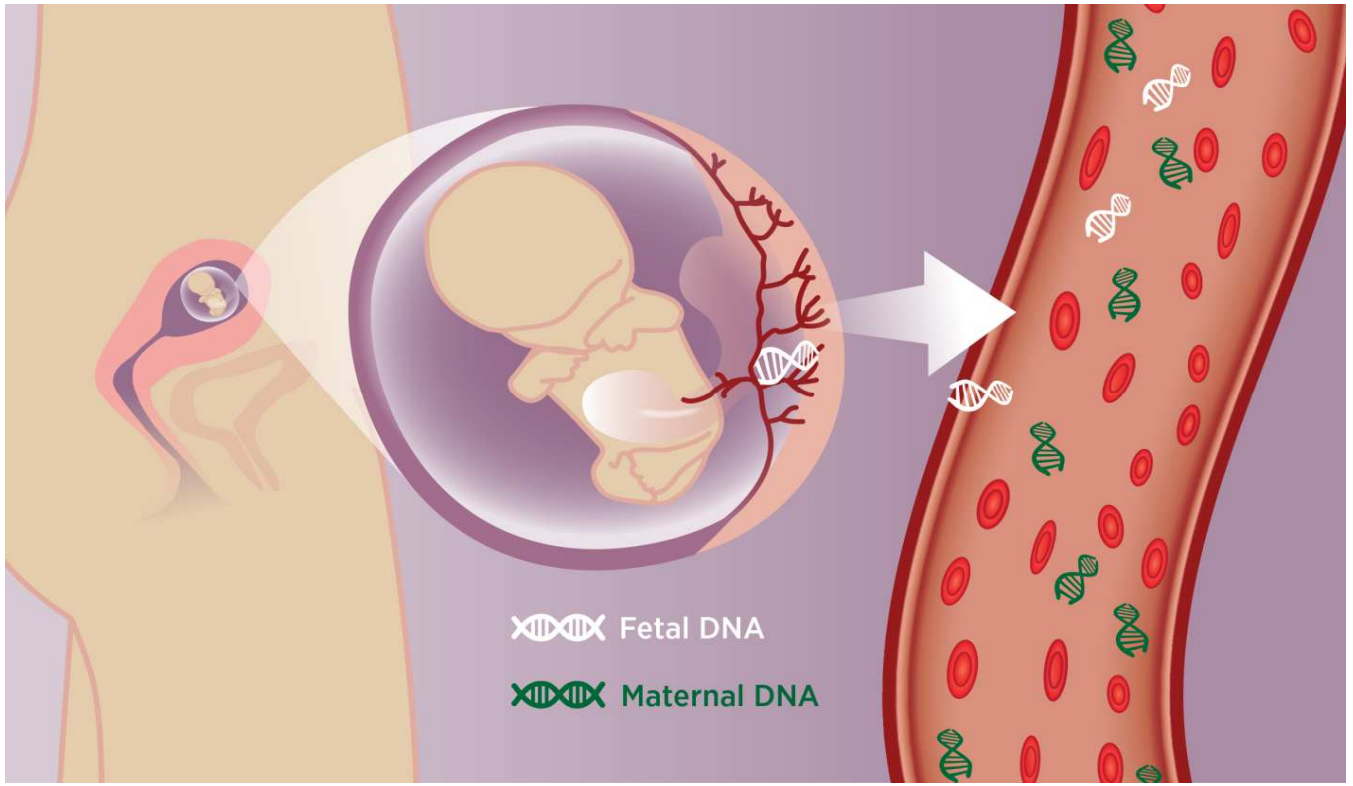
Cell-free DNA en Sangre Materna

- En todas las gestaciones hay cfDNA de madre y feto en circulación materna
- Cell-free DNA (cfDNA) está presente en fragmentos muy cortos
- La cantidad de cfDNA fetal solo es una pequeña fracción del cfDNA materno



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DNA Fetal, de donde viene?

- 1997: Secuencias del cromosoma Y detectadas en plasma y suero de gestantes con fetos masculinos (Lo et al 1997)
- 1998: Cuantificación del ffDNA por Real-Time PCR
  - Porcentaje de ffDNA superior en Plasma
    - Early pregnancy: 0.4 – 11.9% (mean 3.4%)
    - Late pregnancy: 2.3 – 11.4% (mean 6.2%)
- RNA placenta específico (ZFY) en plasma materno (Poon et al. 2000)
- ffDNA desde el trofoblasto (Alberry et al. 2007)

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DNA Fetal, cuanto persiste?

- ffDNA es eliminado muy rápidamente
- ffDNA tiene una vida media de 16,3 min
- A las 2 horas postparto ffDNA no detectable en 7/8 gestantes; en ninguna a los 2 días (Lo 1999).
- Riñón responsable de su eliminación: ffDNA detectado en orina (Botezatu 2000)
- Se estima que ffDNA es liberado a la circulación a 2.24 $x10^4$ copias/min. durante el embarazo

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Noninvasive diagnosis of fetal aneuploidy by shotgun sequencing DNA from maternal blood

H. Christina Fan*, Yair J. Blumenfeld†, Usha Chitkara†, Louanne Hudgins‡, and Stephen R. Quake*§

*Department of Bioengineering, Stanford University and Howard Hughes Medical Institute, 318 Campus Drive, Clark Center, Room E300, Stanford, CA 94305; †Division of Maternal-Fetal Medicine, Department of Obstetrics and Gynecology, Stanford University, 300 Pasteur Drive, Room HH333, Stanford, CA 94305; and ‡Division of Medical Genetics, Department of Pediatrics, Stanford University, 300 Pasteur Drive, Stanford, CA 94305

Communicated by Leonard A. Herzenberg, Stanford University School of Medicine, Stanford, CA, August 22, 2008 (received for review July 13, 2008)

Noninvasive prenatal diagnosis of fetal chromosomal aneuploidy by massively parallel genomic sequencing of DNA in maternal plasma

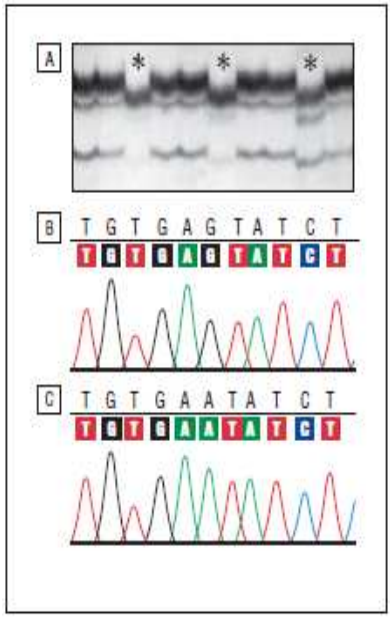
Rossa W. K. Chiu[a,b], K. C. Allen Chan[a,b], Yuan Gao[c,d], Virginia Y. M. Lau[a,b], Wenli Zheng[a,b], Tak Y. Leung[e], Chris H. F. Foo[f], Bin Xie[c], Nancy B. Y. Tsui[a,b], Fiona M. F. Lun[a,b], Benny C. Y. Zee[f], Tze K. Lau[e], Charles R. Cantor[g,1], and Y. M. Dennis Lo[a,b,1]

[a]Centre for Research into Circulating Fetal Nucleic Acids, Li Ka Shing Institute of Health Sciences, Departments of [b]Chemical Pathology and [e]Obstetrics and Gynaecology, and [f]Centre for Clinical Trials, The Chinese University of Hong Kong, Shatin, New Territories, Hong Kong SAR, China; [c]Center for the Study of Biological Complexity and [d]Department of Computer Science, Virginia Commonwealth University, Richmond, VA 23284; and [g]Sequenom, Inc., San Diego, CA 92121

Contributed by Charles R. Cantor, October 22, 2008 (sent for review September 29, 2008)

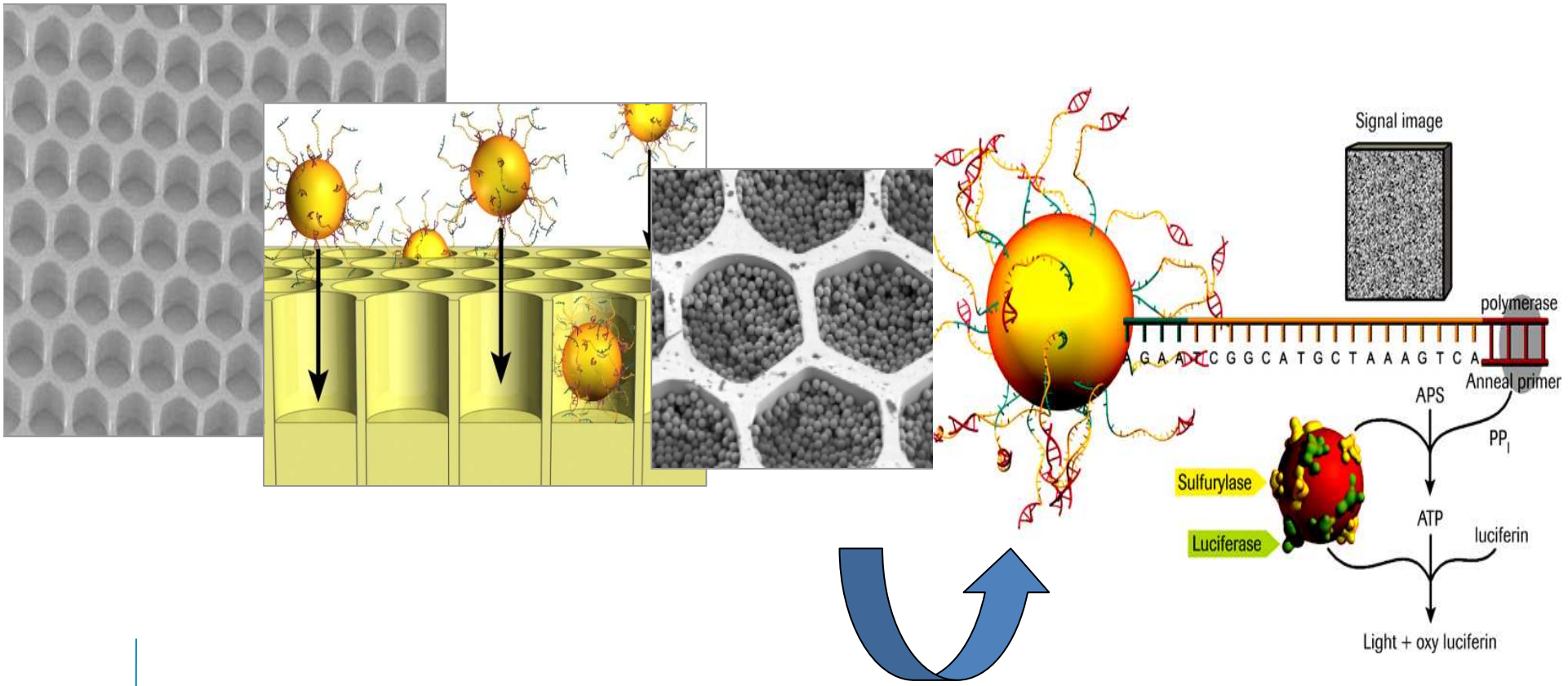
Secuenciación Masiva (NGS)

Secuenciación rápida de gran número de moléculas de ADN (Hasta el momento secuenciación Sanger, desde los 70, secuenciación de un fragmento de ADN cada vez)



Secuenciación de próxima generación

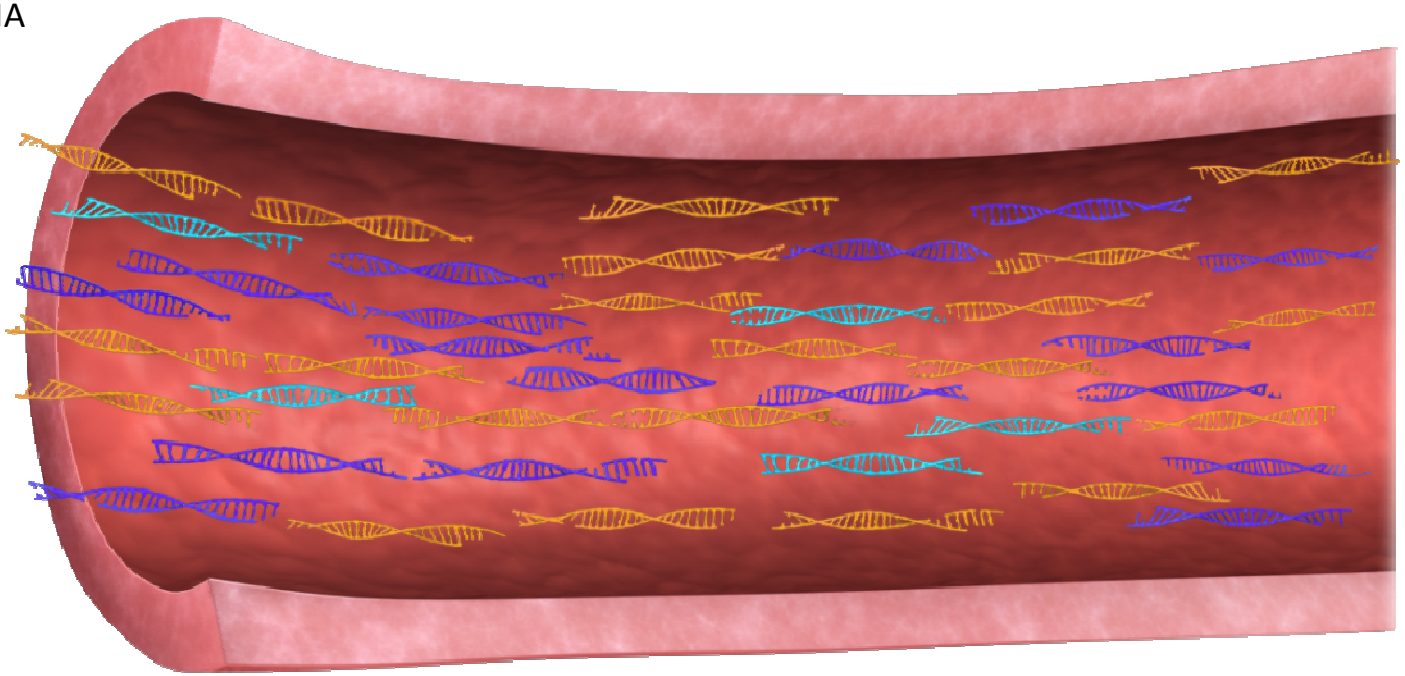
- Se basa en el concepto de reacciones químicas paralelas, masivas, en las cuales millones de secuenciaciones independientes ocurren simultáneamente.

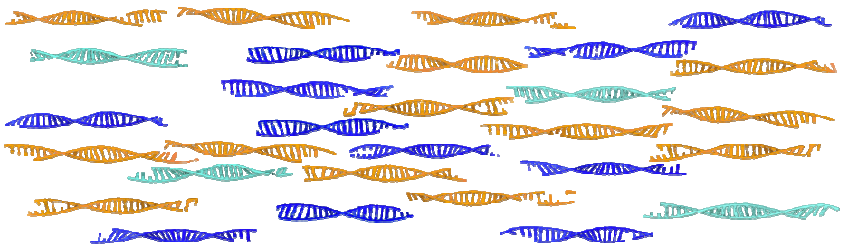


Chr 21, 18, 13, & Y cfDNA

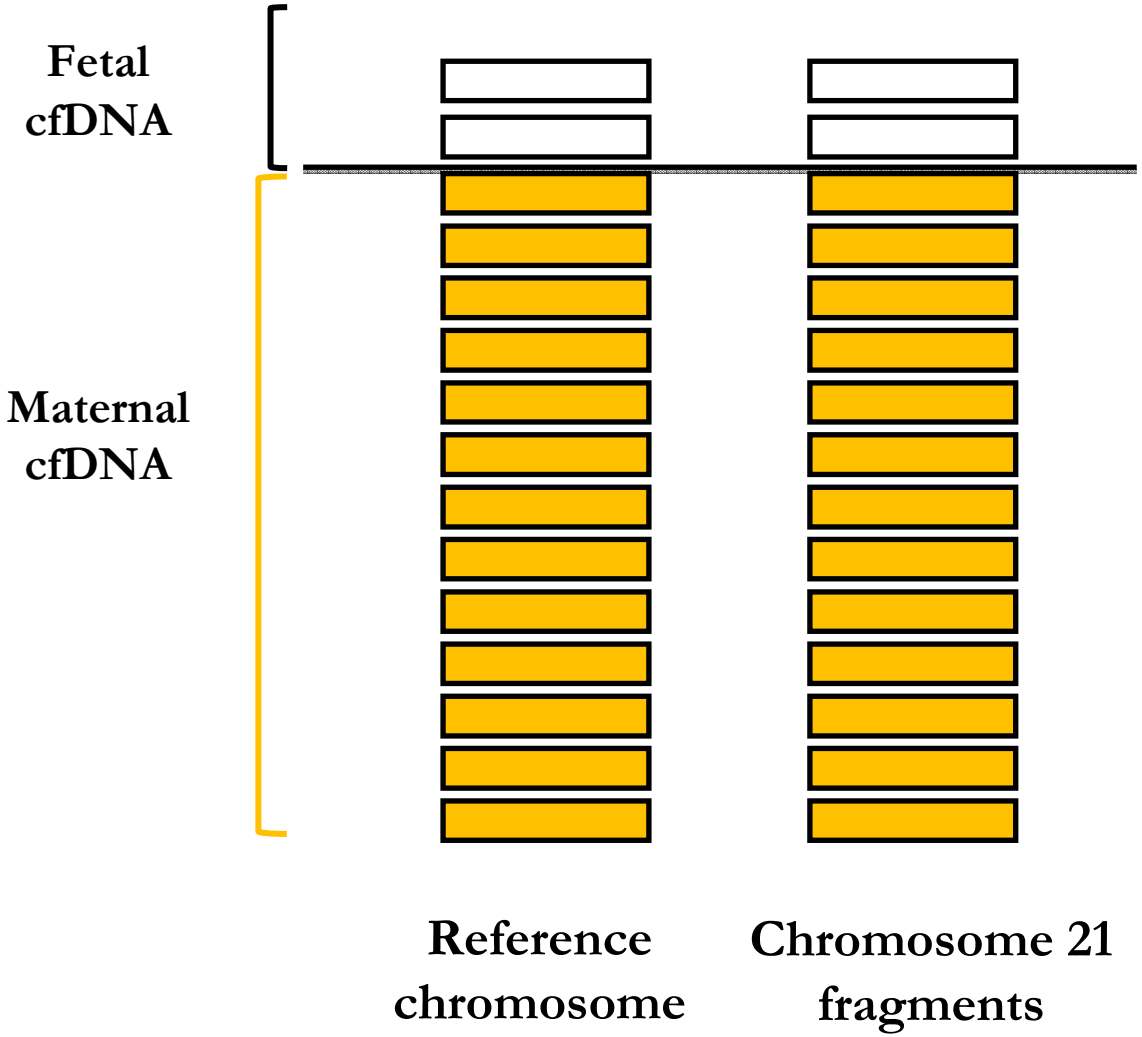
Other Chr cfDNA

Unmapped cfDNA





Fetal Trisomy Detection with cfDNA



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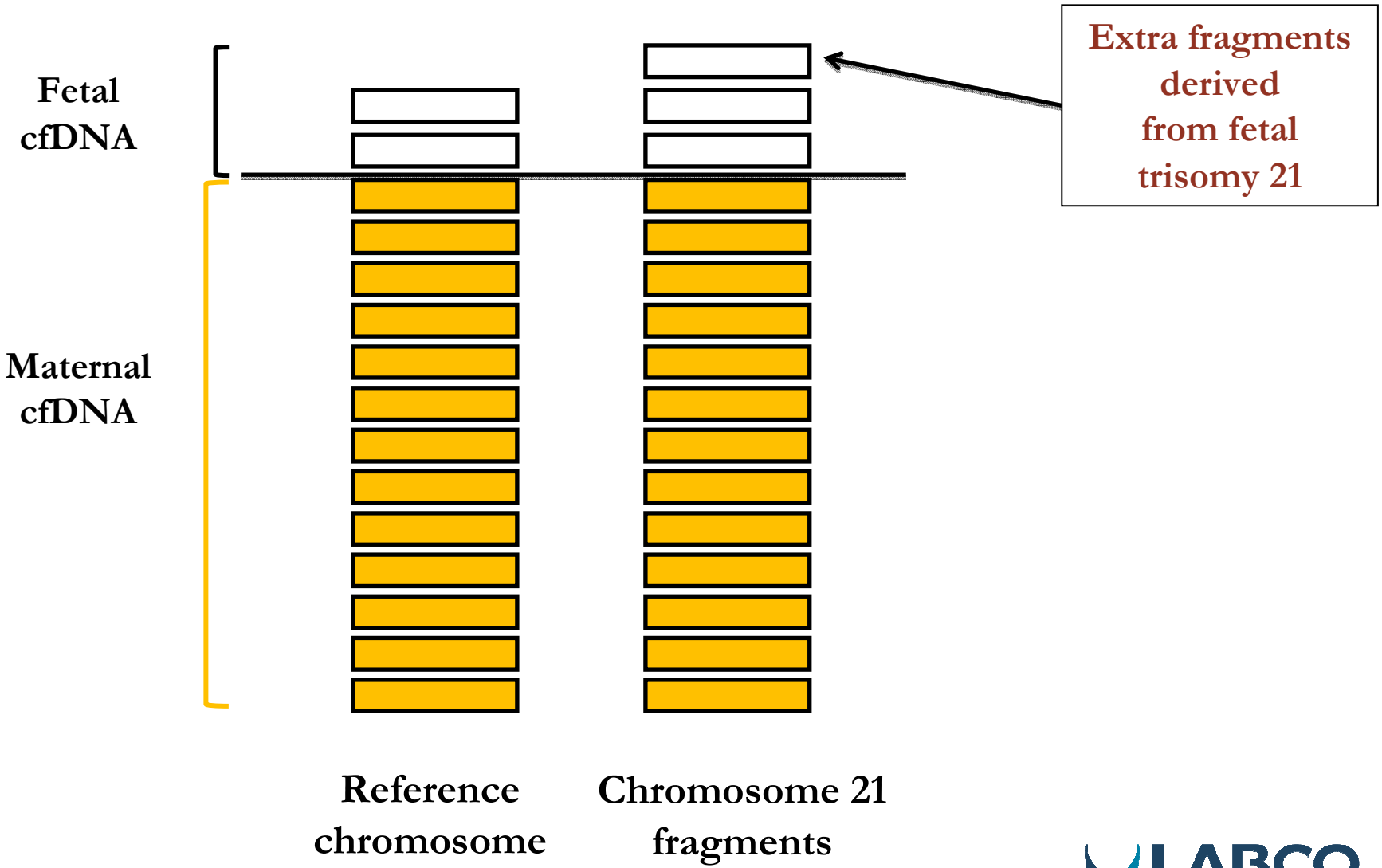
Fetal Trisomy Detection with cfDNA



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Harmony™ PRENATAL TEST

TARGETED NIPT 21, 18, 13

DANSR™

(Digital ANalysis of Selected Regions)

- Directed assay for cfDNA isolation and analysis.
- Targeted method allows for high throughput DNA sequencing

FORTE™

(Fetal-fraction Optimized Risk of Trisomy Evaluation)

- New analysis that provides a trisomy risk score
- Incorporates DANSR assay results (chromosome counts, fetal fraction), maternal and gestational age

High throughput and scalable test
Clinically interpretable results to patients

FORTE Aneuploidy Analysis

Harmony™ PRENATAL TEST

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Inputs

Chr 13, 18 and Chr 21 cfDNA counts

Fetal fraction

Clinical information (e.g. maternal and gestational age)

FORTE

Outputs

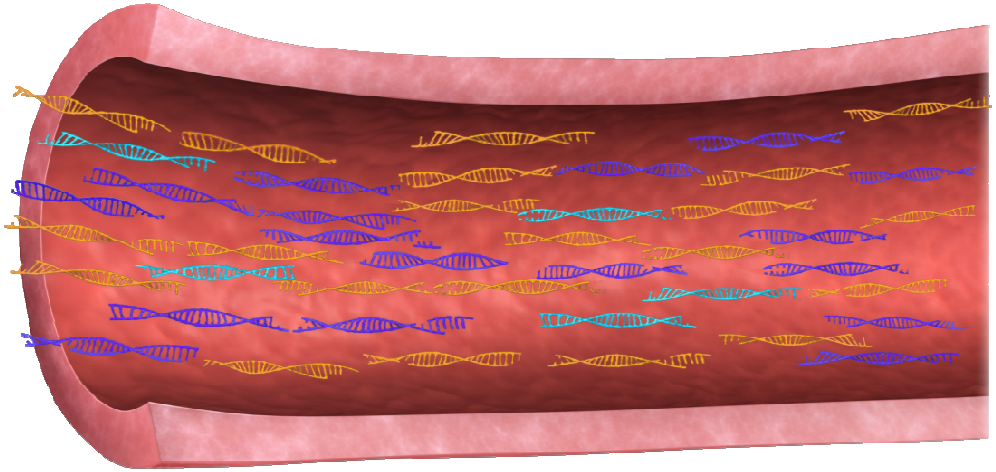
Trisomy 21 risk value

Trisomy 18 risk value

Trisomy 13 risk value

*FORTE - Fetal-fraction Optimized Risk of Trisomy Evaluation

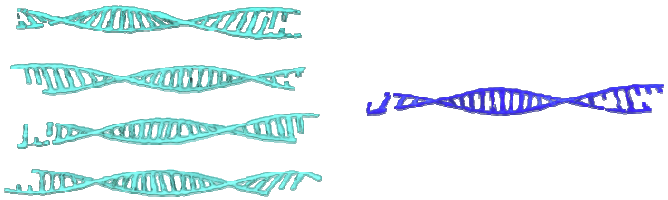
Assay Comparison – Targeted vs MPSS



cfDNA in blood

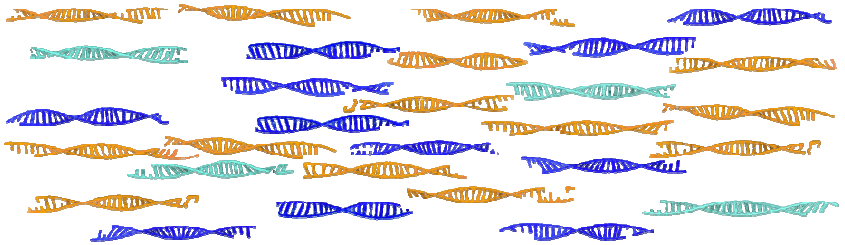
- Chr 21, 18, 13 cfDNA
- Other Chr cfDNA
- Unmapped cfDNA

DANSR™ (Directed)



More efficient

MPSS (shotgun)



Random analysis of cfDNA

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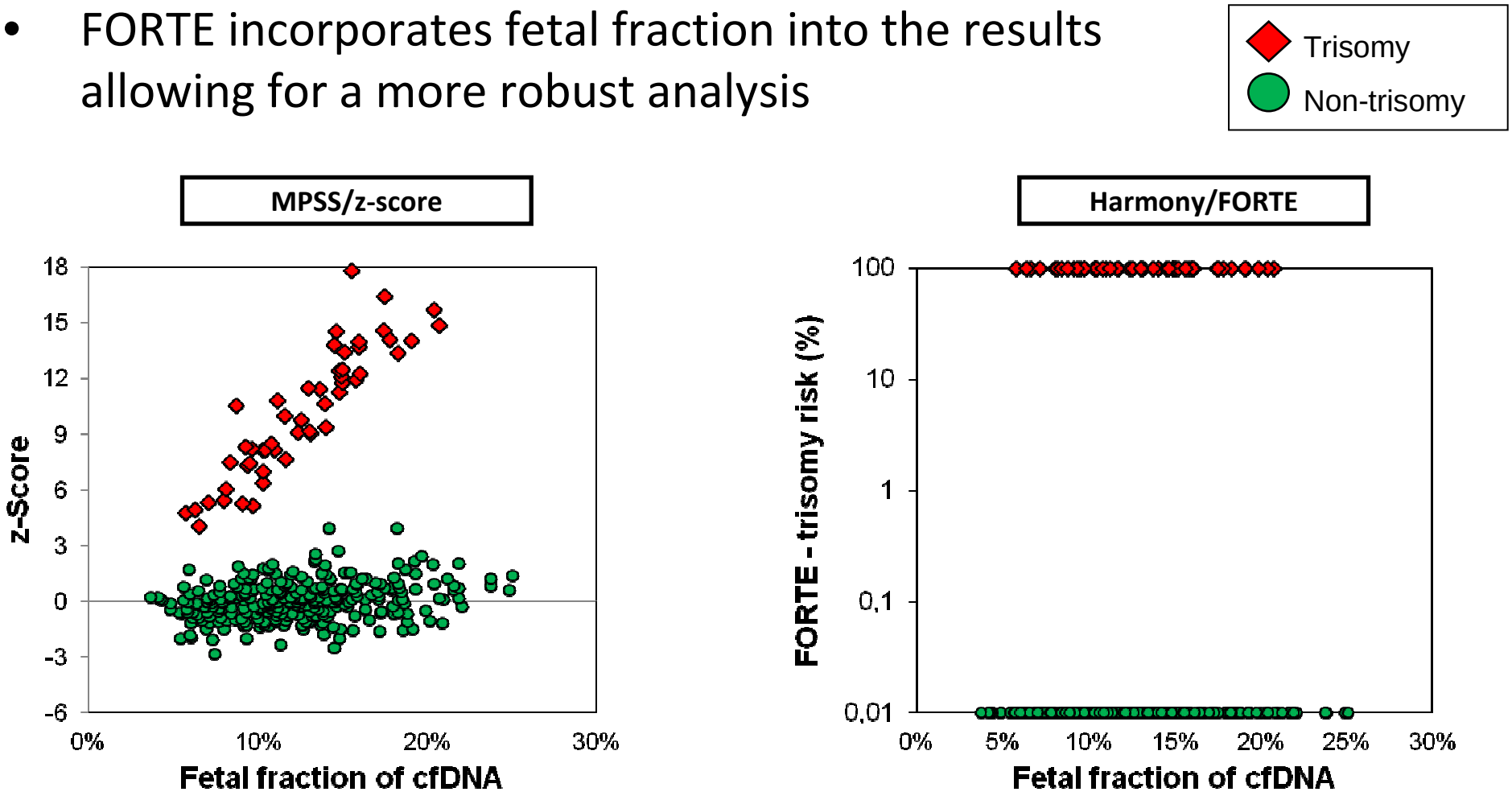
Harmony – Advantages of FORTE

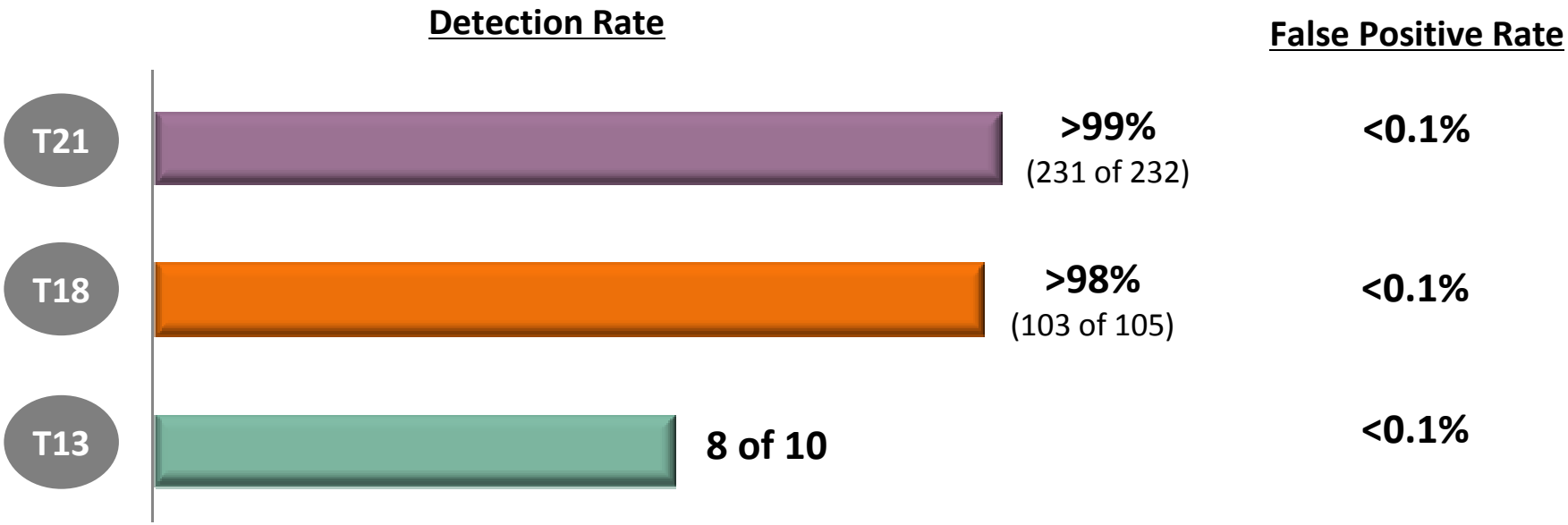
- FORTE incorporates fetal fraction into the results allowing for a more robust analysis



Ashoor G et al., Am J Obstet Gynecol. 2012 Apr;206(4):322.e1-5.

Clinical Performance

Studied in over 6,000 patients, including >2,000 average-risk women



1. Sparks AB et al., Am J Obstet Gynecol. 2012 Apr;206(4):319.e1-9. 2. Ashoor G et al., Am J Obstet Gynecol. 2012 Apr;206(4):322.e1-5. 3. Sparks AB et al., Prenat Diagn. 2012 Jan;32(1):3-9. 4. Norton M et al., Am J Obstet Gynecol. 2012 Aug;207(2):137.e1-8. 5. Nicolaides KH et al., Am J Obstet Gynecol. 2012 Nov;207(5):374.e1-6. 6. Ashoor G et al., Ultrasound Obstet Gynecol. 2013 Jan;41(1):21-5. 7. Data on file

Validacion/Aplicación Clínica

Study	Status	Description
NICE (Non-Invasive Chromosomal Evaluation)	**Published – Editor's choice in The Gray Journal (August 2012)**	**Multi-center (50 sites) clinical validation study, combined high risk and low risk women. Largest NIPT cohort study.**
Average Risk (Nicolaides)	**Published – The Gray Journal (2012, avail online)**	**Exclusive average-risk study of Harmony test in 1st trimester pregnancy**
Ariosa Blinded	**Published – Editor's choice in The Gray Journal (April 2012)**	**Blinded study with risk score reporting**
Nicolaides Blinded	**Published – Editor's choice in The Gray Journal (April 2012)**	**1st trimester blinded study**
Proof of Concept	**Published – cover article Prenatal Diagnosis (Jan 2012)**	**Initial description of directed cfDNA approach with combined average-risk and high-risk women**
Trisomy 13	**Published– The White Journal (2012, avail online)**	**Performance for T13 detection with combined average-risk and high-risk women**
Fetal Fraction – NICE substudy	**Published – J Mat Fet Med (2012, avail online)**	**Fetal fraction same in high-risk and low-risk women**
Fetal Fraction	**Published – Fetal Diagnosis and Therapy (2012)**	**Fetal fraction correlated to placental mass**
NITE (Non-Invasive Trisomy Evaluation)	Enrolled	Multi-center European blinded study
NEXT (Non-invasive EXamination of Trisomy)	Enrolling	Multi-center blinded study of average risk women comparing Harmony to 1st trimester combined screening

NICE Study

RESEARCH www.AJOG.org

GENETICS

Non-Invasive Chromosomal Evaluation (NICE) Study: results of a multicenter prospective cohort study for detection of fetal trisomy 21 and trisomy 18

Mary E. Norton, MD; Herb Brar, MD; Jonathan Weiss, MD; Ardeshir Karimi, MD; Louise C. Laurent, MD, PhD; Aaron B. Caughey, MD, PhD; M. Hellen Rodriguez, MD; John Williams III, MD; Michael E. Mitchell, MD; Charles D. Adair, MD; Hanmin Lee, MD; Bo Jacobsson, MD; Mark W. Tomlinson, MD; Dick Oepkes, MD, PhD; Desiree Hollemon, MSN, MPH; Andrew B. Sparks, PhD; Arnold Oliphant, PhD; Ken Song, MD

OBJECTIVE: We sought to evaluate performance of a noninvasive prenatal test for fetal trisomy 21 (T21) and trisomy 18 (T18).

STUDY DESIGN: A multicenter cohort study was performed whereby cell-free DNA from maternal plasma was analyzed. Chromosome-selective sequencing on chromosomes 21 and 18 was performed with reporting of an aneuploidy risk (High Risk or Low Risk) for each subject.

RESULTS: Of the 81 T21 cases, all were classified as High Risk for T21 and there was 1 false-positive result among the 2888 normal cases, for a sensitivity of 100% (95% confidence interval [CI], 95.5–100%) and a false-positive rate of 0.03% (95% CI, 0.002–0.20%). Of the 38 T18 cases, 37 were classified as High Risk and there were 2 false-positive results among the 2888 normal cases, for a sensitivity of 97.4% (95% CI, 86.5–99.9%) and a false-positive rate of 0.07% (95% CI, 0.02–0.25%).

CONCLUSION: Chromosome-selective sequencing of cell-free DNA and application of an individualized risk algorithm is effective in the detection of fetal T21 and T18.

Key words: aneuploidy detection, cell-free fetal DNA, Down syndrome, noninvasive prenatal diagnosis, trisomy

Cite this article as: Norton ME, Brar H, Weiss J, et al. Non-Invasive Chromosomal Evaluation (NICE) Study: results of a multicenter prospective cohort study for detection of fetal trisomy 21 and trisomy 18. Am J Obstet Gynecol 2012;207:x.ex-x.ex.

Currently, the most effective and commonly used prenatal screening

★ EDITORS' CHOICE ★

ing tests have false-positive rates of 2-3% and false-negative rates of ≥5%.[1-4] Pos

NICE Study

- 50 participating clinical sites in U.S. and Europe
- Largest cohort study to date – All eligible subjects evaluated
- Study population was women undergoing invasive testing for any indication and thus included low risk women

	Sensitivity	Specificity	False Positive Rate
Trisomy 21	**100%** (81/81)	**99.97%** (2887/2888)	**0.03%** (1/2888)
Trisomy 18	**97%** (37/38)	**99.93%** (2886/2888)	**0.07%** (2/2888)

Norton ME et al. (2012) American Journal of Obstetrics and Gynecology

Average Risk Study

REPORTS OF MAJOR IMPACT www.AJOG.org

AQ: 3

Noninvasive prenatal testing for fetal trisomies in a routinely screened first-trimester population

AQ: 1,au Kypros H. Nicolaides, MD; Argyro Syngelaki, RM; Ghalia Ashoor, MD; Cahit Birdir, MD; Gisele Touzet, MD

AQ: 2 **OBJECTIVE:** We sought to assess performance of noninvasive prenatal testing for fetal trisomy in a routinely screened first-trimester pregnancy population.

STUDY DESIGN: This was a cohort study of 2049 pregnant women undergoing routine screening for aneuploidies at 11-13 weeks' gestation. Plasma cell-free DNA analysis using chromosome-selective sequencing was used. Laboratory testing on a single plasma sample of 2 mL was carried out blindly and results were provided as risk score (%) for trisomies 21 and 18.

RESULTS: Trisomy risk scores were given for 95.1% (1949 of 2049) of cases including all 8 with trisomy 21 and 2 of the 3 with trisomy 18. The trisomy risk score was >99% in the 8 cases of trisomy 21 and 2 of trisomy 18 and <1% in 1937 (99.9%) of the 1939 euploid cases.

CONCLUSION: Noninvasive prenatal testing using chromosome-selective sequencing in a routinely screened population identified trisomies 21 and 18 with a false-positive rate of 0.1%.

Key words: first trimester, noninvasive prenatal diagnostics, prenatal screening, trisomy 18, trisomy 21

Cite this article as: Nicolaides KH, Syngelaki A, Ashoor G, et al. Noninvasive prenatal testing for fetal trisomies in a routinely screened first-trimester population. Am J Obstet Gynecol 2012;207:x.ex-x.ex.

In the last 40 years, screening and diagnosis of fetal aneuploidies has

★ EDITORS' CHOICE ★

women with singleton pregnancies attending for their routine first hospital

Average Risk Study

- Independent blinded study
- Patient population:
  - 1st trimester pregnancy (11-13 weeks gestation)
  - General screening population of 2,049 women
- Results
  - NIPT test detected all trisomy cases
    - Trisomy 21: 8 of 8; Trisomy 18: 2 of 2
    - Risk score of >99% given for each trisomy
  - False positive rate
    - NIPT: 2 of 1,939 (0.1%)
      - No false positives for trisomy 21
      - 0.1% false positives for trisomy 18
    - Conventional screening (serum + NT ultrasound): 87 of 1,939 (4.5%)

Nicolaides KH et al. (2012) American Journal of Obstetrics and Gynecology

LABCO Quality Diagnostics

Screening for Aneuploidies by cf-DNA in maternal blood

By Far the best available option for T21 and 18

	100.000 Pregnancies			
	Trisomy 21 N=200		99.800 Normal	
METHOD OF SCREENING	DR	Detected		False Positive
Serum biochemistry at 16 wks	70%	140	5%	4990
Combined test at 12 wks	90%	180	5%	4990
Combined plus at 12 wks	97%	194	3%	2994
Cell-free DNA	>99%	>199	<0.1%	<100

- Can be offered to all women irrespective of risk
- Can provide result in the 1st trimester of pregnancy

K. Nicolaides SMFM SF 2013

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Harmony™ PRENATAL TEST Low False Positives

	False positive rate					List price
	T21	T18	T13	Y	Total	
Harmony™ PRENATAL TEST[1-3]	<0.1%	<0.1%	<0.1%	N/A	**<0.3%**	**$795**
MaterniT21™ PLUS	0.2%	0.28%	0.97%	0.6%	**2.0%**	**~$2,700**

Targeted NIPT shows false positive rates 5-7x lower than MPS

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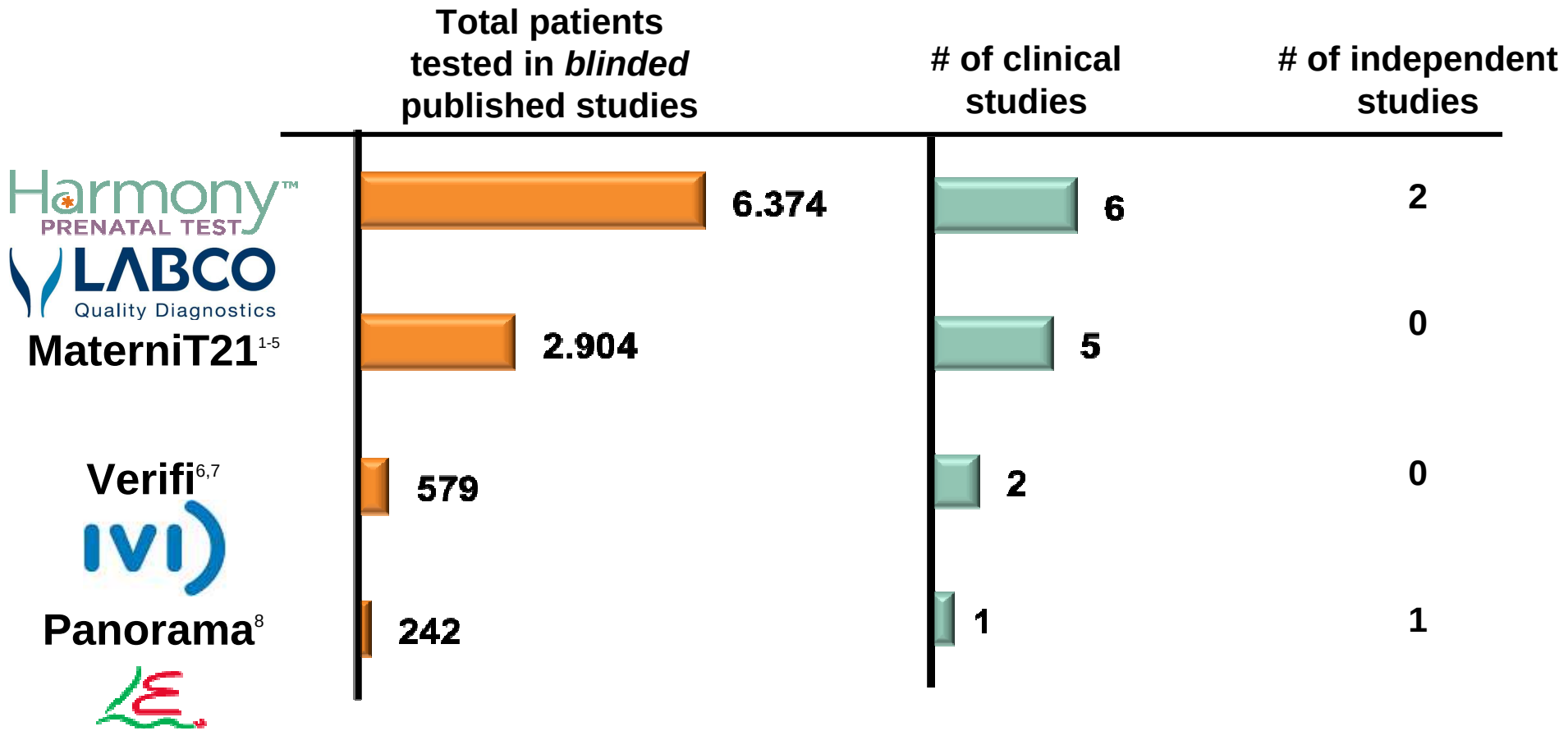
1. Norton et al, *Am J of Obstet and Gyn*, 2012; 2. Nicolaides KH et al, *Am J Obstet Gynecol* 2012; 3. Ashoor G et al., *Ultrasound Obstet Gynecol* 2012 (online); 4. Palomaki GE et al, *Genet Med* 2011; 5. Palomaki et al, *Genet Med* 2012; 6. MaterniT21 report example accessed Aug 2012

Comparison of Harmony Test vs other NIPT tests

	Harmony	MaterniT21+ *(Sequenom)*	verifi *(Verinata)*	NIFTY *(BGI)*	PraenaTest *(Lifecodexx)*	Panorama *(Natera)*
Fraccion Fetal medida para el test	+	+	–	–	+	+
Nivel de éxito	+	+	+	+	+	–
Ovodonación gemelares	+	–	–	–	–	–
Precio	695	–	1.000	820	–	900
Validación en estudios clínicos publicados	+	+	–	–	–	–

Data Comparison – Blinded Clinical Studies



1. Ehrich M et al. Am J Obstet Gynecol. 2011 Mar;204(3):205.e1-11. 2. Palomaki GE et al., Genet Med. 2011 Nov;13(11):913-20, 3. Palomaki GE et al., Genet Med. 2012 Mar;14(3):296-305. 4. Canick JA et al., Prenat Diagn. 2012 Aug;32(8):730-4. 5. Mazloom et al, Prenat Diagn. 2013 Apr 16. [epub ahead of print] 6. Sehnert AJ et al., Clin Chem, 2011 Jul;57(7):1042-9.
7. Bianchi DW et al., Obstet Gynecol, 2012 May;119(5):890-901. 8. Nicolaides et al, Prenat Diagn. 2013 Apr 24:1-5. [Epub ahead of print].

Harmony Test

Validado

- El de mayor número de estudios clínicos
- >100,000 muestras testadas ya en clínica

Respetado

- Acuerdos de investigación con Universidades y centros públicos
- Aprobado y utilizado por los líderes mundiales en medicina fetal

Accessible

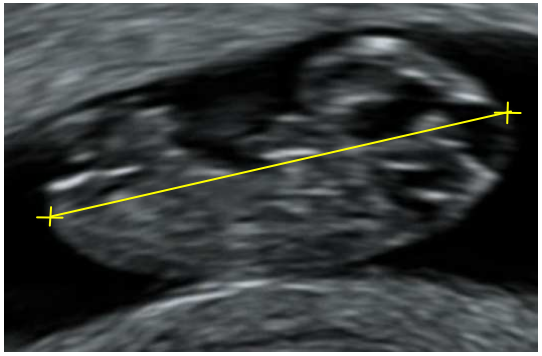
- El de más bajo precio
- Disponible a nivel mundial

Calidad

- Facción fetal medida e incluida en informe
- Monotorización de calidad en cada test

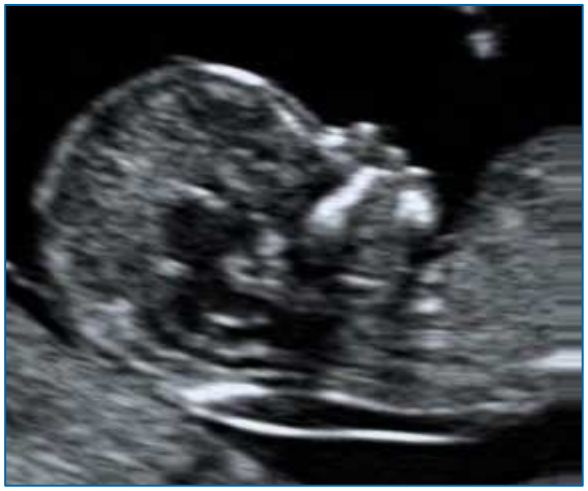
Fetal Medicine Center (Nicolaides): 10wk Implementation Protocol

The Fetal Medicine Centre

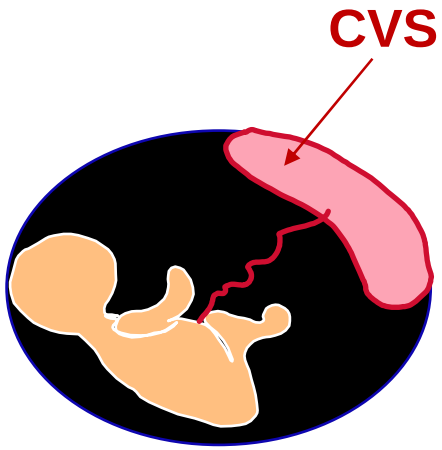


Harmony™ PRENATAL TEST

10 weeks:
- Scan to measure the fetus
- Blood for cfDNA test
- Blood for combined test



12 weeks:
- Detailed ultrasound scan
- Discuss results
- Decide if CVS is necessary



- +ve cfDNA test
- Fetal defects
- NT ≥3.5 mm

SEQUENCES OF PRENATAL TESTS

COUNSELLING (MATERNAL AGE/ HISTORY)

Non Invasive Screening
1st Trimester

mid/low

High/Ultrasound

NIPD

NIPD?

CVS / AF Confirmations

CVS / AF

QF-PCR

QF-PCR

aCGH

?

CYTOGENETICS

laboratorios

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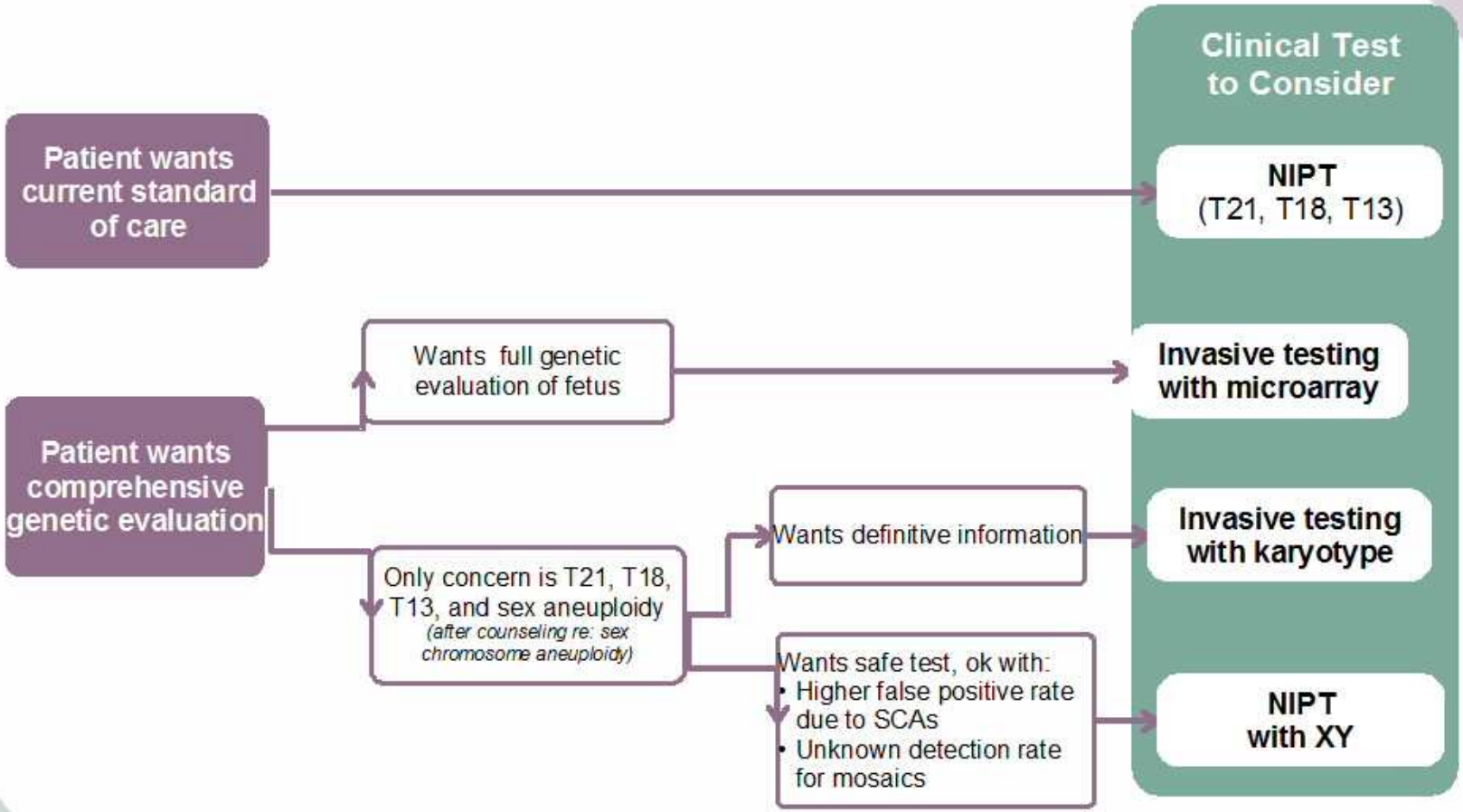
Quality Diagnostics

Testing Beyond Common Trisomies

Harmony™ PRENATAL TEST



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SEQUENCES OF PRENATAL TESTS

COUNSELLING (MATERNAL AGE/ HISTORY)

Low Risk Screening (Anxiety)

NIPD

Common Trisomies
Residual risk 1:1200

CVS / AF

QF-PCR

aCGH

Whole Genome

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Quality Diagnostics

Test Report

LABCO Quality Diagnostics

Harmony™ PRENATAL TEST
www.harmonytest.com

Ariosa DIAGNOSTICS
Ariosa Diagnostics, Inc.
5945 Optical Court
San Jose, CA 95138

For Questions:
clientservices@ariosadx.com
US: (855) 927-4672
Intl: +1 (925) 854-6246

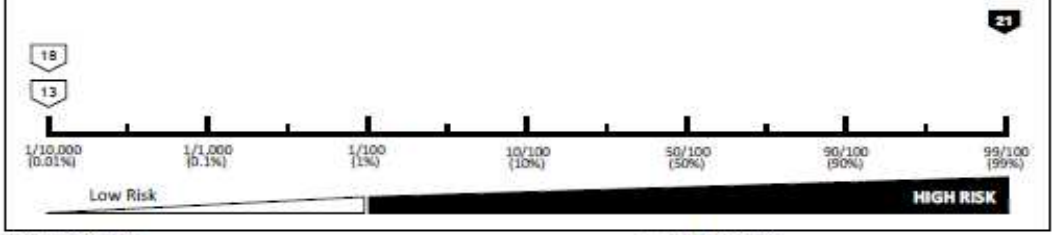
Patient and Provider Information

PATIENT NAME:	Jane Doe	ACCOUNT #:	7654321
DATE OF BIRTH:	01/01/1970	CLINIC NAME:	The Clinic Offering Test
MRN:	1234567890123456789	REFERRING/ORDERING CLINICIAN:	Ordering Physician MD
ARIOSA ID: AD12345678-PAT	OTHER ID: 00123456789012345XZY	REFERRING/ORDERING CLINICIAN FAX #:	123-456-7890
GESTATIONAL AGE:	10 wks 5 days	OTHER CLINICIAN:	Genetic Counselor MA, CGC
# OF FETUSES: 1	IVF STATUS: non-IVF pregnancy	OTHER CLINICIAN FAX #:	987-654-3210
COLLECTION DATE: 01/01/2012	RECEIVED DATE: 01/02/2012	REPORT DATE:	01/10/2012

Test Results

Fetal Fraction: 10.5%

CHROMOSOME	RESULT	PROBABILITY	RECOMMENDATION
Trisomy 21 (T21)	**HIGH RISK**	**Greater than 99/100 (99%)**	**Genetic counseling and additional testing**
Trisomy 18 (T18)	Low Risk	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 13 (T13)	Low Risk	Less than 1/10,000 (0.01%)	Review results with patient
Y Analysis	Male Fetus	Greater than 99/100 (99%)	Review results with patient



TEST DESCRIPTION

Ariosa's laboratory developed tests perform a directed analysis of cell-free DNA (cfDNA) in maternal blood and have been validated in pregnancies of at least 10 weeks gestational age. The probability of autosomal or sex chromosome aneuploidy is based on cfDNA in blood which may not always correlate with fetal genotype. Aneuploidy test results incorporate maternal age (or egg donor age) and gestational age-related risk based on information from the test requisition form. Tests are neither intended nor validated for diagnosis or use in mosaicism, partial chromosomal aneuploidy, translocations, or maternal aneuploidy. Not all aneuploid fetuses will be detected and some euploid fetuses may have high probability for aneuploidy results. Results should be considered with other clinical criteria and communicated in a setting that includes appropriate counseling.

The Harmony Prenatal Test measures the relative proportion of chromosomes to aid in the risk determination of fetal trisomies 21, 18, and 13. This test has only been validated in singleton pregnancies and in twin pregnancies (excluding twins from unrelated egg donors).

Y Analysis provides no information on the X chromosome. A "Female Fetus" result indicates a lack of significant Y chromosome sequences and does not exclude XO. A "Male Fetus" result does not exclude XXY. A Y "Aneuploidy" result indicates two or more fetal Y chromosomes. This test has only been validated in singleton pregnancies. Y "Aneuploidy" is reported only when probability is >99%.

CLINICAL DATA

	Detection Rate	False Positive Rate
T21	>99%	<0.1%
T18	>98%	<0.1%
T13	• Given rarity of condition, limited T13 cases analyzed • Detection rate: 8 of 10 with Harmony • False positive rate: <0.1% (95% CI: 0.0–0.3%)	

Detection and false positive rates based on risk cut-off of 1/100 (1%) and are based on singleton, non egg donor pregnancies.

Y Analysis: >99% accuracy for male or female sex

Y Analysis also provides probability for non-mosaic Y aneuploidy

REFERENCES: Norton ME et al. (2012) Am J Obstet Gyn 207(2):137.e1-8., Nicolaides KH et al. (2012) Am J Obstet Gyn 207(5):374.e1-6 Ashoor G et al. (2012) Ultrasound Obstet Gynecol [epub ahead of print], data on file

The Harmony Prenatal Test is intended for clinical use and should not be regarded as investigational or for research. It was developed, and its performance characteristics determined, by the Ariosa Diagnostics Clinical Laboratory, which is certified under the Clinical Laboratory Improvement Act of 1988 (CLIA) as qualified to perform high complexity clinical testing. The test has not been cleared or approved by the U.S. Food and Drug Administration.

Laboratory Director: M. Junaid Shabbeer, PhD, FACMG

CLIA #05D2032812 STATE # CLF 341864

TP-00115-01 Rev 8.0

Gynea laboratorios

Distinct Individualized Results

99.5% of risk score values are at either extreme

Test Results **Fetal Fraction: 10.5%**

CHROMOSOME	RESULT	PROBABILITY	RECOMMENDATION
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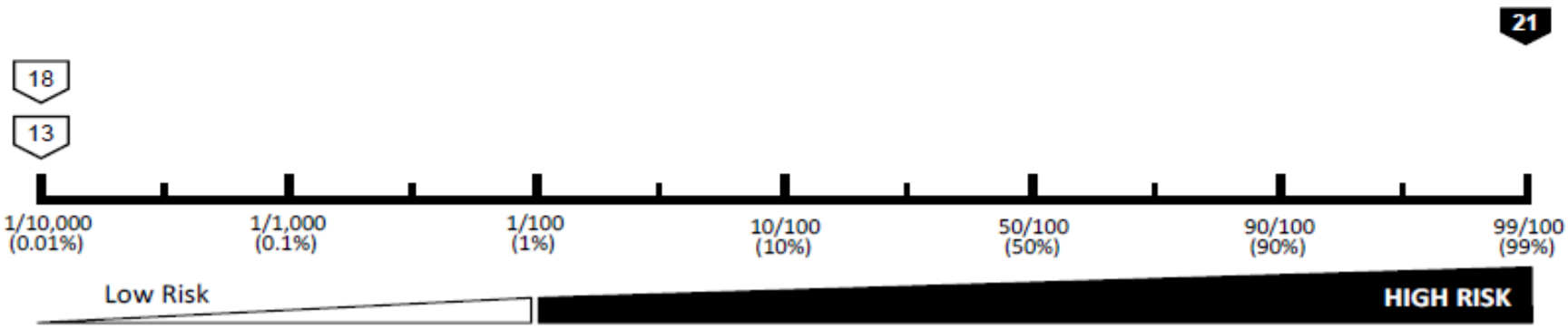
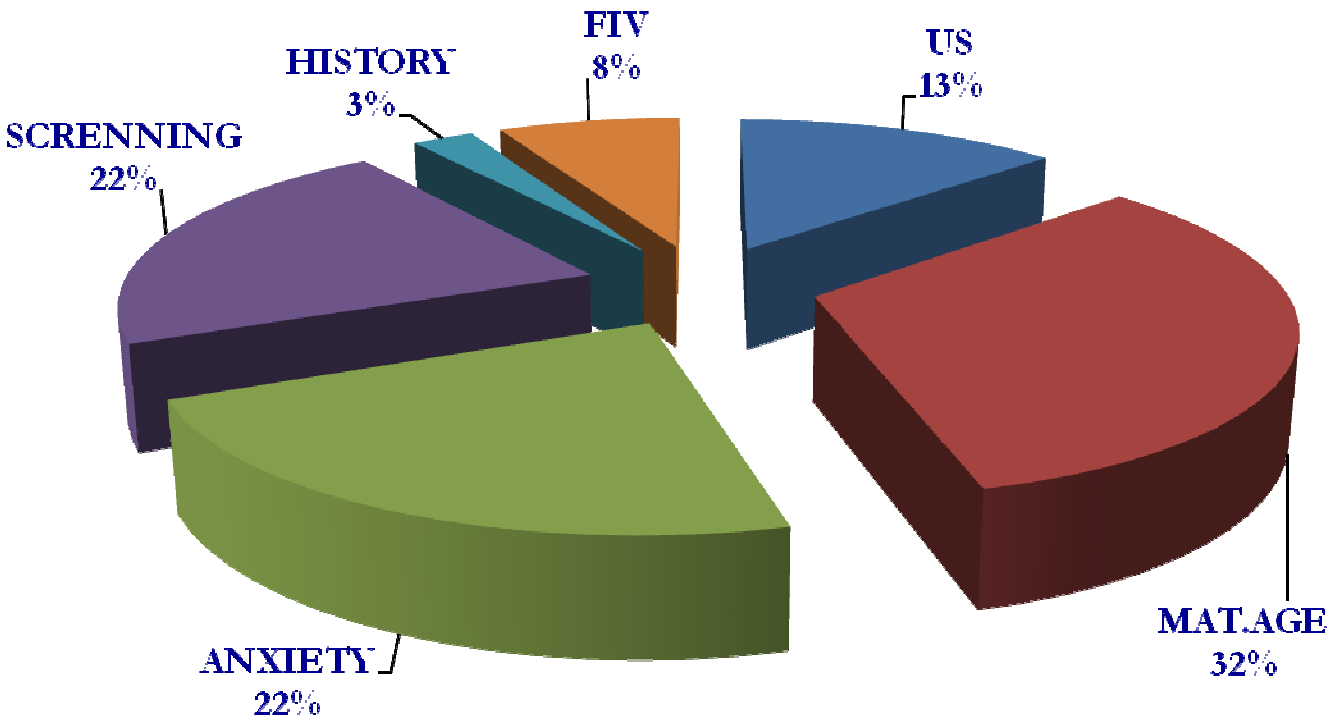


Gynea laboratorios

Harmony Test Indications for 1000 Consecutive Samples



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LABCO
Quality Diagnostics

Experiencia con los primeros 5.000 casos

- A pesar de su muy buen rendimiento es una prueba de cribado no diagnóstica
- Avisar siempre que aunque con poca probabilidad hay pacientes en las que no podremos obtener resultado
- Aumentan considerablemente las sesiones de consejo genético.

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Conclusiones

- En 2013 se han realizado 150.000 NIPD en U.S.
- Se calcula que en 2014 se realizaran 450.000 en U.S.
- En España se han hecho ya más de 10.000, 8.000 Harmony
- El factor limitante actual es el precio
- El test está diseñado y validado para gestantes de bajo riesgo NO de alto riesgo
- Si la TN es mayor de 3 mm hacer directamente procedimiento invasivo

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GRACIAS!

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