



Cell-free DNA and contingent screening our first YEAR

Gomes, H.¹; Lourenço, I.¹; Ribeiro, J.¹; Francisco, C.¹; Marques, J.P.¹

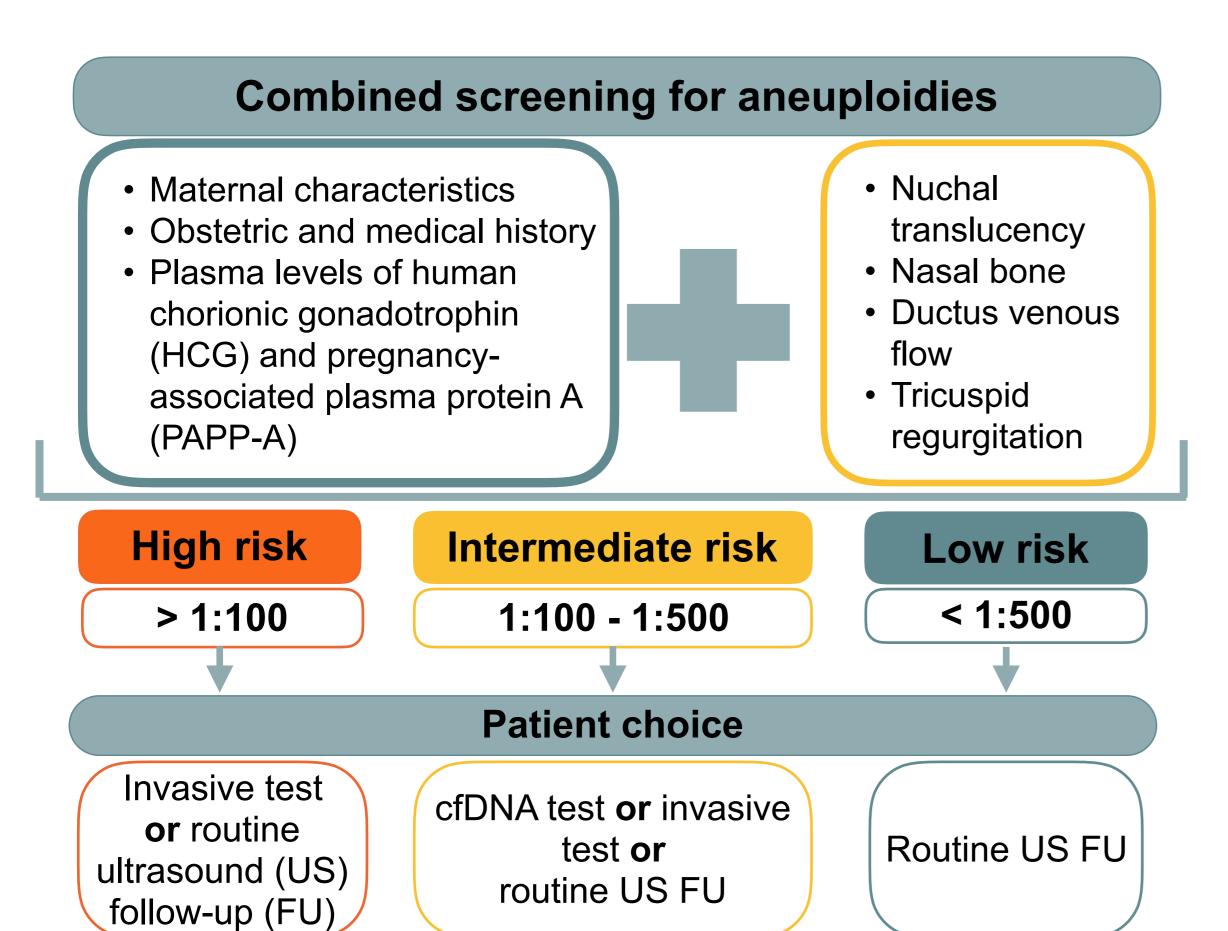
1 - Department of Gynecology and Obstetrics, Hospital Beatriz Ângelo, Loures

Objective

To access the performance of a contingent screening for an euploidies in the first trimester with the introduction of cell-free DNA (cfDNA), one year after the implementation of a new screening protocol in our Hospital.

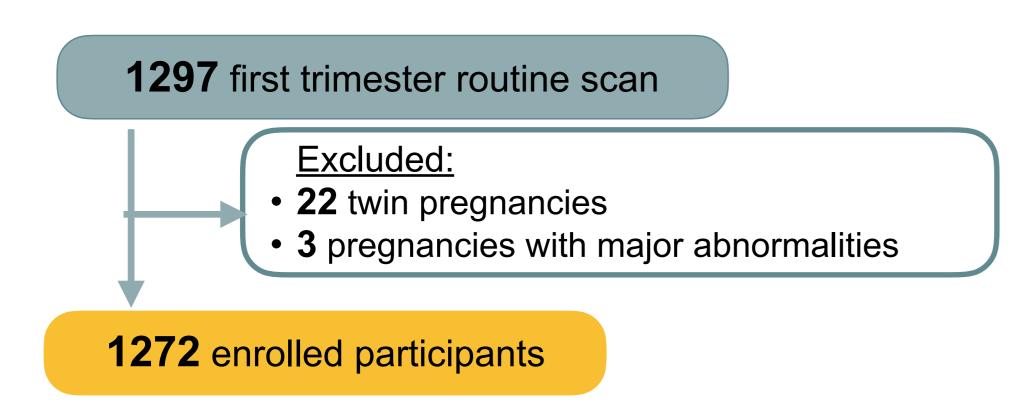
Study Design

- Prospective study
- March 2017 February 2018
- Women attending for their routine 1st trimester scan



Results

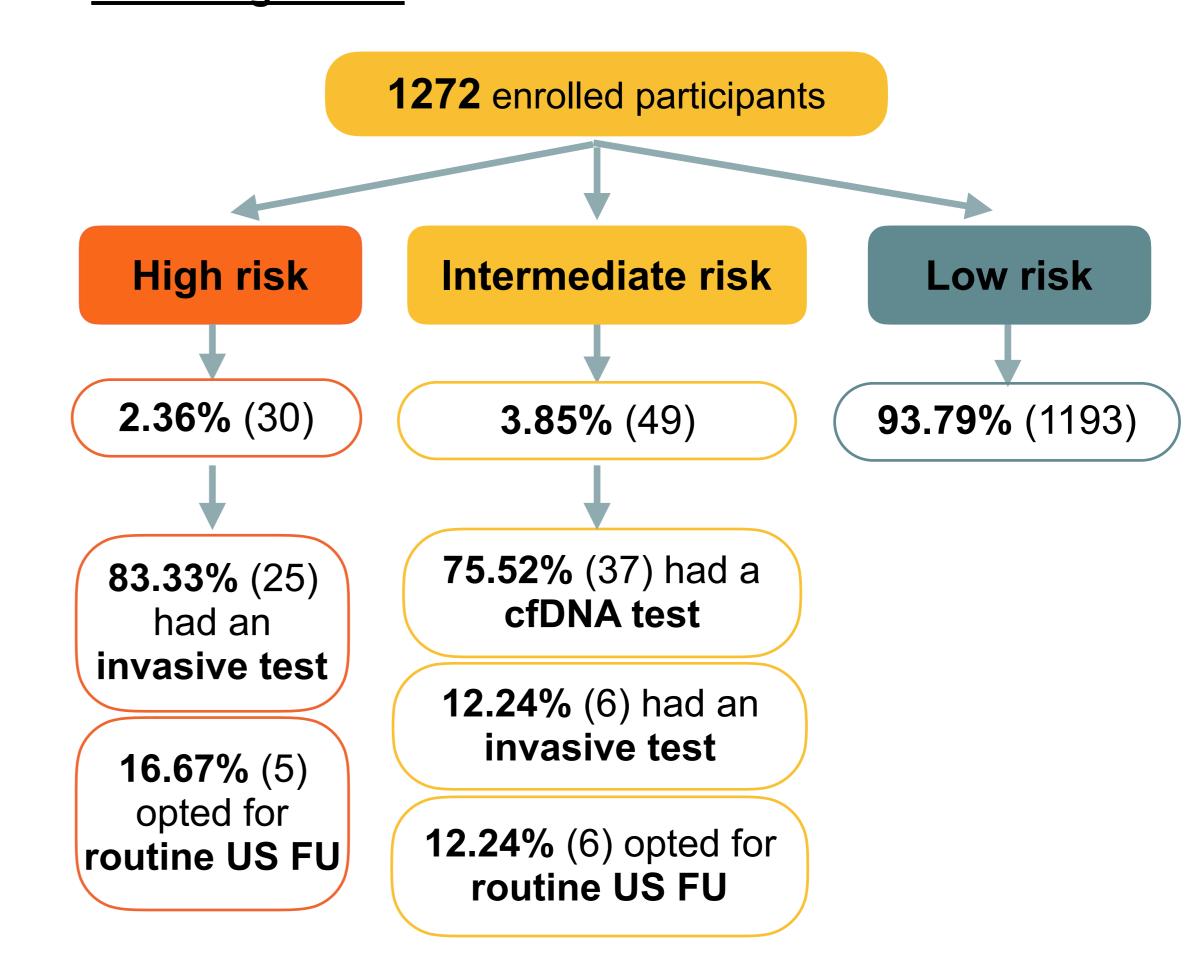
Population



Maternal and pregnancy characteristics	n=1272	
Maternal age (years) (median ± SD)	30.05 ± 5.9	
Maternal BMI (Kg/m²) (median ± SD)	25.06 ± 5.31	
Racial origin - no. (%)		
Caucasian	1051 (82.6%)	
Afro-Caribbean	161 (12.7%)	
South Asian	31 (2.4%)	
East Asian	4 (0.3%)	
Mixed	25 (2.0%)	
Parity - no. (%)		
Nulliparous	614 (48.3%)	
Multiparous	658 (51.7%)	
Obstetric and medical history - no. (%)		
Previous pregnancy with a chromosomal abnormality	3 (0.24%)	
Conception - no. (%)		
Spontaneous	1256 (98.7%)	
Ovulation drugs or IVF	16 (1.3%)	

Results

Screening results



All cfDNA test had a normal result

2.44% (31) of the enrolled participants had an invasive test

3.52% invasive test before implementation of the screening

There was a **significant reduction** in the **rate of invasive test**, (2.44% vs 3.52%, p=0.086), after implementation of continent screening.

<u>Outcomes</u>

	High risk (n=30)	Intermediate risk (n=49)	Low risk (n=1193)
Abnormal Karyotype	7	1	2
Termination	6	0	0
Miscarriage	0	1	0
Live birth	0	0	2
On-going pregnancy	1	0	0
Live birth	15	37	869
Termination	1	0	4
Miscarriage	1	0	2
Fetal death	0	1	1
Missing outcome	2	1	15
On-going pregnancy	4	9	300

Conclusion:

- A contingent screening model, using cfDNA has a place in a routine care setting.
- The introduction of this screening method decreased the rate of invasive tests.
- There were no false negative results on the cfDNA tests.