One clinic's experience with **Cell Free Fetal DNA** testing in **Argentina**

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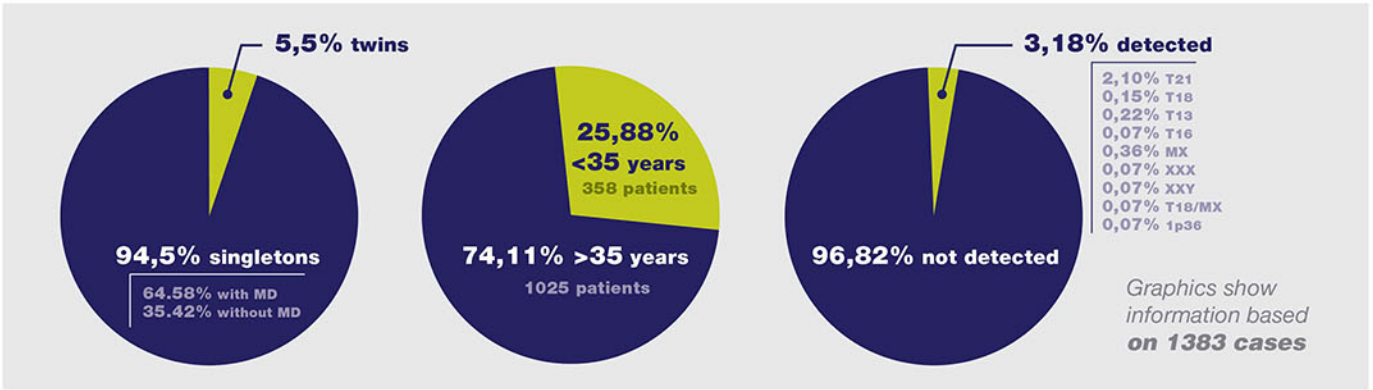
Objective: We have offered cell free fetal DNA (cffDNA) testing in our clinic in **Buenos Aires, Argentina since July 2013.** We want to highlight our experience with cffDNA testing in over 1400 patients, one of the largest cohort reported in LATAM. This is an up to date of the data presented last year at ISDP Meeting in Washington USA.

Method: All samples were shipped to **Illumina's CLIA Laboratory** in USA to perform the **verifi®** prenatal test. Initially, we offered testing for chromosomes 21/18/13/X/Y for singletons and chromosomes 21/18/13/Y for twins. In July 2014, we began to offer additional testing for all eligible singletons, including chromosomes 9/16 and a microdeletion panel. The data here outlines our experience during the period July 2013 – February 2016.



Results: Outcome information was reviewed for a total of 1401 cases. The number of performed tests increased in 761 (121%) since our first report that included 629 samples. Results were reported in **1383 cases (98,71%)**, with an average **turn-around-time of 2.95 business days** from date of sample receipt in lab. The total turnaround time for a patient in ARG is 6.4 days due to logistic conditions. There was **one (0.071%) technical cancellation**, for which a redraw was successful in yielding a result. Of 18 (1.28%) administrative cancellations, all were due to sample arrival in lab beyond stability date (>5 days from date of draw). **Of the 1383 reported cases, 1307 (94,5%) were singletons; 76 (5,5%) were twins.** Initially, we offered cffDNA testing to high risk patients (i.e. patients ≥35 years at delivery, positive first trimester combined test and/or ultrasound findings); however, we recently have noticed a trend in increased use of cffDNA testing in the average risk population. Overall, **aneuploidy was detected (AD) or suspected (AS) in 44 (3.18%) samples, with 42 (3.04%) and 2 (0.14%) reported as AD and AS, respectively.** Of the AD cases, 29 T21 (2.10%), 2 T18 (0.15%), 3 T13 (0.22%), 1 T16 (0.07%), 1 T18/MX (0.07%), 5 MX (0.36%), 1 XXX (0.07%) and 1 XXY (0.07%) were reported. As expected, we saw an increased prevalence of AD cases (28/44) in patients with abnormal ultrasound findings. Of the 2 AS cases, 1 T21 and 1 T13 were reported; AS T21 was confirmed by amniocentesis, and no data is available for the AS T13. Of the eligible singletons, 844 elected to proceed with microdeletion testing, with 1 abnormal result for 1p36 deletion (0.07%) reported. **Outcome information was available in 31/44 (70,45%) aneuploidy cases; two putative false positive aneuploidy cases reported (T18 and double aneuploidy T18/MX).** Of note, for the double aneuploidy case, the patient declined further clinical investigation, including work-up for a possible maternal malignancy. Four cases of XY/XX discordance were reported. The number of invasive procedures performed decreased dramatically and the proportion of low risk patients is approximately ¼ during the entire cohort One putative false positive microdeletion case was reported. **No false negative cases were reported.**

Conclusions: This is an **up to date of our last clinical experience and continues showing the consistence of this technology.** Efforts made in medical education and an a proper patient information opened the possibility to increase the acceptance as a secondary, or primary, screening test for the most prevalent aneuploidies. Continual monitoring of its performance in the clinical setting is important for assuring accurate and reliable results and explore potential application especially with NGS technology.

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