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Benefits of the cell-free fetal DNA protocol for the screening of prenatal aneuploidies in a Catalan population (2022)

Madurga Hernández A (1), Mercadé Salavert I (2), Navarro Badal R (1), Casals Font E (2), Alumà Trullàs A (3).

Laboratori Clínic Territorial Metropolitana Sud (Hospital Universitari de Bellvitge) (1), Servicio de Bioquímica Clínica y Genética Molecular (Hospital Clínic) (2), Laboratori Clínic Territorial Metropolitana Nord (Hospital Germans Trias i Pujol) (3).

INTRODUCTION

The cell-free fetal DNA in maternal blood (cfDNA) protocol, which was established to detect prenatal aneuploidies and decrease the number of invasive tests, has been widely applied to pregnant women in the Catalan public health system.

The laboratories that participate in this protocol are Hospital Vall d'Hebron (Barcelona), Laboratori Clínic Metropolitana Sud (Hospitalet de Llobregat-Barcelona), Laboratori Clínic Metropolitana Nord (Badalona-Barcelona), Hospital Clínic (Barcelona), LRC (Barcelona), Catlab (Terrassa), Hospital de Santa Creu i Sant Pau (Barcelona), Hospital Universitari Dr. Josep Trueta (Girona), Hospital Arnau de Vilanova (Lleida) and Hospital Joan XIII (Tarragona).

OBJECTIVES

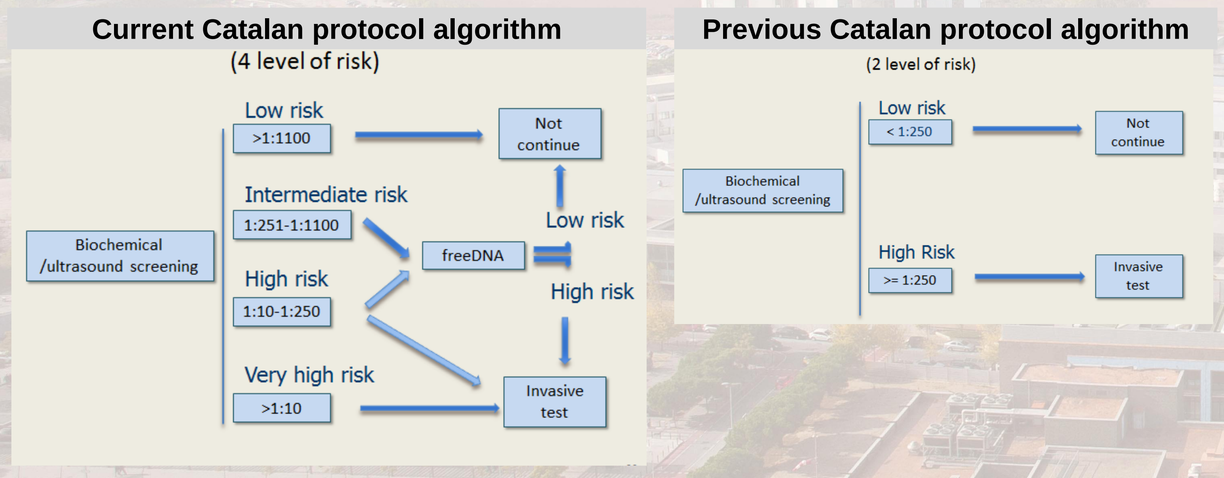
The aim of this study is to review the detection rate of first-trimester trisomy screening during the year 2022 and to evaluate the reduction of invasive procedures after applying the cfDNA protocol compared to the previous protocol.

MATERIALS AND METHODS

The first-trimester screening for aneuploidies involves measuring biochemical markers during the 8th and 13.6th weeks of pregnancy, including Srm-β-free chorionic gonadotropin hormone, c.subst.arb, and Srm-pregnancy-associated plasma protein A, c.subst.arb. Additionally, ultrasound measurement of nuchal translucency (NT) is done during the 11th and 14th weeks of pregnancy.

The current protocol, "*Actualització del Protocol de cribratge prenatal d'anomalies congènites a Catalunya*," classifies results into four risk categories: very high risk (>1/10), high risk (1/10-1/250), intermediate risk (1/251-1/1100), and low risk (<1/1100). To pregnant women with very high risk is offered an invasive procedure, while women with high risk can choose between an invasive procedure or an cfDNA test. Women with intermediate risk have the option of doing an cfDNA test, and women with low risk do not undergo any further testing. In contrast, the previous protocol only offered an invasive procedure to pregnant women with a risk ratio of >1/250 (Figure 1).

1-Comparison of the current Catalan algorithm in regards to the previous protocol:



RESULTS

A total of 34,050 prenatal screening tests were performed during the first trimester of single-gestation pregnancies and analyzed.

Regarding demographics, the mean age of all pregnancies was 32.6 [14-54] years old, and the mode was 34.7 years old. The mean weight was 67.1 kg, with 0.68% of the cases having diabetes and 10.01% being smokers. Concerning origin, 68.5% of the cases were Caucasians.

Regarding Trisomy 21, the current protocol detected 99 cases, resulting in a 100% detection rate. In comparison, the previous protocol detected 91.9% of total cases. For Trisomy 18, 23 cases were detected with the current protocol, with a 95.8% detection rate. In comparison, the previous protocol detected 91.7% of total cases. Finally, for Trisomy 13, the current protocol detected eight cases, with a 100% detection rate, and an equal number was detected using the previous protocol (Table 2).

Regarding invasive procedures, 330 were performed out of a total of 34,050 cases, of which 283 underwent a villous chorionic biopsy, and 47 underwent an amniocentesis. This data is equivalent to 0.97% of total cases. If we compare the number of invasive procedures that would have been performed with the previous protocol (1,303),this is equivalent to 3.83% of total cases.

Therefore, with the new protocol, there has been a reduction of 74.67% of invasive procedures, which is 973 less than with the previous one (Table 3).

2- Description of the detection rate for Trisomies 21, 18 and 13 with the current and previous protocol:

Protocol	Positive screening for Trisomy 21	Total cases of Trisomy 21	Detection rate
Current protocol	99	99	100,0%
Previous protocol	91	99	91,9%

Protocol	Positive screening for Trisomy 18	Total cases of Trisomy 18	Detection rate
Current protocol	23	24	95,8%
Previous protocol	22	24	91,7%

Protocol	Positive screening for Trisomy 13	Total cases of Trisomy 13	Detection rate
Current protocol	8	8	100,0%
Previous protocol	8	8	100,0%

3-Comparison of invasive procedures that were performed with the new protocol during the year 2022 in regards to the previous protocol:

Invasive procedures	Current protocol	Previous protocol
Number of cases	330	1303
Percentage	0,97%	3,83%
Difference of interventions	**-74,67%**	

CONCLUSIONS

The inclusion of the cfDNA in the current protocol of the Catalan public health system has had a very beneficial impact on the detection of first-trimester trisomies, resulting in a very high results for Trisomy 21, 18, and 13 during the year 2022.

It has also had a significant effect on reducing invasive procedures compared to the previous protocol, with a decrease of 74.67% of invasive procedures.

This data supports the benefits of the inclusion of cfDNA in the current Catalan protocol.