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Prenatal invasive diagnostic tests: diagnostic performance according to procedure indication

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Objective

To assess the diagnostic performance of prenatal invasive diagnostic testing according to procedure indication.

Methods

A retrospective cohort of all pregnant women undergoing an invasive test for prenatal diagnosis at Hospital General de Hospitalet between January 2009 and December 2021. Type of procedure, procedure indication, and genetic results were reviewed.

Results

Overall, 1211 prenatal invasive tests were performed, 436 chorionic villus sampling (CVS) (36%), 774 amniocenteses (64%). The number of tests relative to the number of pregnancies remained stable at 4% (CI 3-5%), despite the progressive decrease in the number of pregnancies and tests per year. The main indications for Prenatal invasive testing were: high-risk combined aneuploidy screening (37%), fetal malformations (29%), and increased nuchal translucency (8%). A progressive and significant shift in test justification throughout the years was observed, particularly after 2018, with an increase in invasive tests performed for fetal malformations (from 18% in 2009 to 50% in 2021, $p<0.001$) and a decrease in those performed for high-risk combined aneuploidy screening (from 43% to 10%, $p<0.001$) due to the introduction of cell-free DNA testing in the national health service in those pregnancies at high risk for trisomy 21 at the combined screening The result of genetic testing was normal in 1014 patients (84%), abnormal in 170 patients (14%) and no result was obtained in 27 patients (2%). During the study period the invasive test positive rate steadily increased from 7% in 2009 to 27% in 2021 ($p<0.001$). The positive rate of the prenatal invasive tests according to procedure indication was as follows: increased nuchal translucency (41% abnormal), hereditary disease (39.1% abnormal), first trimester pregnancy loss (42.2% abnormal), second trimester pregnancy loss (22.7% abnormal), fetal malformation (13.5% abnormal), high-risk combined aneuploidy screening (9.5% abnormal), intrauterine growth restriction (6.7% abnormal), third trimester pregnancy loss (5% abnormal), maternal age/anxiety (3.1% abnormal) and previous aneuploidy (2.7% abnormal).

Conclusion

Indications for invasive prenatal testing have changed over the years and so have changed the positive rate of the invasive procedures. The most important reason in our cohort may be the introduction of cel-free DNA testing in the national health service in pregnancies at high risk for trisomy 21, but also the use of more sensitive diagnostic technics such as micro array and NGS.