A retrospective study of Down syndrome in prenatal diagnosis. Did chorionic villus sampling allow a better prevention?

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Introduction

Down syndrome (DS) is the most common single genetic cause of human moderate mental retardation, with an estimated prevalence of 9.2 cases per 10,000 live births. We aimed at analyzing changes in prenatal diagnosis (PND) over time, namely the referral reasons for chromosome analyses and the introduction of chorionic villus sampling (CVS), and its influence on the results obtained in DS cases.

Methods

We retrospectively evaluated the PND results from samples analyzed between 1987 and 2011 (25 years) in our cytogenetic laboratory taking into account the referral reasons, type of sample, karyotype and reporting time.

Results

263 fetuses with a karyotype compatible with DS were identified in a total of 18,107 karyotypes (1.5%). The highest frequencies of DS were found among cases referred because of ultrasonography findigs (namely increased nuchal translucency) or positive first trimester screening and when one parent carries a chromosomal rearrangement. The frequency of recurrence was found to be 1/72.

The increasing use of CVS led to an earlier response in terms of gestational age (mean at diagnosis- 13+4 weeks). In addition, an increased percentage of karyotypes with SD was detected (8.4% of CVS samples). On the other hand, implementation of molecular rapid aneuploidy detection in part of the samples allowed a better report time in DS cases, from 23 days in 1987 to 2 days in 2011.

Discussion

DS detection remains the most important reason for performing PND. The collecting of CVS has been rising over the last years, which has resulted in an increased number of trisomy 21 cases identified in a lower gestational age, allowing a better karyotype-phenotype correlation in earlier pregnancies. Moreover, the use of complementary molecular techniques for the detection of common aneuploidies reduced the mean reporting time and allowed an earlier decision of the couple concerning the future of gestation.