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Prenatal karyotype results of 2169 invasive tests

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Objective

A foetal karyotyping is a basic tool to diagnose the most frequent genetic syndromes. Widely used molecular methods such as FISH, MLPA or QF-PCR deliver a rapid diagnosis of the trisomy 21, 18, 13. These methods are fully available in the almost all centres of prenatal diagnosis. A microarray method of genetic material analysis is a more sensitive method. However it is more expensive, time consuming and difficult to interpret. Therefore, it is still less available. The aim of the study was to analyse the results of a foetal karyotype obtained in the course of prenatal diagnostics.

Methods

We analysed 2169 foetal karyotypes from 2 large referral university centres for prenatal diagnosis in Lodz, Poland.

Results

The invasive procedures were performed when screening methods showed a high risk for chromosomal aberrations or foetal abnormalities. The study was conducted from 2008 to 2017. There were 206 (9, 5%) abnormal foetal karyotypes. Down syndrome was diagnosed in 93 cases. Simple trisomy 21 was found in 91 cases and the Robertsonian translocation in 2 cases of Down syndrome. There were 32 cases of the Edwards syndrome and 18 cases of the Patau syndrome. 2 cases of the Patau syndrome were caused by the Robertsonian translocations. Important notice was that other abnormalities than the trisomy 21, 18 and 13 were found in 30% (n=63) positive cases. Turner syndrome was present in 17 cases, Klinefelter syndrome in 3 cases and other triploidies in 8 cases. 35 cases had a rare structural aberration. Among structural aberrations there were 17 translocations, 9 inversions, 3 deletions and 2 duplications. A marker chromosome was present in 4 cases.

Conclusion

A significant part of positive results in prenatal tests are other chromosomal abnormalities than the most frequent trisomy 21, 18 and 13. The foetal karyotyping still remains an important method for a prenatal diagnosis of chromosomal aberrations.