Does cfDNA testing change the spectrum of detected major chromosomal aberrations?

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Objective

Prior to 2012, detection of major chromosomal aberrations (CHA) had been based mainly on combined first trimester screening, integrated screening tests and ultrasound examinations, all of them covered by national health plans. High - risk cases had been offered targeted invasive diagnostics, followed by karyotype and/or microarray investigation. The implementation of cfDNA testing may lead to decrease in invasive testing resulting in lower accidental detection of other autosomal abnormalities, apart from 21, 18 and 13, in the population. We tested the hypothesis, that the ratio of prenatally diagnosed CHA has been changed since the gradual implementation of cfDNA testing after 2012.

Methods

We used population based data from the National Registry of Congenital Anomalies, stored in the Institute of Health Information and Statistics of the Czech Republic, including the individually collected data on prenatally diagnosed cases. We analyzed the numbers of successfully prenatally diagnosed cases of all CHA (ICD-10 codes: Q90-Q99) during the 2012 - 2015 time period. The numbers were evaluated separately for the group of main autosomal trisomies and “other” CHA. The time trends were statistically evaluated using GraphPad InStat statistical software.

Results

The number of invasive procedures, amniocenteses and chorionic villous samplings decreased from 11407 in the year 2012 to 7680 in 2015 (67,3%). A total of 575 cases of CHA were identified in 2012, of which 329 cases (57.5%) were trisomies 21, 18 and 13. In 2015, 577 cases of all CHA were identified, with major trisomies accounting for 374 cases (64.8%). There was no significant change in the group of major autosomal trisomies, compared to the “other” CHA ($p > 0.05$). During the whole study period (2012 - 2015) the total number of prenatally diagnosed CHA was 2369; major trisomies accounted for 1462 cases (76.3%).

Conclusion

CfDNA testing and the increase in use of first trimester screening has led to a decrease of invasive procedures over the study period. As cfDNA testing is not covered by insurance plans, we only have rough estimation of its use. However, we managed to prove a decrease in invasive procedures with detection rates of both major autosomal and “other” trisomies remaining unchanged.