Screening for chromosomal anomalies – are we doing more for less?

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Objective

To review the performance of screening options regarding clinically significant chromosomal anomalies detection rate.

Methods

Options of screening for chromosomal anomalies are presented from literature reports including clinical, biochemistry, ultrasound, NIPT and combined tests with their performance in terms of detection rates and invasive testing rate. Chromosomal anomalies detected by invasive testing are presented with their distribution depending on the diagnostic test used. Invasive testing can influence detection rate and distribution as well as influence by type of diagnostic test used.

Results

Modern and fashionable non invasive technology that screens for aneuploidy is very important. Better results are expected with future development of this technology both for financial issues and for wider usage. Specific diagnostic tests used also influence the detection rate.

Conclusion

Implementing and practicing a rigourous combined test warrants the best available option in screening for chromosomal anomalies in the population. Together with the genetic counseling for the high risk patients in order to use the proper diagnostic test, we are on track for optimising our resource usage.