

Prenatal diagnosis of major chromosomal aberrations in the era of NIPT (Non-invasive prenatal testing): What could we miss?

**Antonin Sipek Jr.^{1,2}, Vladimir Gregor^{2,3}, Antonin Sipek^{2,3,4},
Jiri Horacek^{2,5} and Pavel Calda⁶**

¹ Institute of Medical Biology and Genetics, 1st Faculty of Medicine, Charles University and General University Hospital, Prague, Czech Republic

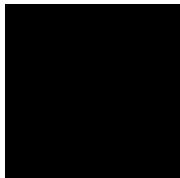
² Department of Medical Genetics, Thomayer Hospital, Prague, Czech Republic

³ Department of Medical Genetics, Pronatal Sanatorium, Prague, Czech Republic

⁴ Institute of Biology and Medical Genetics, 3rd Faculty of Medicine, Charles University, Prague, Czech Republic

⁵ Gennet, Centre for Foetal Medicine and Reproductive Genetics, Prague, Czech Republic

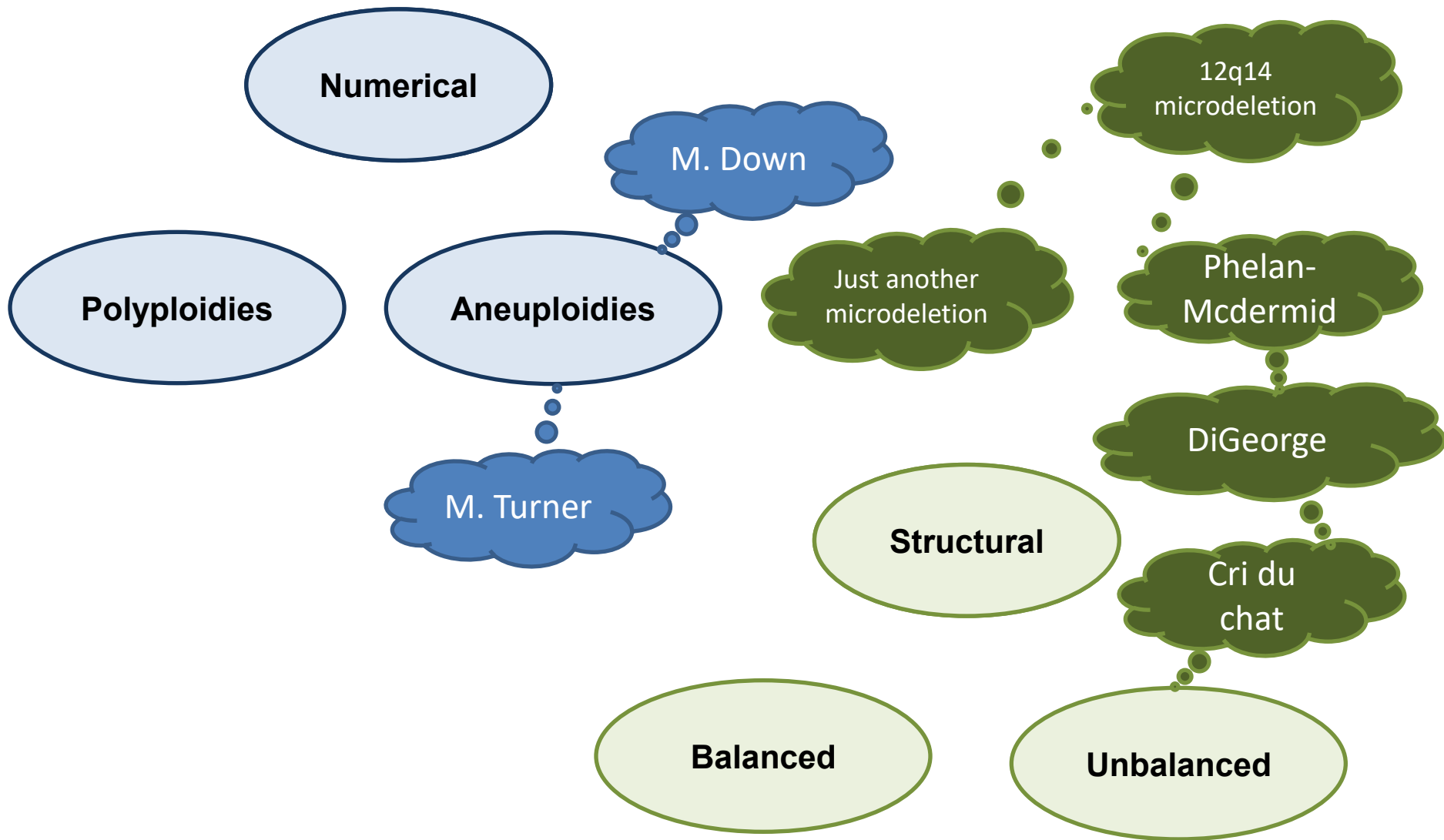
⁶ Department of Gynaecology and Obstetrics, 1st Faculty of Medicine, Charles University and General University Hospital, Prague, Czech Republic



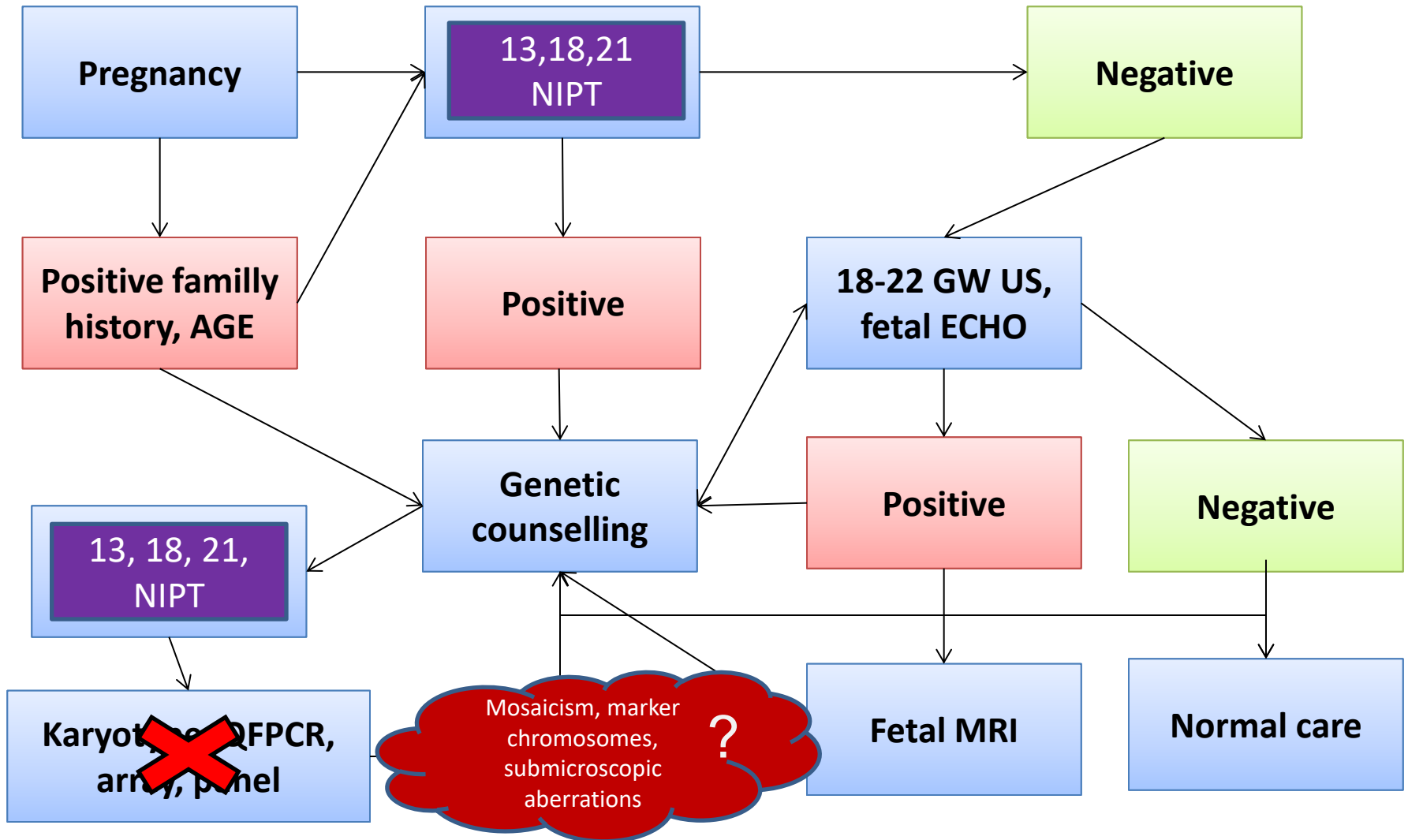
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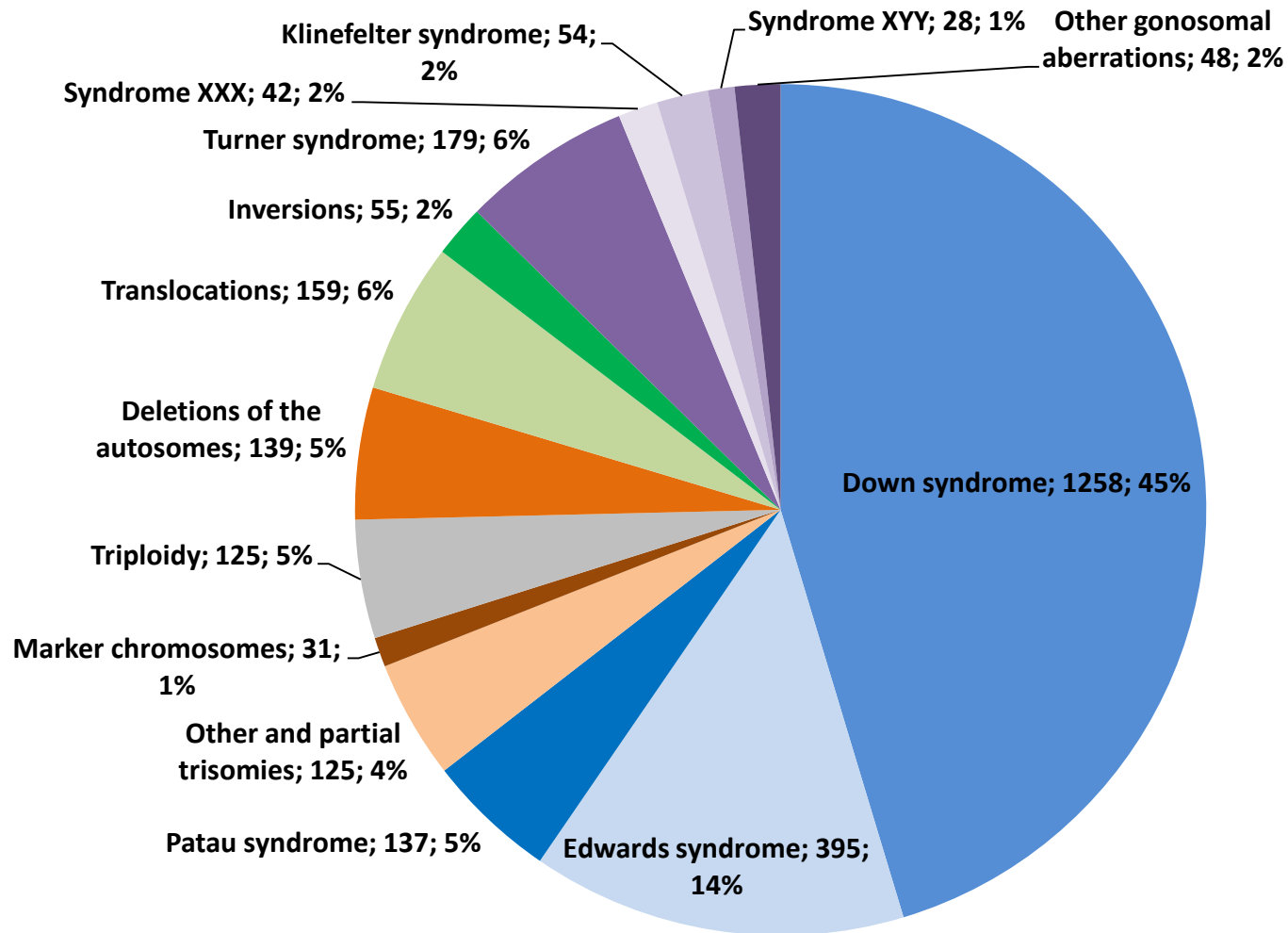
Chromosomal aberrations



Prenatal diagnostics strategy – Czech Republic

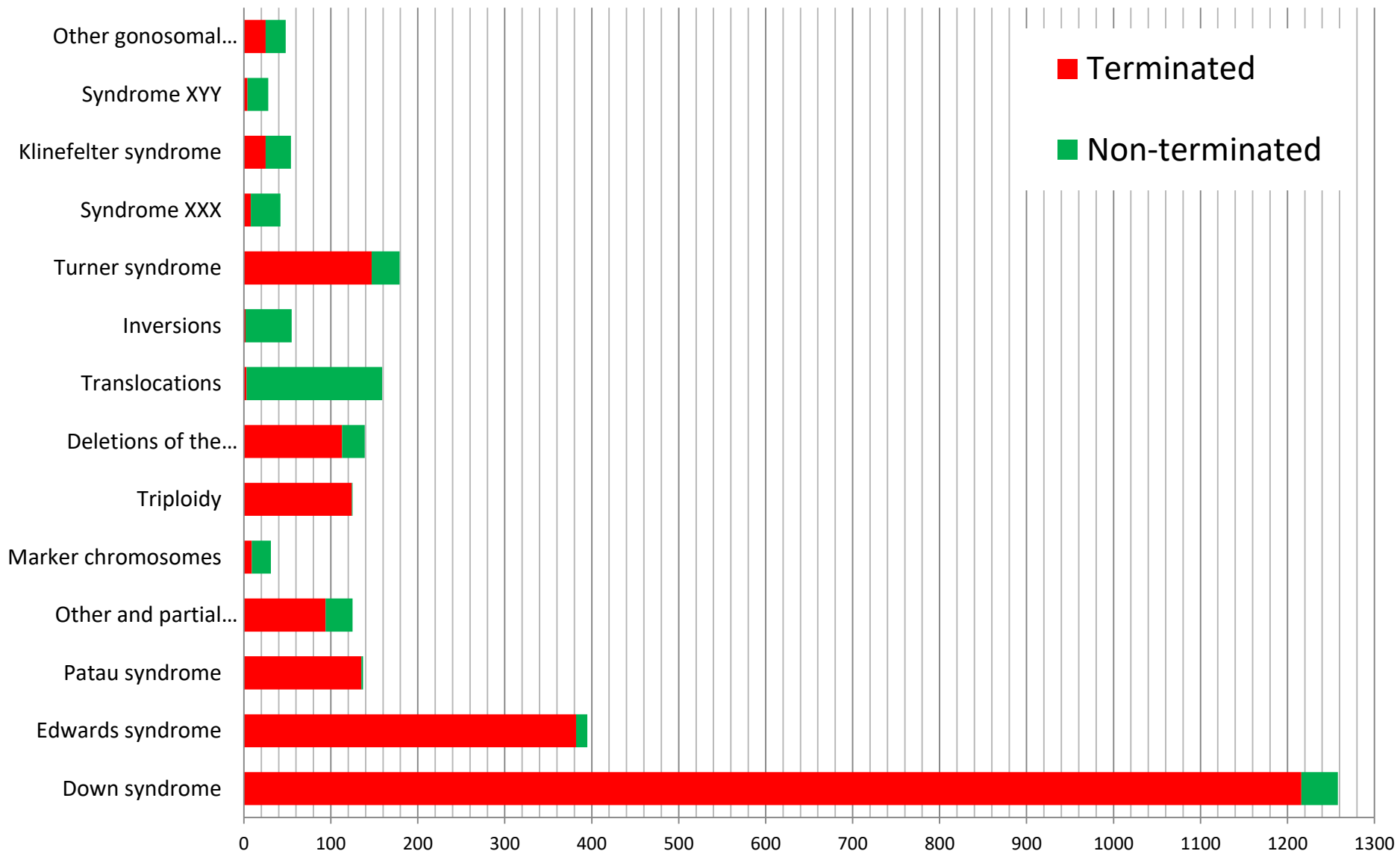


Prenatal diagnostics of chromosomal aberrations in the Czech Republic: 2012-2016; all aberrations

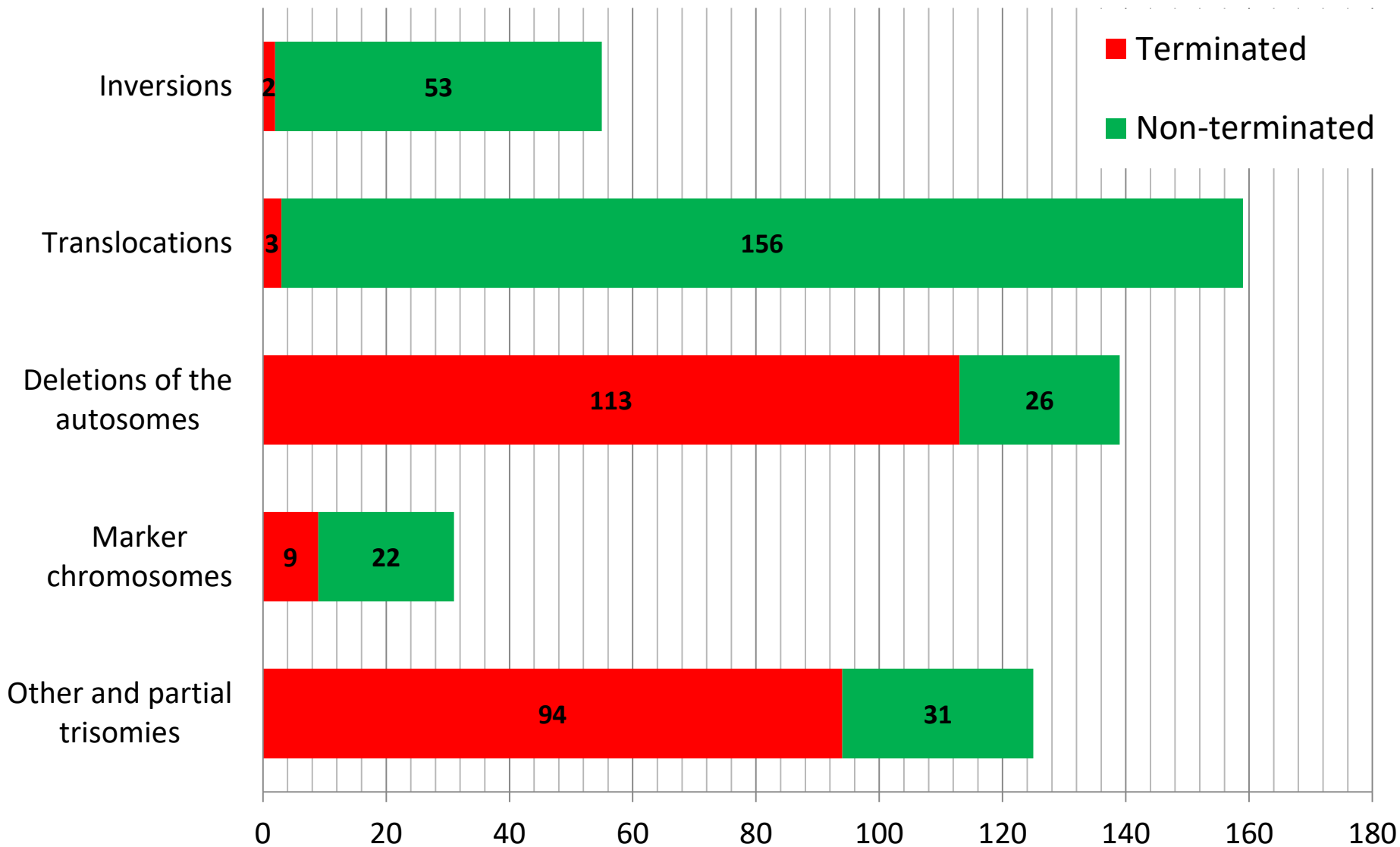




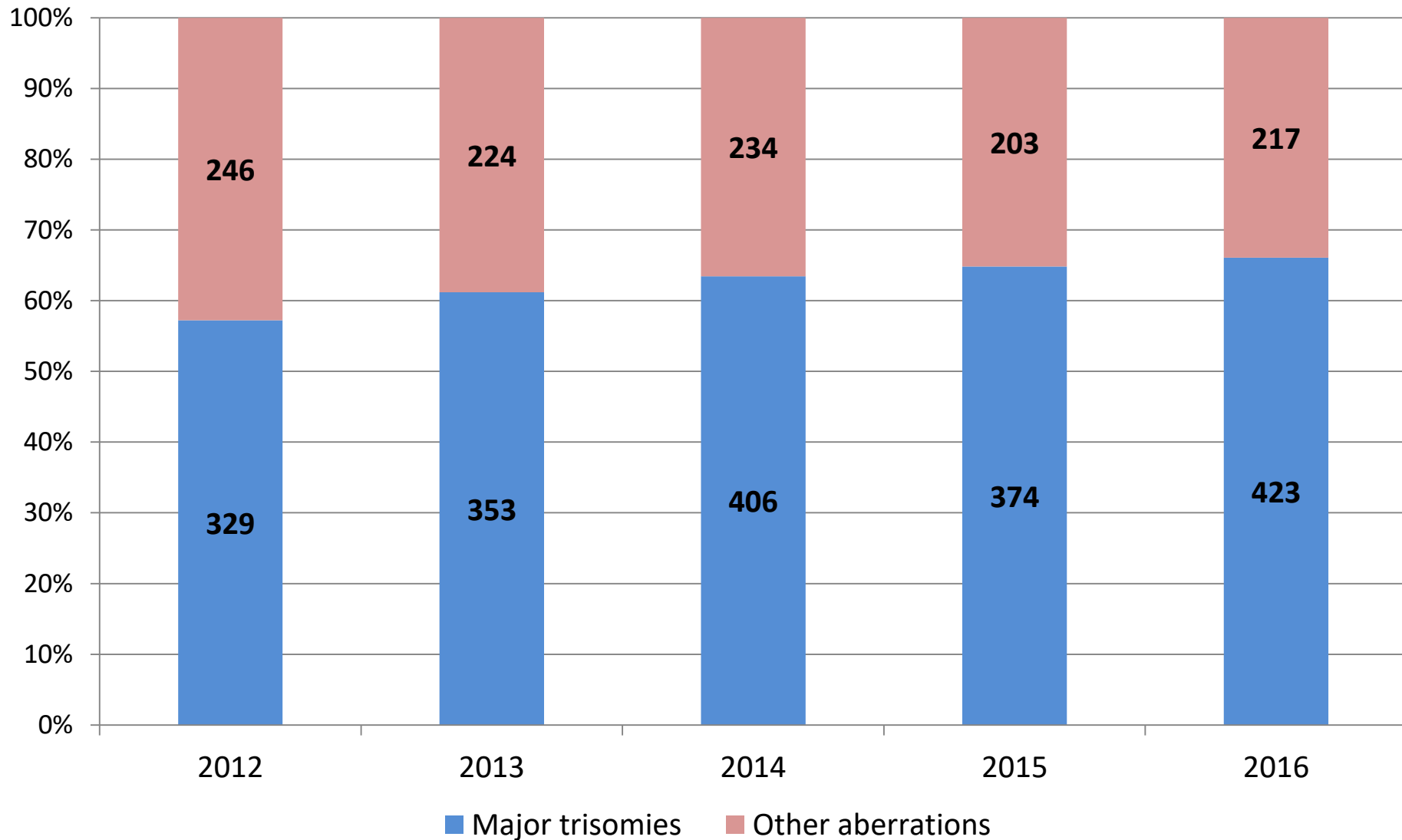
Prenatal diagnostics of chromosomal aberrations in the Czech Republic: 2012-2016; all aberrations - details



Prenatal diagnostics of chromosomal aberrations in the Czech Republic: 2012-2016; structural aberrations - details



Prenatal diagnostics of chromosomal aberrations in the Czech Republic: 2012-2016; Major trisomies vs other aberrations





Prenatal diagnostics of chromosomal aberrations in the Czech Republic: 2012-2016; Major trisomies vs other aberrations



Conclusion

- Nearly 2/3 of all cases of prenatally diagnosed chromosomal aberrations are the trisomies 13, 18 and 21.
- Rest (1/3) of the cases are either the anomalies of the gonosomes, polyploidies or structural chromosomal aberrations.
- Currently, we are not fully able to analyse the group of structural aberrations in detail (microdeletion or microduplication syndrome), this can change in near future.
- The proportion of major trisomies is slowly increasing during last years. The role of NIPT in this trend is tricky to evaluate (other factors: role of increasing maternal age and role of still improving conventional screening methods).

Thank you

Thank you for your attention

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