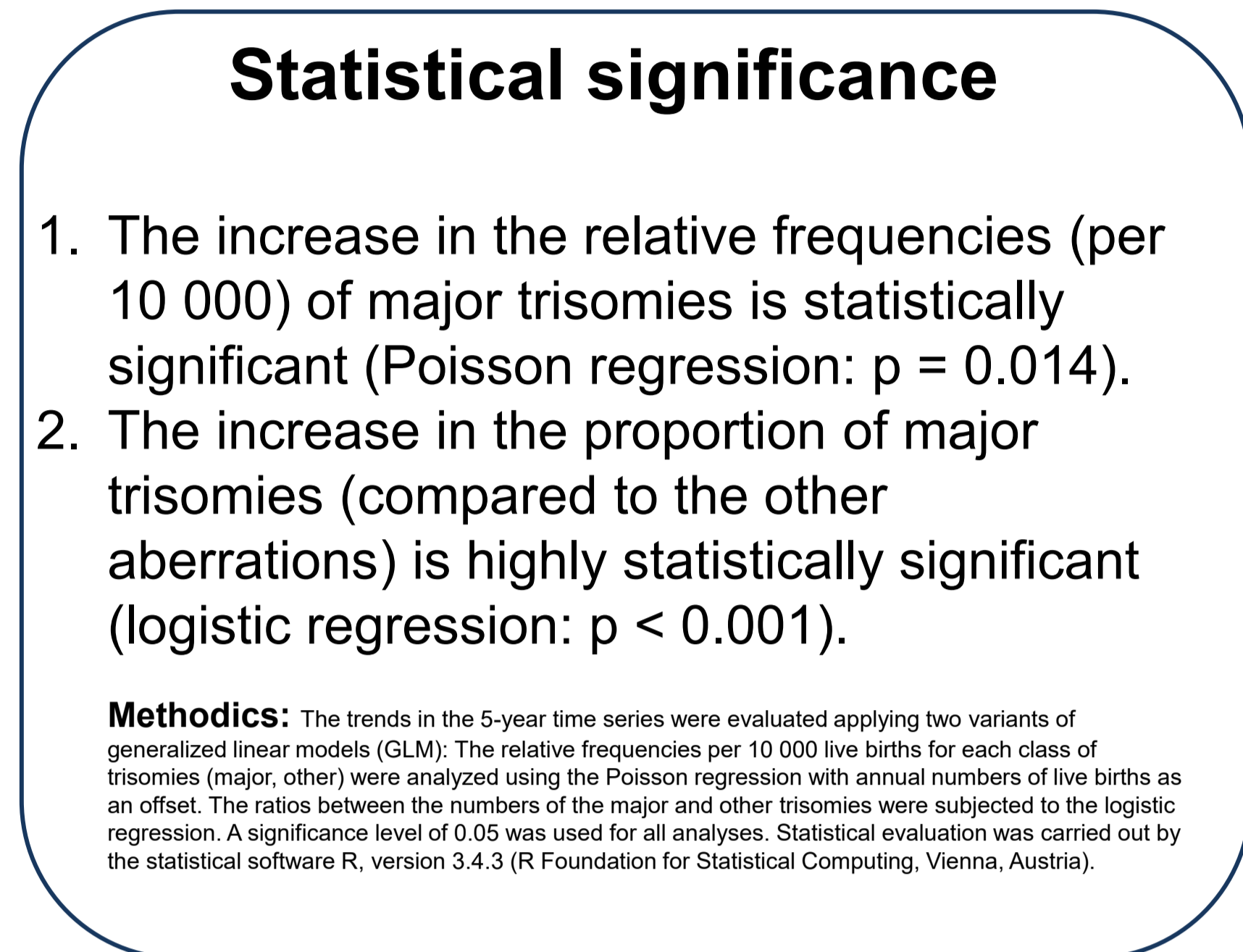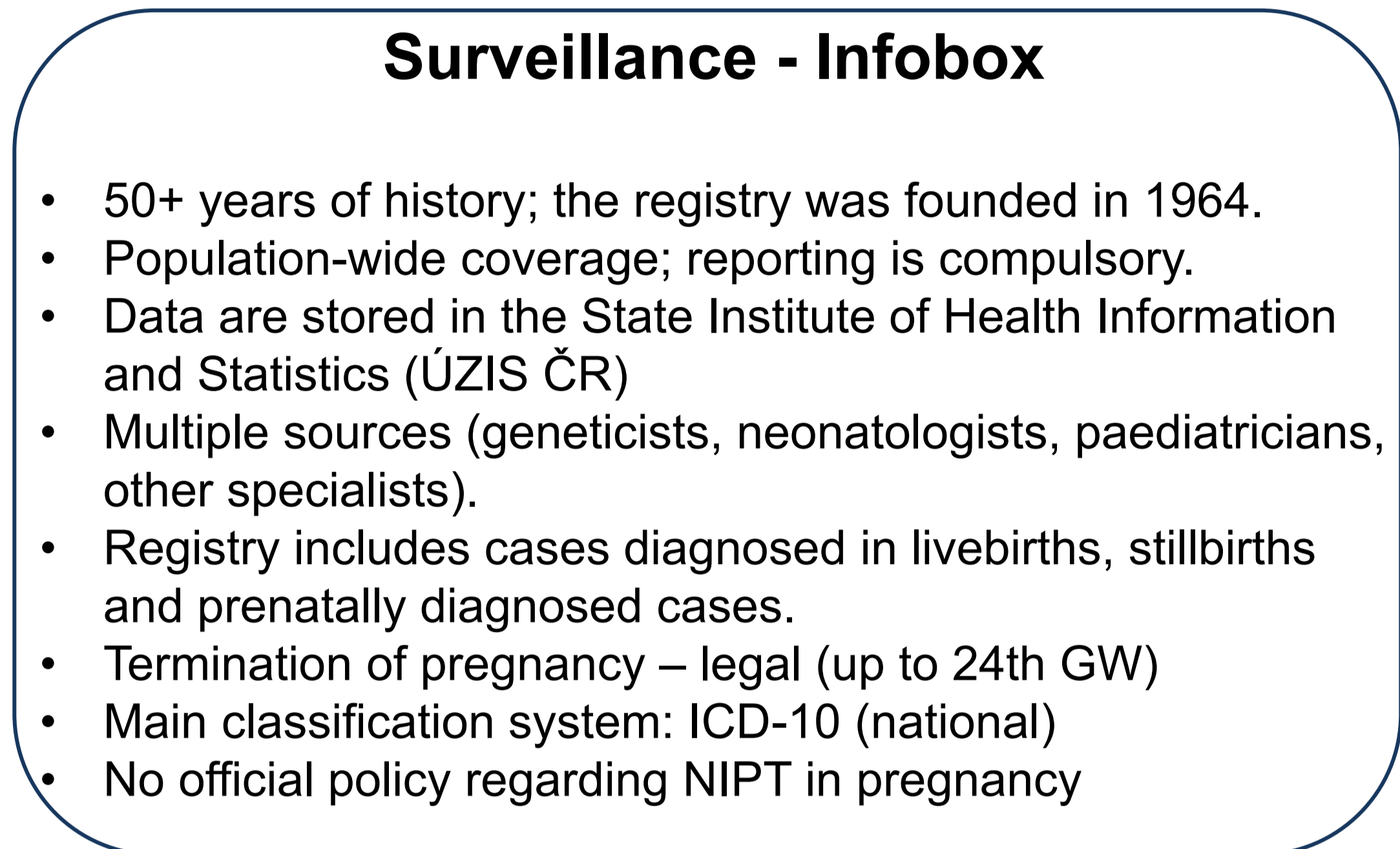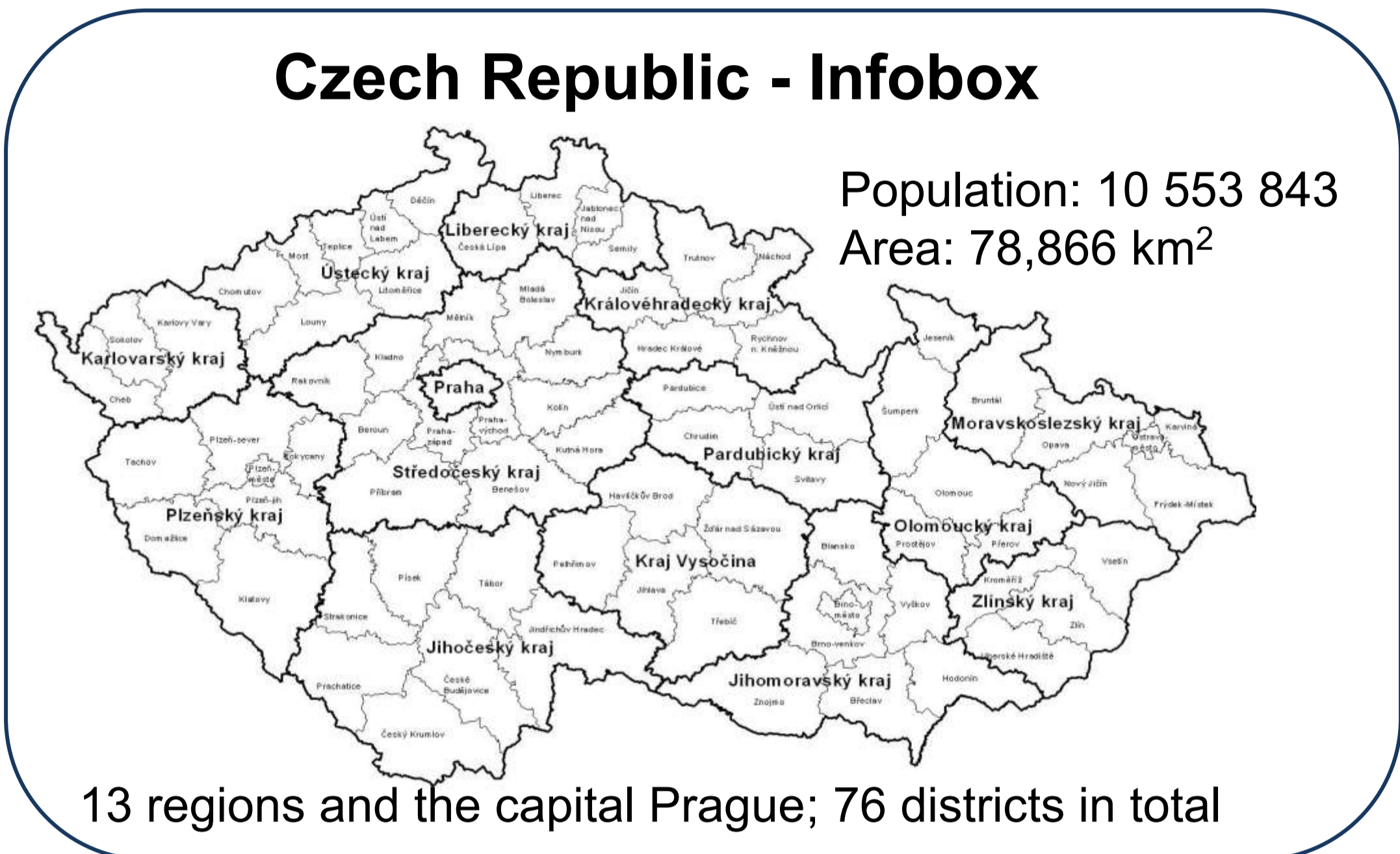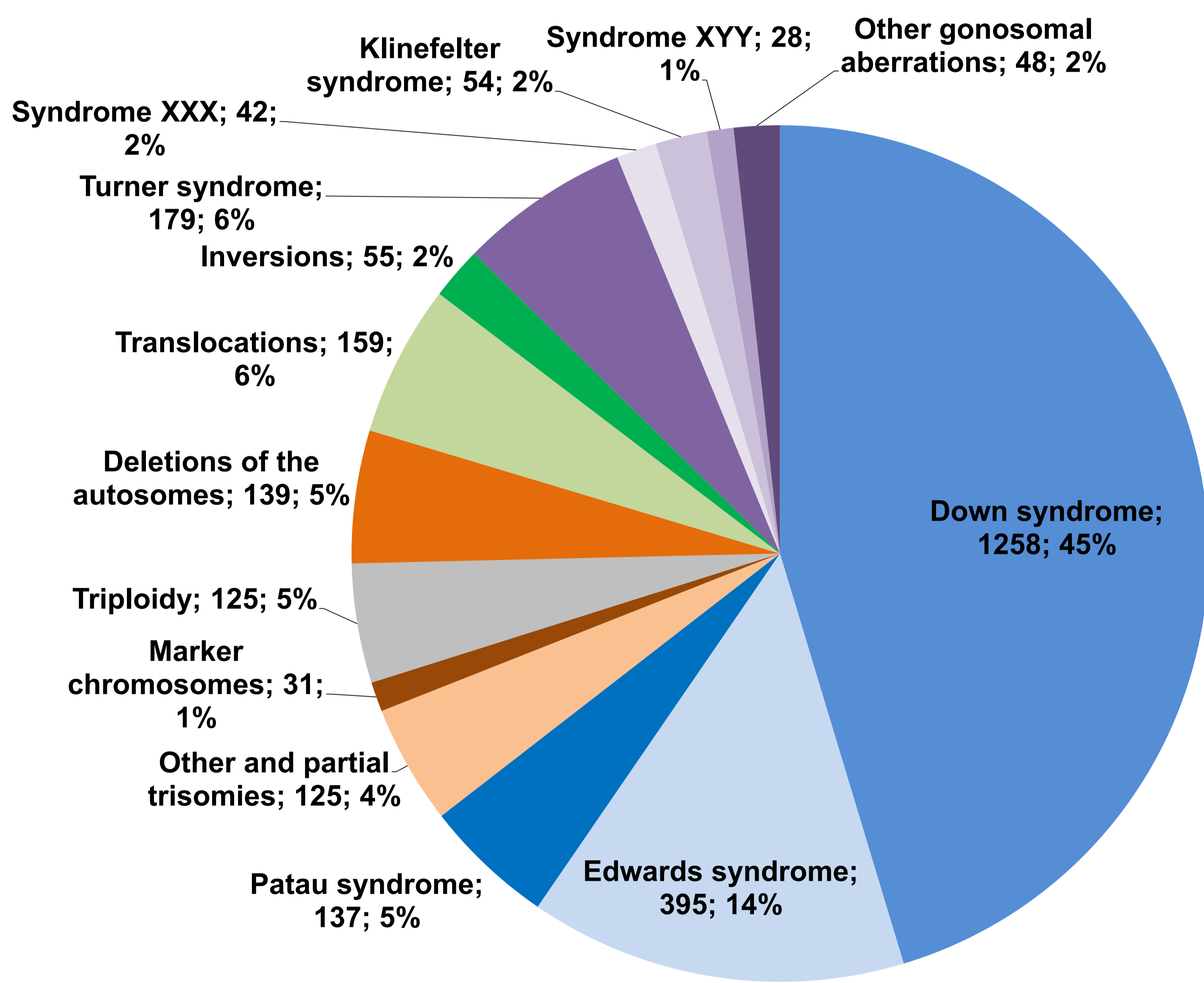# CHANGING FREQUENCIES OF THE MAIN AUTOSOMAL TRISOMIES IN THE NIPT ERA: POPULATION-BASED DATA FROM THE CZECH REPUBLIC.

Antonín Šípek Sr [1,2,3]; Vladimír Gregor [1,2]; Antonín Šípek Jr [1,4]; Jitka Jírová [5]; Jan Klaschka [6,7]; Marek Malý [6,8]

1) Department of Medical Genetics, Thomayer Hospital, Prague, Czech Republic;
2) Department of Medical Genetics, Pronatal Sanatorium, Prague, Czech Republic;
3) Institute of Biology and Medical Genetics, 3rd Faculty of Medicine, Charles University, Prague, Czech Republic;
4) Institute of Medical Biology and Genetics, 1st Faculty of Medicine, Charles University, Prague, Czech Republic;
5) Institute for Health Information and Statistics, Prague, Czech Republic;
6) Institute of Computer Science of the Czech Academy of Sciences, Prague, Czech Republic;
7) Institute of Biophysics and Informatics, 1st Faculty of Medicine, Charles University, Prague, Czech Republic;
8) National Institute of Public Health, Prague, Czech Republic

Czech Republic - Infobox

Population: 10 553 843
Area: 78,866 km^2

13 regions and the capital Prague; 76 districts in total

Surveillance - Infobox

- 50+ years of history; the registry was founded in 1964.
- Population-wide coverage; reporting is compulsory.
- Data are stored in the State Institute of Health Information and Statistics (ÚZIS ČR)
- Multiple sources (geneticists, neonatologists, paediatricians, other specialists).
- Registry includes cases diagnosed in livebirths, stillbirths and prenatally diagnosed cases.
- Termination of pregnancy – legal (up to 24th GW)
- Main classification system: ICD-10 (national)
- No official policy regarding NIPT in pregnancy

Statistical significance

1. The increase in the relative frequencies (per 10 000) of major trisomies is statistically significant (Poisson regression: p = 0.014).
2. The increase in the proportion of major trisomies (compared to the other aberrations) is highly statistically significant (logistic regression: p < 0.001).

Methodics: The trends in the 5-year time series were evaluated applying two variants of generalized linear models (GLM): The relative frequencies per 10 000 live births for each class of trisomies (major, other) were analyzed using the Poisson regression with annual numbers of live births as an offset. The ratios between the numbers of the major and other trisomies were subjected to the logistic regression. A significance level of 0.05 was used for all analyses. Statistical evaluation was carried out by the statistical software R, version 3.4.3 (R Foundation for Statistical Computing, Vienna, Austria).

Prenatal diagnostics of chromosomal aberrations in the Czech Republic 2012-2016: all aberrations (absolute numbers and percentage)

- Klinefelter syndrome; 54; 2%
- Syndrome XYY; 28; 1%
- Other gonosomal aberrations; 48; 2%
- Syndrome XXX; 42; 2%
- Turner syndrome; 179; 6%
- Inversions; 55; 2%
- Translocations; 159; 6%
- Deletions of the autosomes; 139; 5%
- Triploidy; 125; 5%
- Marker chromosomes; 31; 1%
- Other and partial trisomies; 125; 4%
- Patau syndrome; 137; 5%
- Down syndrome; 1258; 45%
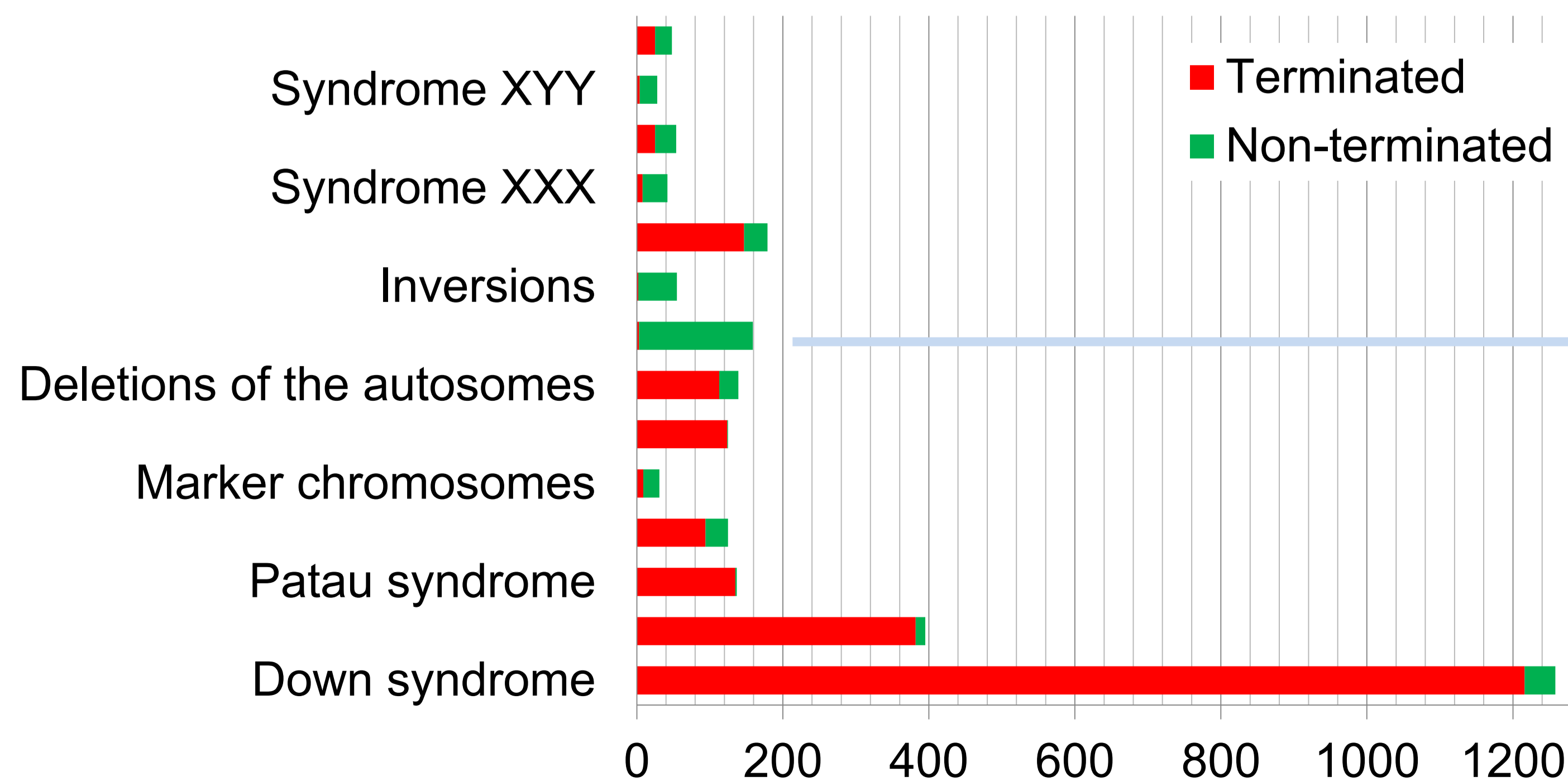- Edwards syndrome; 395; 14%

Prenatal diagnostics of chromosomal aberrations in the Czech Republic: 2012-2016; Major trisomies vs other aberrations (absolute numbers)
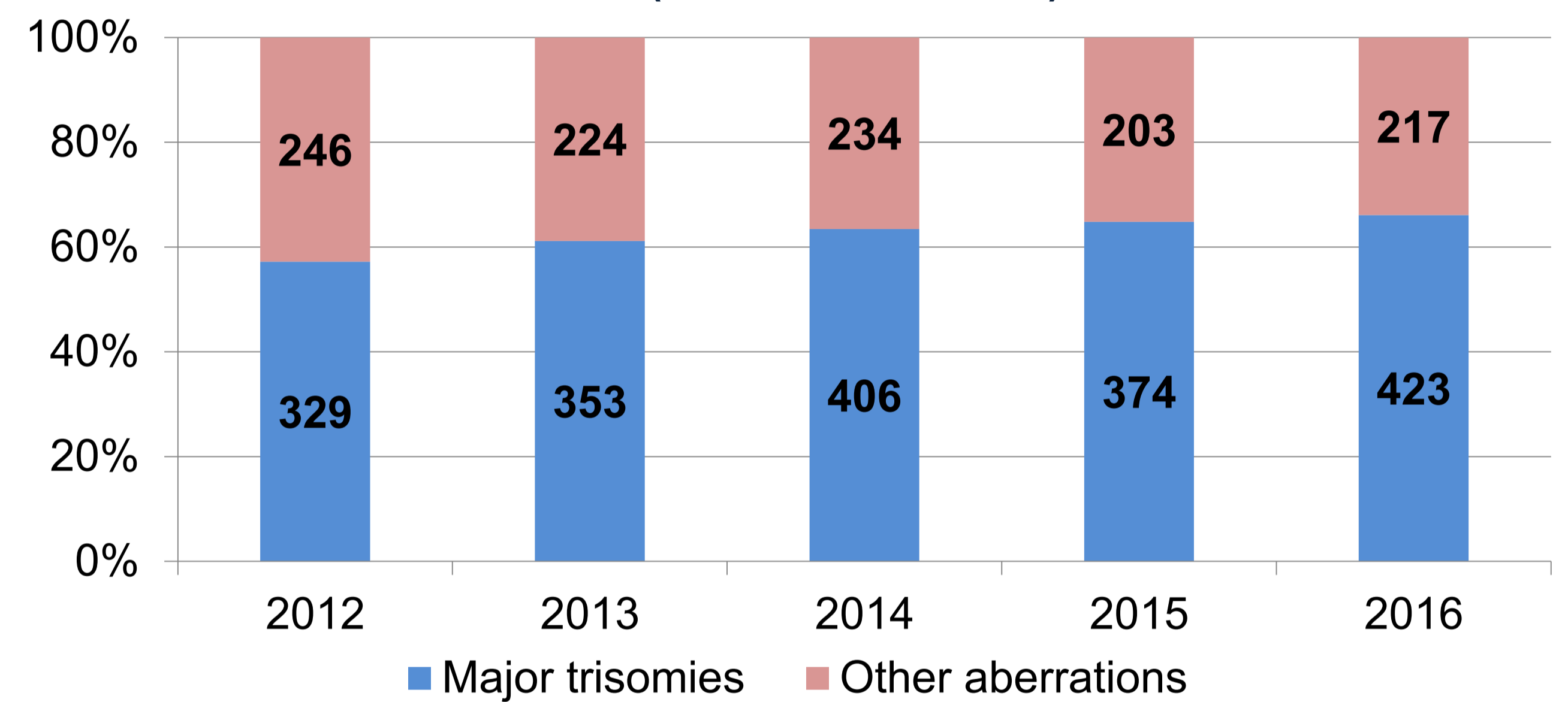
Prenatal diagnostics of chromosomal aberrations in the Czech Republic: 2012-2016; Major trisomies vs other aberrations (relative numbers per 10.000 live births)
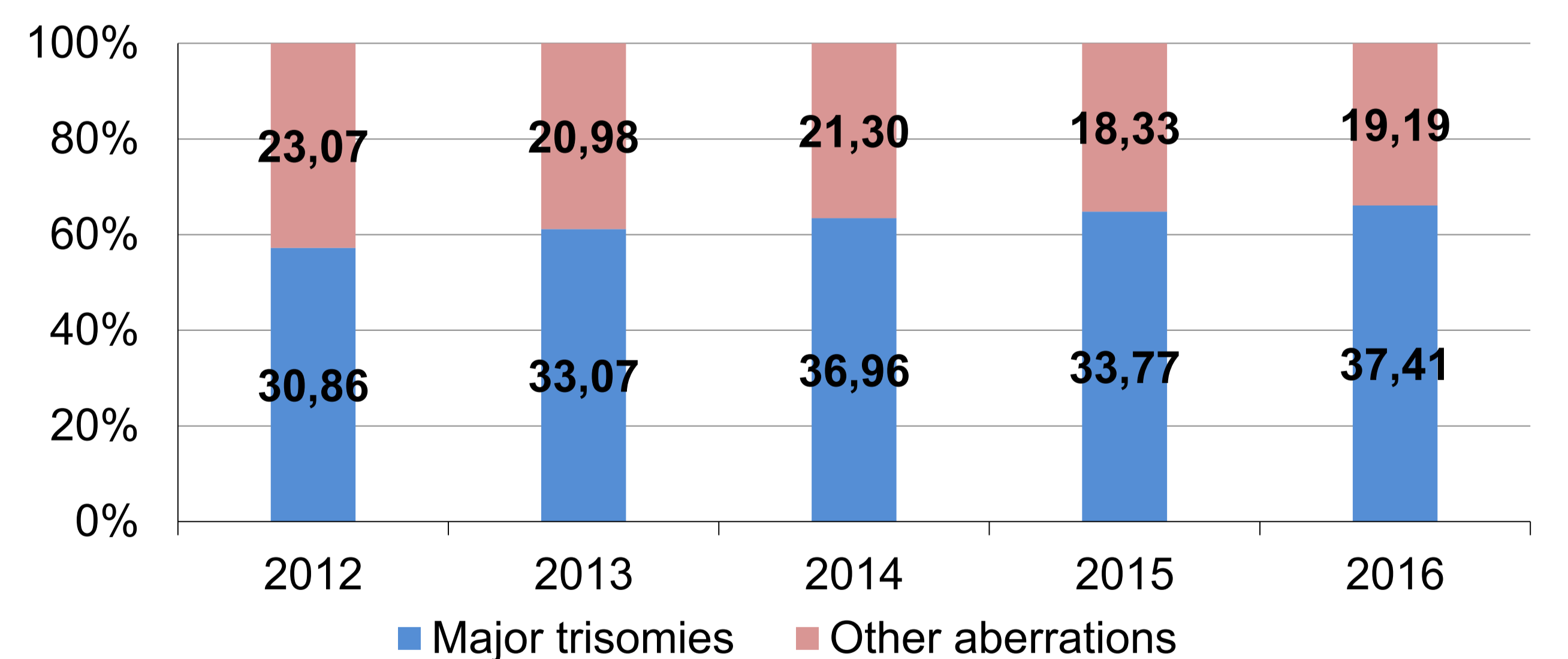
Prenatal diagnostics of chromosomal aberrations in the Czech Republic: 2012-2016; all aberrations – absolute numbers

Prenatal diagnostics of chromosomal aberrations in the Czech Republic: 2012-2016; structural aberrations – details (absolute numbers)

http://www.vrozene-vady.cz/

Contact: registrvvv@seznam.cz