

# Trends in the prenatal diagnostics of the Down syndrome in the Czech Republic: Can we find the association with the maternal age?

**Antonín Šípek Jr<sup>1,2</sup>, Vladimír Gregor<sup>2,3</sup> Jiří Horáček<sup>2,4</sup>  
Ondřej Vencálek<sup>5</sup>, and Antonín Šípek<sup>2,3,6</sup>**

1. *Institute of Biology and Medical Genetics, First Faculty of Medicine, Charles University, Prague, Czech Republic,*
2. *Department of Medical Genetics, Thomayer Hospital, Prague, Czech Republic,*
3. *Department of Medical Genetics, Pronatal Sanatorium, Prague, Czech Republic,*
4. *Gennet, Ltd., Prague, Czech Republic,*
5. *Department of Mathematical Analysis and Applications of Mathematics, Faculty of Science, Palacky University, Olomouc, Czech Republic,*
6. *Department of General Biology and Genetics, 3rd Faculty of Medicine, Charles University, Prague, Czech Republic.*



**<http://www.vrozene-vady.cz/>**

# Czech Republic - Present State

**Registry:** Population based (whole area of the Czech Republic, 100.000 annual births).

**Law:** The registration is compulsory, required by the Internal Law of Ministry of Health (nr. 14/2001). The database is run by the Institute of Health Information and Statistics of the Czech Republic. Additional data on prenatally diagnosed cases are collected thanks to the country-wide cooperation.

**Cases:** All cases in TOPs, live births and stillbirths are reported.

**Coding:** ICD-10 (international), no verbal description.

**Sources:** Multiple sources, including departments of medical genetics, genetic laboratories, pediatric and neonatology departments, delivery units, ultrasound diagnostics departments etc.

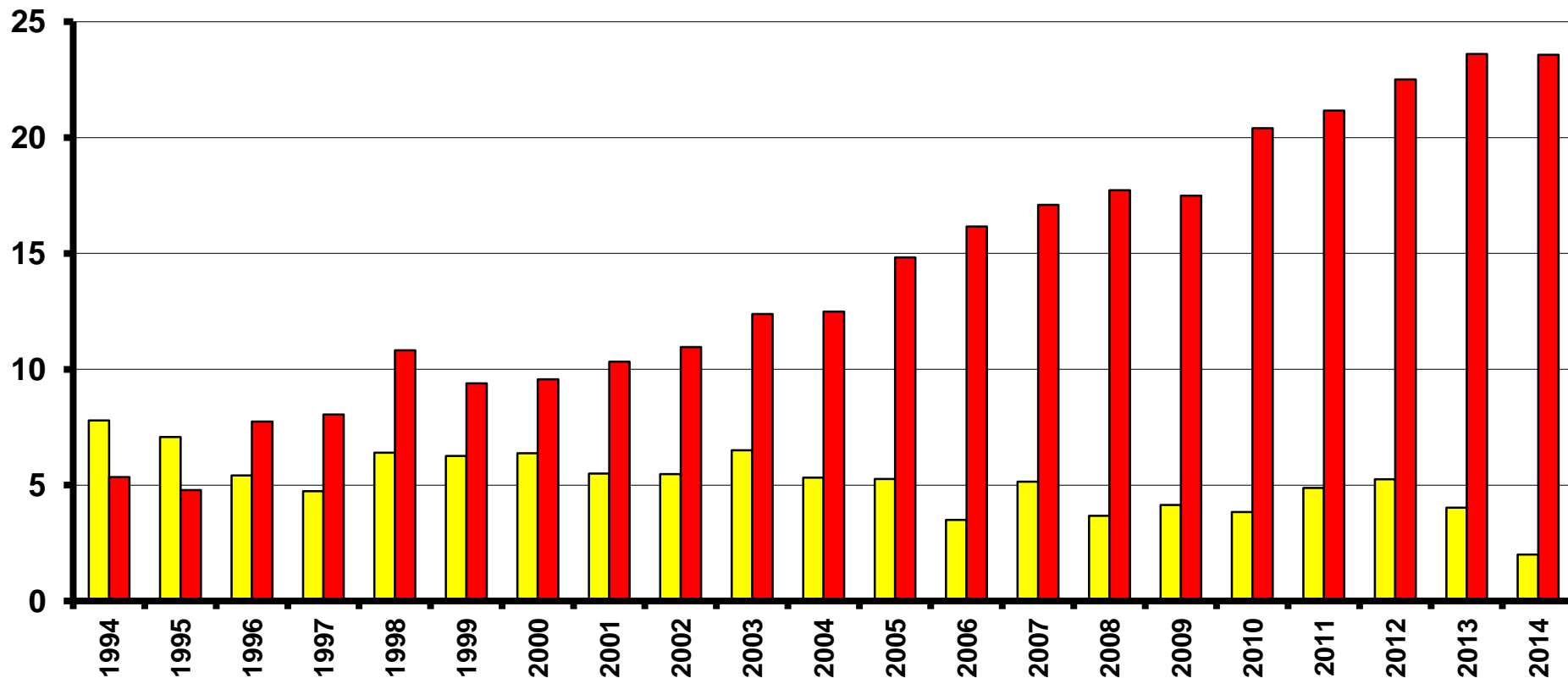
**Termination of Pregnancy:** Legal, up to the 24th week of gestation.

# Down syndrome

Per 10.000 live births

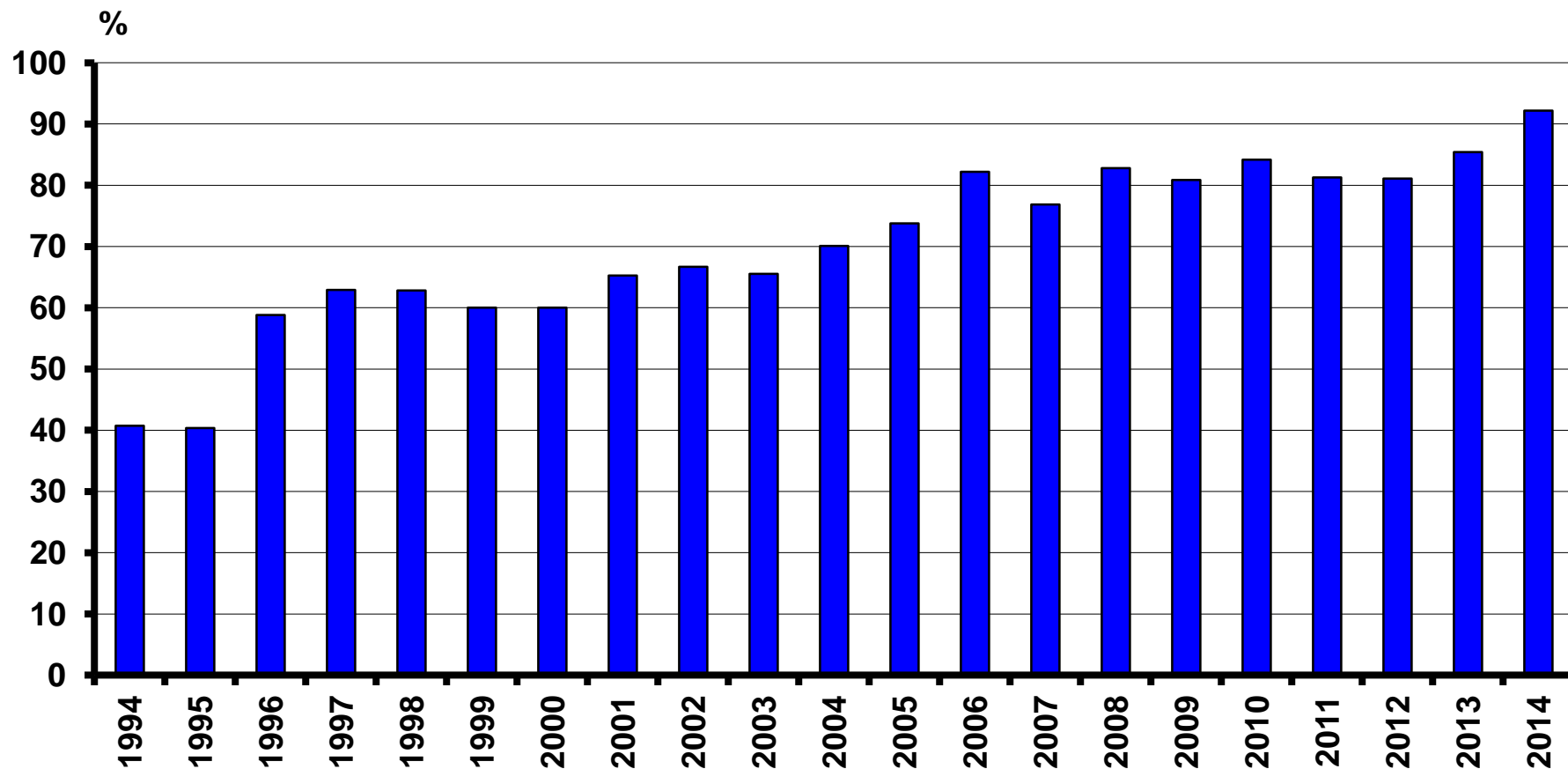
■ Births

■ Prenatal diagnosis



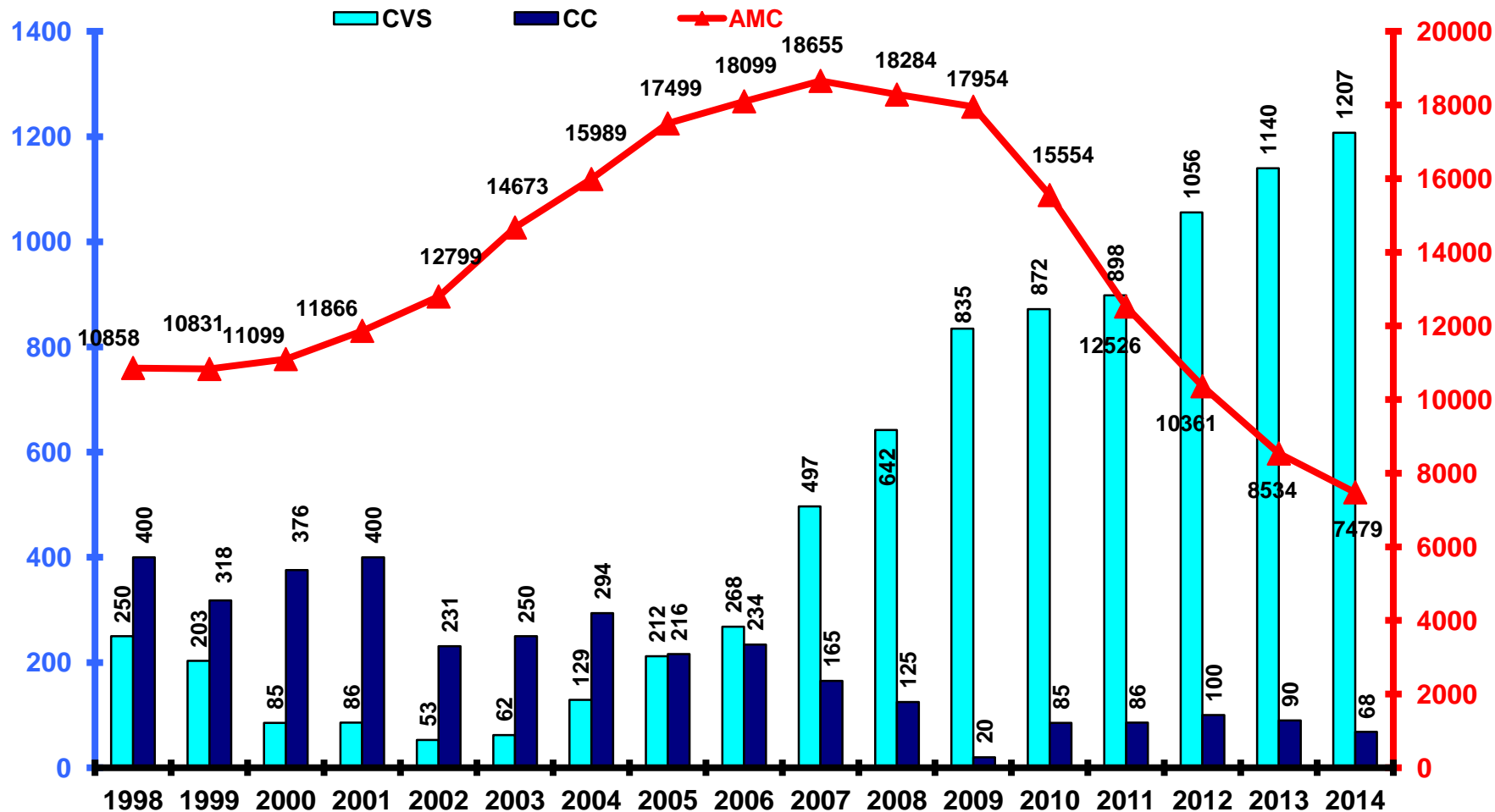
***Down syndrome – All cases in live births and TOPFAs***

# Down syndrome

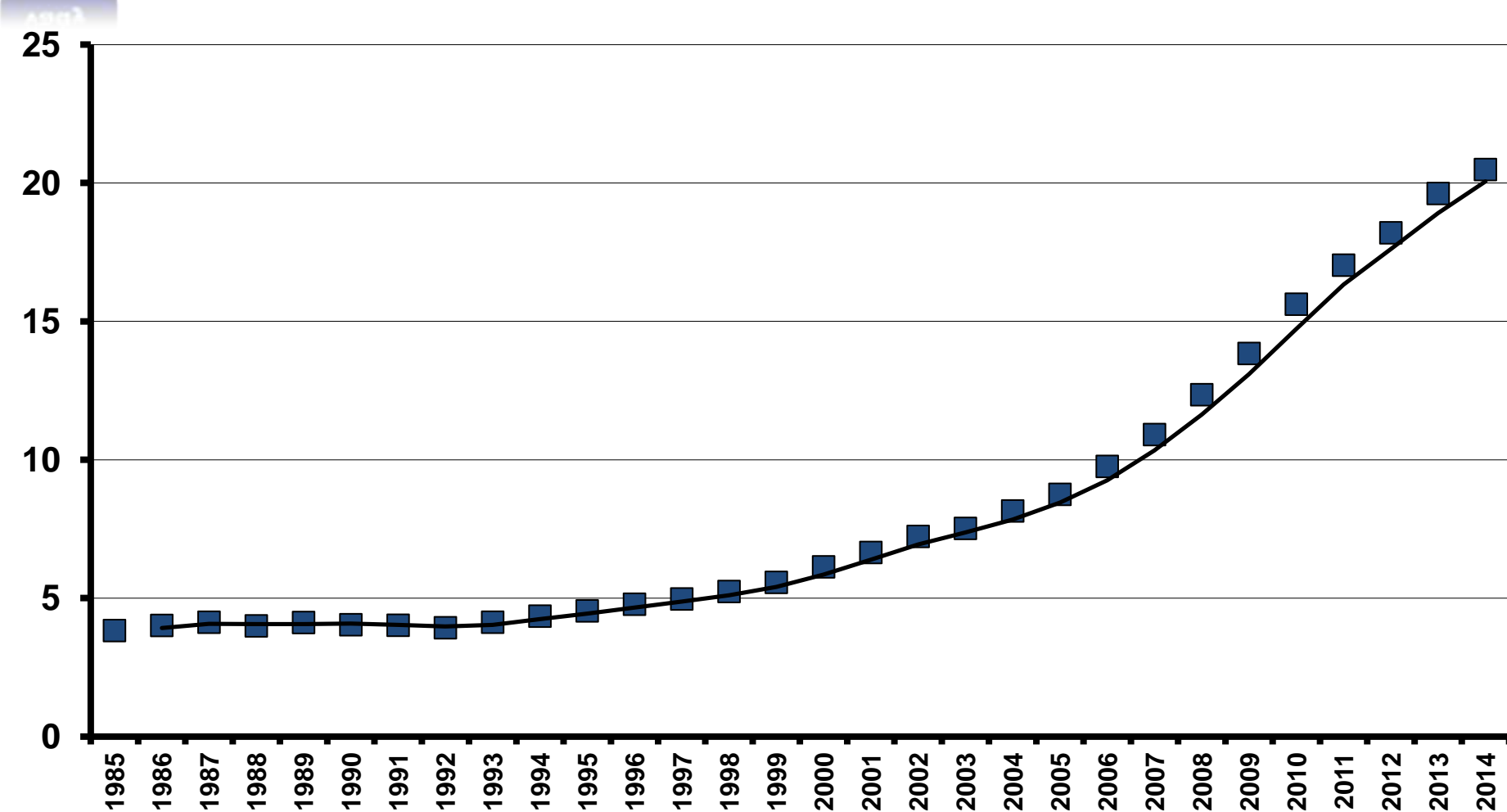


*Down syndrome – relative number of TOPFAs (%)*

# Invasive prenatal diagnosis

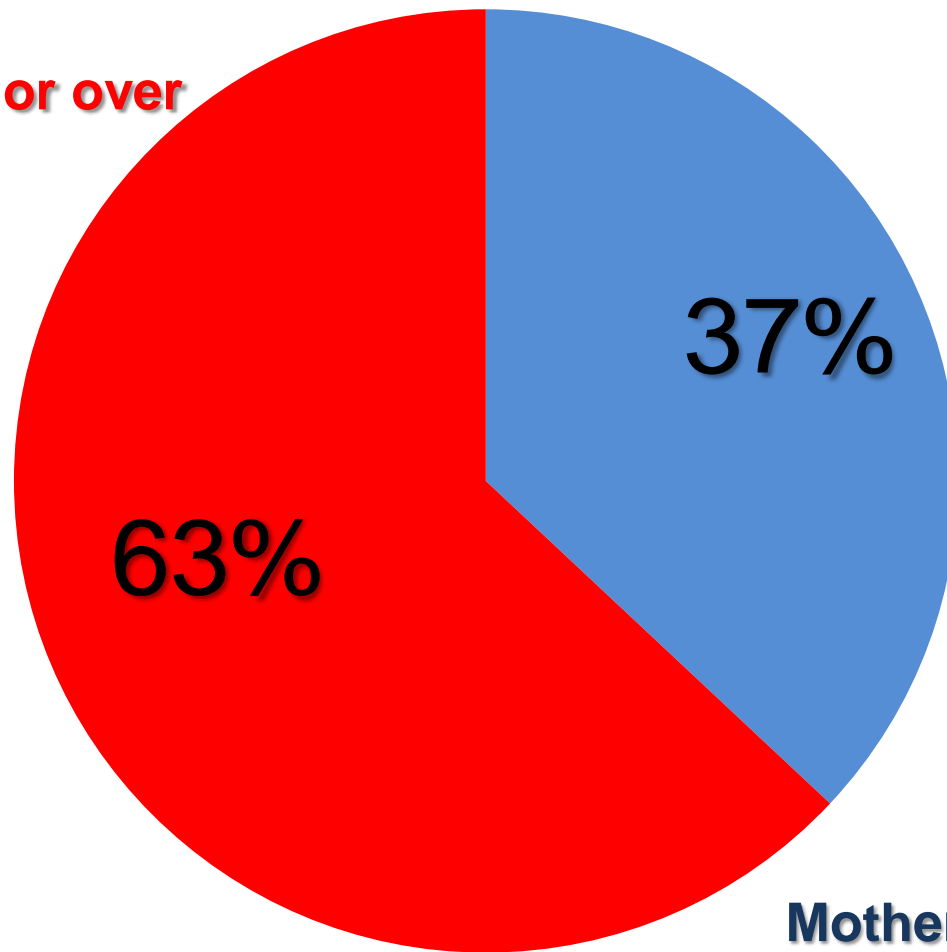


# Maternal age – Time trends



*Proportion of mothers aged 35 years or over*

# Down syndrome (2014)

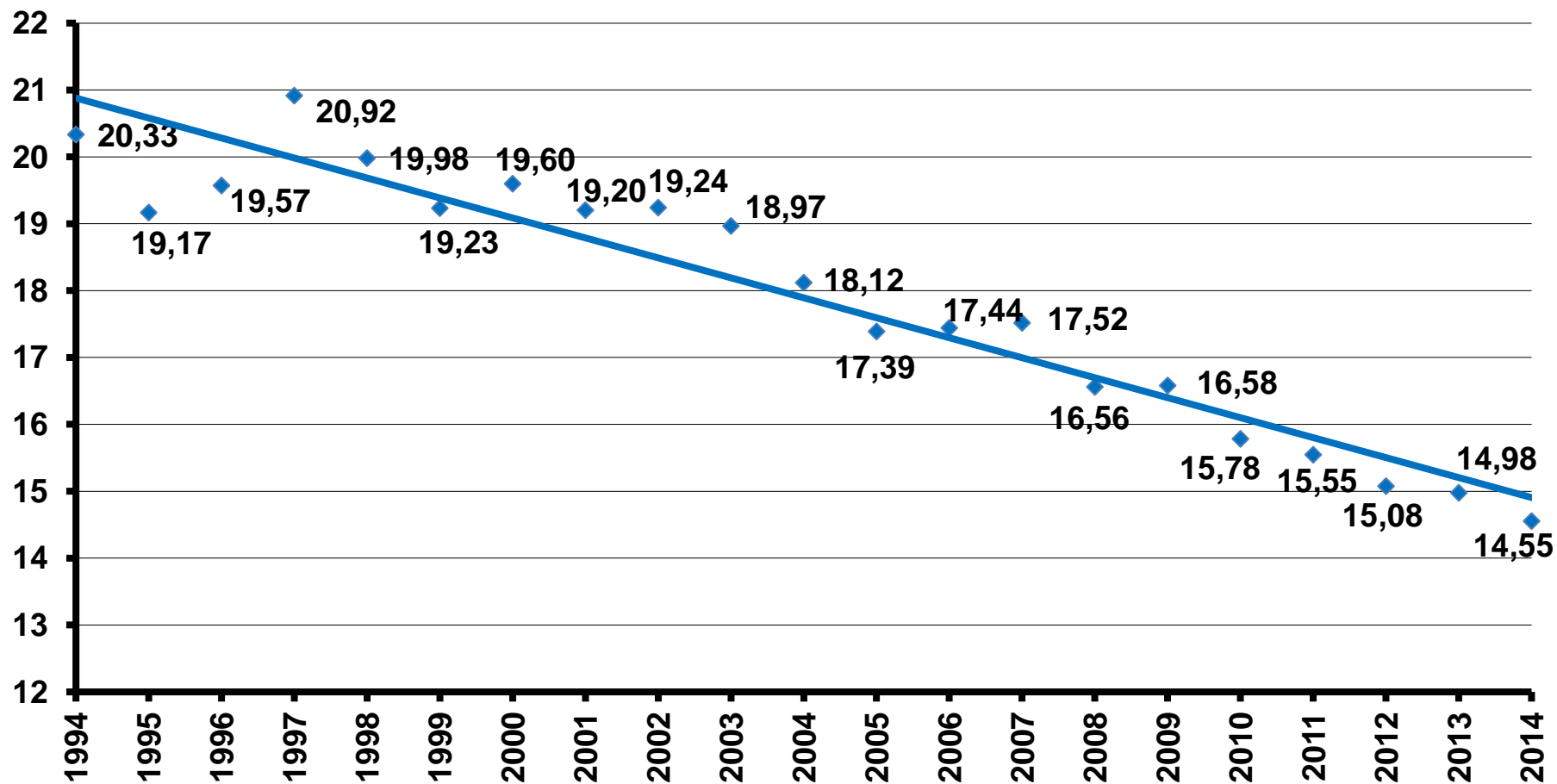


**Mothers aged 35 or over**

**Mothers under 35 years**

*Down syndrome cases (all) according to the maternal age*

# Time of diagnosis

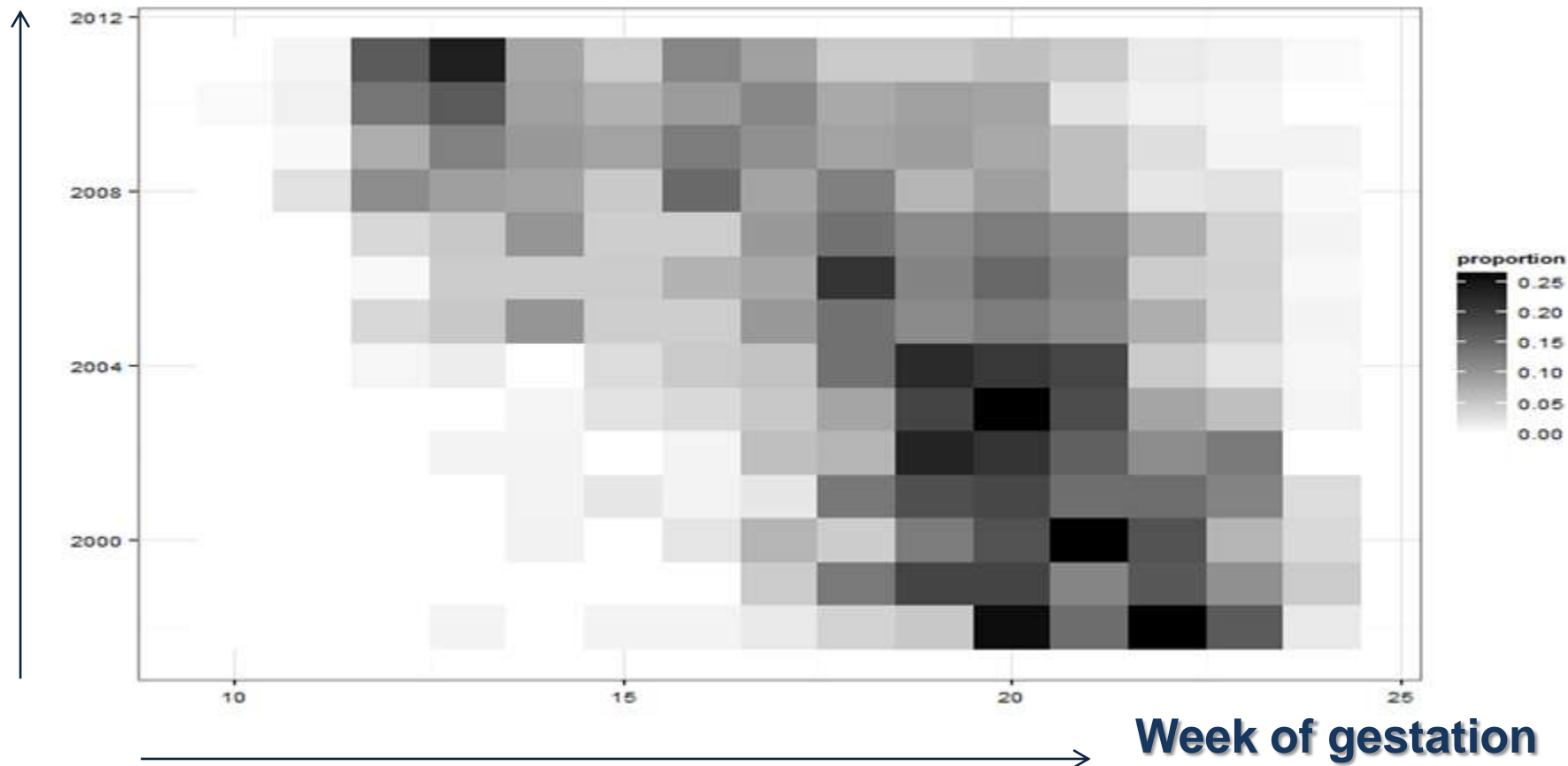


*Average week of gestation at diagnosis of Down syndrome*



# Down syndrome - Diagnostics

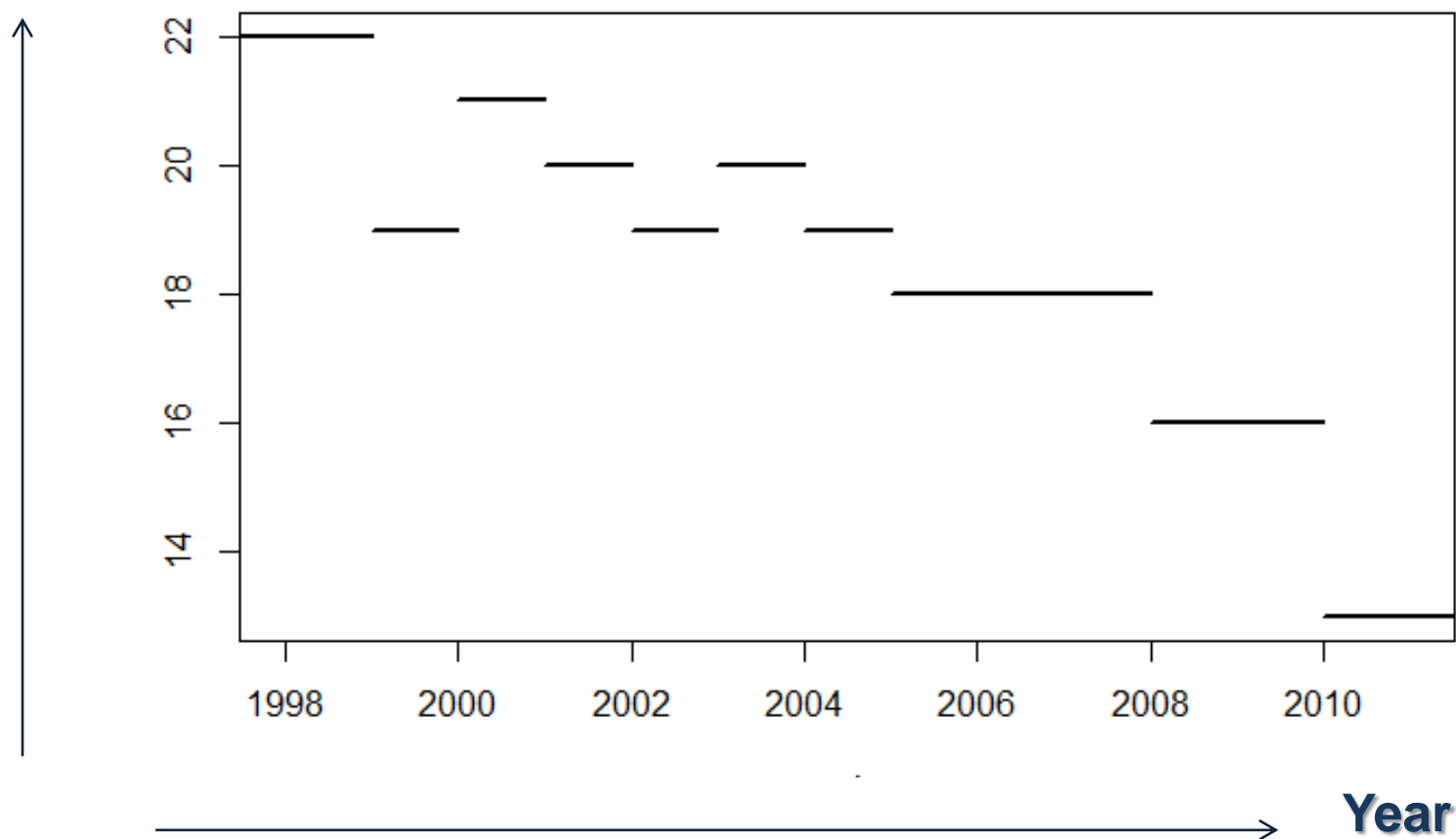
Year



*Time trends – most common week of Down syndrome diagnosis*

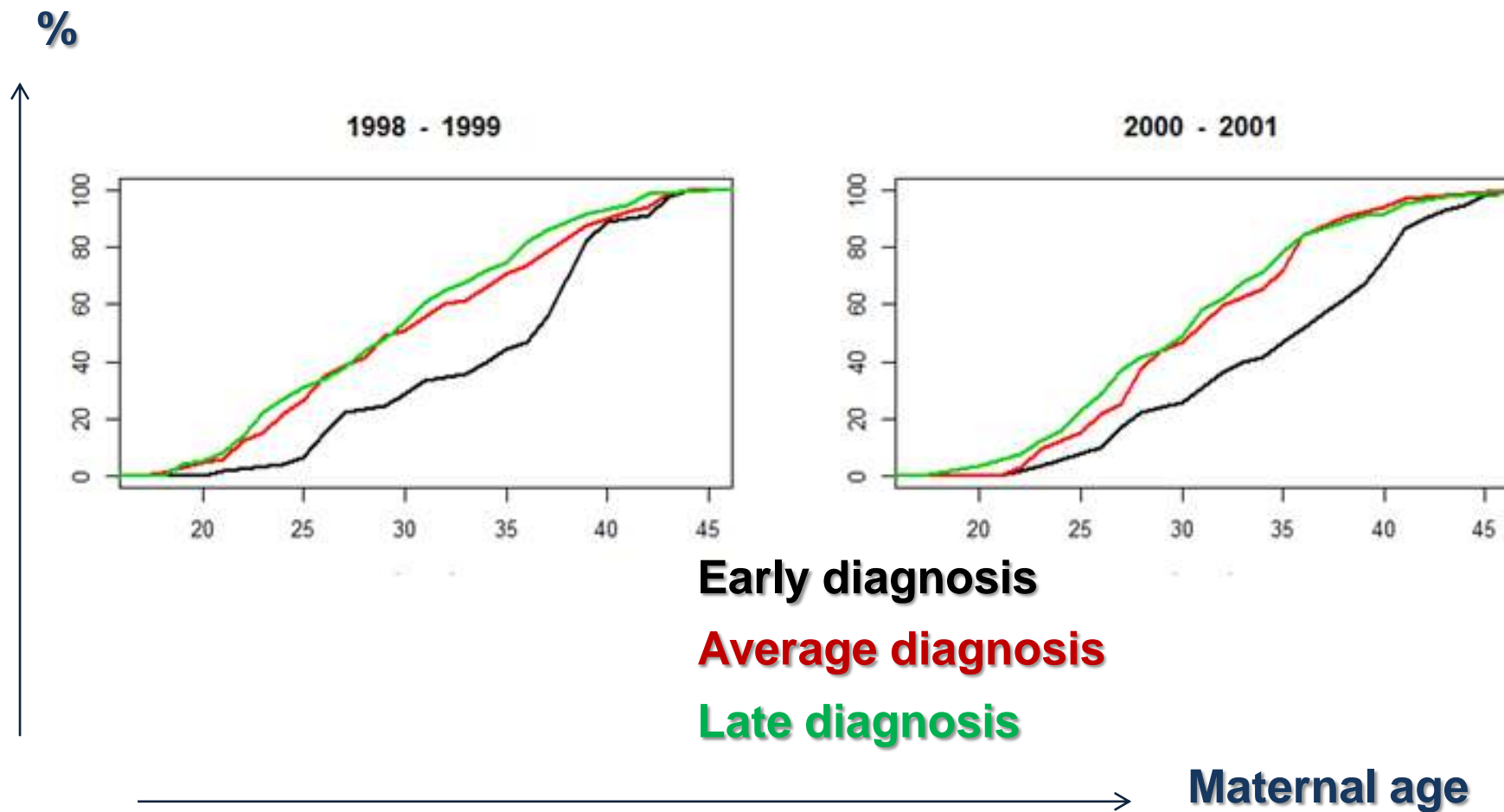
# Down syndrome - Diagnostics

Week of gestation



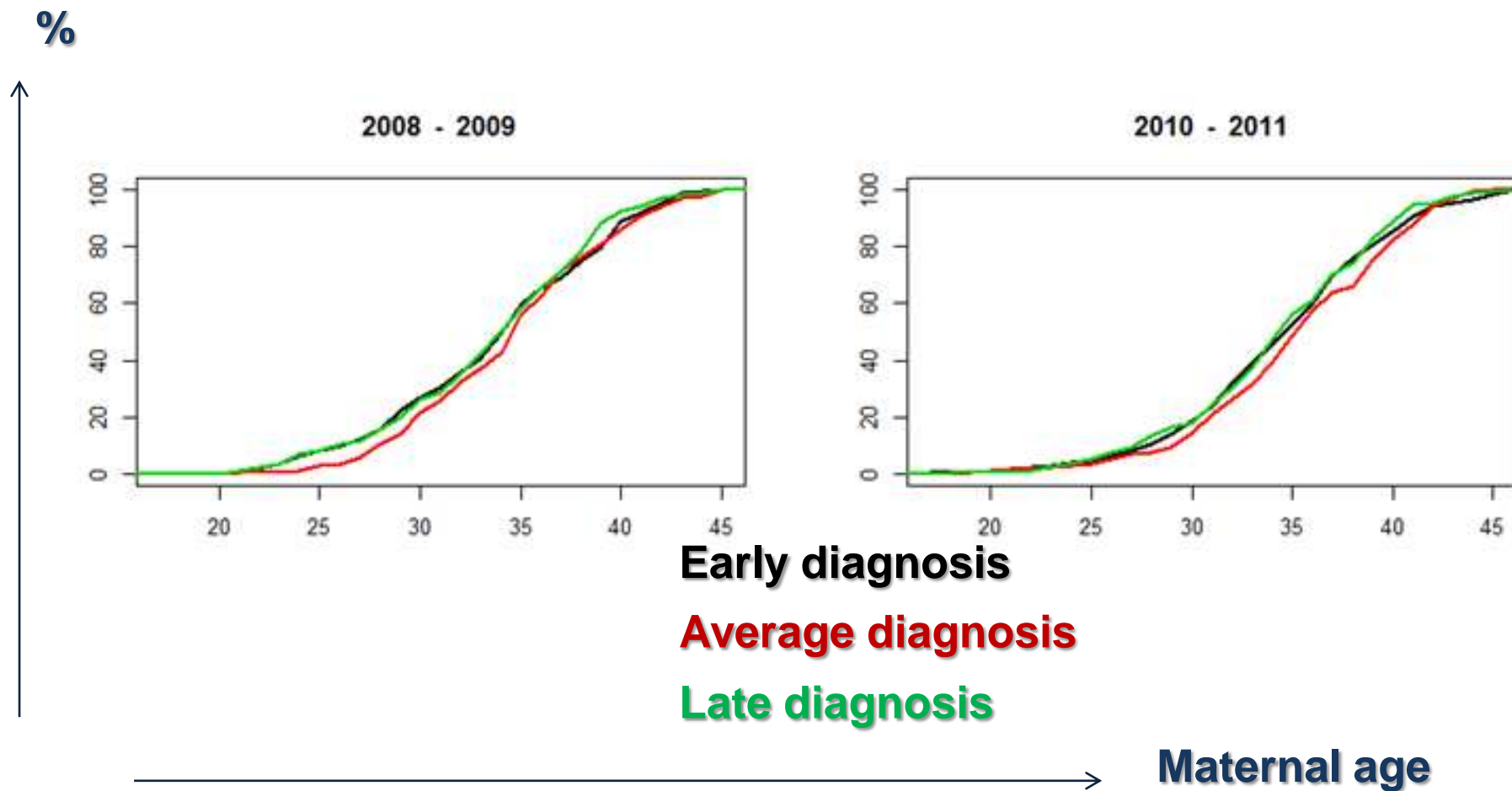
*Time trends – most common week of Down syndrome diagnosis*

# Down syndrome – 1998-2001



*Time trends – Early vs. Late diagnosis regarding the maternal age*

# Down syndrome – 2008-2011



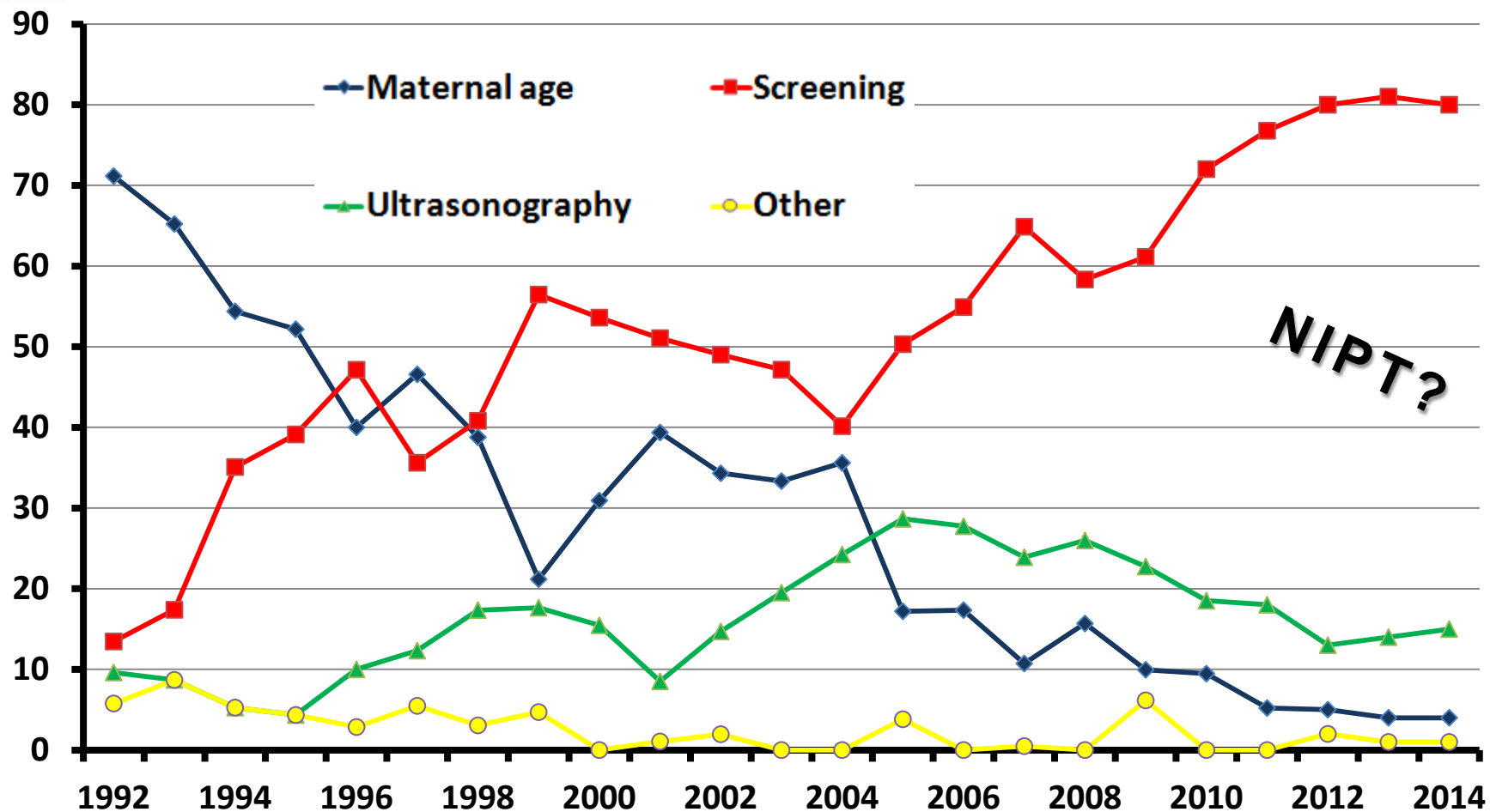
*Time trends – Early vs. Late diagnosis regarding the maternal age*

# What is the significance?

Years	EvSA	AVSL	EvSL
1998–1999	<b>0.039</b>	0.985	<b>0.002</b>
2000–2001	<b>0.025</b>	0.902	<b>0.001</b>
2002–2003	0.083	0.586	<b>0.038</b>
2004–2005	0.827	1.000	0.996
2006–2007	0.103	0.809	0.725
2008–2009	0.792	0.766	0.605
2010–2011	0.412	0.736	1.000

*p values computed using nonparametric Kolmogorov-Smirnov test*

# The role of screening



NIPT?

*Changing role of screening programs in prenatal diagnosis*

# Conclusions

1. *The indication criteria for the invasive prenatal diagnosis are changing.*
2. *The average maternal age is notably increasing.*
3. *The early prenatal diagnosis (during the first trimester of gravidity) is becoming much more important.*
4. *There are no significant changes in the timing of the diagnosis among younger and older mothers.*
5. *However, this trend was visible some 10 years ago.*
6. *May we see it in the future again?*

# Acknowledgements

***We would like to express our thanks to all helpful professionals (geneticists, neonatologists, pediatricians, gynecologists and many others) who are involved in the registration process and provide the data, necessary for the surveillance of the congenital anomalies in the Czech Republic***





*Thank you for your attention!*



antonin.sipek@lf1.cuni.cz

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