

Czech Register of Chromosomal Aberrations

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

1964 50 years anniversary 2014

National Registry of Congenital Anomalies of the Czech Republic

Congenital anomalies surveillance programs in the world - Timeline

- 1962 – Hungary 
- 1963 – Finland 
- **1964 – Czechoslovakia** 
- 1966 – Canada (British Columbia since r. 1952), Israel  
- 1967 – Georgia (USA), Norway, South Africa   
- 1968 – Denmark 
- 1970 – Northern Ireland (Belfast area since 1957) 

Czech Republic - History I

National Registry of Congenital Anomalies of the Czech Republic (NRCA)

- Unofficial monitoring in former Czechoslovakia started in **1961**
- Official monitoring started on 1st of January **1964**
- **First stage (1964 – 1974)** – only **36** selected diagnoses of congenital anomalies (CA) were registered
- **Second stage (1975 – 1993)** – **60** diagnoses of CA registered
- **Present time: (1994 – now)** – **all cases** in terminations of pregnancies (TOPs), stillbirths and live births are registered (age limit for reporting = **15 years**)
- **Near future:** no age limit, additional diagnoses (rare diseases), electronic registration, OMIM and Orphanet codes etc.

Czech Republic - History II

1971 – Cultivation of amniocytes

1971 – First prenatally diagnosed M. Down

1977 – Placentocentesis

1978 – Fetoscopy

1980 – USG Diagnostics

1983 – CVS

1985 – The data on prenatal diagnosis became a part of registration

1987 – Cordocentesis

1988 – Early amniocentesis

1990 – Biochemical screening

2000 – PGD

2012 – NIPST

Czech Republic - Present State

Registry: population based (whole area of the Czech Republic)

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 (from genetic reasons)

International Cooperation

ICBDSR

Established in 1974

World-Wide organization

**CZE: Full membership
(1974)**



<http://www.icbdsr.org>

EUROCAT

Established in 1979

European organization

**CZE: Associate membership
(2009)**



<http://www.eurocat-network.eu/>

Down syndrome

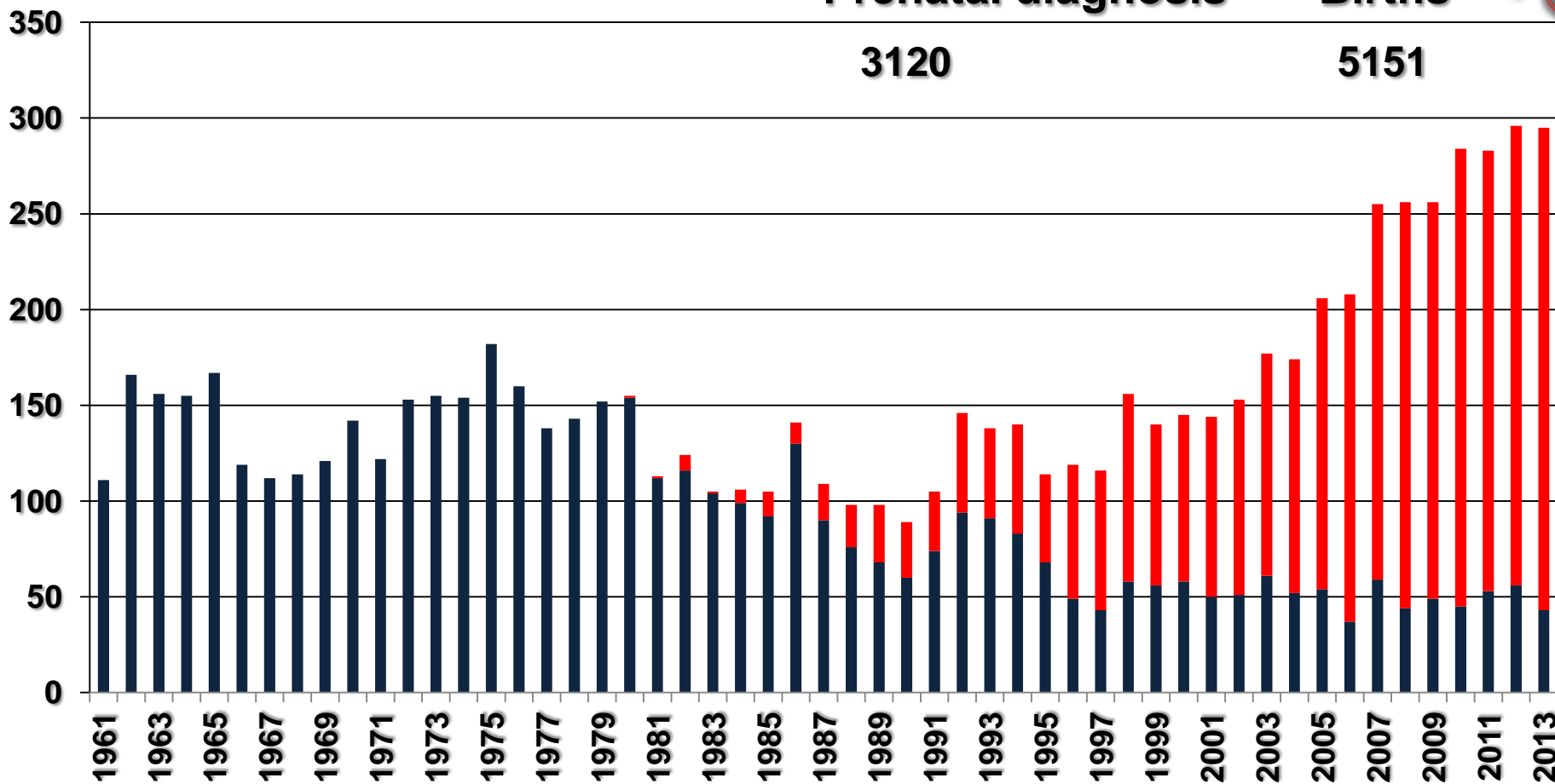
50+ years

Absolute numbers

■ Prenatal diagnosis ■ Births

3120

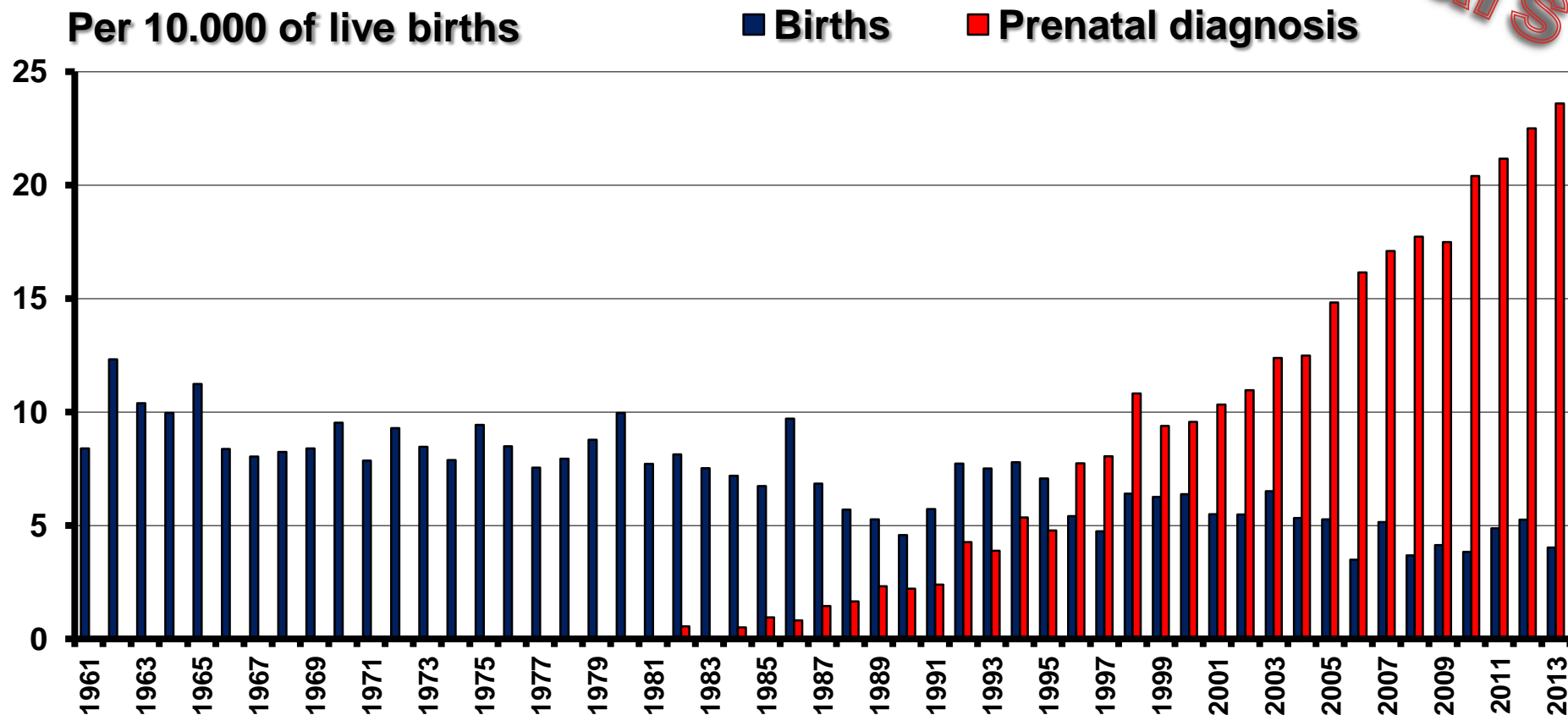
5151



Down syndrome – All cases in live births and prenatal diagnosis

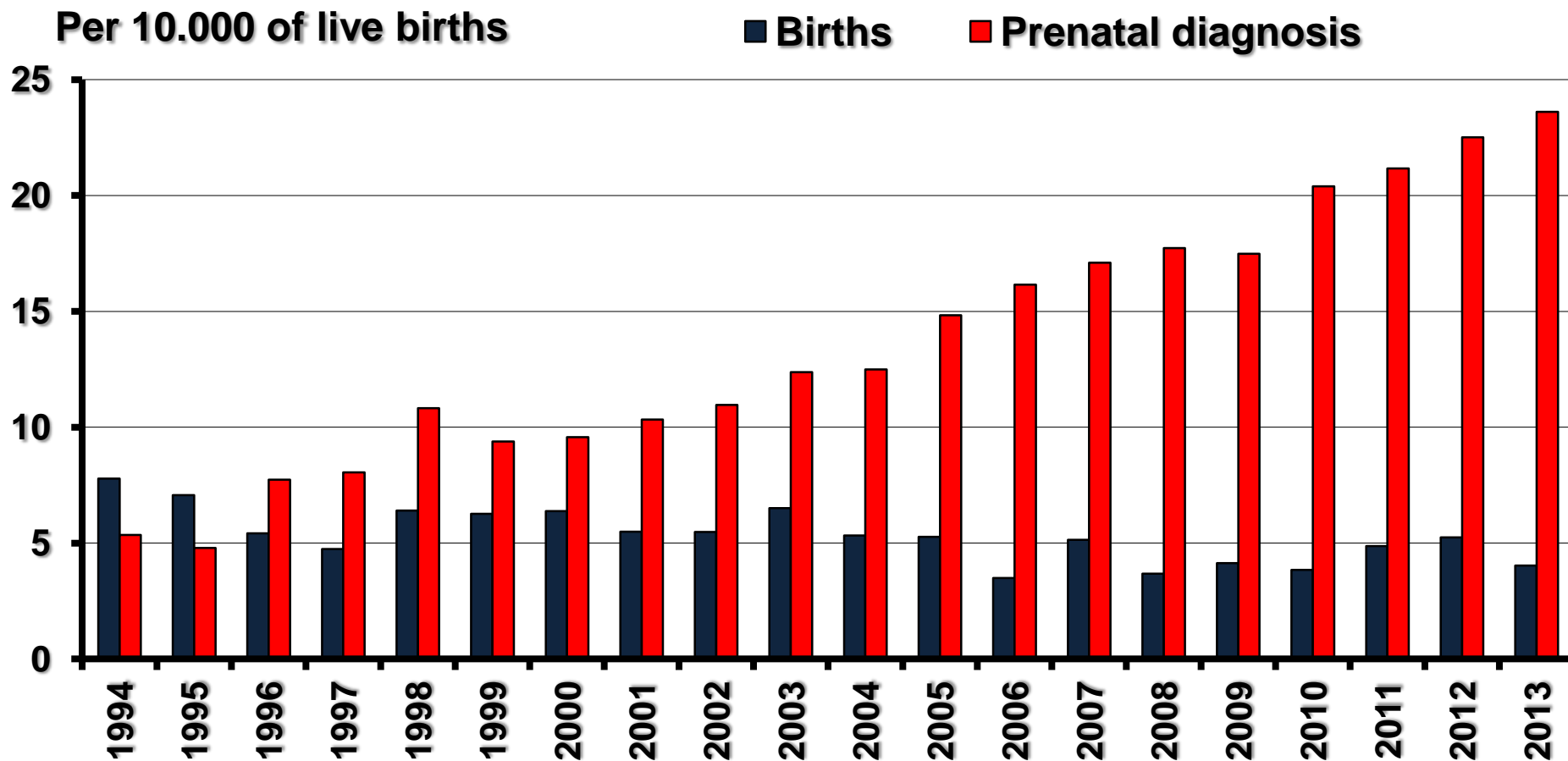
Down syndrome

50+ years



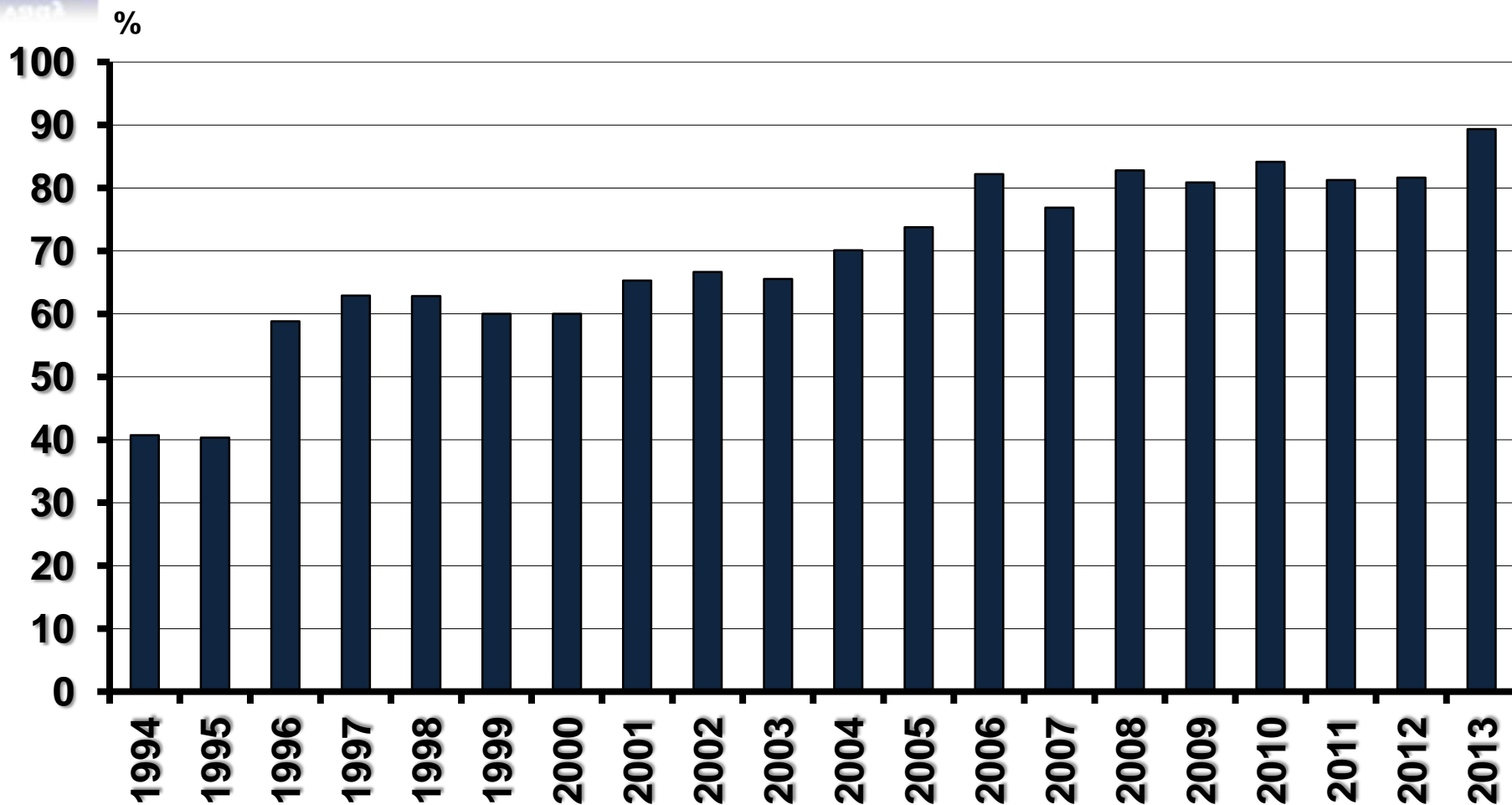
Down syndrome – All cases in live births and prenatal diagnosis

Down syndrome



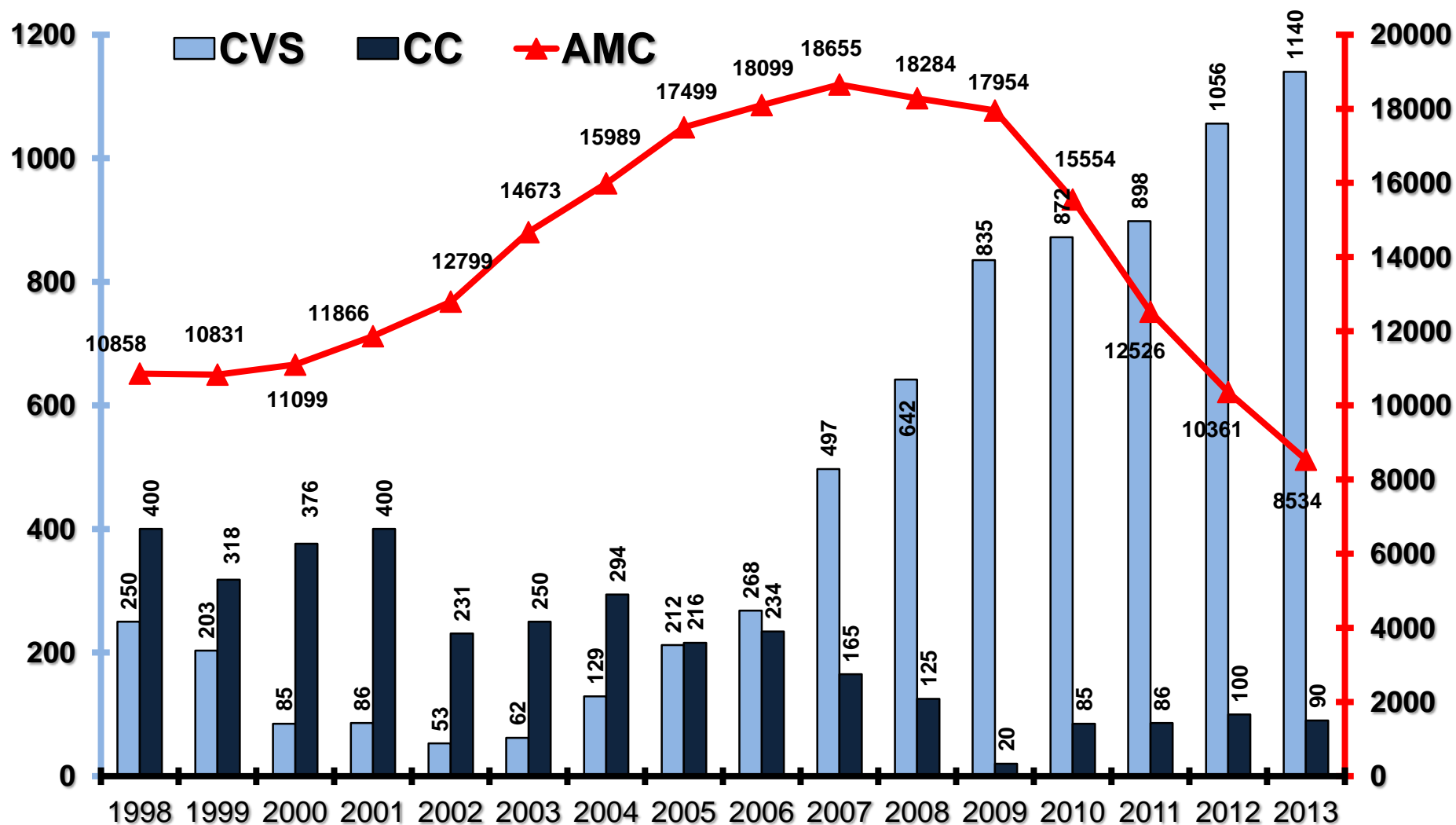
Down syndrome – All cases in live births and prenatal diagnosis

Down syndrome

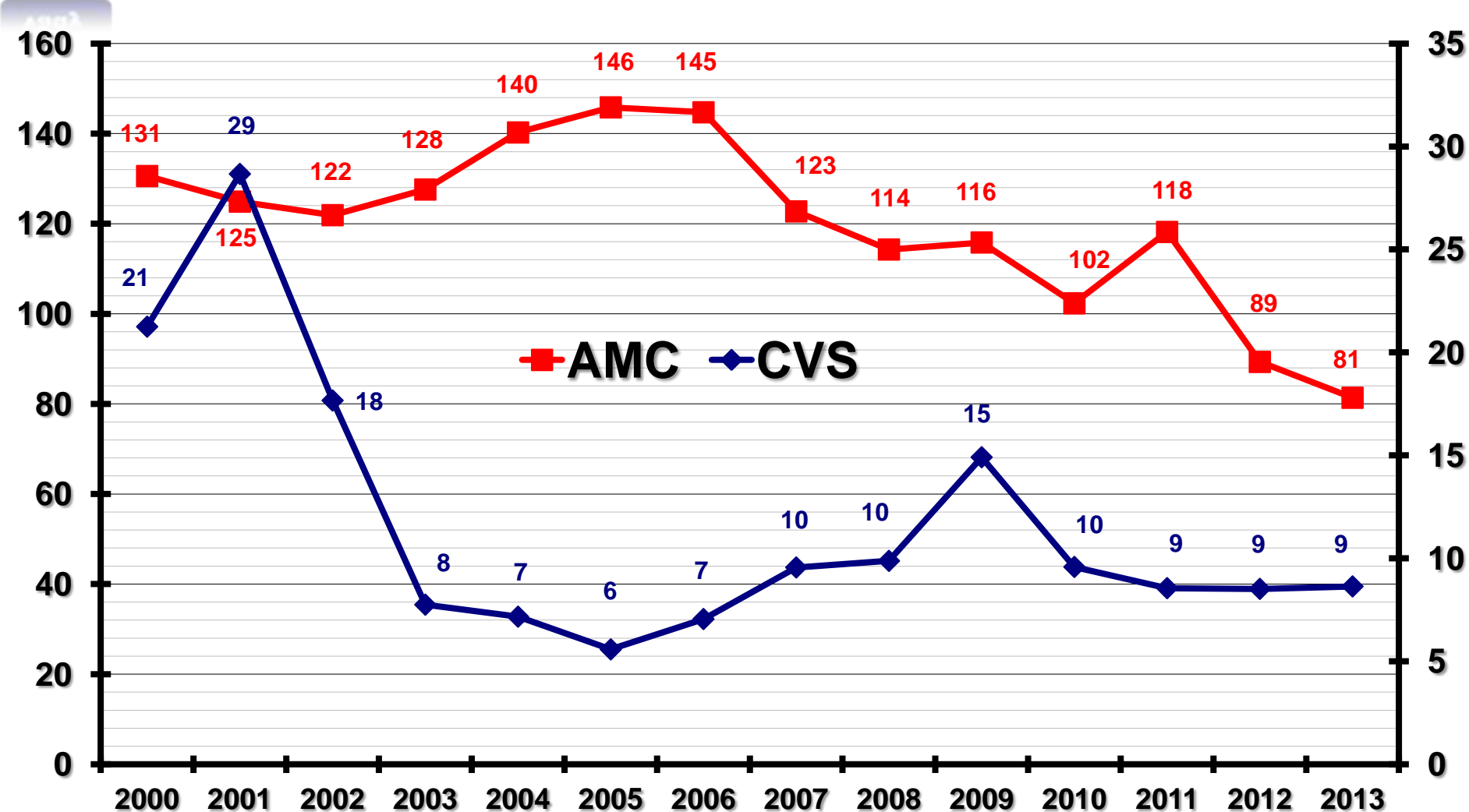


Down syndrome – relative number of prenatally diagnosed cases (%)

Invasive prenatal diagnosis

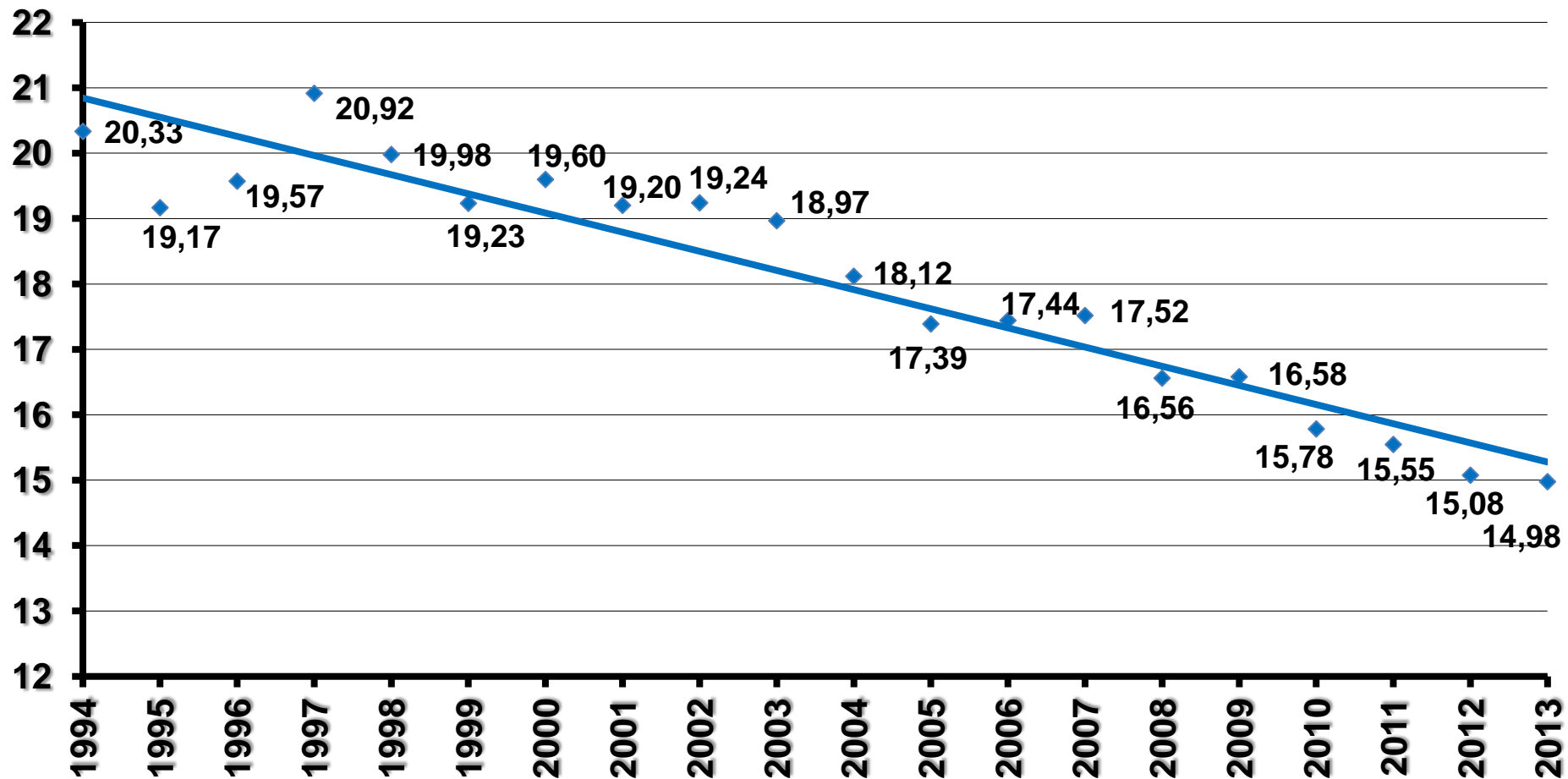


Effectiveness



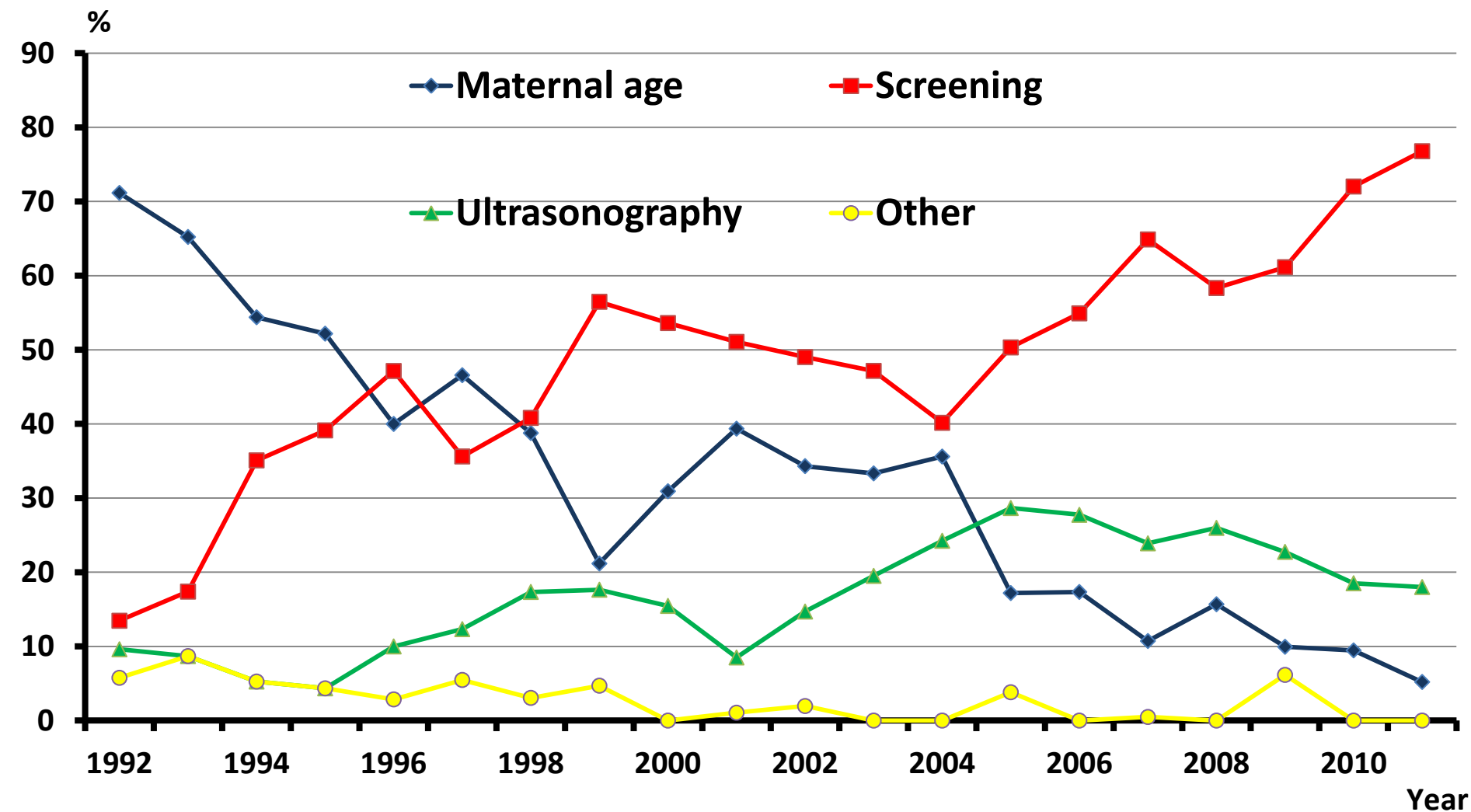
Number of invasive procedures needed for one diagnosis of Down syndrome

Time of diagnosis

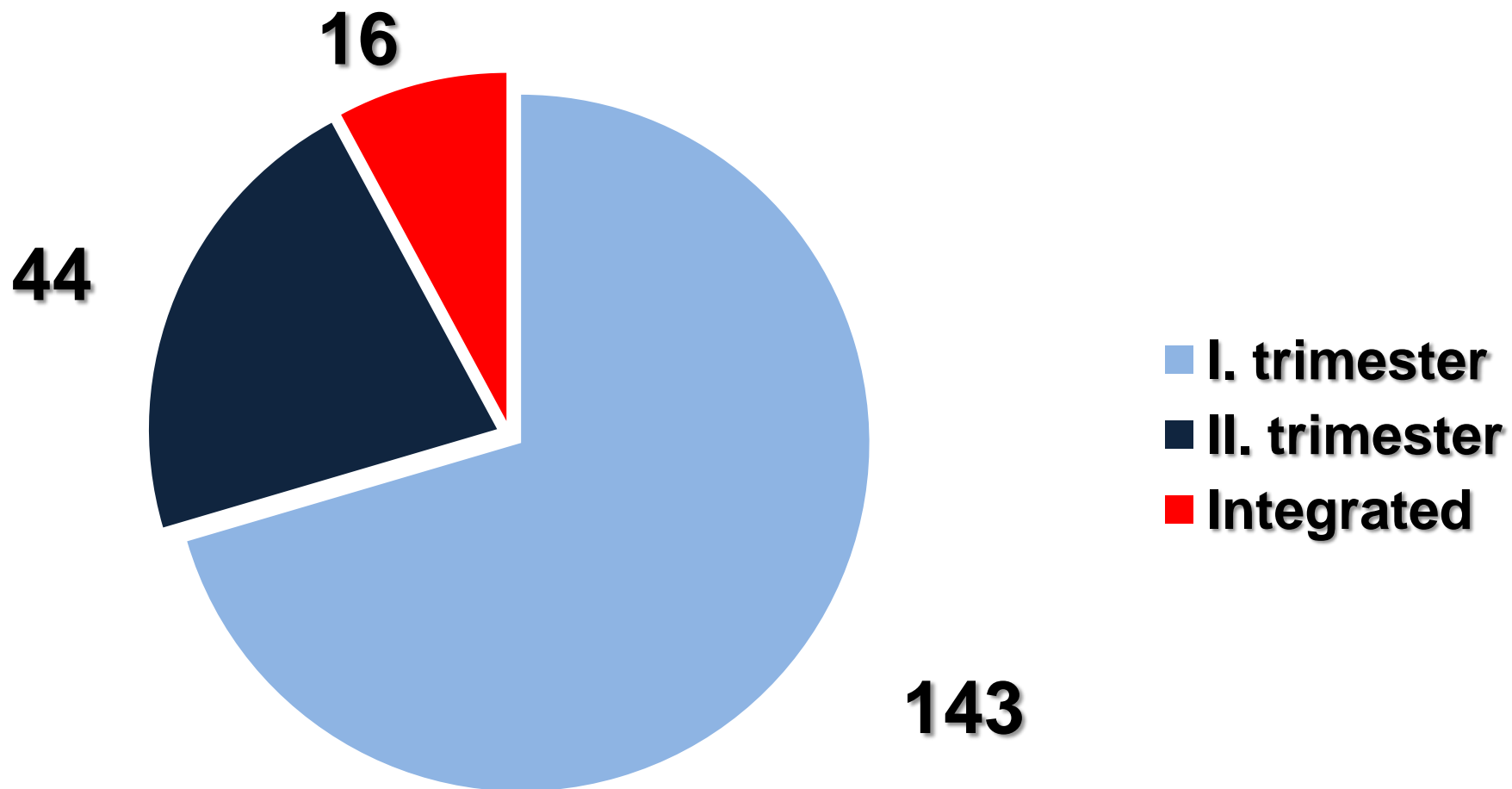


Average week of gestation at diagnosis of Down syndrome

Prenatal diagnosis - Indications



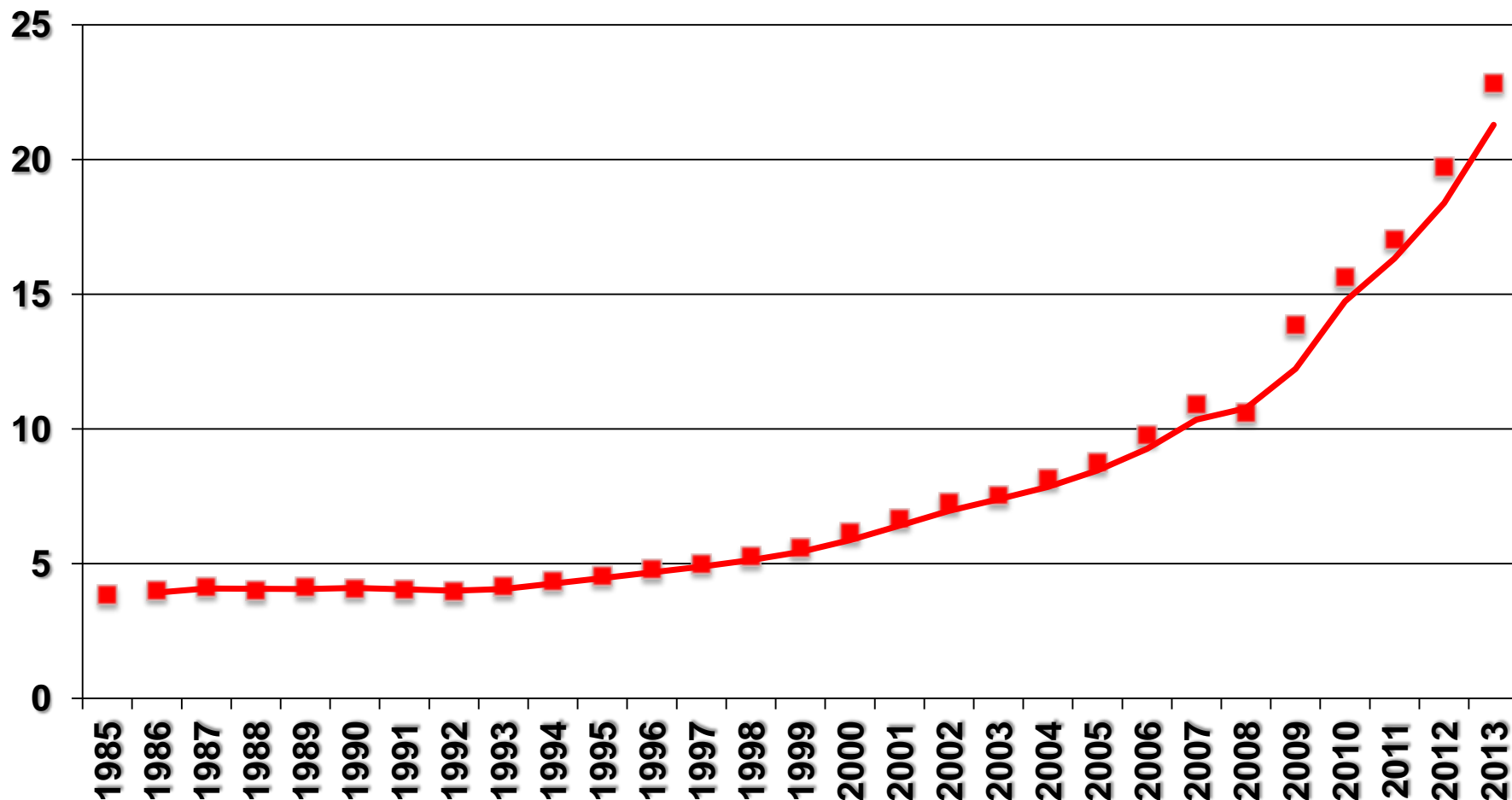
Screening



Proportion of screening programs in prenatal diagnosis (2013)

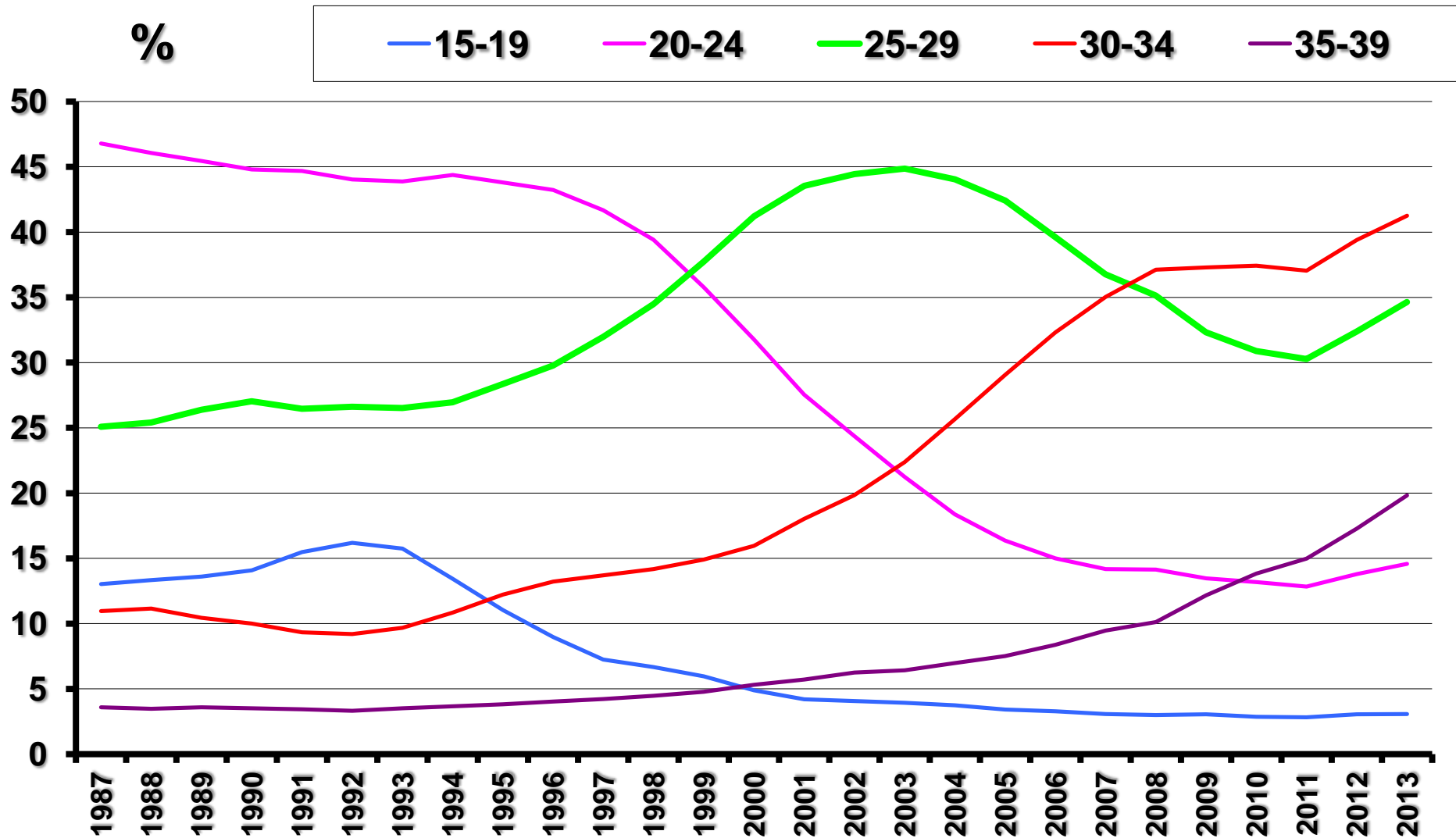
Maternal age – Time trends

%

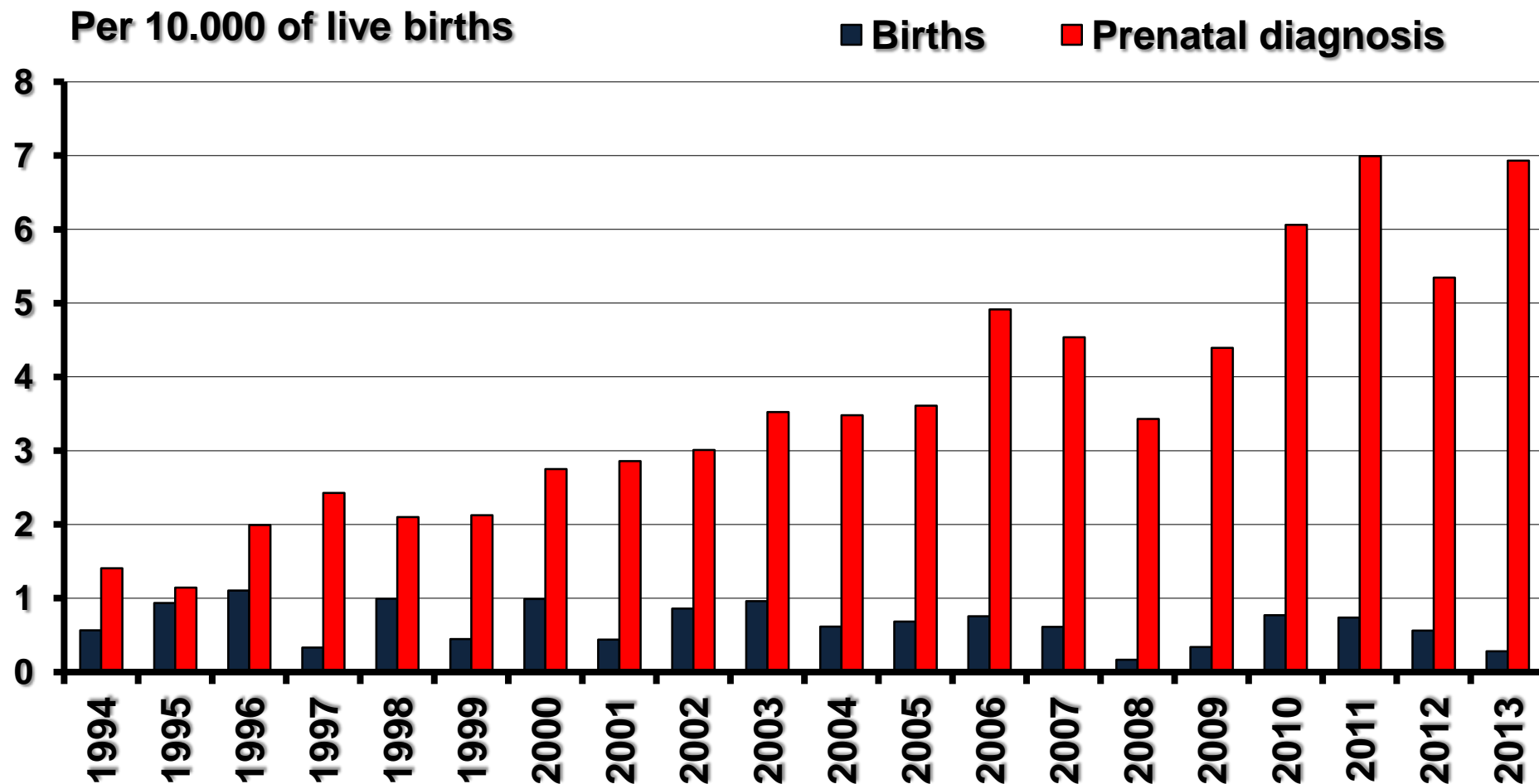


Proportion of 35+ years old mothers

Maternal age – Time trends

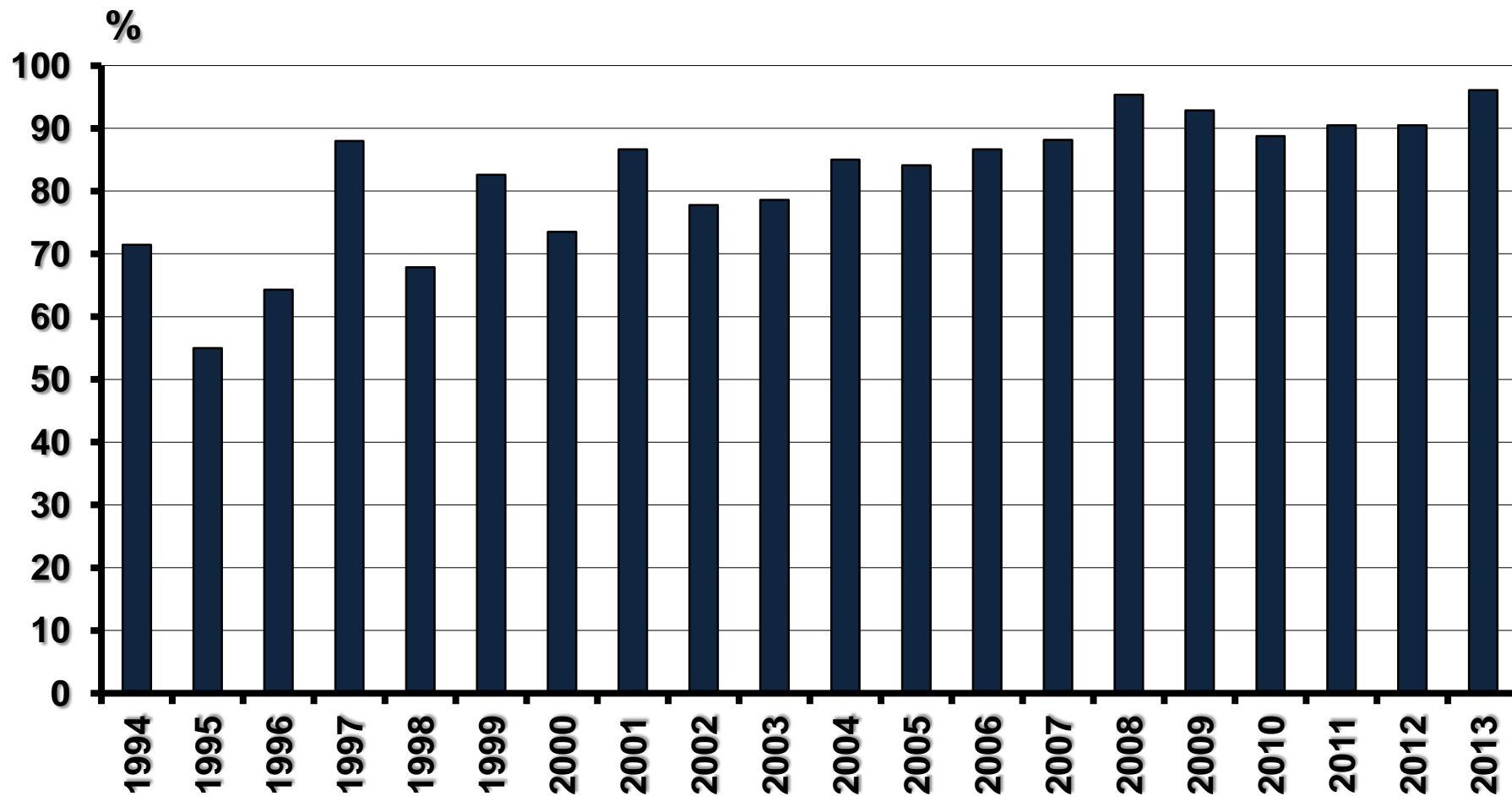


Edwards syndrome



Edwards syndrome – All cases in live births and prenatal diagnosis

Edwards syndrome



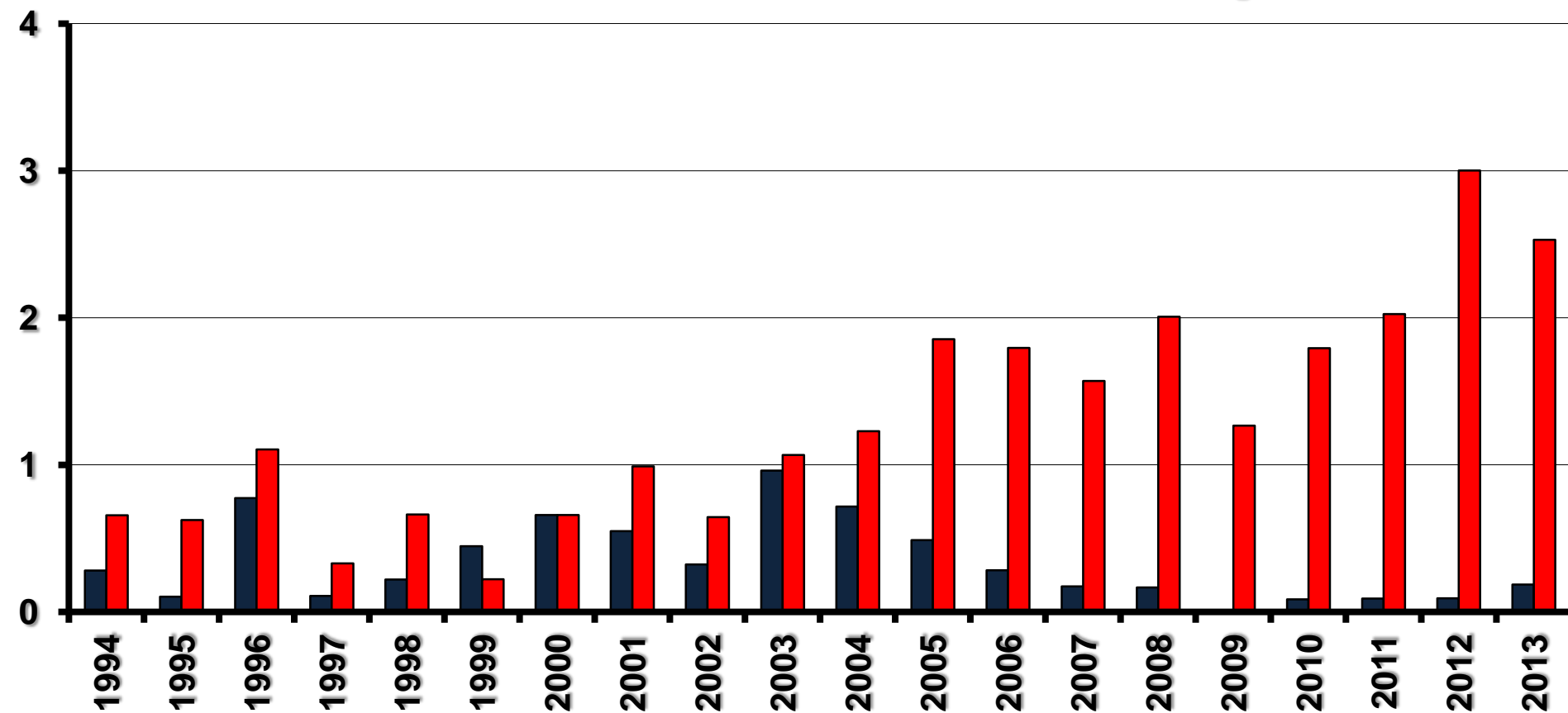
Edwards syndrome – relative number of prenatally diagnosed cases (%)

Patau syndrome

Per 10.000 of live births

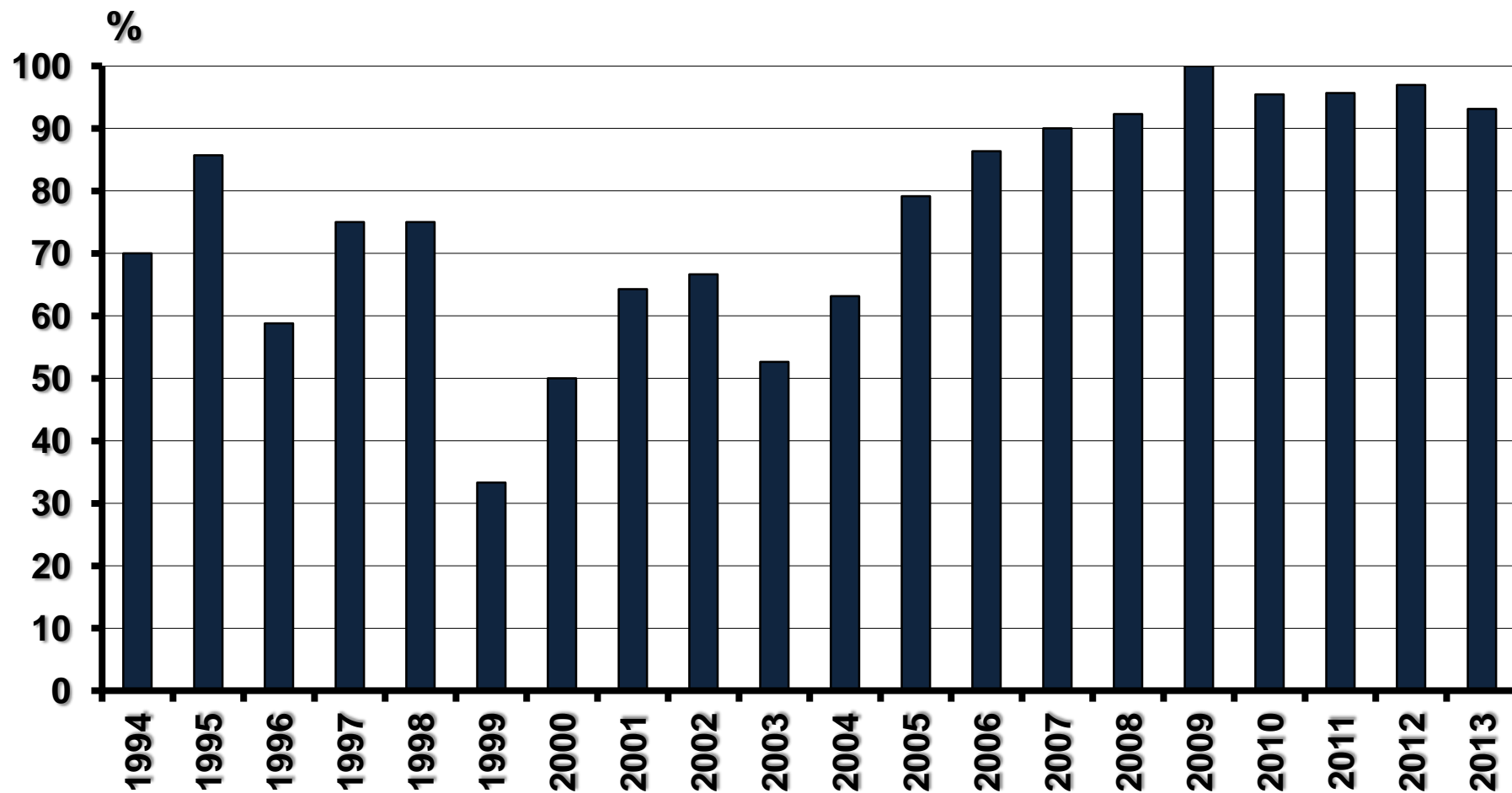
■ Births

■ Prenatal diagnosis



Patau syndrome – All cases in live births and prenatal diagnosis

Patau syndrome



Patau syndrome – relative number of prenatally diagnosed cases (%)

Conclusions

1. *The number of prenatally diagnosed cases of Down syndrome is increasing*
2. *The number of Down syndrome cases in live births is decreasing*
3. *The indication criteria for the invasive prenatal diagnosis are changing during last years*
4. *The average maternal age is notably increasing during last years*
5. *The early prenatal diagnosis (during the first trimester of gravidity) is becoming much more important during last years*

Acknowledgements

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Thank you for your attention!



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