Down syndrome in the Czech Republic in 1961 - 2001

Antonin Sipek[1], Jiri Horacek[2], Vladimir Gregor[2], Dana Masatova[3]

1 Institute for Care of Mother and Child, Prague, Czech Republic
2 Department of Medical Genetics, Postgraduate Medical Institute, Prague, Czech Republic
3 Institute of Health Information and Statistics, Prague, Czech Republic

Down syndrome	Number			Per 10 000 live births	
	Births	Prenatal diagnosis	Efficiency of prenatal diagnosis	Births	Prenatal diagnosis
1961-1965	755			10.47	
1966-1970	608			8.53	
1971-1975	766			8.59	
1976-1980	747	1	0.65	8.50	0.01
1981-1985	523	30	5.42	7.47	0.43
1986-1990	424	111	20.75	6.44	1.69
1991-1995	418	233	35.79	7.27	4.05
1996-2000	255	412	61.77	5.64	9.11
2001	47	94	66.67	5.17	10.33
Total	4543	881	32.61	8.00	3.35

Total Down syndrome 5. 424 cases

In births 4. 543 cases

Prenatally diagnosed 881 cases

male 51.69 %

female 48.31 %

isolated 62.71 %

combinated defects 37.29 %

cardiac malformations	58.4 %
atresia/stenosis of small/large intestine	14.9 %
malformations of the nervous system	5.2 %
malformations of the urinary system	3.4 %
musculoskeletal deformities	7,9 %
malformations of the respiratory system	3.2 %

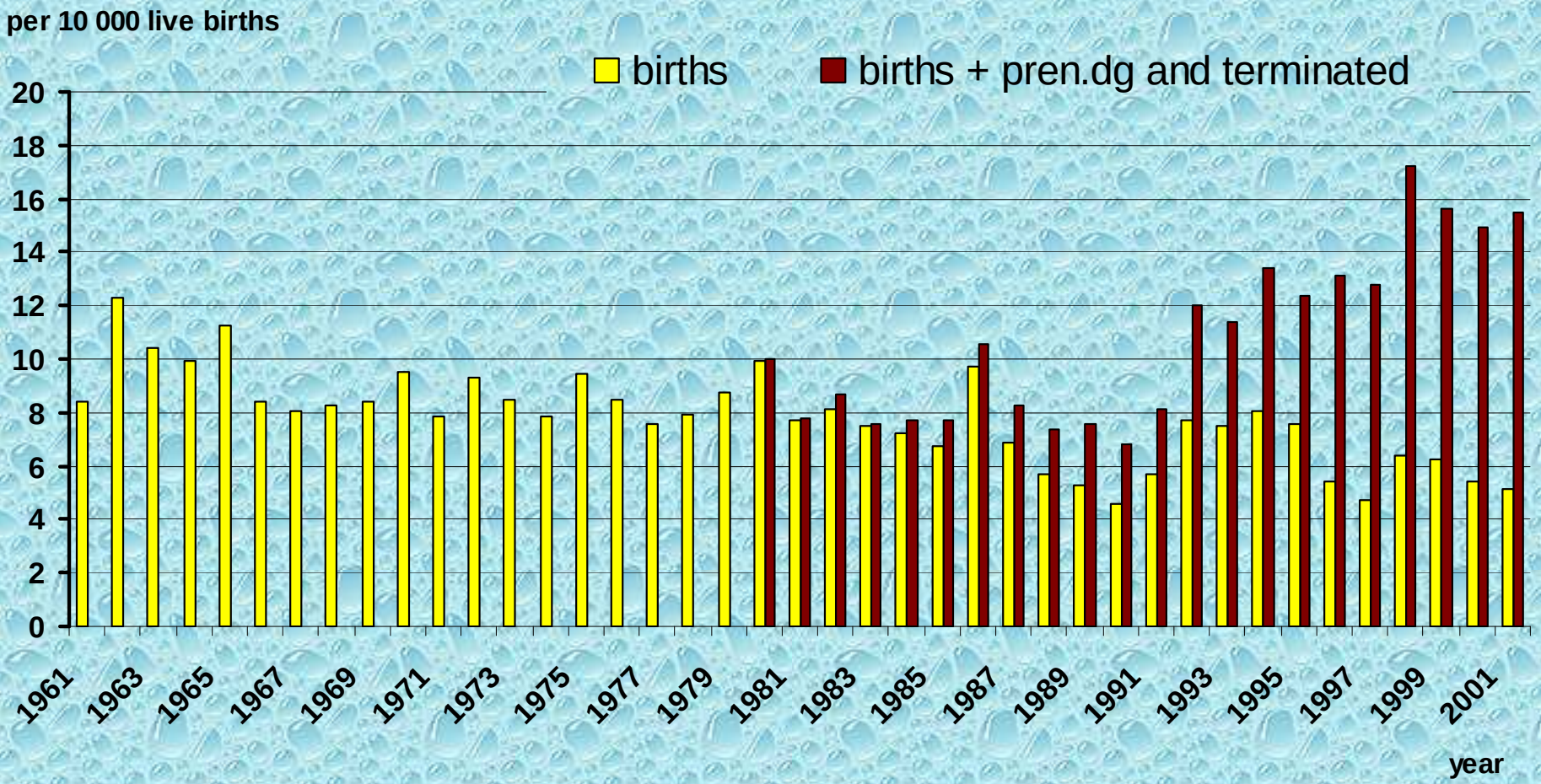
The data on prenatal diagnosis of Down syndrome (DS) have been systematically collected since 1980, when the cytogenetic prenatal diagnosis was started.

Since recently, the maternal age limit for the cytogenetic prenatal diagnosis is 35 years in most of the centres where the prenatal diagnosis is available.

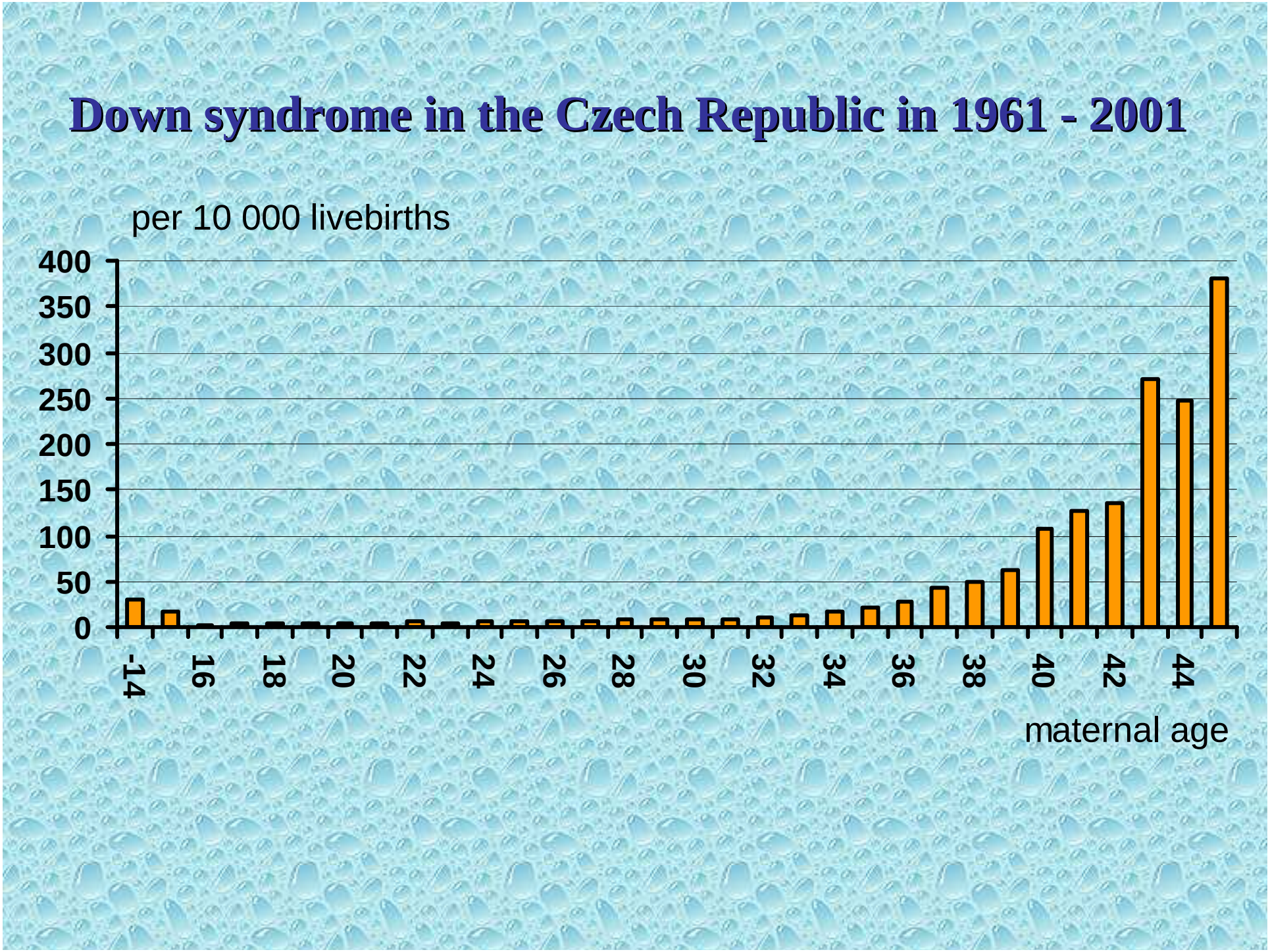
The majority of pregnant women are now being offered a biochemical triple-test screening. Spontaneous abortion cases (< 28 weeks of gestation and/or < 1 000 g of weight) are not registered.

Routine screening techniques for DS, such as ultrasound practice (fetal nuchal translucency) and biochemical screening [serum human Chorionic Gonadotropin (β-hCG), unconjugated estriol (uE_3), and α-fetoprotein (MS-AFP)] are year by year more extensively used. Maternal age was a further criterion.

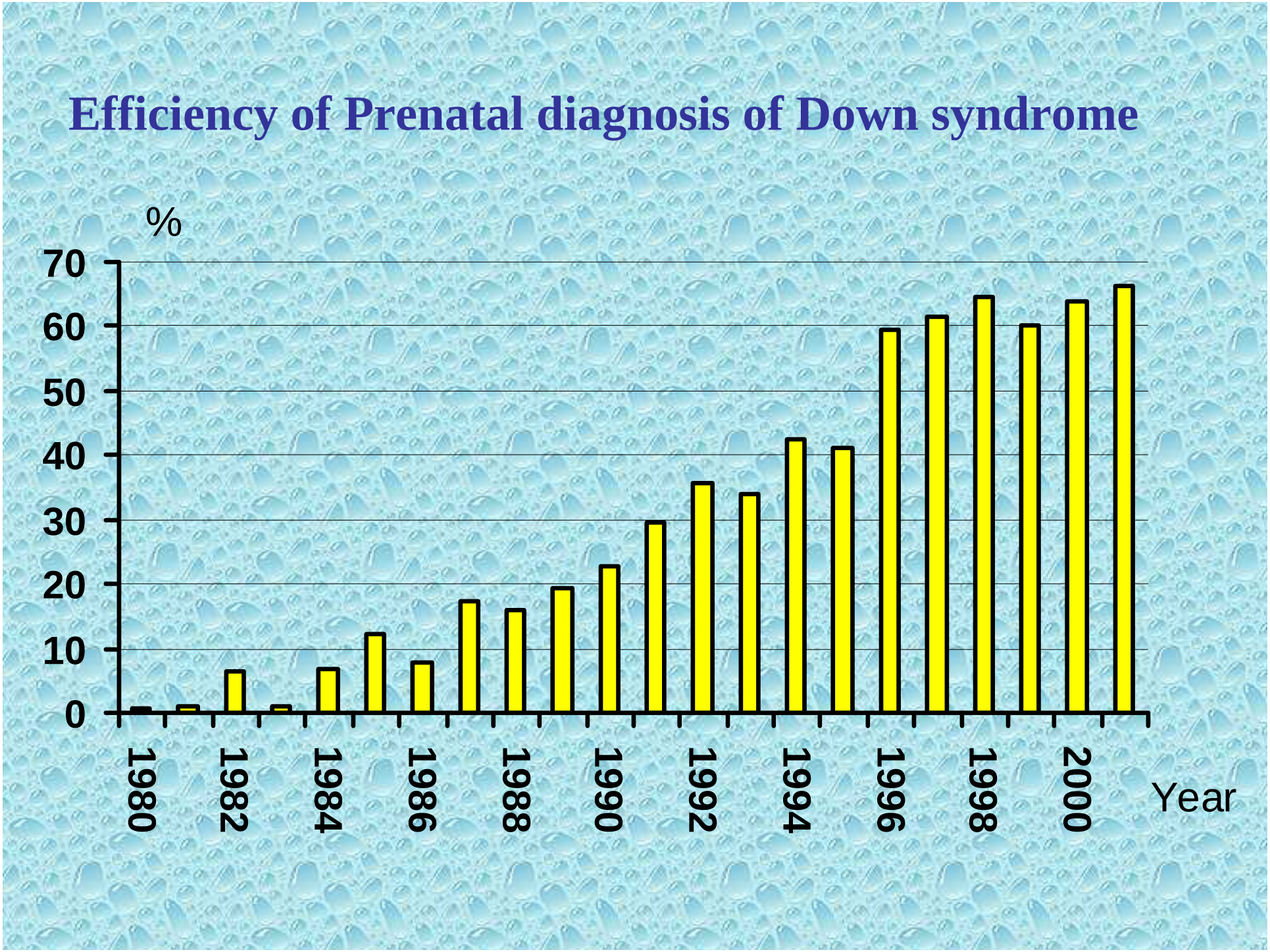
Down syndrome in the Czech Republic in 1961 - 2001



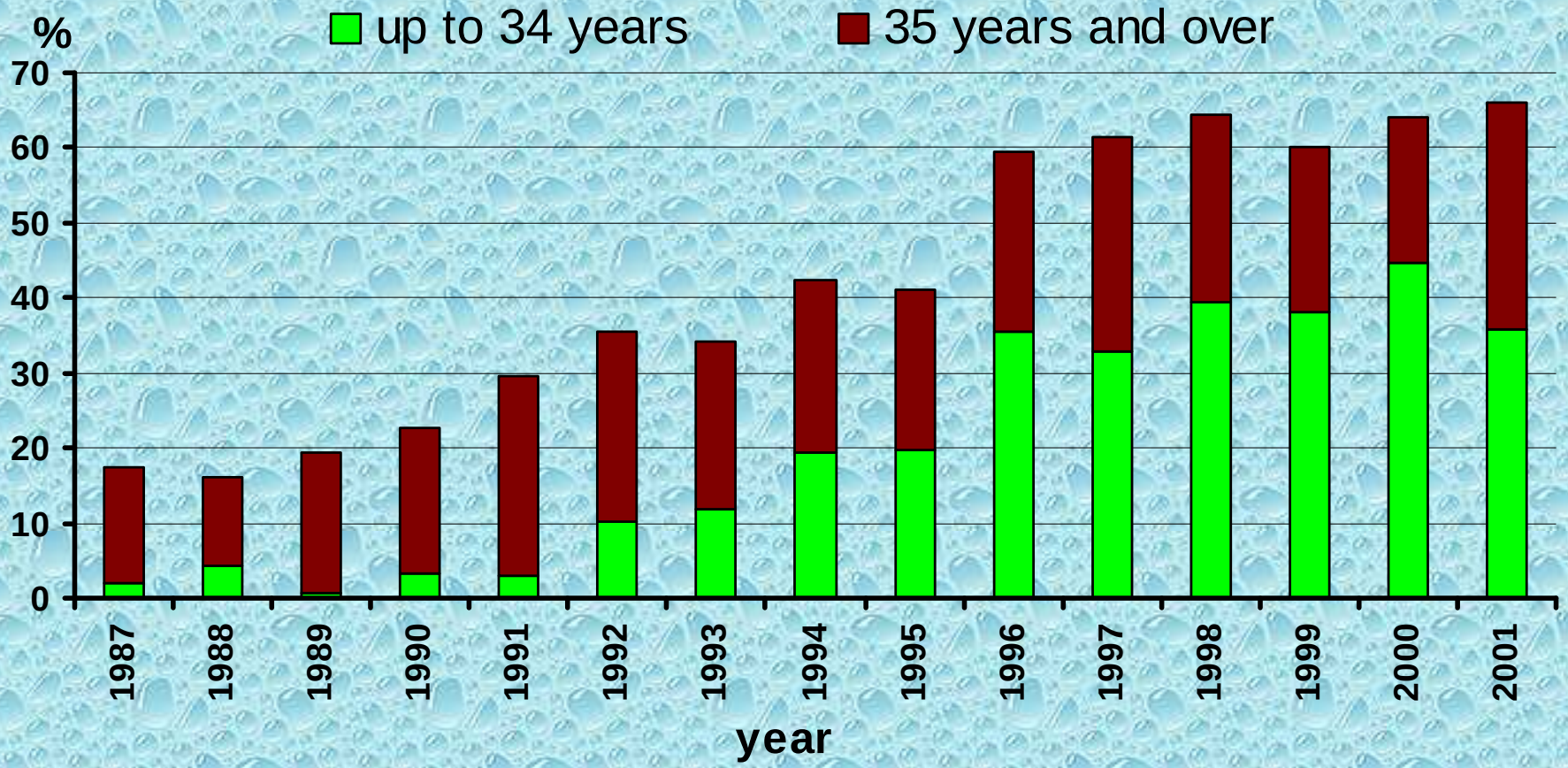
Down syndrome in the Czech Republic in 1961 - 2001



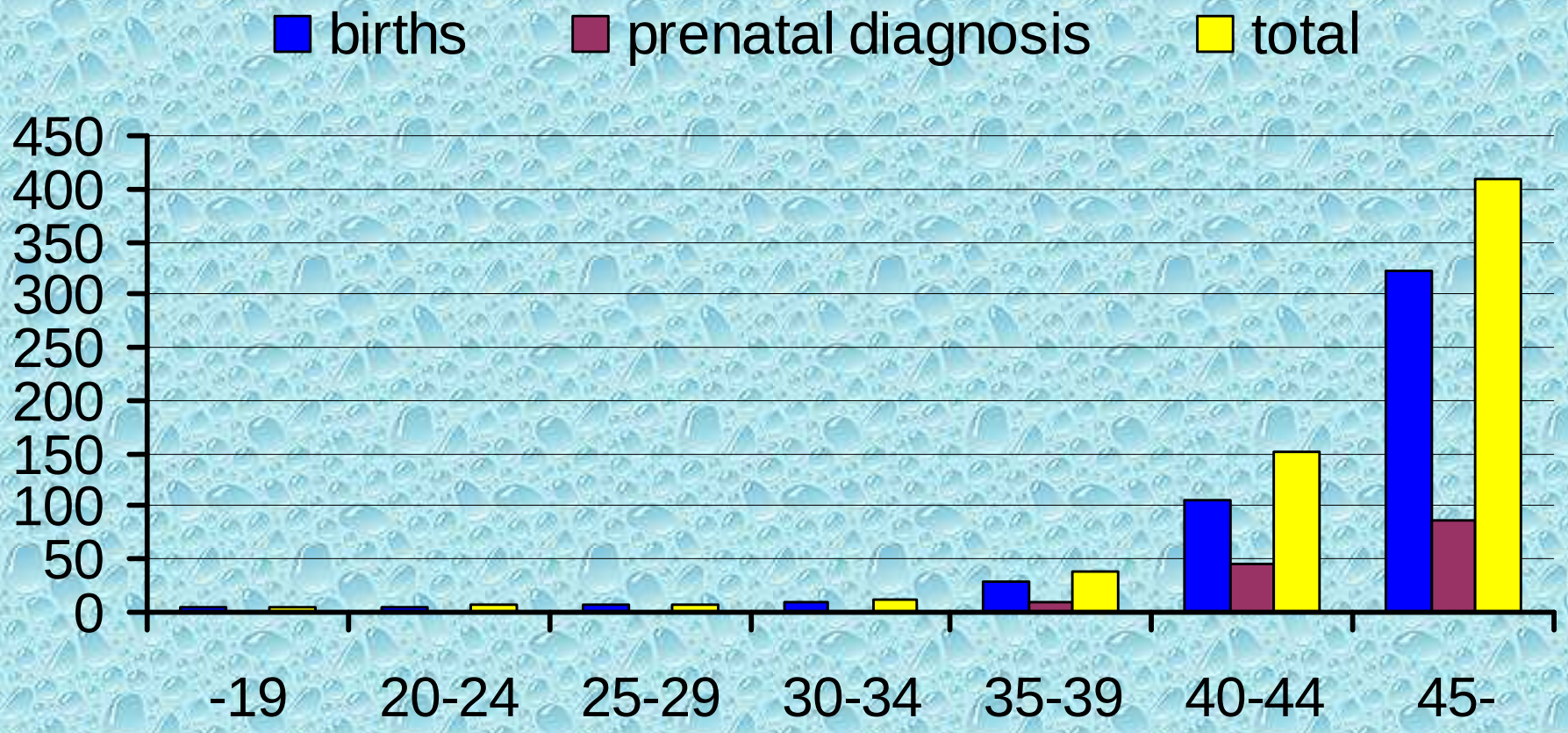
Efficiency of Prenatal diagnosis of Down syndrome



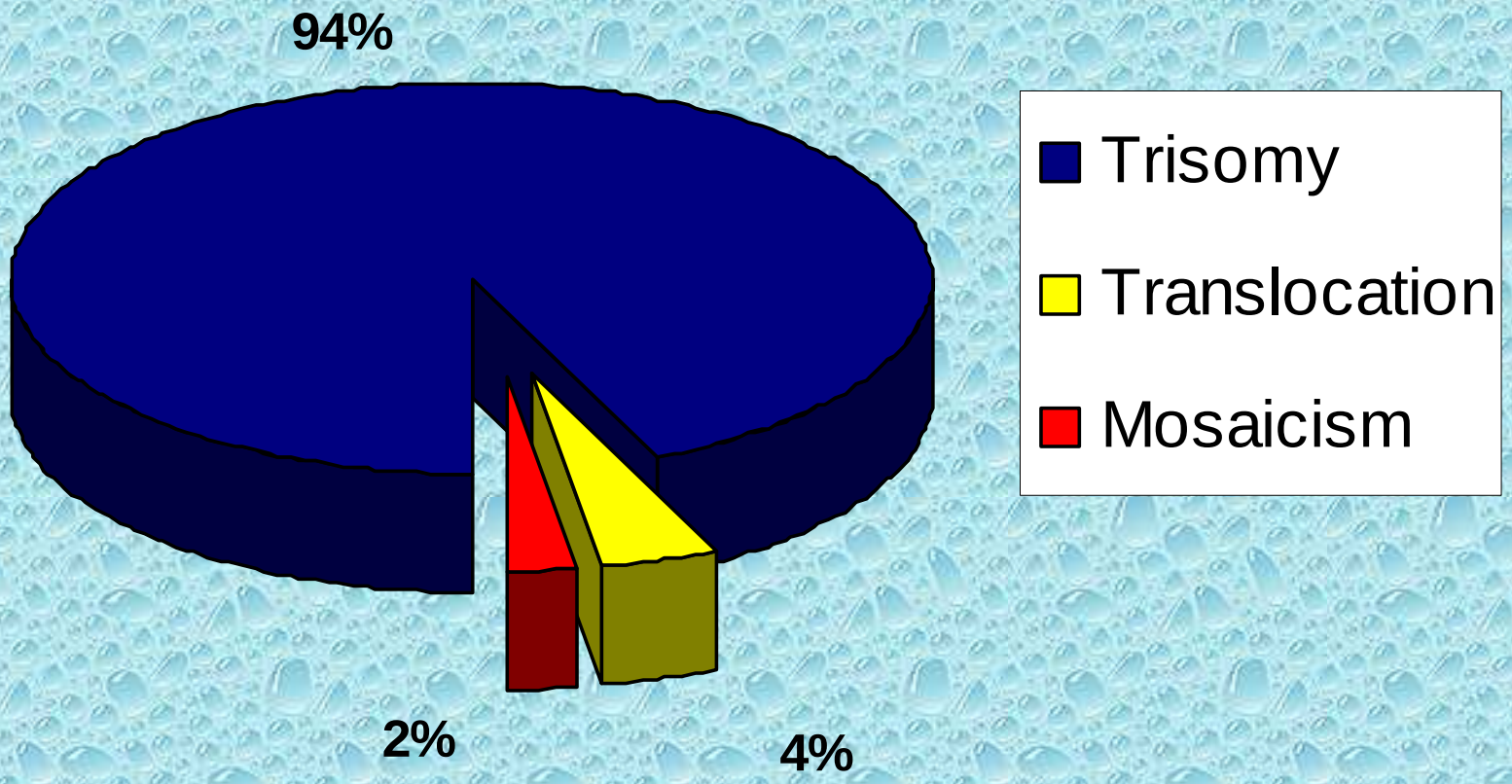
Efficiency of Prenatal diagnosis of Down syndrome according to maternal age



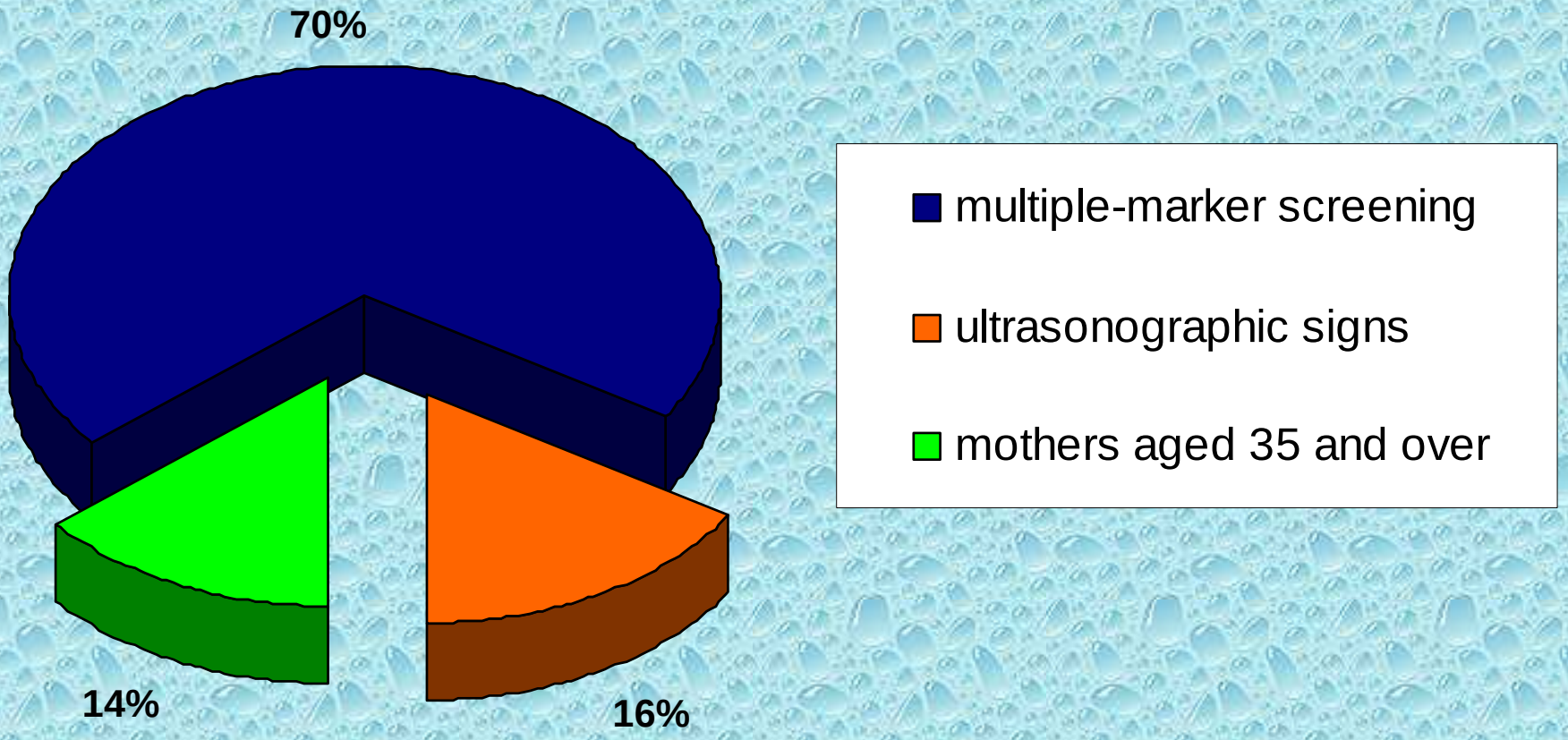
Relative frequency of Down syndrome per 10 000 live births in 1961-2001 according to maternal age



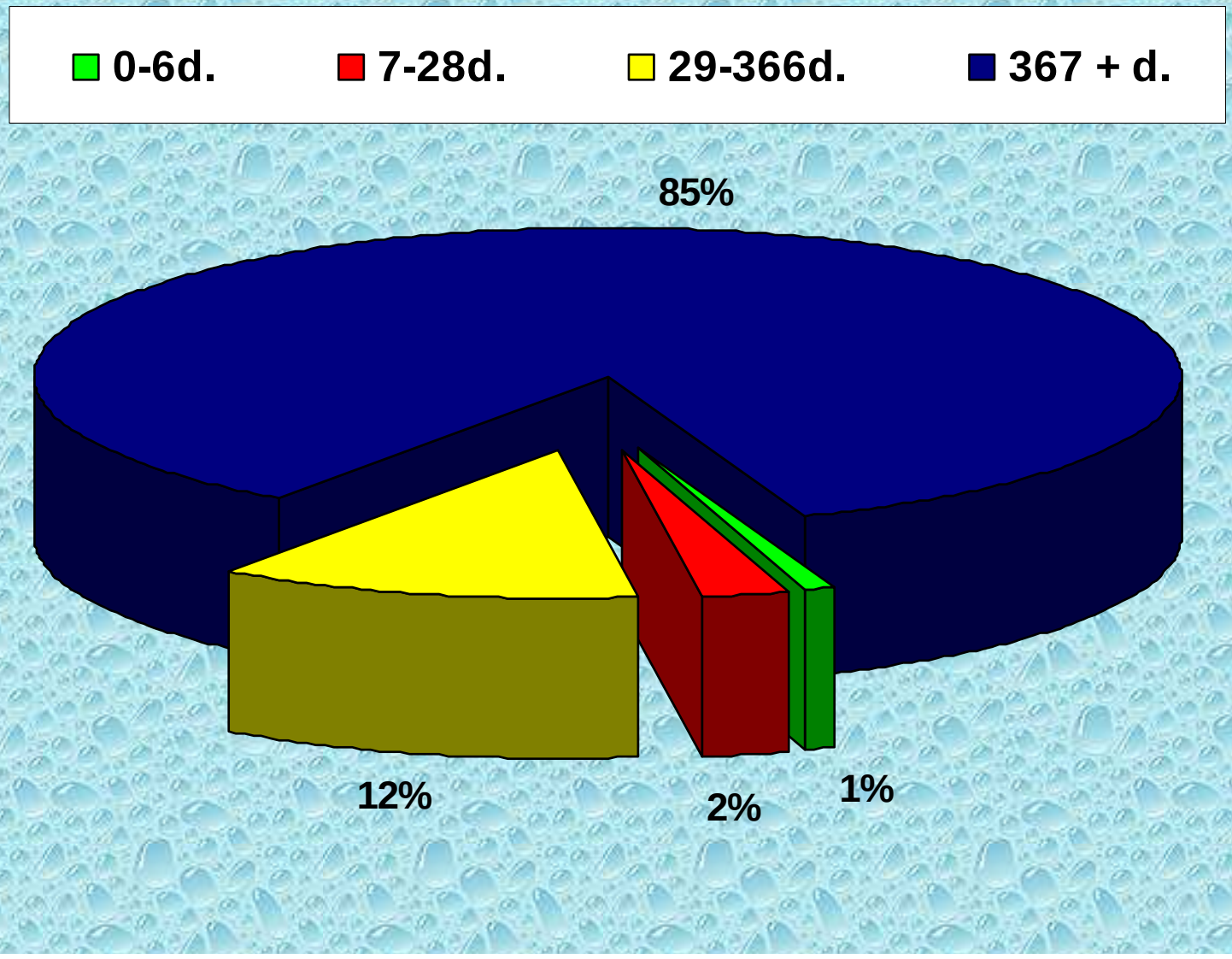
Down syndrome in the Czech Republic in 1994 - 2001, births + prenatal diagnosis



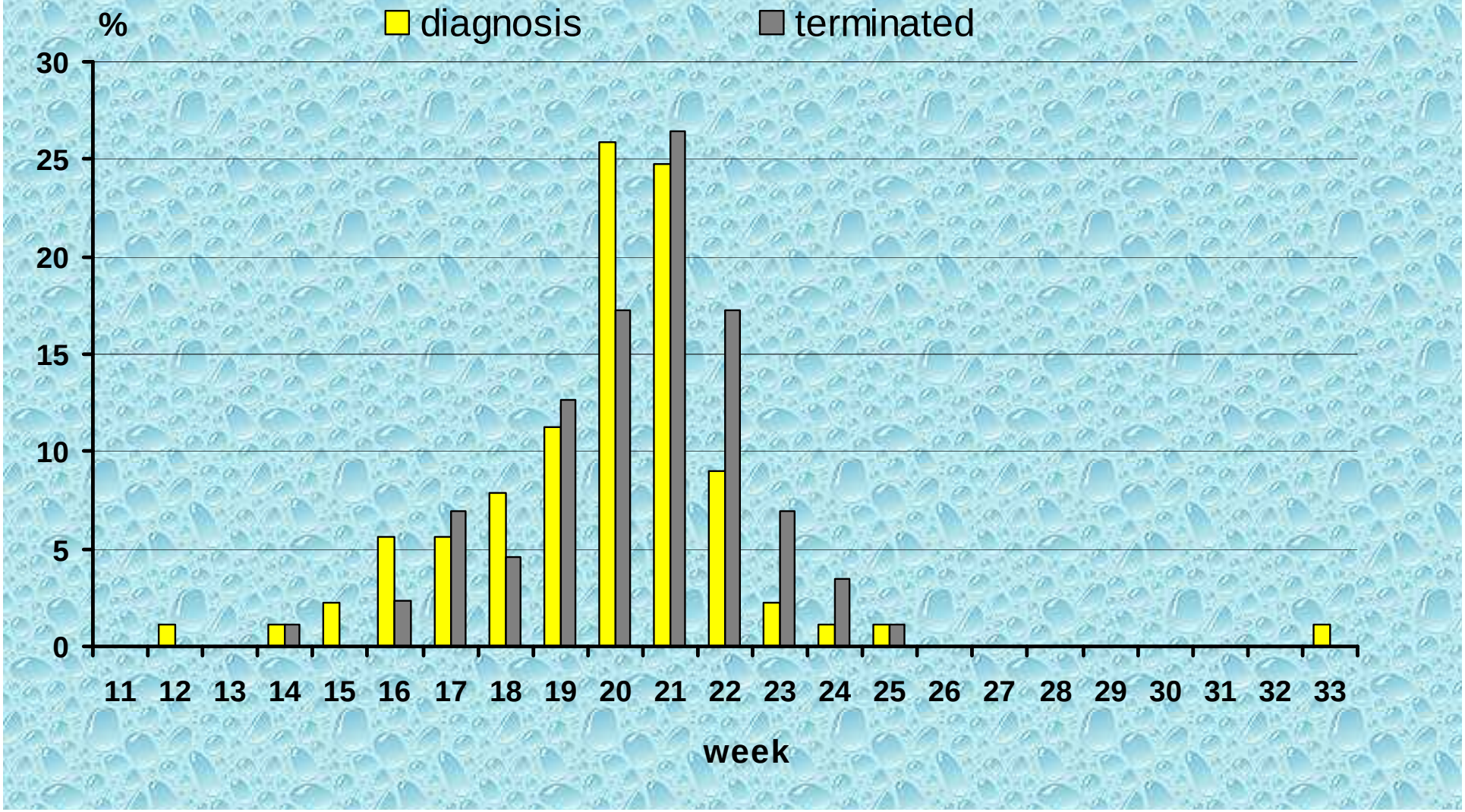
Down syndrome in the Czech Republic in 1994 - 2001, prenatal diagnosis



Survival (in days) of children with Down syndrome, Czech Republic, 1994-1998



Down syndrome in the Czech Republic in 1994 - 2001, prenatal diagnosis



Results 1:

During the period under the study, totally 5 680 904 livebirths were registered in the Czech republic.
Out of this number, 5 424 DS cases were registered in the Czech Birth Defects Register.
Out of this number, 881 cases were diagnosed prenatally and 4 543 cases postnatally.
Mean incidence of DS during the whole period under the study was 8.00 per 10 000 livebirths.
A statistically significant decrease of DS incidences in births has occurred during this 41 years period in the Czech Republic:
from 10.47 per 10 000 livebirths during the 1961 – 1965 period to 5.64 per 10 000 livebirths in 1996 – 2000 and 5.17 per 10 000 in 2001.

Results 2:

An age distribution in mothers of DS babies shows an increased risk in mothers of 34 years of age and over.
A mean number of prenatally diagnosed DS cases out of the total number was 41.1 % during the 1980 – 2001 period, increasing from 5.4 % in 1980 – 1985 to 61.8 % in 1996 – 2000 and 66.7 in 2001.
A mortality of DS babies during the first year of life is about 16 % during the last five years period in the Czech Republic.
In 37.3 % of DS cases in births, an additional associated malformation (congenital heart defects, gastrointestinal or urogenital tracts defects etc.) was found.

Conclusions:

During the 1961 – 2001 period, a statistically significant decrease of DS in births has occurred in the Czech Republic. Along this trend, a number of prenatally diagnosed cases has increased and represents more than 60 % out of reported DS cases recently. This is mostly due to the improvement and availability of both screening and prenatal diagnostics methods. Still, children born with DS as well as with other major birth defects present an important contribution to perinatal, neonatal and infant mortality and morbidity.

The study was supported by IGA MZ CR grant NJ 6214-3

Czech Republic

2001:

10 309 137 inhabitans

90 910 livebirths

3 572 births with birth defetcs

373 deaths under 1 year of age