Czech Register of Chromosomal Aberrations

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http://www.vrozene-vady.cz/
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)
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.
<table>
<thead>
<tr>
<th>Year</th>
<th>Event</th>
</tr>
</thead>
<tbody>
<tr>
<td>1971</td>
<td>Cultivation of amniocytes</td>
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<tr>
<td>1971</td>
<td>First prenatally diagnosed M. Down</td>
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<tr>
<td>1977</td>
<td>Placentocentesis</td>
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<tr>
<td>1978</td>
<td>Fetoscopy</td>
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<tr>
<td>1980</td>
<td>USG Diagnostics</td>
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<tr>
<td>1983</td>
<td>CVS</td>
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<tr>
<td>1985</td>
<td>The data on prenatal diagnosis became a part of registration</td>
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<tr>
<td>1987</td>
<td>Cordocentesis</td>
</tr>
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<td>1988</td>
<td>Early amniocentesis</td>
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<tr>
<td>1990</td>
<td>Biochemical screening</td>
</tr>
<tr>
<td>2000</td>
<td>PGD</td>
</tr>
<tr>
<td>2012</td>
<td>NIPST</td>
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</tbody>
</table>
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)

[ICBDSR logo]

[http://www.icbdsr.org](http://www.icbdsr.org)

**EUROCAT**

- Established in 1979
- European organization
- CZE: Associate membership (2009)

[EUROCAT logo]

Down syndrome

Per 10,000 of live births

Births

Prenatal diagnosis

Down syndrome – All cases in live births and prenatal diagnosis
Down syndrome – All cases in live births and prenatal diagnosis
Down syndrome – relative number of prenatally diagnosed cases (%)
Invasive prenatal diagnosis

![Graph showing invasive prenatal diagnosis data for CVS, CC, and AMC from 1998 to 2013.]
Effectiveness

Number of invasive procedures needed for one diagnosis of Down syndrome
Time of diagnosis

Average week of gestation at diagnosis of Down syndrome

- 1994: 20.33
- 1995: 20.92
- 1996: 19.57
- 1997: 19.98
- 1998: 19.60
- 1999: 19.20
- 2000: 19.24
- 2001: 18.97
- 2002: 18.12
- 2003: 17.44
- 2004: 17.52
- 2005: 17.39
- 2006: 16.56
- 2007: 16.58
- 2008: 15.78
- 2009: 15.55
- 2010: 15.08
- 2011: 14.98
- 2012: 14.98
- 2013: 14.98
Screening

Proportion of screening programs in prenatal diagnosis (2013)

- 143 programs
- 44 in I. trimester
- 16 in II. trimester
- 16 integrated
Maternal age – Time trends

Proportion of 35+ years old mothers
Edwards syndrome – All cases in live births and prenatal diagnosis
Edwards syndrome – relative number of prenatally diagnosed cases (%)
Patau syndrome – All cases in live births and prenatal diagnosis

Per 10,000 of live births

Births
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
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!

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