Current Results of Prenatal Diagnosis in the Czech Republic

Antonin Sipek jr.1, Vladimir Gregor2,3, Antonin Sipek2,3
Jiri Horacek4 and Pavel Langhammer5

1 – Institute of Biology and Medical Genetics, Charles University, 1st Faculty of Medicine and General University Hospital, Prague, Czech Republic
2 – Department of Medical Genetics, Thomayer Hospital, Prague, Czech Republic
3 – Screening centre, Pronatal Sanatory, Prague, Czech Republic
4 – Gennet Ltd, Prague, Czech Republic
5 – Institute for Health Information and Statistics of the Czech Republic

http://www.vrozenevady.cz/
Czech Republic

Population: 10 548 527 (in 2011)
Area: 78,866 km²
Regions: 13 regions and the capital Prague; 76 districts in total
Population density: 133/km²
Annual births: approx. 100 000 (119 570 in 2008)
Capitol and largest city: Prague (1 241 000 inhabitants)
Life expectancy (years): Males - 73.54; Females - 80.28
Language: Czech
Ethnicity: Czech 90.4%, Moravian 3.7%, Slovak 1.9%, other 4%
Religions: Roman Catholic 27%, Protestant 2%, unaffiliated 59%
History and Current State 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)

Cooperating centre:
**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

**Cases:** all cases in TOPs, live births and stillbirths (above 1000g) 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)
Prenatal diagnosis - Numbers

Terminations of pregnancies
Continuing pregnancies
Invasive diagnostics

per 10 000 live births

year

Terminations of pregnancies
Continuing pregnancies
Invasive diagnostics

per 10 000 live births

year
Invasive prenatal diagnosis

- CVS
- CC
- AMC

Number of diagnoses over the years:
- 1998: 250
- 1999: 318
- 2000: 376
- 2001: 400
- 2002: 53
- 2003: 62
- 2004: 129
- 2005: 212
- 2006: 216
- 2007: 234
- 2008: 165
- 2009: 642
- 2010: 835
- 2011: 88
Effectiveness

Number of invasive procedures needed for one diagnosis of Down syndrome
Prenatal diagnosis - Indications

Maternal age

Screening

Ultrasonography

Other
Maternal age – Time trends

- 15-19
- 20-24
- 25-29
- 30-34
- 35-39

%
Anencephaly

per 10,000 live births

- Births
- Prenatal diagnosis

Year

Anencephaly – All cases in live births and prenatal diagnosis
Anencephaly – relative number of prenatally diagnosed cases (%)
Spina bifida – All cases in live births and prenatal diagnosis
Spina bifida

Spina bifida – relative number of prenatally diagnosed cases (%)
Omphalocele – All cases in live births and prenatal diagnosis
Omphalocele – relative number of prenatally diagnosed cases (%)
Gastroschisis – All cases in live births and prenatal diagnosis
Gastroschisis – relative number of prenatally diagnosed cases (%)
Down syndrome – All cases in live births and prenatal diagnosis
Down syndrome – relative number of prenatally diagnosed cases (%)
Conclusions

1. The number of prenatally diagnosed cases of congenital anomalies is increasing
2. The number of congenital anomalies 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
Thank you for your attention!

antonin.sipek@lf1.cuni.cz

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