ULTRASOUND PRENATAL DIAGNOSTICS OF CONGENITAL ANOMALIES IN THE CZECH REPUBLIC

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Aim and type of study: A retrospective study with data analysis of prenatal occurrence of selected types of congenital anomalies (CA) in the Czech Republic in 1994 – 2006. An analysis of prenatal diagnostics contribution to the decrease of CA frequency in the Czech Republic with a special attention to ultrasound diagnostic techniques.

Material and methods: A retrospective epidemiological analysis of incidence of prenatal diagnosed cases of selected types of CAs during 1994 – 2006 in the Czech Republic was performed along with an analysis of their proportion in corresponding total numbers of CAs. An analysis on gestational weeks at the time of diagnosis and/or pregnancy termination was also accomplished.

Results: In 1994 – 2006, there was a total of 8,705 CAs prenantly diagnosed in the Czech Republic, which led to 6,118 pregnancy terminations. Out of all CAs, 54% were diagnosed by means of ultrasound, 44% cytogenetically and 2% by molecular genetic techniques. In chromosomal aberrations, between 20 – 30% were referred to karyotyping by an pathological ultrasound finding/marker (25% in Down syndrome). The highest rate of prenatal diagnosis was in anencephaly (100%), omphalocele (100%), gastroschisis (92%), spina bifida (81%), Down syndrome (74%), congenital hydrocephalus, omphalocele (87%), cystic kidneys (59%), renal agenesis (20%) and diaphragmatic hernia 35%. The mean gestational week at the time of diagnosis was 19.5 for total birth defects and 20.1 for pregnancy termination. Corresponding values (diagnosis x termination) in gestational weeks were as follows: Anencephaly 17.2 and 17.8, spina bifida 19.3 and 19.7, omphalocele 17.6 and 18.0, gastroschisis 19.0 and 18.7 and Down syndrome 19.8 and 20.4.

Conclusions: In the period under the study, the number of prenatally diagnosed cases was increasing and the number of diagnosed, but non-terminated cases increased as well (late diagnosis, multiple pregnancies i. a.). The rate of prenatal diagnostics varied in the study period mostly between 50 – 100% according to the type of defect. A proportion of ultrasound techniques also increased in this period along with their contribution to further diagnostics (karyotyping e. g.). Prenatal diagnostics also helps in finding associated defects and deciding on a prompt and adequate prenatal health care in compatible defects with consequences on perinatal mortality and morbidity rates.

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