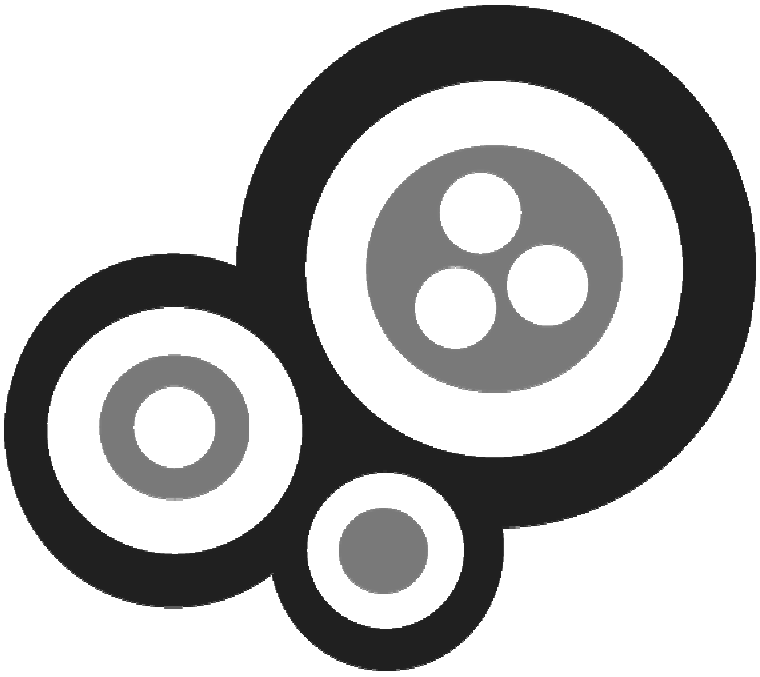

BREAKOUT 4

BIRTH DEFECTS SURVEILLANCE AND RESEARCH

Moderators:
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OMNI-NET BIRTH DEFECTS UKRAINIAN CONSORTIUM: IMPACT AND LESSONS LEARNED FROM "DE-CENTRALIZED" STRATEGIES

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Background: Following the Chornobyl disaster (1986), it was determined that the regions most impacted by ionizing radiation were in the Northwest of Ukraine. Public concerns about the potential teratogenic and genetic impacts of Chornobyl were acute. In 1991, Ukraine separated from the Soviet Union and in 1996 our program was launched in response to a suggestion from the then Minister of Health, Dr. Andryj Serdyuk. The planning, implementation and results are reported in J Appl Genet 47(2), 2006, pp.143-149; the program's website (http://ibis-birthdefects.org/) and 12 companion abstracts.

Method: Core strategies stressed include: 1, implementations in provinces (oblasts); 2, birth defects (BD) teams inclusive of medical geneticists, neonatologists, English-competent information - telecommunications coordinators and other specialists; 3, stress on links and partnerships with local, regional, national and international counterparts including parental support groups.

Results: Initial international funding of a Ukrainian-American BD program evolved into the current OMNI-Net partnerships formalized in 2004 as a Ukrainian not-for-profit organization registered in Kyiv. Resource centers (RC) sustain population-based neonatal as well as population-based BD registries in five regions and are located in provincial capital cities (Lutsk, Rivne, Khmelnytsky, Kherson, Simferopil). Currently, a population-based pregnancy registry is under development in Rivne. OMNI-Net partners rely on advice and exchanges with experts from Ukraine, UK, France, Italy and the U.S. BD data is reported to the European Registries of Congenital Anomalies and the International Clearinghouse BD Surveillance and Research Systems. Social and public health impact of OMNI-Net is illustrated by the creation by the city of Rivne of a model center for early interventions for infants with special healthcare needs, which offers its services to children identified by an international research program focused on alcohol impacts on the unborn.

Conclusion: OMNI-Net demonstrates the capacity and effectiveness of provincial healthcare systems to engage in complex partnerships and to enhance BD care and prevention policies. OMNI-Net experience with "de-centralized" province centered implementations offers an alternative to "capital city centered" international initiatives that may be of interest to other Eastern European countries.

ROLE OF THE HUNGARIAN CONGENITAL ABNORMALITY REGISTRY IN PREVENTION OF BIRTH DEFECTS

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Congenital anomalies (CAs) have great importance for public health. The estimated prevalence of CAs is 6-7% and they constitute the major cause of fetal death, perinatal and infant mortality, childhood morbidity, and long-term disability. Complete recovery is rare for major defects, and therefore, the optimal solution is the prevention.

The Hungarian Congenital Abnormality Registry (HCAR) collects, records, and keeps personal and health data of cases with CAs diagnosed until the age of one year (including stillbirths and terminations) covering all births in Hungary: approximately 100.000 births annually. Reporting CAs is compulsory by medical doctors.

The aims of the HCAR are: to assess the frequency of each CA; to help establish the number of handicapped persons; to promote primary prevention; to provide materials for scientific research; and to enable international cooperation.

The HCAR analyzes the spatial distribution and time-trend for some selected CAs. We present the results of our analysis on the prevalence of two CAs, which were in the focus of the past couple of years.

The prevalence of neural tube defects (NTDs) among live births declined during 1996-2006, but when taking into account terminations, it was around 6.5‰, and did not change much over time. The effectiveness of prenatal diagnosis in detecting NTDs, which might influence the rates, did not change much in the examined period, either.

Considering the prevalence of isolated cleft lip with or without cleft palate (0.3‰) and posterior cleft palate (0.7‰) during 2000-2006, there was a slightly increasing trend for both types of clefts.

Significant proportion of some congenital abnormalities (among them NTDs) could be prevented by periconceptional folic acid/multivitamin supplementation. Folic acid supplementation has been recommended in Hungary since the early 1990s, but it does not seem to influence the prevalence of NTDs and isolated orofacial clefts. Based on our previous studies, folic acid supplementation during the periconceptional period is low. Practical realization and working out the method of primary prevention is an actual national and public health care task in Hungary.

Summary: The HCAR provides a centralized surveillance, and can play a significant role in monitoring and supporting prevention efforts of CAs in Hungary but more resources are needed to expand its activities.

CONGENITAL MALFORMATIONS IN CHILDREN LIVE BORN DURING 1998-2002 IN POLAND - DATA FROM THE POLISH REGISTRY OF CONGENITAL MALFORMATIONS (PRCM)

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AIMS: The aim of this study was to determine the prevalence and structure of major congenital malformations (CM) identified in live born children aged 0-2 years in Poland during 1998-2002 and reported to the Polish Registry of Congenital Malformations (PRCM).

Methods: Population-based registry study of 2,902,452 live births (LB) of mothers resident in eight Polish provinces in years 1998-2000, nine provinces in 2001 and eleven provinces in 2002. Main outcome measures were: total LB prevalence of major CM, LB prevalence of selected groups and subgroups of CM (according to ICD-10 coding system), secular trends and comparison with EUROCAT data.

Results: The total number of live born children with major CM in the area covered by the PRCM during 1998-2002 was 17,781, giving a total prevalence of 197 per 10,000 LB. Significant variation in rates of several CM between the PRCM and the EUROCAT was present: prevalence of spina bifida in LB in Poland was 6.21 (significantly higher than its prevalence in total births - LB and stillbirths - in the EUROCAT), anencephaly - 1.52 (more than threefold higher prevalence in LB and significantly lower prevalence in total births in comparison with the EUROCAT), cleft palate - 7.01, cleft lip with or without cleft palate - 10.74, polydactyly - 11.1, upper limb reduction defects - 4.78 (prevalence of all these malformations is significantly higher than their prevalence in total births in EUROCAT), Down syndrome - 15.07 (significantly higher than its prevalence in LB in EUROCAT and lower than its prevalence in total births in the EUROCAT).

Conclusions: The study presents the very first data on CM in Poland, collected according to recognized standards using population based registry. Differences in prevalence rates of selected CM in LB between the PRCM and the EUROCAT can be partially explained by ascertainment variation as well as Polish law regulations concerning termination of pregnancy. Higher prevalence rates of selected CM in LB in Poland than in total births in EUROCAT countries require further analysis, including detailed geographic distribution and suspected risk factors.

Key words: congenital malformations, registry, epidemiology

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FOLLICULAR CYST OF OVARY, CLOMIPHENE TREATMENT AND THE RISK OF NEURAL-TUBE DEFECTS

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An association was found between preconceptional follicular cyst of ovary (FCO) in pregnant women and a higher risk for neural-tube defects in their offspring, but this association was not confirmed after the inclusion of clomiphene treatment among confounders. Another association was found between clomiphene treatment and a higher risk for neural-tube defects as well, but this association was not confirmed after the inclusion of FCO among confounders. Thus, predisposition for FCO due to clomiphene treatment may have some role in the origin of neural-tube defects.

POLISH REGISTRY OF CONGENITAL MALFORMATIONS – BENEFIT FOR MEDICAL GENETICS

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Congenital malformations present one of the most important problems in medicine. In about 50% of the cases, etiology of congenital malformations remains obscure. However, in 80% of the malformations of known etiology, genetic factors play a vital role. Children with congenital malformations comprise one of the main group of the patients admitted to genetic clinics all over the world.

The Polish Registry of Congenital Malformations (PRCM), the largest EUROCAT registry to date, has operated since 1997. The PRCM is currently covering the whole area of Poland and monitoring over 370,000 births each year. The most important aims of the PRCM constitute improvement of genetic care in Poland and integration of the clinicians and the scientists dealing with genetic background of congenital malformations. To fulfill these and other objectives, PRCM developed special organization (for details see J.Appl.Genet. 46, 341-348, 2005).

The PRCM has several merits for medical genetics, such as: monitoring the prevalence of particular types of congenital malformations and some rare malformation syndromes, evaluation of the state of genetic care and the state of prenatal diagnosis, monitoring of folic acid supplementation in prevention of neural tube defects and some other malformations, evaluation of demand for genetic service in Poland. The PRCM is also used for research purposes, especially for investigation of molecular background of congenital malformations (limb malformations; genitourinary malformations). It is a starting point for many other research projects in medical genetics (partner in SLOS, NBS, CdLS projects) and is also used for disseminating the knowledge on clinical genetics and genetic counseling among the physicians and society.

Conclusion: the PRCM proves that although the registry of congenital malformations is a real challenge, it is worth trouble, since the advantages for medical genetics cannot be overestimated.

** Wiśniewska K, Mejnartowicz J, Czerwionka-Szaflarska M, Gajewska E, Godula–Stuglik U, Kondała-Chojnacka A, Kornacka M, Krawczyński M, Sawulicka-Oleszczuk H, Rusin J, Stańczyk J, Szwałkiewicz-Warowicka E, Walczak M, BrzozowskaD, Glazar R, Krawczyński MR, Wierzba J, Chrzanowska K, Midro A, Sąsiadek M, Zajączek S, Kałużewski B, Lassota M, Pyrkosz A, Jakubowski L, Bocian E, Obersztyn E, Zaremba J,*
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INFANT MORTALITY DUE TO BIRTH DEFECTS IN THE CZECH REPUBLIC IN 1994 – 2006

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Aim and type of study: A retrospective data study with an analysis of pre- and postnatal occurrence of selected types of birth defects in the Czech Republic during 1994–2006 period. An analysis of infant mortality rate in children with selected types of birth defects during their first year of life.

Methodology: Data on prenatal diagnostics were obtained from particular departments of medical genetics. Data on birth defects incidences were obtained from National Health registers - Institute of Health Information and Statistics (National Register of Congenital Anomalies and National Newborns Register) from the 1996–2006 period were used. A case analysis of incidences of 14 types of pre- and postnatally diagnosed birth defects was performed for the period 1994–2006. In postnatally diagnosed cases, an analysis of survival and extinction of particular types of defects during the first year of life was also performed.

Results: During 1994–2006, there were 1 132 567 children born in the Czech Republic. Out of this number, more than 42 000 children were born with at least one birth defect in this period (mean 339 per 10 000 live births). Including prenatally diagnosed cases, we registered 345 cases of anencephaly, 515 spina bifida, 107 encephalocele, 538 cases of congenital hydrocephalus, 314 omphalocele and 361 cases of gastroschisis, 241 oesophageal defects, 302 anorectal malformations, 294 cases of diaphragmatic hernia, 606 renal agenesis/hypoplasia cases, 627 cases of cystic kidneys and 1 974 Down syndrome cases. According to a type of a defect, perinatal mortality can be as high as 100% in anencephaly, infant mortality ranges from 5 to 25%.

Conclusions: Birth defects present a major contribution to infant mortality and morbidity. Prematurity in a combination with a birth defect prolongs impatient days and increases a need of a total parenteral feeding in a newborn; it also makes a worse prognosis for the infants with birth defects. An improvement of prenatal diagnostics with an early reveal of the most severe types along with the associated ones could decrease incidences of such cases. A progress in surgical techniques and a special neonatal and infant intensive health care would also contribute an improvement of survival.

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