



## **POSTER SESSION II**





## P - II/1

### **SURVEY OF PRE-CONCEPTION HEALTH AND BIRTH DEFECTS PREVENTION KNOWLEDGE AND ATTITUDES IN UKRAINE**

<sup>1</sup>Erika Patskun, <sup>2</sup>Svitlana Kalynka, <sup>3</sup>Svitlana Onishchenko  
<sup>4</sup>Oksana Semenenko, <sup>5</sup>Lyubov Yevtushok, <sup>6</sup>Nataliya Zymak-Zakutnya

<sup>1</sup>*Uzhhorod OMNI-Net and Regional Medical Genetics Service*

<sup>2</sup>*Volyn OMNI-Net and Regional Medical Genetics Center*

<sup>3</sup>*Kherson OMNI-Net and Children's Regional Hospital*

<sup>4</sup>*Cherkasy Perinatal and Medical Genetics Regional Center*

<sup>5</sup>*Rivne OMNI-Net and Regional Diagnostic Center*

<sup>6</sup>*Khmelnytsky OMNI-Net and City Perinatal Center  
Ukraine*

**Background:** To assess knowledge and attitudes regarding pre-conception health and birth defects prevention in Ukraine, we initiated a pilot survey in six heterogeneous regions (northwest, Rivne and Volyn oblasts; west, Transcarpathia oblast; south, Kherson oblast; and central-west, Khmelnytsky and Cherkasy oblasts).

**Method:** A questionnaire was designed by OMNI-Net partners with consideration to previous surveys conducted by the March of Dimes and other agencies. Pregnant women seeking family planning or medical genetic services were asked to volunteer anonymously information about their ethnicity, health, previous pregnancies and negative pregnancy outcome risk factors. Information was recorded by health care personnel.

**Results:** Preliminary analyses indicate that pregnant women are willing to provide information more readily about medical risk factors and less so about risky lifestyles. Most pregnancies were planned (81%); 27% consulted a physician; 68% knew about the birth defects preventive effects of folic acid; 10% smoked before pregnancy; 5% did when already pregnant; 16% consumed alcohol and most of believed that in small quantities it was harmless.

**Conclusions:** OMNI-Net partners concluded that the preliminary data warrants small adjustments to the contents of the inquiry protocol and that it should be implemented in multiple regions because the information will be valuable to design context specific approaches to improve pre-conception health and prevention of birth defects.



---

P - II/2

**NEUROTOXIC EFFECTS IN HUMAN FETUSES EXPOSED TO LARGE DOSES OF  
TARDYL AS THE RESULT OF SUICIDE ATTEMPT DURING PREGNANCY**

Dora Petik

*Szent Istvan Hospital, Budapest, Hungary*

The objective of the study was to evaluate the effects on fetal development of very large doses of a drug combination (amobarbital, glutethimide and promethazine in Tardyl®, one of the most popular drug for insomnia in Hungary) that were used for suicide attempts during pregnancy. Pregnant women who attempted suicide with drugs and admitted to Department of Toxicology Internal Medicine, Korányi Hospital from Budapest and surrounding region including 3 million people. Congenital abnormalities, mental retardation, intrauterine development based on pregnancy age at delivery and birth weight, cognitive-behavioral status were compared in exposed children born to mothers who attempted suicide with Tardyl® during pregnancy and in their sib controls. 74 of the 1044 pregnant women who attempted suicide during pregnancy with drugs between 1960 and 1993 used Tardyl® for self-poisoning. Of these 74 women, 27 delivered live-born babies. The dose of Tardyl® used for suicide attempt was 24-fold of clinical dose. Of the 27 exposed children, 2 (7.4%) had congenital abnormalities: undescended testes and fetal alcohol syndrome. Of 48 sibs, 3 (6.3%) had CAs (oesophageal atresia, undescended testes, fetal alcohol syndrome). Thus, there was no higher risk for congenital abnormalities after the large doses of Tardyl®. However, of 27 exposed children, 6 (22.2%) were mentally retarded, and an additional 2 exposed children had a very low cognitive status (their IQ was about 75). These exposed children were born to mothers who attempted suicide with Tardyl® between the 13th and 21st postconceptional week. Two other exposed children had moderate behavioral deviation. If we evaluate these exposed children together, compared with their unexposed sibs, the neurotoxicity of Tardyl® associated with 22-fold higher risk for mental retardation and behavioral deviation (10 vs. 1, OR with 95% CI: 27.6, 3.3–232.4). The conclusion of the study was that the large doses of Tardyl did not increase the risk for congenital abnormalities, however, the interaction of amobarbital, glutethimide and promethazine associated with a high risk for mental retardation, particularly when exposure occurred during the second trimester of pregnancy. Finally our experiences have shown the feasibility and benefits of self-poisoning model for the estimation of human teratogenic risk of drugs.



**P - II/3**

**COMPARISON OF BIRTH DEFECTS DETECTION  
TO ANTENATAL MONITORING**

**<sup>1</sup>Manuela Russu, <sup>2</sup>Maria Paun, <sup>1</sup>C. Posea, <sup>1</sup>H. Rahimian, <sup>1</sup>J.A Marin**

***"Carol Davila" University of Medicine and Pharmacy***

***<sup>1</sup>"Dr. I. Cantacuzino" Clinic of Obstetrics and Gynecology***

***<sup>2</sup>"Dr. I. Cantacuzino" Department of Neonatology***

***Bucharest, Romania***

**Objective:** Neonatal comparison of birth defects to antenatal detected abnormalities

**Material and method:** comparison of birth defects registered in the Department of Neonatology to the congenital abnormalities detected by ultrasound monitoring around 20<sup>th</sup> week gestation, or to serological markers from the 13<sup>th</sup> week's gestation. There are analyzed: maternal age, GA at delivery, smoking, alcohol, illicit drug consumption, preconceptional use of folic acid, vitamins or other drugs/procedures, rate of TORCH syndrome, birth weight, perinatal outcome.

**Results:** During January-December 2007, in the Neonatology Department there were recorded 54 malformed alive new born among 2656 deliveries. There were 6 cases Down Syndrome (2 discovered by ultrasound near term), 5 neural tube defects (2 spina bifida, 1 spina bifida and encephalocele, 2 hydrocephalia - not recorded at ultrasound), 11 cardiovascular abnormalities (3 ASD, 3 VSD, 2 large vessels transposition plus VSD, 1 common arterial trunk - half registered at ultrasound); 11 digestive abnormalities: 3 omphalocele (1 ultrasound discovered at the onset of labour), 2 diaphragmatic hernia (ultrasound depicted), 1 keylognato-palatoskisis, 1 anal imperforation, 1 stomach and 1 esophagus atresia; 7 genital&urinary tract defects (2 kidneys cysts, 5 hypospadias - all registered ultrasonical); 16 limbs defects (13 club foot, 1 hip dysplasia; 1 osteogenesis imperfecta & 1 arthrogriposis ultrasound depicted); 9 abnormalities of hands & digits (1 complex hand malformation, 6 polydactyly of the hand, 1 with cutaneous syndactyly (all not ultrasound evaluated).

We registered: average maternal age:  $26.9 \pm 4.77$  yrs; gestational age at delivery: 28 to 40 weeks; 2 cases of amniocentesis for CMV infection (negative), no chorionic villous sample; alcohol consumption >50 ml/day in 4 cases with limb defects; no recognized illicit drug consumption, smoking of >5 cigarettes in 10 cases; 5 cases of preconceptional folic acid use; birth weight:  $3250 \pm 537.27$  g (limits 900 to 4100 g); 6 neonatal death from 6 hours (neural tube defects, complex cardiovascular defects) to 10 days (after cardiac surgery).

**Conclusion:** During 12 months between 2656 registered deliveries are recorded 54 malformed new life born babies, 21 are antepartum depicted by ultrasound, amniocentesis and serological markers for aneuploidies. A significant majority (35-67%) were not antenatal monitored for malformations, either preconceptional treated with folic acids, vitamins. Gestational age and birth weight in severe/complex malformations are influenced; the short time from presentation to delivery was not favorable medical/surgical interventions.



---

P - II/4

**IMPROVING SURVIVAL OF INFANTS WITH BILIARY ATRESIA IN ATLANTA**

<sup>1,2</sup>Csaba Siffel, <sup>1</sup>Chengxing Lu, <sup>1</sup>Assia Miller, <sup>1</sup>Richard Olney, <sup>1</sup>Adolfo Correa

<sup>1</sup>*National Center on Birth Defects and Developmental Disabilities,  
Centers for Disease Control and Prevention*

<sup>2</sup>*Computer Sciences Corporation, Atlanta, Georgia, USA*

**Background:** Biliary atresia (BA) is a progressive condition manifested shortly after birth that without surgical intervention leads to early death. It has been suggested that survival of infants with BA has improved in recent years, but population-based information on long-term survival and prognostic significance of demographic and clinical characteristics at birth is limited. We aimed to examine the long-term survival of infants with BA and whether demographic and infant clinical characteristics influence such survival.

**Methods:** The study cohort consisted of infants with extrahepatic BA (n=75) born between 1979 and 2003 and identified through the Metropolitan Atlanta Congenital Defects Program. Vital status was ascertained through 2004 through linkage with state vital records and the National Death Index (19 deaths). We estimated Kaplan-Meier survival probabilities stratified by birth period, sex, race, maternal and paternal age, socioeconomic status, birth weight, gestational age, method of delivery, pediatric hospital admission, presence of multiple major defects, and time of diagnosis. Adjusted hazard ratios (aHR) and 95% confidence intervals (CI) were estimated using Cox proportional hazards models.

**Results:** The overall survival probability of infants with BA (71%) significantly improved over time: 1979-1984, 40%; 1985-1990, 72%; 1991-1996, 87%; 1997-2003, 89% (test for trend: p=0.0036). Infants born in recent time periods (1994 and later) had better survival than those born before 1994 (aHR=0.25, CI 0.82-0.76). Time of diagnosis seemed to affect survival: survival was better when the diagnosis was made less than 60 days after birth (aHR=0.34, CI 0.13-0.84). None of the other factors examined showed a significant effect on survival.

**Conclusions:** Survival probability of infants with BA improved during the last three decades in metropolitan Atlanta. Time of diagnosis may be an important prognostic factor but further studies with larger sample sizes are needed to corroborate this finding.



**P - II/5**

**PERINATOLOGIC OUTCOMES OF CHILDREN BORN AS A RESULT OF  
IN VITRO FERTILISATION IN THE CZECH REPUBLIC: PERINATAL  
AND NEONATAL MORBIDITY AND MORTALITY**

<sup>1,2</sup>Antonin Sipek, <sup>2</sup>A. Mardesic, <sup>1,3</sup>J. Horacek, <sup>1,2</sup>V. Gregor, <sup>5</sup>A. Sipek jr, <sup>6</sup>P. Langhammer

<sup>1</sup>*Department of Medical Genetics, Thomayer's University Hospital, Prague, Czech Republic*

<sup>2</sup>*Sanatorium Pronatal, Prague, Czech Republic*

<sup>3</sup>*Clinic Gennet, Prague, Czech Republic*

<sup>4</sup>*Chair of Medical Genetics, Postgraduate Medical Institute, Prague, Czech Republic*

<sup>5</sup>*1st Medical Faculty, Charles University, Prague, Czech Republic*

<sup>6</sup>*Institute of Health Information and Statistics, Prague, Czech Republic*

**Aim and type of study:** Neonatal data on a cohort of 22,949 children born as a results of In Vitro Fertilisation (IVF) in the Czech Republic (CR) compared with a cohort of 649,595 naturally conceived (NC) children. Analysis of perinatal and neonatal morbidity and mortality according to birth weight, gestational age, and fetal number.

**Material and methods:** A retrospective epidemiological analysis of perinatal data from the National Registry of Births and the National Registry of Congenital Anomalies in CR during 2000–2006.

**Results:** During the period between 2000 and 2006, there were 22,949 children born as a result of IVF with a sex ratio M/F of 1.03. From this total number there were 46.88% twins and 53.12% singletons. During the same period 649,595 NC children were born, with a sex ratio M/F of 1.06. From this total number there were 96.94% singletons and 3.06% twins.

The incidence of stillbirths in the IVF/non-IVF cohort was 4.14‰/2.13‰ for the whole cohort, 5.13‰/2.03‰ for the singletons, 3.09‰/4.90‰ for the twins, 3.73‰/2.37‰ for the singletons with CA and 7.34‰/6.49‰ for the twins with CA. Perinatal mortality in the IVF/non-IVF cohort was 8.78‰/3.26‰ for the whole cohort, 6.82‰/2.91‰ for the children without CA, 23.44‰/12.03‰ for the children with CA, 6.67‰/3.03‰ for singletons and 11.49‰/10.24‰ for twins.

In the case of neonatal morbidity, the most frequent complications in the singletons in the IVF cohort were from the P50-P61 group according to ICD X (Haemorrhagic and haematological disorders). The highest perinatal mortality was found in the P90-P96 group (Other disorders originating in the perinatal period) – 89.77‰ and in the P20-P29 group (Respiratory and cardiovascular disorders) – 32.75‰. In the twins from the IVF cohort, the most frequent complications were from the P00-P04 group (Foetus and newborn affected by maternal factors) and from the P05-P08 group (Disorders related to length of gestation and foetal growth).

**Conclusion:** In the IVF children cohort we found a higher incidence of stillbirths, perinatal mortality and infant mortality, compared to the control cohort of NC children.



**P - II/6**

**PERINATOLOGIC OUTCOMES OF CHILDREN BORN AS A RESULT OF  
IN VITRO FERTILISATION IN THE CZECH REPUBLIC:  
CONGENITAL ANOMALIES AND BIOSOCIAL FACTORS**

<sup>1,2</sup>Antonin Sipek, <sup>2</sup>A. Mardesic, <sup>1,3</sup>J. Horacek, <sup>1,2</sup>V. Gregor, <sup>5</sup>A. Sipek jr, <sup>6</sup>P. Langhammer

<sup>1</sup>*Department of Medical Genetics, Thomayer's University Hospital, Prague, Czech Republic*

<sup>2</sup>*Sanatorium Pronatal, Prague, Czech Republic*

<sup>3</sup>*Clinic Gennet, Prague, Czech Republic*

<sup>4</sup>*Chair of Medical Genetics, Postgraduate Medical Institute, Prague, Czech Republic*

<sup>5</sup>*1<sup>st</sup> Medical Faculty, Charles University, Prague, Czech Republic*

<sup>6</sup>*Institute of Health Information and Statistics, Prague, Czech Republic*

**Aim and type of study:** Neonatal data on a cohort of 22,949 children born as a result of In Vitro Fertilisation (IVF) in the Czech Republic (CR) compared with a cohort of 649,595 naturally conceived (NC) children. Analysis of perinatal data – birth weight, gestational age, maternal age, fetal number, sex ratio and the occurrence of congenital anomalies (CA).

**Results:** During this period, there were 22,949 children born as a result of IVF with a sex ratio M/F of 1.03. From this total number there were 46.88% twins and 53.12% singletons. During the same period 649,595 NC children were born, with a sex ratio M/F of 1.06. From this total number there were 96.94% singletons and 3.06% twins. In IVF/non-IVF cohort the average gestational age in weeks was 37.32/39.37; the average birth weight was 2796.72 g/3321.71 g and the average maternal age in years was 30.66/27.43. Prematurity in the IVF/non-IVF cohort (not distinguishing the fetal number) was 1.80%/0.27% for prematurity below the 28<sup>th</sup> week and 19.62%/3.58% for prematurity between the 28<sup>th</sup> and 35<sup>th</sup> week. Low birth weight (1000–2499 g - LBW) in the IVF/non-IVF cohort was found in 27.72%/5.91% of the whole cohort. The incidence of CA was 464.06 in 10,000 live births in the IVF children cohort and 369.27 in 10,000 live births in the NC children cohort. In the IVF group, the incidence of CA per 10,000 live births was 544.06 in singletons and 362.80 in twins. In the NC children cohort, the incidence of CA per 10,000 live births was 349.95 in singletons and 425.57 in twins.

**Conclusion:** In the cohort of IVF children we found a significantly lower birth weight and gestational age at birth, further a higher occurrence of twins and increased maternal age. After dividing the cohort into singleton and twin groups - we found no statistically significant differences in birth weight and gestational age between the IVF and NC children. In the IVF cohort there was a higher occurrence of some CA of the central nervous system (CNS), heart, lower respiratory tract and oesophagus, selected anomalies of female and male genital organs, selected anomalies of extremities and balanced chromosomal rearrangements.





**P - II/7**

**OCCUPATIONAL EXPOSURES AND BIRTH DEFECTS  
IN THE SLOVAK REPUBLIC**

**Elena Szabova, Dagmar Zeljenkova, and the \*SMU collaborative group**

*Slovak Medical University, Bratislava, Slovak Republic*

**Introduction:** Case-control study to collect workplace and other data in mothers of children with a select list of congenital malformations (CM) in Slovakia. Using standardized questionnaires, mothers of offspring with at least one major CM of multifactorial origin, and two mothers of healthy control children matched for place and time of birth were interviewed to obtain information on occupation during pregnancy. During 5 years physicians from maternity hospitals and obstetric clinics covering about one third (cca 20,000 per year) of total births in the Slovak Republic participated in the study and contributed cases during the study period.

**Material and Methods:** According to the common European protocol, cases were defined as alive or stillborn babies, or fetuses resulting from a therapeutic abortion, with a confirmed major congenital morphological defect, discovered prenatally, at birth, or within the first week of life and of unknown cause. Congenital malformations of known environmental origin or associated with chromosomal or monogenic syndromes were excluded. Occupations and industrial activities were coded using the ILO codes for occupations and the UN codes for industrial activity.

**Results:** Cases were more likely than controls to have reported any infection before and in the first trimester of pregnancy, mainly due to reporting of influenza. This is reflected in increased use of anti-infection drugs before pregnancy among cases and during pregnancy. As expected mean birth weight, and duration of gestation was lower in cases than in controls. Cases were significantly more likely to be unemployed than controls during pregnancy. There were no differences between cases and controls for microbial contaminants, biocides, antibiotics, solvents, food products, inks, etc. However cases have significantly elevated crude odds ratios for all organic dust, cleaning agents, inorganic acids and hypochlorites.

**Conclusions:** Our preliminary findings do not suggest a significant risk of occupational factors to birth defects, however, further in-depth analysis is needed to clarify the significant differences of some risk factors at the working environment.

---

*\*Members of the SMU collaborative group: E. Véghová, S. Plačková, D. Brašeňová, E. Nesčáková, A. Číková, E. Bieliková, E. Horáková, D. Mračnová, I. Kolosziová, R. Sečkárová, M. Borgulová, I. Moravcová, P. Košík, M. Kvasnicová, E. Čarnoká, M. Molčáková, H. Sedláčková, D. Dragijská, N. Mišovicová, M. Hodálová, E. Koubeková, V. Ostatník, E. Nemcová, K. Tancárová, J. Juričová, J. Koblík, Šmelková, K. Tancárová, M. Sopková, Pšeničková, K. Sterczová, M. Vasil, E. Nebesňáková, O. Pirochová, E. Gnothová*



**P - II/8**

**THE OUTCOME OF PATIENTS WITH SPINA BIFIDA TREATED  
IN LVIV CITY CHILDREN'S HOSPITAL IN 2005-2007**

**\*Yuriy Korzhynskyy, K. Bihanych, Olga Tychkivska, A. Saviv**

***\*Danylo Halytskyy Lviv National Medical University, Lviv, Ukraine***

Spina bifida is one of the most deleterious birth defects. The vast majority of all the infants born with spina bifida in Lviv region are transferred to Lviv City Children's Hospital.

An *objective* of present study was to analyze the outcome of infants born with spina bifida admitted to the City Children's Hospital during 2005-2007 years.

*Methods:* Case histories of patients with spina bifida from NICU, division for sick neonates, neurology department and neurosurgery division of surgical department of Lviv City Children's Hospital were reviewed as well as their outpatient medical cards. These data were supplemented with interviews with parents of the patients. The outcome of the patients as it looked in April 2008 was evaluated.

*Results:* 41 patients admitted for the first time with spina bifida to Lviv City Children's Hospital were identified. They included Arnold-Chiari malformation type II – 18 cases, meningocele – 11 cases, meningocele – 12 cases. Surgical closure of meningocele and meningocele was performed in 23 children (6 of them required also ventriculoperitoneal shunting). In one child only ventriculoperitoneal shunting was performed without closure of the defect. Only one child was operated on the 1<sup>st</sup> day of life, the other – at different age during the first year of life. In one anecdotic case a girl with small meningocele was operated at 13 years of life. 17 children were not operated because of different causes.

7 out of 17 non-operated children died because of complications. Survivors are significantly handicapped. In the group of children who underwent surgery one patient died. In 16 out of 23 children who survived after surgery we observed lower extremities weakness, bowel and bladder dysfunction, mental retardation or combination of these symptoms; 6 children mentally and neurologically did well.

None of the mothers of the patients took prophylactic folate before or during pregnancy.

*Conclusions:* most of the followed spina bifida patients had unfavorable outcome. Efforts should be made towards earlier surgical repair of skin defect in patients with spina bifida. The opportunity of preconceptional folic acid prophylaxis was missed in studied cases. That is why prophylaxis on the population level may be a good alternative.



**P - II/9**

**DYSMORPHOLOGY PLATFORM – A WEB-BASED ELECTRONIC SYSTEM  
OF DYSMORPHOLOGY CONSULTATION IN POLAND**

<sup>1</sup>Marzena Wisniewska, <sup>1</sup>Magdalena Badura-Stronka,  
<sup>2</sup>Bartosz Brodecki, <sup>2</sup>Jacek Richter, <sup>1</sup>Anna Materna-Kirylyk, <sup>2</sup>Jerzy Brzezinski,  
<sup>1</sup>Anna Latos-Bielenska

*<sup>1</sup>Department of Medical Genetics, Poznan University of Medical Sciences, Poznan, Poland*

*<sup>2</sup>Institute of Computing Science, Poznan University of Technology*

Computer-based diagnosis is a modern useful tool for disease diagnosis, especially regarding genetic syndromes. Correct diagnosis in malformed patients is very important for genetic prognosis and counselling in each concrete family.

Dysmorphic syndromes are a large group of difficult and often undiagnosed cases in clinical genetics. Some of them are quite rare and it happens that although all possible methods of diagnosis have been done, they are still undiagnosed. Syndrome identification and correct diagnosis is one goal of genetic evaluation because it permits precise genetic counselling. Correct diagnosis is the cornerstone of genetic counselling and patient management.

In this paper we present the Dysmorphology Platform which is a web-based electronic system, created in Poland. The aim of this Platform is to provide a mutual consultation of clinical geneticists to diagnose unknown dysmorphic cases. It enables dysmorphologists throughout Poland to submit undiagnosed cases to other dysmorphologists for their opinion and diagnosis. The Dysmorphology Platform is available only for chosen group of geneticists and all the data including photographs of patients are fully protected.

We hope that the Dysmorphology Platform will be a great help for dysmorphologists in their work and it will develop better, faster and so necessary cooperation in making diagnosis of dysmorphic syndromes among geneticists in Poland. This computer-assisted diagnosis could be the fastest way to have diagnosis in rare genetic syndromes.

*The work is a part of the project of the Polish Registry of Congenital Malformations (PRCM) and is financed by the Polish Ministry of Health.*



## P - II/10

### **FOLIC ACID SUPPLEMENTATION AND RISK OF ISOLATED CONGENITAL MALFORMATIONS (DATA FROM THE POLISH REGISTRY OF CONGENITAL MALFORMATIONS - PRCM)**

<sup>1,2</sup> Katarzyna Wisniewska, <sup>1</sup>A. Materna-Kiryluk, <sup>1</sup>M. Badura-Stronka, <sup>1</sup>J. Mejnartowicz, <sup>3</sup>B. Wieckowska, <sup>2</sup>J. Wysocki, <sup>1</sup>A. Latos-Bielenska and other members of the \*PRCM Working Group

*University of Medical Sciences, Poznan, Poland*

*<sup>1</sup> Department of Medical Genetics*

*<sup>2</sup> Department of Preventive Medicine*

*<sup>3</sup> Department of Computer Science and Statistics  
Poland*

Congenital malformations represent a considerable public health problem by virtue of their mortality, morbidity, social cost, and human suffering. The discovery that folic acid supplementation before pregnancy or in early pregnancy reduces the risk of several congenital malformations is one of important public health advances of recent years. Preventive effect of folic acid on the occurrence of neural tube defects is well documented. Similar protective effect has been suggested for other congenital malformations, for example congenital heart defects, orofacial clefts, limb reduction defects, anal atresia, and urinary tract defects, but the evidence for this is not as consistent or as strong as for neural tube defects.

The aim of the study was to analyze the association between the risk of selected isolated congenital malformations and maternal use of folic acid ( $\geq 0.4$  mg/day), alone or in multivitamin supplements, before conception and during the first trimester of pregnancy.

The study was based on the data from the Polish Registry of Congenital Malformations (PRCM) and included 8,346 mothers of live born infants with major isolated congenital malformations in the years 2001-2006 in thirteen administrative regions of Poland.

Folic acid supplementation before conception and during the first trimester of pregnancy was associated with decreased risk of isolated neural tube defects, congenital heart defects, hypospadias, renal agenesis or hypoplasia, and polydactyly. No significant relationship between maternal folic acid intake and the following congenital malformations was detected: hydrocephaly, cleft palate, cleft lip with or without cleft palate, oesophageal atresia, atresia or stenosis of small and/or large intestine, ano-rectal atresia or stenosis, polycystic kidney diseases, atresia or stenosis of ureter, syndactyly, limb reduction defects, diaphragmatic hernia, omphalocele and gastroschisis.

Our results confirm the observation of other authors that the primary prevention of some congenital malformations by folic acid supplementation is of great importance for public health. Knowledge about folic acid role in preventing congenital malformations should be spread among young people.

*The Project is financed by the Polish Ministry of Health.*

---

\*A. Balcar-Boron, M. Borszewska-Kornacka, M. Czerwionka-Szaflarska, E. Gajewska, U. Godula-Stuglik, M. Krawczynski, J. Limon, J. Rusin, H. Sawulicka-Oleszczuk, J. Stanczyk, E. Szwałkiewicz-Warowicka, M. Walczak



---

**P - II/11**

**SEASONAL VARIATION OF NTD RATES IN RIVNE – UKRAINE**

**Bohdana Yevtushok**

*Medical Student, Ternopil Medical University, Ukraine*

**Background:** Neural Tube Defects (NTD) are reduced by supplements of folic acid, which sustains the view that folate deficiency may be a cause of these highly lethal and unusually common malformations in Ukraine, particularly in the Chornobyl impacted Ukrainian Polissia region.

**Method:** We analyzed estimated times of conception by season. The data analyzed was from Rivne: 217 NTD patients born between 2000-2006, of which 179 (82%) were isolated NTDs.

**Results:** Most conceptions of isolated NTD cases (29%) occurred during winter (December, January, February); 22% in spring; 22% in the summer (June, July, August); and 22% in the fall. The seasonality trend for complex NTDs was stronger, 39% and 26% in the winter – spring period and a decrease to 21% and 13% during summer – autumn.

**Conclusion:** The seasonal variations in prevalence of NTD rates have been noted in other regions of the world. Because the Polissia region in Rivne oblast, as Polissia regions elsewhere, is a unique ecological and demographic zone, the higher rates of NTD in the Rivne – Polissia population may also be elevated in Polissia populations in other areas of Ukraine and Belarus. The reliance of the “Polischuks” (woodland people) on home and locally grown nutrients (see companion abstract by L. Yevtushok), the seasonal NTD variations may be indicative, at least in part, of folate deficiencies related to the winter and spring seasons within the overall possible confounding effects of chronic low dose radiation since DNA repair is folate dependent.



## P - II/12

### DIETARY AND ACTIVITY PATTERNS AND IMPLICATIONS ON BIRTH DEFECTS IN THE CHORNOBYL IMPACTED RIVNE-POLISSIA REGION IN UKRAINE

<sup>1</sup>Lyubov Yevtushok, <sup>2</sup>K. Needham, <sup>3</sup>Serhiy Lapchenko, <sup>4</sup>Wladimir Wertelecki,  
<sup>2</sup>R. Garruto

<sup>1</sup>*Rivne OMNI-Net and Regional Diagnostic Center Medical Genetics Center, Ukraine*

<sup>2</sup>*State University of New York at Binghamton, USA*

<sup>3</sup>*Volyn OMNI-Net Center and Regional Children's Clinical Hospital, Ukraine*

<sup>4</sup>*Ukraine OMNI-Net and University of South Alabama, USA*

After the Chernobyl disaster in 1986, population-based birth defects monitoring was initiated in 2000 in five oblasts (provinces) in Ukraine. The northern half of Rivne province, a region known as Polissia, is contaminated by radiation. Systematic monitoring shows consistently high rates of neural tube defects and other developmental disorders. These observations prompted a dietary and subsistence activities survey about consumption of Cs137 contaminated foodstuffs, water, smoke, and agricultural and gardening exposure through dust and soil. Contamination levels measured from one up to 1590 Bq/l in milk, 129 Bq/kg in potatoes, and 146 Bq/kg in soil.

We report preliminary information provided by 99 of 350 surveyed pregnant women residing in the Polissia region of Ukraine. Information gathered concerned consumption of local and imported meat, dairy, fruit, vegetable, and grain products, as well as water use and beverages, including alcohol consumption during pregnancy. We also gathered information about activities likely to expose individuals to radiation through contact with soil, such as working in fields and collecting wild foods or firewood. Results indicate that most foods consumed in Rivne-Polissia are local or wild and thus contaminated with Cs-137. The government has recommended that milk be imported because of the risk of contamination of local milk. However, less than 5% of individuals reported consuming imported milk. All participants reported consuming local vegetables (none imported), and 97% of the major fruits consumed were locally grown. Furthermore, 73% reported obtaining water from highly contaminated wells or brooks, the rest consumed piped water. In addition, 30% drank alcoholic beverages often as homemade or local beer and liquor. Information about outdoor activities, particularly agricultural fields, and contact with contaminated soil or smoke from burning biomass. Overall, our data indicate that some people in Polissia consume diets that may be deficient in micronutrients, consume alcohol during pregnancy and are exposed to chronic radiation. The extent to which each may be involved in birth defects and developmental nervous system disorders in the Polissia region is unknown and calls for further investigations.



**P - II/13**

**PREDICTORS OF BINGE DRINKING DURING PREGNANCY  
AMONG WOMEN IN UKRAINE**

<sup>1</sup>Lyubov Yevtushok, <sup>1</sup>Svitlana Shevchenko, <sup>2</sup>Svitlana Onishchenko, <sup>2</sup>Iryna Drapkina,  
<sup>3</sup>Wladimir Wertelecki, <sup>4</sup>Ludmila Bakhireva, <sup>5</sup>Christina Chambers

<sup>1</sup>*Omni-Net Birth Defects Center, Rivne Clinical Diagnostic Center, Rivne, Ukraine*  
<sup>2</sup>*Omni-Net Birth Defects Center, Kherson Oblast Children's Hospital, Kherson, Ukraine*  
<sup>3</sup>*University of South Alabama, Mobile, AL, USA*  
<sup>4</sup>*University of New Mexico, Albuquerque, NM, USA*  
<sup>5</sup>*University of California, La Jolla, CA, USA*

**Background:** Fetal Alcohol Spectrum Disorders (FASD) are among the most pervasive childhood developmental disorders worldwide. A comprehensive program for prevention of FASD calls for targeted intervention/prevention among women in the preconception period who are current risky drinkers. An understanding of the characteristics of reproductive aged women who engage in risky drinking is essential for the development of a targeted prevention program, and these characteristics may vary by culture.

**Methods:** We screened pregnant women in two oblasts in Ukraine over a two year period and recruited 84 women who reported binge and/or frequent drinking early in pregnancy and 82 women who reported little to no alcohol consumption during early pregnancy. Using a standard in-person interview, we collected information on demographics, pregnancy and medical history, current and past maternal and paternal drinking habits, and knowledge of FASD. We compared these groups to identify predictors of heavy episodic or binge drinking in early pregnancy.

**Results:** Similar to findings in other countries, lower maternal socioeconomic status, tobacco use, unplanned pregnancy, no use of prenatal or multivitamins, and lack of accurate knowledge regarding the effects of alcohol on the developing fetus were all significant predictors of binge drinking by mothers early in pregnancy ( $p < 0.05$ ). Frequent and/or heavy paternal drinking was also strongly associated with maternal binge drinking. Fathers who drank 2-4 times per week, relative to those who drank less than 1 time per week, were 32 times more likely (95% CI 8.9-118.6) to have female partners who binge drank in pregnancy. Similarly, fathers who consumed 5 or more drinks per occasion, relative to those who drank only 1-2 drinks per occasion, were 39 times more likely (95% CI 12.2-124.7) to have female partners who binge drank in pregnancy.

**Conclusions:** These findings suggest that interventions among Ukrainian women who drink alcohol should be targeted to women in the preconception period with other high risk behaviors and poor knowledge regarding FASD, and that family drinking culture may represent another opportunity for intervention.

*This work was supported by a grant from NIAAA-U01-AA014835-05.*



## P - II/14

### **BANKING OF BIOLOGICAL AND ENVIRONMENTAL SAMPLES**

Dagmar Zeljenkova, Elena Szabova, Jevgenij Kovriznych

*Slovak Medical University, Bratislava, Slovak Republic*

Specimen banking is a complex program for systematic monitoring and long term archiving selected matrices under very low temperatures under exactly defined conditions. This is the condition for internationally accepted comparison and analyses of contamination trends. Method is based on systematic collection of selected samples in given intervals, kryogenic pretreatment of samples at place of their collecting, and its transport, kryogenic homogenization, preparation of samples and their long term archiving over liquid nitrogen level at temperatures under  $-160^{\circ}\text{C}$ . An important part is the complex analytical and statistical characterization of archived samples and undertaking retrospective analyses in exactly defined time intervals. Specimen banking is an important tool in contaminant monitoring and ecotoxicological and medical research for more than one decade.

Banking specimen plays an important role in evaluating trends in the population loads due to negative factors. It was shown that its application is not only possible but, also necessary to be used in the field of toxicological research, in the study of the biomarkers, mycotoxins and in the persisting endocrine disruptors.

The last few years have witnessed an important expansion of human DNA sampling and data collecting in order to exploit and study the genetic information collected. DNA analysis is an increasingly important source of medically useful information. Banking for the preservation of DNA needed for analysis at future time is becoming more widespread. DNA analysis for clinical purpose differs from many other clinical genetics tests in several ways: The long - term stability of DNA may permit questions to be answered later that were not envisioned at the time of its procurement.

Our Institute has collected samples through research activities and monitoring programs. They preserve a variety of environmental samples (placenta, mother blood, cord blood and breast milk). Our specimen bank has been built from large cohort studies provide the opportunity to design and carry out very cost-effective nested case control studies.

The logistics of shipment from the respective countries has been developed. Partners have been trained in samples collection in compliance with the internationally recognized standard operation procedures. The stored sample can be reanalyzed when upgraded or completely new analytical methods will be available. The stored samples can also be used for assessment of long term trends of environmental contamination. This approach is essential for evaluation of environmental influences on the health state of the population. Establishment of this cohort and the collection and storage of biological samples for later use will provide the basis for long-term follow-up and the application of technologies to be developed in the 21<sup>st</sup> century.





## P - II/15

### REDUCTION OF INFANT MORBIDITY AND MORTALITY – GASTROSCHISIS AS ONE INDEX

<sup>1</sup>Natalia Zymak-Zakutnya, <sup>1</sup>A. Ropotan, <sup>2</sup>I. Shumlyanski, <sup>3</sup>H. Vashchylin, <sup>4</sup>L. Lyatetska,  
<sup>5</sup>T. Tekuchenko, <sup>2</sup>V. Yenykeyeva, <sup>2</sup>Z. Sosynyuk, <sup>2</sup>L. Yevtushok, <sup>3</sup>S. Kalynka,  
<sup>4</sup>S. Onishchenko, <sup>5</sup>N. Afanasyeva, <sup>6</sup>W. Wertelecki

<sup>1</sup>*Khmelnysky OMNI-Net, City Children's Hospital and Perinatal Center, Ukraine*

<sup>2</sup>*Rivne OMNI-Net, Diagnostic Center,*

*Children's Hospital and Medical Genetics Department, Ukraine*

<sup>3</sup>*Volyn OMNI-Net and Regional Children's Territorial Medical Center, Ukraine*

<sup>4</sup>*Kherson OMNI-Net and Children's Regional Hospital, Ukraine*

<sup>5</sup>*Crimea Autonomous Republic OMNI-Net, Ministry of Health Medical Care of Child  
and Maternity Division and Medical Genetics Center, Ukraine*

<sup>6</sup>*Ukraine OMNI-Net and University of South Alabama, USA*

**Background:** Gastroschisis (GS) is a malformation that can often be detected prenatally and when treated early and effectively is often compatible with survival. The GS rates reported by OMNI-Net to EUROCAT for 2005-2006 were 6.27 per 10,000 live births. GS can serve as a sentinel anomaly to assess the effectiveness of prenatal diagnostic services, neonatal care and surgical interventions. This project, among others, seeks to provide input toward fact-based prospective enhancements of maternal child health policies seeking to reduce infant mortality, morbidity and developmental disorders in Ukraine. (See companion abstract by Korzhynskyy and by Afanasyeva).

**Method:** Analysis of 143 instances of GS recorded by OMNI-Net registries and the corresponding medical records and other sources of information.

**Results:** Preliminary analysis showed that in Ukraine GS is often detected prenatally (71%); termination of pregnancy due to GS is high (54% vs. 14% in Europe as per EUROCAT 2001-2006); GS, in live-born Ukrainian infants, is generally an isolated malformation (80%); and that in Ukraine GS is a highly lethal malformation (59% compared to 13% in the U.S.). The main factor contributing to the high mortality was delayed transfer of infants to specialized facilities followed by a high prevalence of post-surgical complications.

**Conclusion:** The preliminary analysis provides data confirming informal reports that hundreds of Ukrainian infant-lives can be saved with enhancements of existing infant transportation systems and a better regionalization and experience by pediatric surgical teams focused on the care of infants. Enhancements in this domain will also impact the care of infants with spina bifida, cardiac and other non-lethal malformations. Therefore, this and other companion studies by OMNI-Net will continue and be expanded to four additional oblasts. (See companion abstract by Korzhynskyy.)