

**2000-2001  
SUMMARY OF SCHOLARLY ACTIVITIES  
DEPARTMENT OF MEDICAL GENETICS**

**I. ARTICLES PUBLISHED IN BOOKS AND JOURNALS**

**A. Full-length published articles.**

A. Narayan, C.M. Tuck-Muller, K. Weissbecker, D. Smeets, and M. Ehrlich. Hypersensitivity to Radiation-Induced Non-Apoptotic and Apoptotic Death in Cell Lines From Patients With the ICF Chromosome Instability Syndrome. *Mutat. Res.* 456:1-15 (2000).

C.M. Tuck-Muller, B.K. Goodman, J.E. Martinez, J.R. Korenberg, X.N. Chen, W. Wertelecki, and G. Stetten. Partial Trisomy 7p Defined by a Complex Chromosome Rearrangement (CCR) Using a BAC Clone Panel. *Genet. Med.* 3:126-131 (2001).

L.J. Wong, P. Dai, D. Tau, M. Lipson, A. Grix, M. Sitry-Platt, A. Gropman, and T.J. Chen. Severe Lactic Acidosis Caused by a Novel Frame Shift Mutation in Mitochondrial-Encoded Cytochrome C Oxidase Subunit II. *Am. J. Med. Genet.* 102:95-99 (2001).

L.J. Wong, W.L. Hwu, P. Dai, and T.J. Chen. Molecular Genetics of Glycogen-Storage Disease Type 1a in Chinese Patients of Taiwan. *Mol. Genet. Metab.* 72:175-180 (2001).

**B. Articles in press.**

W. He, C.M. Tuck-Muller, J.E. Martinez, S. Li, E.R. Rowley, and W. Wertelecki. Molecular Characterization of a Ring Chromosome 16 From a Patient With Bilateral Cataracts. *Am. J. Med. Genet.* (2001)

J.G. Scammell, J.L. Wright, and C.M. Tuck-Muller. The Origin of Four Squirrel Monkey Cell Lines Established by Karyotype Analysis. *Cytogenet. Cell Genet.* (2001).

L.J. Wong, T.J. Chen, P. Dai, L. Bird, and M. Mueuke. Novel SNP at the Common Primer Site of Exon IIIa of FGFR2 Gene Cause Error in Molecular Diagnosis of Craniosynostosis Syndrome. *Am. J. Med. Genet.* (2001).

W. Wertelecki. The World Wide Web and Birth Defects Prevention. *Proc., World Alliance of Organizations for the Prevention of Birth Defects* (2000).

W. Wertelecki, O. Sulima, I. Baryliak. Implementations of Core Notions of the World Alliance of Organizations for the Prevention of Birth Defects in Ukraine. Proceedings, World Alliance of Organizations for the Prevention Birth Defects (2000).

## II. PUBLISHED ABSTRACTS.

I. Baryliak, I. Kharytonova, and W. Wertelecki. Promotion of Birth Defects Prevention Alliances. Proc., 10<sup>th</sup> Int. Cong. Human Genet., HG01-2189-WMA (2000).

F. Tsien, P. Laski, M. Park, C.M. Tuck-Muller, and M. Ehrlich. Hypomethylation, Heterochromatin Decondensation, and Rearrangements Targeted to the Pericentromeric Regions of Chromosomes 1 and 16 in Cell Lines From ICF Syndrome Patients and in Chorionic Villus Cultures From Normal Fetuses. Am. J. Hum. Genet. 67(4):150 (2000).

T. Vihovska, O. Shevchuk, O. Sulima, H. Vashchylin, W. Wertelecki, and L. Yevtushok. The Role of Neonatologists in Birth Defects (BD) Monitoring in Ukraine. Proc., 10<sup>th</sup> Int. Cong. Human Genet., HG01-1423-WMA (2000).

W. Wertelecki. Birth Defects Monitoring in Ukraine. Proc., 28<sup>th</sup> Annu. Mtg., Int. Birth Defects Clearinghouse of Monitoring Systems, IBDCMS1-28-BD (2001).

W. Wertelecki. International Birth Defects Information System (IBIS) – A Prevention Tool. Proc., 10<sup>th</sup> Int. Cong. Human Genet., HG01-1725-WMA (2000).

L. Yevtushok, I. Baryliak, S. Lapchenko, T. Vihovska, and W. Wertelecki. Role of the "Web-Internet" and Birth Defects (BD) in Ukraine. Proc., 10<sup>th</sup> Int. Cong., Human Genet., HG01-1416-WMA (2000).

## III. BOOKS PUBLISHED.

None.

## IV. PRESENTATIONS.

J.E. Martinez. The Genetics Lab to the Rescue: Solving Clinical Dilemmas. Ala. State Perinatal Conf., Gulf Shores, AL (2001).

J.E. Martinez. Common Genetic Syndromes. Providence Hospital Noon Conf., Mobile, AL (2000).

F. Tsien, P. Laski, M. Park, C.M. Tuck-Muller, and M. Ehrlich. Poster. Hypomethylation, Heterochromatin Decondensation, and Rearrangements Targeted to the Pericentromeric Regions of Chromosomes 1 and 16 in Cell Lines From ICF Syndrome Patients and in Chorionic Villus Cultures From Normal Fetuses. 50<sup>th</sup> Annu. Mtg., Am. Soc. Human Genet., Philadelphia, PA (2000).

W. Wertelecki. Lessons From a Prevention of Birth Defects Program in Ukraine. Plenary Session Key Note Speaker, 1<sup>st</sup> Int. Cong., Prevention of Birth Defects in the Developing World, Johannesburg, South Africa (2001).

W. Wertelecki. Population Based Birth Defects Surveillance in Two Regions (Oblasts) of Ukraine. 28<sup>th</sup> Annu. Mtg., Int. Birth Defects Monitoring Clearinghouse, Groningen, The Netherlands (2001).

W. Wertelecki. Neural Tube Defects – Realities in Ukraine. Key Note Speaker. Special Session., Ukrainian Academy of Medicine, Folic Acid – From Scientific Research to Health Care Practice, Kyiv, Ukraine (2001).

W. Wertelecki. Folic Acid – From Scientific Research to Health Care Practice. Academy of Medical Sciences, Ministry of Public Health of Ukraine, Kyiv, Ukraine (2001).

W. Wertelecki. OMNI Information Centers – A Strategy to Prevent Birth Defects. National Birth Defects Center, Havana, Cuba (2000).

## **V. NATIONAL PROFESSIONAL RECOGNITION**

Dr. W. Wertelecki was re-elected Secretary-Treasurer of the World Alliance for the Prevention of Birth Defects. He was awarded a Certificate of Appreciation from the Regional Perinatal Advisory Committee. Dr. W. Wertelecki was awarded the Franklin Smith Award for Distinguished Service, from the State of Alabama.

## **VI. BRIEF SUMMARY OF DEPARTMENT ACTIVITIES AND PROGRESS.**

The COM II Medical Genetics Course expanded its emphasis on case-based learning and introduced a web-based assignment. In short, every student was asked to explore and analyze our International Birth Defects Information Systems (IBIS) site, developed as one component of the USAID-sponsored program to provide an informational resource to Ukraine. This exercise required each student to engage in several hours of web explorations, focused on a specific domain of human developmental pathology.

Clinical training was provided to pediatric residents during elective rotations, and to an international fellow. Increasing demands for clinical services called for an expansion of the frequency of outreach clinics held in Dothan, Alabama.

The Cytogenetic Laboratories were CLIA and CAP certified. The scope of FISH and neoplasia DNA cytogenetic probes were expanded to meet growing demands. New collaborative investigations of primate chromosomes were initiated by Dr. C. Tuck-Muller.

The Medical DNA Diagnosis Laboratory directed by Dr. T.J. Chen was CLIA and CAP certified. Parentage testing capabilities are completed and this service is now offered.

The Alabama Birth Defects (BD) Surveillance and Prevention program received funding from the CDC to expand its scope to include prevention of NTD. Additional "genetic" nurses were trained and deployed in Dothan, Alabama, and Montgomery, Alabama. Voluntary agreements with hospitals were signed and population-based BD surveillance has expanded to Montgomery, Alabama, and Dothan, Alabama, regions. The BD program is a "de facto" partner of the Alabama Department of Rehabilitation Services (ADRS) since the outreach Genetic BD Surveillance Nurses are housed in ADRS facilities. Integrating BD surveillance with care and prevention is a national health priority and, to this end, we received a Special Project of Regional and National Significance (SPRANS) grant, which led to the development of strategies now being integrated into the BD program.

The Department continues to foster close links with community organizations. Our programs continue to receive financial assistance from the Alabama Department of Mental Health/Mental Retardation and ADRS. Also, Dr. W. Wertelecki was awarded a Certificate of Appreciation from the Regional Perinatal Advisor Committee for community services.

The USAID funding for the Ukrainian-USA Birth Defects Program was renewed. The territory impacted by the program was doubled and an "Omni" Information-Resource Center was established in Kyiv, Ukraine. The program was admitted as a member of the International Birth Defects Monitoring Systems Clearinghouse. Program results were also reported at the International Congress of Human Genetics. The Ukrainian Academy of Medicine and the Mohyla Universities became partners for two core prevention implementations, namely the 1<sup>st</sup> National Congress on Bioethics (held September 19, 2001), and to hold a World Health Organization/Europe (WHO/EURO) BD prevention meeting dedicated to Eastern Europe (to take place in late November 2001).