1999-2000
SUMMARY OF SCHOLARLY ACTIVITIES
DEPARTMENT OF MEDICAL GENETICS

I. ARTICLES PUBLISHED IN BOOKS AND JOURNALS

A. Full-length published articles.

H. Chen, X. Mu, T. Sonoda, K.C. Kim, R. Fan, K. Dailey, J.M. Martinez, C.M. Tuck-Muller, and W. Wertelecki. A FGFR3 Mutation (Gly380Arg) in a Patient With Achondroplasia and i(21q) Down Syndrome. South. Med. J. 93:622-624 (2000).

T.J. Chen, R.G. Boles, and L-J C. Wong. Detection of Mitochondrial DNA Mutations by Temporal Temperature Gradient Gel Electrophoresis. Clin. Chemistry 45:1162-1167 (1999).

C.M. Tuck-Muller, A. Narayan, F. Tsien, J. Sawyer, D. Smeets, E. Fiala, O.S. Sohn, and M. Ehrlich. DNA Hypomethylation and Unusual Chromosome Instability in Cell Lines From ICF Syndrome Patients. Cytogenet. Cell. Genet. 89:121-128 (2000).

B. Articles in press.

T.J. Chen, D.G. Hu, and C. Gao. Hereditary Neurological and Neuromuscular Disorders. IN: G. Lu and W. Li (Eds.) Prenatal Diagnosis of Genetic Diseases, Guangdong Press of Science and Technology, Guangzhou, China (2000)

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. (2000).

L.J. Wong, T.J. Chen, P. Dia, L. Bird, and M. Muenke. A Novel SNP at the Common Primer Site of Exon IIIa of FGFR2 Gene Causes Error in Molecular Diagnosis of Craniosynostosis Syndromes. Am. J. Hum. Genet. (2000).

II. PUBLISHED ABSTRACTS.

I. Barylyak, M. Dumanovska, H. Mikulska, S. Polishchuk, O. Shevchuk, T. Vihovska, W. Wertelecki, and L. Yevtushok. Introduction of International Standards to Ukraine -Birth Defects Surveillance - Care - Prevention. Special Report - 1st Int. Symp., Prevention and Epidemiology of Congenital Malformations (2000).

T.J. Chen, W. Fan, and L.J. Wong. A Novel 2bp Deletion in Cytochrome C Oxidase Sub-unit II. Am. J. Hum. Genet. 65(4):A267 (1999).

T.J. Chen and L.J. Wong. Detection of Heteroplasmic Mitochondrial DNA Mutations by Temporal Temperature Gradient Gel Electrophoresis. Bio-Rad, Tech Note #2450 (1999).

M.H. Liang, J.J. Wong, T.J. Chen, W. Fan, and L.J. Wong. Novel mtDNA Mutations Detected by TTGE. Am. J. Hum. Genet. 65(4):A458 (1999).

J. Meck, T.J. Chen, L.J. Wong, et. al. Cytogenetic and Molecular Evidence of Constitutional Mosaic Trisomy 8 and Hematologic Abnormalities in a Phenotypically Normal Woman. Am. J. Hum. Genet. 65(4):A138 (1999).

M.C. Prieto, S. Kahn, C.M. Tuck-Muller, S. Li, R. Gordon, and M. Marble. Reciprocal 1;17 Translocation in a Ppatient With Apparent Aicardi Syndrome Plus Complex Congenital Heart Disease and Limb Malformation. Am. J. Hum. Genet. 65(4):A355 (1999).

C.M. Tuck-Muller, A. Narayan, F. Tsien, D. Smeets, E. Fiala, O.S. Sohn, and M. Ehrlich. DNA Demethylation, Heterochromatin Decondensation, and Generation of Pericentromeric Rearrangements in Chromosomes 1 and 16 in B Cell Lines From Patients With ICF Syndrome. Am. J. Hum. Genet. 65(4):A361 (1999).

L-J.C. Wong, T.J. Chen, S. Agnor, and K. Cleverley. The Polymorphic Polyglutamine Tract of AIB1 Gene and Breast Cancer. Am. Assn. Cancer Res. 41:91 #583 (2000).

III. BOOKS PUBLISHED.

None.

IV. PRESENTATIONS.

J.E. Martinez. The Role of Medical Genetics in Rehabilitation Services. ADRS Conf., Mobile, AL (2000).

J.E. Martinez. Integration of Genetics Into the Alabama Department of Rehabilitation Services. ADRS Conf., Perdido Beach Resort, AL (1999).

J.E. Martinez. Common Genetic Syndromes: Advances in Diagnosis and Genetics in the New Millennium. Int. Symp., Genet. Health Dis., Fundacion de Genetica Medica Gregorio Mendel, Cordoba, Argentina (1999).

C.M. Tuck-Muller. DNA Demethylation, Heterochromatin Decondensation, and Generation of Pericentromeric Rearrangements in Chromosomes 1 and 16 in B Cell Lines From Patients With ICF Syndrome. 49th Annu. Mtg., Am. Soc. Human Genet., San Francisco, CA (1999).

C.M. Tuck-Muller. Karyotyping: the Who, What, Why, and How of Chromosome Studies. Ala. State Soc., Clin. Lab. Sci. Annu. Prof. Conf., Mobile, AL (2000).

W. Wertelecki. Telemedicine and Birth Defects Diagnosis and Care. Ukrainian American Birth Defects Program, USAID Mission, Kyiv, Ukraine (1999).

W. Wertelecki. (1) Analytical Approaches to Birth Defects Data; (2) Gene-Environment Considerations Regarding Birth Defects (1999); and (3) Birth Defects Surveillance Translation into Care. Ukrainian American Birth Defects Program. Mohyla Academy University, Kyiv, Ukraine (2000).

W. Wertelecki. Alabama Birth Defects Surveillance - Strategies and Implementations. Natl. Birth Defects Prevention Network 3rd Annu. Mtg., New Orleans, LA (2000).

W. Wertelecki. Birth Defects Prevention. Ala. Southern Rural Access Prog., Montgomery, AL (2000).

W. Wertelecki. Programs for Children with Genetic Disorders and Complex Medical Needs in Alabama. Special Project of Regional and National Significance Development Advisory Committee, Mobile, AL (2000).

W. Wertelecki. Prevention of Birth Defects. Ala. Assn., Operating Room Nurses, Annu. Educ. Sem., Gulf Shores, AL (2000).

W. Wertelecki. Birth Defects and Health Care Networks. Cincinnati-Kharkiv Sister City Project, Cincinnati Medical Center, Cincinnati, OH (2000).

W. Wertelecki. Birth Defects - Ideas Implementations and Results. University of Warsaw Child Health Center, Warsaw, Poland (2000).

W. Wertelecki. Birth Defects Data Coding and Management. Ukrainian American Birth Defects Program, Lutsk Medical Center, Lutsk, Volyn, Ukraine (2000).

W. Wertelecki. Birth Defects in Ukraine - Preliminary Findings. 1st Int. Symp., Prevention of Congenital Malformations (ISPECOM), Cardiff, UK (2000).

W. Wertelecki. (1) Ideas and Implementations in Birth Defects; and (2) Birth Defects Information and the Role of the Web. World Alliance for the Prevention of Birth Defects, Amsterdam, The Netherlands (2000).

V. NATIONAL PROFESSIONAL RECOGNITION

Dr. W. Wertelecki was re-elected Secretary-Treasurer of the World Alliance for the Prevention of Birth Defects.

VI. BRIEF SUMMARY OF DEPARTMENT ACTIVITIES AND PROGRESS.

The COM II Medical Genetics Course has been modified to provide a greater emphasis on case-based teaching. The mandatory practicum (review of clinical materials) has also been expanded. A web site with over 3000 links to selected sources of information has been launched. Teaching of nursing students, medical technology students, and residents continues.

Clinical core services are augmented by regional outreach clinics located in Brewton, Dothan, Montgomery, and the Children's Rehabilitation Centers in Mobile. The Clinical Cytogenetic Laboratory has experienced a considerable growth in cancer cell studies. The Molecular Genetics Diagnostic Laboratory has a new Director and Faculty member, Dr. Tian-Jian Chen. Dr. Chen is a Board Certified, recognized expert in medical DNA diagnostic procedures, particularly mitochondiral disorders. Demand for the recently expanded array of DNA diagnostic tests (HFE, BRCA1/2, Connexin26, SMN genes among others) and FISH studies continues to grow. Several research projects on cancer genetics have been initiated.

Our Alabama Birth Defects Surveillance team has compiled the first population-based data in Alabama and joined the National Birth Defects Prevention Network. For the purpose of better translating birth defects surveillance into care and prevention, our team was awarded a Special Project of Regional and National Significance Grant. USA surveillance activities will be expanded to most South Alabama Counties, and health and prevention implementations will be funded by a grant from the Centers for Disease Control and Prevention. Funding by USAID for USA to continue leading the Ukrainian Birth Defects Surveillance Project has been extended. Population-based data for the year 2000 will provide a basis for studies to assess environmental impacts on the development of children in Ukraine (including Chornobyl).