ROBERTO T. ZORI, M.D.
CURRICULUM VITAE

ADDRESS:

Roberto T. Zori, M.D.
Professor and Chief
Division of Genetics
Departments of Pediatrics and Genetics
University of Florida
Box 100296
Gainesville, Florida 32610-0296

1754 NW 61st Terrace
Gainesville, FL 32605

BORN:

April 28, 1952, Genoa, Italy

CITIZENSHIP:

U.S.A.

EDUCATION:

High school diploma, Regis High School
New York City, New York, 1966-1970
B.S. in Psychology and Zoology, Duke University,
North Carolina, 1970-1973
M.D., Odense University School of Medicine,
Odense University, Denmark, 1974-1979

PROFESSIONAL TRAINING:

Resident, Department of Neurology, Odense University Hospital,
Denmark, July 1979
Resident, Department of Neurophysiology and the Epilepsy Clinic,
Odense University Hospital, Denmark, August 1979 - May 1980
Resident, Department of Pediatrics, Baystate Medical Center,
Springfield, MA (major affiliation with Tufts University and the
University of Massachusetts, Boston, MA), June 1980 – June
1983
Fellow, Division of Genetics, Department of Pediatrics, University of
Florida, June 1988-1990
Resident, Department of Surgery (K), Odense University Hospital,
Denmark, June 1992 – November 1992

COURSES:

Eurocat/NPEU/WHO course on applications of perinatal epidemiology, (course by appointment), Oxford, England, June 1984

Medical and Experimental Mammalian Genetics, Jackson Laboratory, (course by appointment), Bar Harbor, Maine, July 17-28, 1989

First Nordic Hugo Workshop on Genetic Mapping (Family studies and linkage analysis), (course by appointment), Aarhus, Denmark, September 21-23, 1992

FISH: Clinical Applications in Cancer and Genetics, Lake Tahoe, California, February 8-11, 1994

College of American Pathologists Laboratory Inspector Training Course, Memphis, Tennessee, May 1996

APPOINTMENTS:

Staff Pediatrician, Department of Pediatrics, Odense University Hospital, Denmark, July 1983 – September 1985

Project leader, Denmark, Eurocat (European Registry of Congenital Anomalies and Twins) Project, funded by the European Economic Community and one of their major research endeavors, 1984-1988

Member, European Community Low Birth Weight Study Group, funded by the European Economic Community, 1984-1988 Clinical Lecturer, Pediatrics, Odense University, Denmark, 1986-1988

Staff Pediatrician, Department of Pediatrics, Odense University Hospital, Denmark, April 1986 – June 1988

Instructor, Division of Genetics, Department of Pediatrics, University of Florida College of Medicine, Gainesville, Florida, July 1990 – July 1991

Assistant Professor, Division of Genetics, Department of Pediatrics, University of Florida College of Medicine, Gainesville, Florida, July 1991-July 1992; July 1993-July 1997

Visiting Assistant Professor, Division of Genetics, Department of Pediatrics, University of Florida, College of Medicine, Gainesville, Florida, July 1992-June 1993

Director, Cytogenetics Laboratory, R.C. Phillips Unit, University of Florida College of Medicine, Gainesville, Florida, April 1992-July 1992; December 1992-present

Director, Genetics Services, University Medical Center, Jacksonville, Florida, December 1992-September 1993

Assistant Professor, Department of Pathology, University of Florida College of Medicine, Gainesville, Florida, June 1994-July 1997

Director, Cytogenetics Technologist Training Program, University of Florida Cytogenetics Laboratory, Gainesville, Florida, March 1995-present

APPOINTMENTS (continued):

Associate Professor, Division of Genetics, Department of Pediatrics and the Department of Pathology, University of Florida College of Medicine, Gainesville, Florida, July 1997-present
Chief, Division of Genetics, Department of Pediatrics, University of Florida College of Medicine, Gainesville, Florida, 2000 to present
Director, University of Florida Clinical Genetics Residency Program, 2004-present
Director, Cytogentics Fellowship Program University of Florida 2005-present
External Lecturer D'Annunzio University, Chieti, Italy, 2006-2008
Professor, Division of Genetics, Department of Pediatrics and the Department of Pathology. University of Florida, College of Medicine, Gainesville, Fl. June 20007-Present.
Abstract Reviewer, 2008 Pediatric Academic Society and Asian Society for Pediatric Research Joint Meeting Honolulu Hawaii, May 2-6, 2008

HOSPITAL STAFF PRIVILEGES:

Shands Teaching Hospital at the University of Florida, Gainesville, Florida, 1990-present
Shands at Alachua General Hospital, Gainesville, Florida, 1989-1990; 1994-present
North Florida Regional Medical Center, Gainesville, Florida, 1994-2002
Shands at Jacksonville, University Medical Center Teaching Hospital, Jacksonville, Florida, 1993-1997
Sacred Heart Hospital, Pensacola, Florida, 2001-2002
Sacred Heart Hospital, Pensacola, Florida, 2005-Present.

COMMITTEES/SERVICES:

Danish Representative to EURACAT Collaborative Study funded by the European Economic Community, Denmark, 1986-1988
Member, Eurocat Collaborative Study on Congenital Heart Disease, funded by the European Economic Community, Denmark, 1986-1988
Member, Pediatric Postoperative Cardiac Care Group, Odense University, Odense, Denmark, 1987-1988
Member, Pediatric Quality Assurance Committee, University of Florida, Gainesville, Florida, 1990-July 1992; 1999-present
Site Reviewer, University of Miami Regional Genetics Program, Department of Health and Rehabilitative Services, State of Florida, Miami, Florida, February 11, 1992; February 17, 1995
Reviewer, Shands Hospital at the University of Florida Guide to Clinical Dietetics, Gainesville, Florida, 1992

COMMITTEES/SERVICES (continued):

Member, Curriculum Committee for Graduate Education, The Center for Mammalian Genetics, University of Florida, Gainesville, Florida, July 1992-present
Faculty Advisor, Joyce de Geus, M.D., Pediatric resident 1993-1996
Physician Advisor, Florida Medical Quality Assurance, Inc., 1994-2000
Member, Pharmacy and Therapeutics Committee, Shands Hospital at the University of Florida, Gainesville, Florida, 1994-2001
Reviewer for:
Journal of Pediatric Neurology
American Journal of Medical Genetics
Journal of Pediatrics
Lancet
Journal of Clinical Genetics
Clinical Pediatrics
Cancer Letters
Journal of Medical Retardation
International Journal of Cancer
Genetic Testing
Human Genetics
Member, Supervisory Committee for Tiffany Alley, Ph.D. in Biochemistry and Molecular Biology, 1994-1997
Member, the Neurobiology Advanced Program of the University of Florida College of Medicine Interdisciplinary Program for Graduate Education, 1995-present
Member, the Genetics Advanced Program of the University of Florida College of Medicine Interdisciplinary Program for Graduate Education, 1995-present
Inspector, College of American Pathologists Volunteer National Laboratory (Inspector number 1011515), 1996-present
Site Reviewer, University of South Florida Regional Genetics Program, Department of Health and Rehabilitative Services, Tampa, Florida, September 1996
Site Reviewer, Cytogenetics, Inc. Laboratory, College of American Pathologists Laboratory Accreditation Program, Nashville, Tennessee, December 1996
Editor for Natural Standard, 2001-present
Member, Florida Infant Screening Task Force, 2002
Consultant, University of Florida Craniofacial Clinic, Gainesville, Florida, 2002-present
Member, Florida Infant Screening Advisory Committee, 2003-Present
Member, Association of Professors of Human and Medical Genetics, 2002- present.
Reviewer, NCI site review, Myeloproliferative Disorders Research Consortium, Dr. Hoffman, University of Chicago, Michigan March 1-2 2004

COMMITTEES/SERVICES (continued):

Newborn Screening Program Implementation Plan Workgroup, Tallahassee Florida, October 7-8, 2004
Biochemical consultant for State of Florida expanded newborn biochemical screening 2004-Present
Member of Safety Monitoring Board 2006 Urea cycle defects clinical trials (phase III clinical study "UP1204-003 A Phase 3, Open-Label, Switch-Over Study of the Safety, Tolerability, and Efficacy of Glyceryl Tri (4-Phenylbutyrate) (GT4P) Compared to Buphenyl® (Sodium Phenylbutyrate) in Patients with Urea Cycle Disorders
Site Reviewer, College of Pathology, Ameripath Cytogenetic Laboratory, Orlando, Florida, April 2006
Reviewer, University of Florida Cancer Center Pilot Grant 2005
Member, Department of Pediatrics Clinical Space Committee 2006
Member, PhD Supervisory Committee for Maggie Kelogg, Ph.D. in Biochemistry and Molecular Biology, 2005-2007
Member, Newborn Screening Laboratory, Directors and Genetics Coordinators Workgroup, Southeastern Regional Genetics Group. 2005-2008.
Member, Southeastern Regional Genetics Disaster Preparedness Workgroup, New Orleans, LA. August 2006, and Atlanta, Ga. May 2008.
Member of PhD Committee for Joe Kramer, Department of Animal Science. 2007-Present
Member, Advisory Committee, Region 3 Collaborative Regional Genetics and Newborn Screening services, HRSA Grant, July 2007-present
Member, Information Technology Committee, Department of Pediatrics, University of Florida, 2005-2007
Chairman, Space Committee, Department of Pediatrics, University of Florida, 2008-Present

BOARDS AND LICENSES:

ECFMG: #310-540-0, 1979
Danish medical license, 1979
Temporary license, State of Massachusetts, 1980-1983
State of Florida, Board of Medical Examiners License, #ME0054105, 1988
FLEX: Florida, 1988
Diplomate, American Board of Pediatrics, 1990, recertification 1995, 2002
Diplomate, American Board of Medical Genetics, certified in Clinical Genetics, 1990, and in Clinical Cytogenetics, 1993, recertification 2002, 2005,2007,2009.
Danish certification in Pediatrics, valid in all European community countries (Danish Board of Health), 1992
Danish Medical Association, 1979-present

BOARDS AND LICENSES (continued):

Section of Genetics and Birth Defects, American Academy of Pediatrics, October 1991-1997
American College of Medical Genetics, 1993-1997; 2002-2004.
American Medical Association, 1993-1996

MEMBERSHIP:

Federation of Young Danish Physicians, 1979-2000
Angelman Research Group, 1988-present
American Academy of Pediatrics, Fellow, 1989-1997
Alachua County (FL) Medical Society, 1990-2002, 2005-Present
American Society of Human Genetics, 1990-1997. 1999-2004
Florida Medical Association, 1990-2002
Florida Pediatric Society, 1991-2002, 2005-Present
Florida Pediatric Alumni Association, 1991-2002

TEACHING:

Clinical Lecturer, Odense University Medical School, Odense, Denmark, 1983-1988
Lecturer in Pediatrics at Odense School of Nursing, Odense, Denmark, 1983-1988
Workshop Leader, "Neurologically Impaired Children", Postgraduate Teacher Education, Odense, Denmark, 1984
Lecturer, postgraduate course "Neonatal Intensive Care", Copenhagen, Denmark, 1985
Presenter, Workshops for Parents "Sleep Disorders" and "Seizures", 1st North American Angelman Support and Research Conference, Orlando, Florida, August, 1990
Presenter, "Rett Syndrome" and "Teratogens, Cocaine, Fetal Alcohol Syndrome, Anticonvulsants", Nursing Education Workshop on Genetics and Developmental Delay, University of Florida, Gainesville, Florida, August 9-10, 1990
Instructor, Medical Genetics Course (BMS 5202), University of Florida College of Medicine, Gainesville, Florida, 1990-1992; 1994-1998, 2003, 2004,2007, 2008, 2009
Lecturer, PL1-PL3 core rotation, Department of Pediatrics, University of Florida, Gainesville, Florida, 1990-1992; 1994; 1998; 2006
General Pediatric Teaching Attending, (one month per year) Department of Pediatrics, University of Florida, Gainesville, Florida, 1990-1992; 1994-1998
Presenter, Workshops for Parents "Sleep Disorders" and "Seizures", 2nd North American Angelman Support and Research Group Conference, Orlando, Florida, August, 1991
Medical Staff, North Florida Area Health Education Centers Program, Gainesville, Florida, 1991-1992
Science Fair Judge, Westside Middle School, Gainesville, Florida, 1992;

TEACHING (continued):

1994; 1996-1997

Genetics lectures to Pediatric Housestaff, University Medical Center, Jacksonville, Florida, 1992-1994

Small group instructor for Medical Student Pediatric Clinical Diagnosis Course, University of Florida College of Medicine, Gainesville, Florida, 1990-1992; 1994; 1995; 1997; 1998; 2007

Lecturer, Junior Honors Genetics Seminar Course, University of Florida, (BMS 4010), Gainesville, Florida, Spring 1995-1997

Pediatric Grand Rounds, "Clinical and Molecular Analysis: X-linked Arthrogryposis", University of Florida, Gainesville, Florida, June 2, 1995

Pediatric Grand Rounds, "Clinical Genetics and Clinical Cytogenetics", Sacred Heart Hospital, Pensacola, Florida, January 23, 1996

Lecturer, Genetics Course (ARG 6353), University of Florida College of Agriculture, Gainesville, Florida, 1997

Presenter, "Medical Cytogenetics Workshop", National Cytogenetics Technologists Meeting, Orlando, Florida, August, 1999

Faculty, Pathology Board Review Course, Osler Institute, Tampa, Florida, April 2000; April 2001

Participant, Master of Education Program, University of Florida, 2000-2002

Presenter, University of Florida on-line web course MEL5011, "Introduction to the Profession of Medicine", Pediatrics, 2002

Pediatric Grand Rounds, "Genetic Conditions with Imprinting Defects", University Hospital, Jacksonville, Florida, September, 2003

Grand Rounds, "Non-Mendelian Inheritance", Nemours Children's Clinic, Jacksonville, Florida, July, 2003

Presenter, Gene Therapy, Florida Annual Meeting, Associations of Retarded Citizens, Gainesville, Florida, July 11, 2003

IDP graduate student rotation, University of Florida, Cytogenetics Laboratory, February 2004

Director of Department of Pathology Residency Program Cytogenetics Rotation 2000-present

Designed Genetics Medical Residency Program, University of Florida, Accepted by ACGME 2004

Designed Cytogenetics and PhD Medical Genetics Fellowship Program Submitted to ABMG 2003

Developed and provide services for telemedicine program between University of Florida Division of Genetics and Sacred Heart NICU Sacred Heart Hospital, Pensecola Florida 2004-present

Developed telemedicine program between University of Florida Division of Genetics and Childrens Medical Services, Department of Health Florida Pensecola, Florida 2004-present

Writer, Questions for Clinical Genetics Board Examination, American College of Medical Genetics, 2005

TEACHING (continued):

Grand Rounds, Metabolic Disease of Children, Sacred Heart Hospital, Department of Pediatrics, Pensacola, Florida, June 2007.

Visiting Professor including Grand Rounds, Metabolic Disease of Children, Sacred Heart Hospital, Department of Pediatrics, Pensacola, Florida, June 11-12, 2009

INVITED MEETINGS/PRESENTATIONS:

Instructor, Undergraduate Course HSC 3801, "Clinical Observation" Spring 2005

Pediatric Grand Rounds, "Expanded Biochemical Newborn Screening in Florida", Department of Pediatrics,Sacred Heart Hospital, Pensacola, Florida, October 24, 2005

Genetic Residency Site Visit by RRC. April 2007 – Approved.

Danish Registry of Congenital Anomalies: Review of collected data, Eurocat (European registry of congenital anomalies and twins) project symposium and meeting on congenital anomalies, Perugia, Italy, May 1983

"Very Low Birth Weight Infant Mortality and Morbidity Rates in the Region of Funen, Denmark, 1979-1984", European Community Low Birth Weight Project Meeting, Glasgow, Scotland, May 1985

"Proposals for Future Collaborative Studies", European Community Low Birth Weight Project Meeting, Leiden, The Netherlands, October 1985

Very Low Birth Weight Infant Mortality and Morbidity Rates in the Region of Funen, Denmark, 1979-1985", European Community Low Birth Weight Project Meeting, Leiden, The Netherlands, February 1986

"Proposals for Future Collaborative Studies", European Community Low Birth Weight Project Meeting, Brussels, Belgium, December 1987

Genetics and Infant Screening Advisory Council of Florida Meeting, representing Division of Genetics of the University of Florida, Tallahassee, Florida, May 21 1992

"Imprinting in Angelman Syndrome and Prader-Willi Syndrome", Department of Pediatrics, University of Odense, Denmark, August 1992

"Structural Chromosome 15 Aberrations and Imprinting", Giannina Gaslini Institute, Genoa, Italy, July 6-9 1998

"Genetic Therapy for PKU" IX Course on Genetic Diseases, Università "G. d'Annunzio", Chieti, Italy, July 15-16 2004

"Imprinting in genetic diseases" IX Course on Genetic Diseases, Università "G. d'Annunzio", Chieti, Italy, July 15-16 2004

Grand Rounds, Expanded Biochemical Newborn Screening in Florida, Sacred Heart Hospital, Department of Pediatrics, Pensacola Florida, October 24,2005.

INVITED MEETINGS/PRESENTATIONS (continued):

Medical Genetics Course to Medical Genetics Residents and PhD Genetic Candidates (15-20 lecture hours), Universita G. d'Annunzio, Chieti, Italy, June-July 2005, June-July 2006, June-July 2007. June-July 2008, June-July 2009.

Genetic Rounds, Imprinting In Genetic Disease, Department of Genetics, Bambino Gesu Children's Hospital, Vatican city, Rome, Italy December 2008.

Grand Rounds, Metabolic Disease of Children, Sacred Heart Hospital, Department of Pediatrics, Pensacola, Florida, May 2009

GRANTS/AWARDS:

Principal Investigator, Children's Miracle Network Telethon Award, "Fluorescence Microscope", May 1994, funding: $12,063

Principal Investigator, Hayward Foundation, "Mapping an X-linked Mental Retardation Gene", July 1994, funding: $10,000

Principal Investigator, Hayward Foundation, "Diagnosis of Fragile X by Fluorescence *In Situ* Hybridization", May 1996, funding: $7500

Principal Investigator, Vysis, "Prenatal Aneuploidy Detection Kit CEP 18/X/Y, and LSI 13/21 Aqua, Green, and Orange for Direct Enumeration of Chromosomes 18, X, Y, 13, and 21 by Fluorescence *In Situ* Hybridization, Sensitivity, and Specificity", 1996, funding: $4,425

Principal Investigator, Hayward Foundation, "Fiber Fluorescent In Situ Hybridization Techniques", May 1997, funding: $5,000

Principal Investigator, Children's Miracle Network Telethon Award, October 1997, "DNA Probe Labeling For Use in FISH Assessment of Genetic Disorders," October 1997, funding: $10,928.25

Center for Autism and Related Disabilities, Jacksonville, "Telomeric Microdeletions in Autism", 1999, funding: $8,000

The following annual Children's Medical Services contracts were under my directorship from 2000-present as Chief of Genetics:
Regional Genetics Program, 2000-2004, yearly funding: $273,000
Raymond C. Philips Research and Education Unit, 2000-2004, yearly funding: $260,000

Teratogen Information Service, 2000-2002, yearly funding: $68,000; 2000-2003, funding: $50,000

Principal Investigator, Genetic Services via Telemedicine, Children's Medical Services Grant, Florida, 2002 funding: $24,000

Development and Assessment of a Pediatric Instructional Web Site based on "Pediatric Morning Report at the University of Florida." COMEC Research and Development Program, August 20 2001 through May 30 2002, funding: $3,675

Best oral presentation "Web Based Morning Report" May 2002

GRANTS/AWARDS (continued):

Univeristy of Florida College of Medicine Society of Teaching Scholars award for oral presentation May 2002, Middelton M, Zori RT

Principal Investigator, Telemedicine Development Grant, Children's Medical Service, Florida, 2003, funding: $119,000 (grant includes programs to delivery services to Sacred Heart Hospital NICU, and Children's Medical Services, Pensecola, Florida, Florida School of Deaf and Blind, Saint Augustine, Florida, and the PKU community North Florida)

Exemplary Teacher in Clinical Science, 2005, University of Florida

Co-Principal Investigator, Center for Instructional Technology and Training Innovations in Uses of Technology in Instruction Grant 2004, Interactive website for children with Phenylketonuria $10,000

Consultant for Core C, Program project grant – NIH/NIDDK – Grant #DK58327, PI – Dr. Terrence Flotte, University of Florida, 2005, 2006 funding: $4,145.

Society of Pediatric Research 2005.

Principal Investigator, March of Dimes Chapter Grant, Interactive educational website for children with Phenylketonuria (PKU) $15,000

Principal Investigator, Expanded Newborn Screening Services Children's Medical Services, 2004-present $151,600 yearly

Principal Investigator, Genetics Infant Hearing Program, Children's Medical Services, 2006-present $37.967 yearly

Principal Investigator, March of Dimes Grant, Enhanced Genetic Resources and Services for families affected by birth defects and rare inherited diseases, $5,043.

Co-Chair Telegenetics/Telemedicine project. Southeaster Regional Genetics Group. Health Resources and Services Administration Grant/U.S. Department of Health and Human Services. 2007.

Genzyme, Education and Research Grant $25,000/year 2006-2009.

Hayward Research Grant, , Genetic Service Projects $25,000 2008-2009.

Hayward Research Grant, , Genetic Service Projects $50,000 2009-2010.

PUBLICATIONS:

1. Rowland T, Zori RT, Lefleur W, Reiter EO: Malnutrition and hypernatremic dehydration in breast-fed infants. JAMA, 19;247(7):1016-7, 1982.

2. Hansen LP, Brandrup F, Zori RT: Erythema toxicum neonatorum mit pustulation versus transitorische neonatale pustulose melanose. Hautarzt, 36;(8):475-7, 1985.

3. Zori RT and Kofoed P: Persisterende pulmonale hypertension af nyfodte. Ugeskrft for Laeger (Danish Medical Weekly), Jan 20;148(4):168-70, 1986.

PUBLICATIONS (continued):

4. De la Mata I, De Wals P, Dolk H, Lechat M F, Beckers R, Borlee I, Lys F, Zori RT, Goujard J, Stoll C, Ayme S, Hansen-Koenig D, Karkut G, Galanti C, Marchi M, Bianchi F, Calabro A, Lungarotti S, Calzolari E, Radic A, Lillis F, Stone D, Harris F, Nevin N, Ten Kate D, Svel I, Ligutic I, Cuschieri A: Incidence of congenital Rubella syndrome in 19 regions of Europe in 1980-1986. Eur J Epidemiology, 5(1)106-109, 1989.

5. Williams CA, Zori RT, Stone JW, Gray BA, Cantú ES, Ostrer H: Maternal origin of 15q11-q13 deletions in Angelman syndrome suggests a role for genomic imprinting. Am J Med Genet, 35:350-353, 1990.
6. Zori RT, Schatz D, Ostrer H, Williams CA, Riley WJ: The relationship of autoimmunity to thyroid dysfunction in children and adults with Down syndrome. Am J Med Genet, 7(suppl):238-241, 1990.

7. Zori RT, Williams C, Mattei JF, Moncla A: Letter to the Editor: Parental origin of del(15)(q11-q13) in Angelman and Prader-Willi syndromes. Am J Med Genet, 37:294-295, 1990.

8. Zori RT, and Williams C: Neonatal hemangiomatosis with hydrops fetalis and nuchal cystic hygroma. Dysmorph and Clin Genet, 4(4):138-140, 1990.

9. McIlwaine GM, Verloove-Vanhorick SP, Mutch L, Schmidt E, Spitz B, Zori RT, Adam H, Cussen G, Paludetto R, Lloyd D: European community collaborative study of outcome of pregnancy between 22 and 28 weeks gestation. The Lancet, 336:782-784, 1990.

10. Dolk H, De Wals P, Gillerot Y, Lechat MF, Aymé S, Beckers R, Bianchi F, Borlée I, Calabro A, Calzolari E, Cuschieri A, Galanti C, Goujard J, Hansen-Koenig D, Harris F, Karkut G, Lillis DF, Lungarotti S, Lys F, Marchi M, Nevin NC, Radic A, Stoll C, Stone D, Svel I, Ten Kate LP, Zori RT: The prevalence at birth of Down syndrome in 19 regions of Europe 1980-86. In: Key issues in mental retardation research, I. William. FRASER, Routledge, 1990: 3-11.

11. EUROCAT Working Group: Dolk H, De Wals P, Lechat MF, Beckers R, Borlée I, Lys F, Zori RT, Goujard J, Stoll C, Tenconi R, Calzolari E, Galanti C, Marchi M, Bianchi F, Calabro A, Lungarotti MS, Radic A, Lillis DF, Hansen-Koenig D, Nevin N, Stone D, Harris F, Laurence KM, Svel I, Cuschieri A. Prevalence of neural tube defects in 20 regions of Europe and impact of prenatal diagnosis, 1980-1986. Journal of Epidemiology and Community Health 1991; 45:52-58.

12. Williams CA and Zori RT: Response to the letter by Kenji Naritomi: On the genetic imprinting suggested in Angelman syndrome. Am J Med Genet, 39:497, 1991.

PUBLICATIONS (continued):

13. Zori RT, and Williams CA: Phenocopy versus genocopy. Am J Med Genet, 40:248, 1991.

14. Zori RT, Nicholls R, Driscoll DJ, Williams CA: Clinical findings in Angelman individuals without a molecular deletion or uniparental disomy. In "Proceedings of the International Conference on Prader-Willi Syndrome and Other Chromosome 15q Deletion Disorder" (S. Cassidy, ED.), NATO Advanced Studies Institutes Series, Springer-Verlag, Heidelberg, 61:237, 1992.

15. Nicholls RD, Gottlieb W, Zori RT, Driscoll DJ, Zackowski J, Waters M, Williams CA: Molecular analysis in Angelman syndrome. In "Proceedings of the International Conference on Prader-Willi Syndrome and Other Chromosome 15q Deletion Disorder" (S. Cassidy, ED.), NATO Advanced Studies Institutes Series, Springer-Verlag, Heidelberg, 61:41-51, 1992.

16. Driscoll DJ, Waters MF, Williams CA, Zori RT, Avidano KM, Nicholls RD: A DNA methylation imprint, determined by the sex-of-parent, distinguishes the Angelman and Prader-Willi syndromes. Genomics, 13:917-924, 1992.

17. Zori RT, Hendrickson J, Woolven S, Whidden EM, Gray B, Williams CA: Angelman syndrome: Clinical profile. J Child Neurol, 7:270-280, 1992.

18. Zori RT, Gray BA, Bent-Williams A, Driscoll DJ, Williams CA, Zackowski JL: Preaxial acrofacial dysostosis (Nager syndrome) associated with an inherited and apparently balanced X;9 translocation: Prenatal and postnatal late replication studies. Am J Med Genet, 46:379-383, 1993.

19. Zackowski JL, Nicholls RD, Gray BA, Bent-Williams A, Gottlieb W, Harris PJ, Waters MF, Driscoll DJ, Zori RT, Williams CA: Cytogenetic and molecular analysis in Angelman syndrome. Am J Med Genet, 46:7-11, 1993.

20. Zori RT, Lupski JR, Zhang H, Gray BA, Driscoll DJ, Patel PI, Zackowski JL: An infant with Smith-Magenis syndrome born from a mother having a mosaic 17p11.2 p12 deletion. Am J Med Genet, 47:504-511, 1993.

21. Zori RT, Stalker H, Williams CA: A syndrome of familial megaloencephaly, early developmental delay, short stature, pectus abnormalities and dysplastic nails. Dysmorph and Clin Genet, 6(3):116-122, 1994.

22. Wallace M, Zori RT, Alley T, Whidden E, Gray BA, Williams CA: Smith-Lemli-Opitz syndrome in a female with a de novo, balanced translocation involving 7q32: probable disruption of an SLOS Gene. Am J Med Genet, 50:368-374, 1994.

23. Williams CA, Angelman H, Clayton-Smith J, Driscoll DJ, Hendrickson JE, Knoll

PUBLICATIONS (continued):

JHM, Magenis RE, Schinzel A, Wagstaff J, Whidden EM, Zori RT: Angelman syndrome: consensus for diagnostic criteria. Am J Med Genet, 56:001-008, 1995.

24. Alley T, Gray BA, Lee S, Scherer S, Tsui L-C, Tint S, Zori RT, Williams CA, Wallace MR: Identification of a yeast artificial chromosome clone spanning a translocation breakpoint at 7q32.1 in a Smith-Lemli-Opitz syndrome patient. Am J. Hum Genet, 56:1411-1416, 1995.

25. Sandler D, Mancuso A, Becker T, Zori RT, Hellrung J, Silverstein J, Burton V, Hamosh A, Williams C: Association of anophthalmia and esophageal atresia. Am J Med Genet, 59:484-491, 1995.

26. Williams CA, Zori RT, Hendrickson J, Stalker H, Marum T, Whidden E, Driscoll DJ: Angelman Syndrome. Current Problems in Pediatrics, 25:213-244, 1995.

27. Rasmussen SA, Williams CA, Ayoub EM, Sleasman JW, Gray BA, Bent-Williams A, Stalker HJ, Zori RT: Juvenile Rheumatoid Arthritis (JRA) in two individuals with Velo-Cardio-Facial syndrome. Am J Med Genet, 64:546-550, 1996.

28. Migeon BR, Jeppesen P, Torchia BE, Melanie SF, Dunn MA, Axelman J, Schmeckpeper BJ, Fantes J, Zori RT, Driscoll DJ: Lack of X inactivation associated with maternal X isodisomy: evidence for a counting mechanism prior to X inactivation during human embryogenesis. Am J Hum Genet, 58:161-170, 1996.

29. Zori RT, Gardner JL, Mullan MJ, Osborn AR, Houlden H, Wallace MR, Roberts S, Yang TP: A novel form of X-linked arthrogryposis maps to the distal long arm of the human X chromosome. Am J Med Genet, in press, 1998.

30. Abernathy CA, Rasmussen SA, Stalker HJ, Zori RT Driscoll DJ, Williams CA, Kousseff BG, Wallace MR: NF1 mutation analysis using a combined heteroduplex/ SSCP approach. Human Mutation, 9:548-554, 1997.

31. Zori RT, Boyar FZ, Williams WN, Gray BA, Bent-Williams A, Stalker H, Rimer L, Nackashi JA, Driscoll DJ, Rasmussen S, Dixon-Wood V, Williams CA: Prevalence of 22q11 region deletions in patients with velopharyngeal insufficiency. Am J Med Genet, 77:8-11, 1998.

32. Gray B, Bent-Williams A, Wadsworth J, Maiese RL, Zori RT: Characterization of jumping 1q translocations by FISH in a case of aggressive B-cell non-Hodgkin's lymphoma/leukemia. Cancer Genetics and Cytogenetics, 98:20-27, 1997.

PUBLICATIONS (continued):

33. Stalker H, Zori RT: A family with scapular defects, cleft palate, rib anomalies and heart defects. Am J Med Genet, 73:247-250, 1997.

34. Krklus S, Johnson J, Abernathy CR, Williams CA, Driscoll DJ, Zori RT, Stalker H, Whidden E, Kousseff BG, Baumbach L, Wallace MR: Analysis of CpG C to T mutations in the NF1 gene. Human Mutation, Mutation in Brief (#129) (1997) on-line.

35. Arn PH, Williams CA, Zori RT, Driscoll DJ, Rosenblatt DS: Methylene tetrahydrofolate reductase (MTHFR) deficiency in a patient with phenotypic features of Angelman syndrome. Am J Med Genet, 77:198-200, 1998.

36. Moreb JS, Schweder M, Gray B, Zucali J, Zori RT: In vitro selection for K562 cells with higher retrovirally mediated copy number of aldehyde dehydrogenase class-1 (ALDH-1) and higher resistance to 4-hydroperoxcyclophamide (4-HC). Human Gene Therapy, 9:611-619, 1998.

37. Zori RT Marsh DJ, Marliss EB, Eng C: Germline PTEN mutation in a family with Cowden syndrome and Bannayan Riley Ruvacalba Syndrome. Am J Med Genet, 80:399-402, 1998.

38. Marsh DJ, Kum JB, Lunetta KL, Bennett MJ, Gorlin RJ, Ahmed SF, Bodurtha J, Crowe C, Curtis MA, Dasouki M, Dunn T, Feit H, Geraghty MT, Graham DM, Hodgson SV, Hunter A, Korf BR, Manchester D, Miesfeldt S, Murday VA, Nathanson KL, Parisi M, Pober B, Romano C, Tolmie JL, Trembath R, Winter RM, Zackai EH, Zori RT, Weng L, Dahia PL, Eng C: PTEN mutation spectrum and genotype-phenotype correlations in Bannayan-Riley-Ruvalcaba syndrome suggest a single entity with Cowden syndrome. Hum Mol Genet, 8(8):1461-1472, 1999.

39. Wallace MR, Rasmusssen SA, Lim IT, Gray BA, Zori RT, Muir D. Culture of cytogenetically abnormal Schwann cells from benign and malignant NF1 tumors. *Genes, Chromosomes, and Cancer.* Feb:27(2):117-23, 2000.

40. Boyadjiev SA, Jabs EW, LaBuda M, Jamal JE, Torbergsen T, Ptacek LJ 2nd, Rogers RC, Nyberg-Hansen R, Opjordsmoen S, Zeller CB, Stine OC, Stalker HJ, Zori RT, Shapiro RE. Linkage analysis narrows the critical region for oculodentodigital dysplasia to chromosome 6q22-q23. Genomics, 15;58(1):34-40, 1999.

41. Lossie AC, Whitney MM, Amidon D, Dong HJ, Chen P, Theriaque D, Hutson A, Nicholls RD, Zori RT, Williams CA and Driscoll DJ: Distinct phenotypes distinguish the molecular classes of Angelman syndrome. J Med Genet. December; 38(12): 834-845, 2001.

42. Boyar FZ, Whitney MM, Lossie AC, Gray BA, Keller KL, Stalker HJ, Zori RT,

PUBLICATIONS (continued):

Geffken G, Mutch J, Edge PJ, Voeller KS, Williams CA and Driscoll DJ: A family with a grand-maternally derived duplication of proximal 15q. Clin Genet. December; 60(6): 421-430.

43. Gray B, Bent-Williams A, Wolff D, Zori RT. A non-sex chromosome marker in a patient with an atypical Ullrich-Turner phenotype and mosaicism of 46,X,mar/46,XX. Clin Genet 2001 July; 60(1): 73-76.

44. Stalker H, Bent-Williams A, Gray B, Zori RT. LTE: Dominant Transmission of a Previously Unidentified 13/17 Translocation in a 5-generation Family with Robin Cleft and Other Skeletal Defects. Am J Med Genet. Nov 1; 103(4): 339-341, 2001.

45. Imermch B, Caruso LJ, Zori RT [Commentator]: Prolonged Neuromuscular Block in Patient Undergoing Renal Transplantation. J Clin Anesth. Nov; 13(7): 540-544, 2001.

46. Marsh D, Zori RT. Genetic insights into familial cancers—update and recent discoveries. Cancer Lett. Jul 26; 181(2): 125-164, 2002.

47. Keller K, Williams C, Wharton P, Paulk M, Bent-Williams A, Gray BA, Ward A, Stalker H, Wallace M, Zori RT. Routine cytogenetic, FISH studies for 17p12/15q11 duplictions and subtelomeric rearrangement studies in children with autistic spectrum disorder Am J Med Genet. 2003 Mar 1; 117A(2): 105-11.

48. Gray BA, Zori RT, McGuire P, Felton S, Bonde R. Cytogenetic Characterization of the Florida Manatee (Trichechus Manatus Latirostris) By Chromosome Banding Techniques, Hereditas. 2002 Dec 135(3): 215-223.

49. Stalker H, Zori RT. Concurrence of Fragile X Syndrome and 47, XYY in an individual with a Prader-Willi –like Phenotype. Am J Med Genet. 2003 Jan 15; 116(2): 176-8.

50. Gray BA, Cornfield D, Bent-Williams A, Zori RT. Translocation (X;20)(q13.1; q13.3) as a primary chromosomal finding in two patients with myeloid disorders. Cancer Genet Cytogenet. 2003 Mar 141(2): 169-74.

51. Howell VM, Zori RT, Stalker HJ, Williams CA, Jesse N, Nelson AE, Robinson BG, Marsh DJ. A molecular diagnosis of hyperparathyroidism-Jaw tumor syndrome in an adolescent with recurrent kidney stones. J Pediatr. 2004 Oct; 145(4):567.

52. Williams CA, Wallace MR, Drury KC, Kipersztok S, Edwards RK, Williams RS, Haller MJ, Schats DA, Silverstein JH, Gray BA, Zori RT. Blood Lymphocyte

chimerism associated with IVF and monochorionic dizygous twinning: Case
PUBLICATIONS (continued):
report. Hum Reprod. 2004 Sep 16.

53. Stevenson DA, Anaya TM, Clayton-Smith J, Hall BD, Van Allen MI, Zori RT, Zackai EH, Frank G, Clericuzio CL. Unexpected death and critical illness in Prader-Willi syndrome: report of ten individuals. Am J Med Genet. 2004 Jan 15; 124A(2):158-164.

54. Hagler KT, Zori RT, Yuan CM, Gray BA, Moreb JS. Diagnosis of an early precursor-B ALL presenting with hypereosinophilia using FISH on immunomagnetically selected CD19+ cells. Clin Adv Hematol Oncol. 2005 3(1): 62-63.

55. Kang SH, Levings, PP Andersen F, Laipis PJ, Berns KI, Zori RT, Bungert J. Locus control region elements HS2 and HS3 in combination with chromatin boundaries confer high level expression of a human beta-globin transgene in a centromeric region. Genes Cells Nov 9(11): 1043-53.

56. Hill-Harfe KL, Kaplan L, Stalker HJ, Zori RT, Pop R, Scherer G, Wallace MR. Fine mapping of chromosome 17 translocation breakpoints $\geq$ 99kb upstream of SOX 9 in acampomelic campomelic dyslplasis and a mild familial skeletal dysplasia. Am J Hum Gen. 2005 Apr; 76(4): 663-71.

57. Stalker HJ, Zori RT, Wallace MR, Hill-Harfe KL, Kaplan L. Reply to Unger: The Mildest Form of Campomelic Dysplasia. Am J Med Gen. 2005 Jan 1; 132A: 114-115.

58. Zhuang H, Kosboth M, Lee P, Rice A, Driscoll DJ, Zori RT, Narain S, Lyons R, Satoh M, Sobel E, Reeves WH. Lupus-like disease and high interferon levels corresponding to trisomy of the type I interferon cluster on chromosome 9p. Arthritis Rheum. 2006 May;54(5):1573-9.

59. Pause KC, Bonde RK, McGuire PM, Zori RT, Gray BA. G-Banded Karyotype and Ideogram for the North Atlantic Right Whale (Eubalaena glacialis). J Hered. 2006 May-June, 97(3):303-6.

60. Stalker HJ, Wilson R, McCune H, Gonzalez J, Moffitt M, Zori RT. Tele-genetic medicine: establishing enhanced services in an underserved area. Journal of Telemedicine and Telecare 2006; 12(4):182-5.

61. Stalker HJ, Gray BA, Bent-Williams A, Zori RT. High cognitive functioning and behavioral phenotype in Pallister Killian syndrome. American Journal of Medical Genetics. 2006 Sept 15; 140(18):1950-4.

62. Erger KE, Conlon TJ, Leal NA, Zori RT, Bobik TA, Flotte TR. *In Vivo* Expression of Human ATP:Cob(I)alamin Adenosyltransferase (ATR) using

PUBLICATIONS (continued):

Recombinant Adeno Associated Virus (rAAV) serotypes 2 and 8. J Gene Med. 2007 June;9(6)462-9.

63. Kellogg ME, Burkett S, Dennis TR, Stone G, Gray BA, McGuire PM, Zori RT, Stanyon R. Chromosome painting in the manatee supports Afrotheria and Paenungulata. BMC Evol Biol. 2007 Jan 23;7:6.

64. Dagli A, Heese BH, Zori RT. Testing Strategy for Inborn Errors of Metabolism in the Neonate. NeoReviews, Vol. 9, No. 7. July 2008.

65. Milunsky JM, Maher TA, Zhao G, Roberts AE, Stalker HJ, Zori RT, Burch MN, Clemens M, Mulliken JB, Smith R, Lin AE. TFAP2A mutations result in banchio-oculo-facial syndrome. Am J Hum Genet. 2008 May; 82(5):1171-7. Erratum in: Am J Hum Genet. 2009 Feb;84(2):301

66. Garner R, Li Y, Gray B, Zori R, Braylan R, Wall J, Hunger SP. Long-term disease control of refractory anaplastic large cell lymphoma with vinblastine. J Pediatr Hematol Oncol. 2009 Feb;31(2):145-7

67. Dennis TR, Raptoulis GN, Stalker HJ, Boles D, Meck JM, Krasnewich DM, Schiffmann R, Zori RT. Molecular cytogenetic characterization of two small supernumerary marker chromosomes derived from chromosome 19. Am J Med Genet A. 2009 Feb;149A(2):262-5.

68. Ucar D, Cogle CR, Zucali JR, Ostmark B, Scott EW, Zori R, Gray BA, Moreb JS. Aldehyde dehydrogenase activity as a functional marker for lung cancer. Chem Biol Interact. 2009 Mar 16;178(1-3):48-55. Epub 2008 Oct 5.

69. Dagli AI, Zori RT, McCune H, Ivsic T, Maisenbacher MK, Weinstein DA. Reversal of glycogen storage disease type IIIa-related cardiomyopathy with modification of diet. J Inherit Metab Dis, 2009 Mar 30. [Epub ahead of print].

70. Laipis PJ, Charron CE, Perera OP, Embury JE, Alexander JJ, Hurt CB, Steele HA, Song S, Berns KI, Zori RT, Flotte TR. Gene Therapy for PKU: Negative Interference in a Recessive Disease. (In preparation).

71. Heggestad, AD; J Dostie; TR Dennis; J Dekker; S Rodriguez-Jato; BA Gray; Zori RT, DJ Driscoll; TP Yang. Analysis of Allele-Specific Long-Range Interactions in the Imprinted Angelman/Prader-Willi Syndrome Domain. Human Molecular Genetics (In preparation).

72. Wat, M, Shchelochkov O, Holder A, Breman A, Dagli A, Scaglia F, Zori R, Cheung S, Scott D, Kang S. Chromosome 8p23.1 Deletions as a Cause of

Complex Congenital Heart Defects and Diaphragmatic Hernia. Am J Med Genet: Part A. 2009 Aug;149A(8):1661-77.

CHAPTERS/SECTIONS:

1. Zori RT, Williams CA: Angelman Syndrome. (CDROM Version 1997, Updated Print Version 1998) In Adelman G, Smith BH (eds.): Encyclopedia of Neuroscience, 2nd ED. 2001, Elsevier Science, Amsterdam, the Netherlands.

2. Marsh DJ, Zori RT: Cowden Syndrome, 2000. Encyclopedia of Cancer, Manfred Schwab Editor, Springer, Heidelburg, Germany.

3. Zori RT Genetic Review questions in 2004 Pedaitric Board Questions. Medtext.

4. Marsh DJ, Zori RT: Cowden Syndrome, 2004. Marcel Dekker Encyclopedia of Medical Genomics & Proteomics.

5. Marsh DJ, Zori RT, Cowden Syndrome, 2007. Encyclopedia of Cancer 2nd Edition, Manfred Schwab Editor, Heidelburg, Germany.

6. Heese BA, Zori RT: Genetics in Critical Care Medicine, 2007. Civetta, Taylor, Kirby: Critical Care, 4th Ed. Gabrielli A, Yu M, and Layan AJ, editors.

7. Heese B, Zori R, Medical Genetics and Inherited Metabolic Disorders, Critical Care, 4th Ed. Editors Civetta JM, Taylor RW, Kirby RR. 2008, Lippincott-Ravin, Philadelphia

ABSTRACTS:

1. Zori RT and Kamper J: Respiratory insufficiency in newborn with a birthweight of less than 1500 g.; 7 years experience with low intensity treatment. Meeting of Danish Pediatric Society, Copenhagen, Denmark, April 1987 (presented).

2. Kamper J, Nielsen G, Zori RT: Early treatment of idiopathic respiratory distress syndrome with binasal continuous positive airway pressure. Meeting of Danish Pediatric Society, Copenhagen, Denmark, April 1987 (presented).

3. Kamper J, Nielsen G, Zori RT: Early treatment with binasal CPAP in idiopathic respiratory distress syndrome; 5-7 year prognosis. Meeting of Danish Pediatric Society, Copenhagen, Denmark, April 1987 (presented).

4. De Wals P, Dolk H, Bertrand F, de la Mata I, LeChat M F, Beckers R, Borlee I, Lys F, Zori RT, Goujard J, Stoll C, Tenconi R, Calzolari E, Galanti C, Marchi M, Bianchi F, Calabro A, Lungarotti S, Radic A, Lillis F, Hansen-Koenig D, Ten Kate L, Nevin N, Stone D, Harris F, Laurence KM, Svel I, Ligutic I, Cuschieri A, Ayme S, Mastroiacovi P: Evaluation of the mutagenic and teratogenic effects of

the Chernobyl radiological contamination in regions of Western Europe. XI European Congress of Perinatal Medicine, April 1988 (presented).

ABSTRACTS (continued):

5. Dolk H, De Wals P, Gillerot Y, Lechat MF, Aymes S, Beckers R, Bianchi F, Borlee I, Calabro A, Calzolari E, Cuschieri A, Galanti C, Goujard J, Hansen-Koenig D, Harris F, Karkut G, Lillis DF, Lungarotti MS, Lys F, Marchi M, Nevin N C, Radic A, Stoll C, Stone D, Svel I, Ten Kate LP, Zori RT: The prevalence at birth of Down syndrome in 19 regions of Europe 1980-1986. 8th International Association of the Scientific Study of Mental Deficiency, Dublin, Ireland, August 1988 (presented).

6. Zori RT, Schatz D, Ostrer H, Williams C, Riley W: The relationship of autoimmunity to thyroid dysfunction in children and adults with Down syndrome. International meeting on Down syndrome, Rome, Italy, 1989.

7. Zori RT, Stone JW, Gray BA, Cantú S, Ostrer H, Williams C: Parental origin of *de novo* 15(q11-q13) deletions in Angelman syndrome. 7th Annual Pediatric Science Day, University of Florida, Gainesville, Florida, April 1989 (presented).

8. Zori RT, Papenhausen P, Essig YP, Tedesco TA: Centromeric heteromorphism of chromosome 20. Twelfth summer meeting, Southern Genetics Group, Navarre Beach, Florida, July 1989.

9. Strickland TS, Zori RT, Williams CA: Atypical neuromuscular disorders associated with neurofibromatosis. Annual Clinical Care Symposium of the National Neurofibromatosis Foundation, Inc., Baltimore, Maryland, November 1989.

10. Zori RT and Williams CA: An overgrowth disorder involving lipoid, sebaceous and fibrous elements. Additional comments on the Proteus syndrome, Neurofibromatosis (NF) and the "Elephant Man Disorder." Annual Clinical Care Symposium of the National Neurofibromatosis Foundation, Inc., Baltimore, Maryland, November 1989 (presented).

11. Zori RT, Hendrickson J, Stone J, Whidden E, Gray B, Williams C: 15(q11-13) deletion syndromes: Are Angelman and Prader-Willi syndromes different expressions of a similar CNS defect? 8th Annual Pediatric Science Day, University of Florida, Gainesville, Florida, April 1990 (presented).

12. Zori RT: Clinical findings in Angelman syndrome. 1st North American Angelman Support and Research Group Conference, August 1990 (presented).

13. Zori RT, Hendrickson J, Woolven S, Williams CA: Angelman syndrome: Further characterization of the clinical phenotype and natural history. American

Society of Human Genetics Meeting, Baltimore, Maryland, October 1990. Am J Hum Genet, 47:A85, 1990.

14. Zackowski JL, Driscoll DJ, Gray BA, Zori RT: Probable Smith Magenis ABSTRACTS (continued):
syndrome resulting from maternal mosaicism for an interstitial deletion of 17p. 29th Annual American Cytogenetic Conference, Tahoe City, California, February 1991 (presented), Cytogenet Cell, 56:230, 1991.

15. Nicholls RD, Gottlieb W, Zori RT, Driscoll DJ, Zackowski J, Waters M, Williams CA: Molecular analysis in Angelman syndrome. International Conference on Prader-Willi Syndrome and other Chromosome 15q Deletion Disorders, De Leewenhorst Congress Center, Noordwijkerhout, the Netherlands, May 1991 (presented). Am J Med Genet, 42:235-236, 1992.

16. Zori RT, Nicholls R, Driscoll DJ, Williams CA: Clinical findings in Angelman individuals without a molecular deletion or uniparental disomy. International Conference on Prader-Willi Syndrome and other Chromosome 15q Deletion Disorders, De Leewenhorst Congress Center, Noordwijkerhout, the Netherlands, May 1991 (presented). Am J Med Genet, 42:249-250, 1992.

17. Zori RT: Natural history of Angelman syndrome, Scientific Session of the 2nd North American Angelman Support and Research Group Conference, Orlando, Florida, August 1991 (presented).

18. Driscoll DJ, Waters MF, Gottlieb W, Williams CA, Zori RT, Nicholls RD: Differences in DNA methylation in the Angelman and Prader-Willi syndromes. 8th International Congress of Human Genetics, Washington, D.C., USA, October 1991 (presented). Am J Hum Genet, 49A:2224, 1991.

19. Gustafson SR, Humphreys WF, Zori RT: Pure X-linked hereditary spastic paraplegia. 8th International Congress of Human Genetics, Washington, D.C., USA, October 1991. Am J Hum Genet, 49A:708, 1991.

20. Gray BA, Driscoll DJ, Zori RT, Bent-Williams A, Zackowski JL: Abnormal phenotype associated with an inherited and apparently balanced X/autosome translocation: Prenatal and postnatal late X-replication studies. 8th International Congress of Human Genetics, Washington, D.C., USA, October 1991. Am J Hum Genet, 49A:1457, 1991.

21. Zori RT and Williams CA: Ruvalcaba-Myhre-Smith syndrome: Nosology and inheritance pattern revisited. 8th International Congress of Human Genetics, Washington, D.C., USA, October 1991. Am J Hum Genet, 49A:907, 1991.

22. Williams CA, Zori RT, Nicholls R, Hendrickson J, Driscoll DJ: Angelman syndrome patients with and without 15q11-13 deletions have similar phenotype

suggesting little or no genetics heterogeneity outside region 15q11-13. 8th International Congress of Human Genetics, Washington, D.C., USA, October 1991 (presented). Am J Human Genet, 49A:1116, 1991.

ABSTRACTS (continued):

23. Zackowski JL, Cantú ES, Driscoll DJ, Ostrer H, Zori RT, Gray BA: X-Inactivation patterns in multiple tissues of three cases of apparently balanced X/autosome translocations with abnormal phenotypes. 8th Annual American Cytogenetics Conference, Virginia Beach, Virginia, March 1992 (presented). Cytogenetic Cell Genet, 60:264.

24. Driscoll DJ, Waters MF, Williams CA, Zori RT, Glenn CC, Avidano K, Nicholls RD: DNA methylation and genomic imprinting: The Angelman and Prader-Willi syndromes as models. N.I.C.H.H.D. "Genomic Imprinting: Consequences for Human Disease," Bethesda, Maryland, April 1992.

25. Driscoll DJ, Waters MF, Glenn CC, William CA, Zori RT, Avidano KM, Nicholls RD: A DNA methylation imprint distinguishes the Angelman and Prader-Willi syndrome. American Pediatric Society, Society for Pediatric Research and Ambulatory Pediatric Association Meeting, Baltimore, Maryland, May 1992 (presented).

26. Nicholls RD, Rinchik EM, Gottlieb W, Avidano K, Jong M, Horsthemke B, Russell LB, Bultman SJ, Spritz RA, Rogan PK, Mascari MJ, Ladda RL, Waters MF, Glenn CC, Williams CA, Zori RT, Driscoll DJ: Candidate genes, mechanisms and comparative mapping of loci from the PWCR/ANCR in 15q11-q13. Physical and Genetic Mapping of Chromosome 15 Workshop, June 1992 (presented). Cytogenetic Cell Genet 61.

27. Nicholls RD, Rinchik EM, Gottlieb W, Avidano K, Jong M, Horsthemke B, Russell LB, Bultman SJ, Spritz RA, Rogan PK, Mascari MJ, Ladda RL, Waters MF, Glenn CC, Williams CA, Zori RT, Driscoll DJ: Molecular characterization of candidate genes and mechanisms for clinical features of Prader-Willi and Angelman syndromes. Prader-Willi Syndrome Association (U.S.A.), July 1992, (presented). Dysmorphol Clin Genet J.

28. Patel PI, Zhang H, Zori RT, Zackowski JL, Greenberg F, Lupski JR: Mosaicism for del (17) (p11.2): Clinical, cytogenetic and molecular analysis of two cases. 42nd Annual American Society of Human Genetics Meeting, San Francisco, California, November 1992. Am J Hum Genet, 51A:36.

29. Zori RT, Stalker HJ, Williams CA: Familial megaloencephaly, early developmental delay and short stature, pectus abnormalities and dysplastic nails: A new syndrome. 42nd Annual American Society of Human Genetics Meeting, San Francisco, California, November 1992. Am J Hum Genet, 51A:429.

30. Wallace M, Driscoll DJ, Zori RT, Williams CA: NF1 mutation and polymorphisms. College of Medicine, Faculty Research Day, University of Florida, Gainesville, Florida, April 21, 1993.

ABSTRACTS (continued):

31. Zori RT, Gardner JL, Mullan M, Roberts S, Wallace MR, Yang TP: Linkage analysis of a novel form of X-linked arthrogryposis. American Society of Human Genetics, New Orleans, Louisiana, October 1993. Am J Hum Genet, 53A:1003.

32. Driscoll DJ, Porter KA, Glenn CC, Williams CA, Zori RT, Whidden E, Gottlieb W, Filbrandt M, Bowers L, Gray B, Nicholls RD: Molecular, clinical and cytogenetic studies of the Angelman and Prader-Willi syndromes. American Society of Human Genetics, New Orleans, Louisiana, October 1993. Am J Hum Genet, 53A:255.

33. Colman S, Abernathy C, Ho V, Kamath R, Kousseff B, Williams C, Driscoll DJ, Zori RT, Collins F, Wallace MR: Heteroduplex detection and RNA analysis of NF1 mutations. American Society of Human Genetics, New Orleans, Louisiana, October 1993. Am J Hum Genet, 53A:1145.

34. Wallace M, Zori RT, Alley T, Whidden E, Gray B, Williams C: Characterization of a balanced chromosome translocation in Smith-Lemli-Opitz syndrome. Florida Association of Biochemists and Molecular Biologists, March 12-14, 1994.

35. Williams CA, Wallace M, Zori RT, Alley T, Whidden EM, Gray BA: Smith-Lemli-Opitz syndrome in a female with a *de novo*, balanced translocation involving 7q32: probable disruption of an SLOS gene. 25th March of Dimes Clinical Genetics Conference, Orlando, Florida, March 13-15, 1994.

36. Zori RT, Sandler DL, Mancuso AA, Becker TA, Williams CA: The association of anophthalmia, esophageal atresia, tracheoesophageal fistula and midbrain anomalies. XV David W. Smith Workshop on Malformations and Morphogenesis, Tampa, Florida, August 4-9, 1994. Proceedings of the Greenwood Genetics Center, 14:74.

37. Alley TL, Gray BA, Lee S, Zhang J, Zori RT, Williams CA, Scherer S, Tsui L-C, Wallace MR: Localization of a translocation breakpoint involved in Smith- Lemli-Opitz syndrome. 44th Annual American Society of Human Genetics Meeting, Montreal, Quebec, Canada, October 18-22, 1994. Am J Hum Genet, 55A:1481.

38. Becker TA, Zori RT, Williams CA: Genetic counseling considerations in clinical anophthalmia. 44th Annual American Society of Human Genetics Meeting, Montreal, Quebec, Canada, October 18-22, 1994. Am J Hum Genet, 55A:803.

39. Bent-Williams AH, Felton SM, Driscoll DJ, Gray BA, Zori RT: Presence of *XIST* specific sequences and apparent failure of X dosage compensation by inactivation in a patient with severe Turner phenotype and mosaicism for X chromosome abnormalities. 44th Annual American Society of Human Genetics

ABSTRACTS (continued):

Meeting, Montreal, Quebec, Canada, October 18-22, 1994. Am J Hum Genet, 55A:557.

40. Victorica BE, Kumar A, Zori RT: Tetralogy of Fallot in monozygotic triplets. 44th Annual American Society of Human Genetics Meeting, Montreal, Quebec, Canada, October 18-22, 1994. Am J Hum Genet, 55A:537.

41. Whidden EM, Kubilis P, Marum T, Driscoll DJ, Zori RT, Williams CA: Growth in individuals with Angelman syndrome. 44th Annual American Society of Human Genetics Meeting, Montreal, Quebec, Canada, October 18-22, 1994. Am J Hum Genet, 55A:540.

42. Williams CA, Angelman H, Clayton-Smith J, Driscoll DJ, Hendrickson JE, Knoll JMH, Magenis RE, Schinzel A, Wagstaff J, Whidden EM, Zori RT: Angelman syndrome: Consensus for Diagnostic Criteria. 44th Annual American Society of Human Genetics Meeting, Montreal, Quebec, Canada, October 18-22, 1994. Am J Hum Genet, 55A:541.

43. Zori RT, Stalker HJ, Driscoll DJ: Variable expression of vertebral, rib, pectus and scapular anomalies with Robin-type cleft palate in a 5 generation family: A new syndrome? 44th Annual American Society of Human Genetics Meeting, Montreal, Quebec, Canada, October 18-22, 1994. Am J Hum Genet, 55A:545.

44. Stalker HJ, Driscoll DJ, Zori RT, Hendrickson JE, Williams CA: Genetic counseling in Angelman syndrome: The importance of molecular studies and clinical judgement in determination of recurrence risk. 44th Annual American Society of Human Genetics Meeting, Montreal, Quebec, Canada, October 18-22, 1994. Am J Hum Genet, 55A:827.

45. Driscoll DJ, Filbrandt M, Glenn CC, Gray BA, Blaydes SM, Nicholls RD, Whidden EW, Zori RT, Williams CA: Distribution of genotypic classes in Angelman syndrome. American Society of Human Genetics, Minneapolis, Minnesota, October 1995. Am J Hum Genet, 57A:238.

46. Zori RT, Rasmussen SA, Ayoub EM, Gray BA, Bent-Williams A, Stalker HJ, Williams CA: Juvenile Rheumatoid Arthritis (JRA) in two individuals with Velo-Cardio-Facial syndrome. American Society of Human Genetics, Minneapolis, Minnesota, October 1995. Am J Hum Genet, 57A:106.

47. Alley T, Scherer S, Gray B, Krkljus S, Kamath R, Zori RT, Williams C, Tsui LC, Wallace M: Molecular mapping of the chromosome 7 Smith-Lemli-Opitz

syndrome (SLOS) t(7;20) breakpoint. RSH/SLO meeting, NIH, Baltimore, Maryland, September 25-27, 1995.

ABSTRACTS (continued):

48. Flick K, Gray BA, Zori RT: Cytogenetic and molecular investigation of two cases of Wolf-Hirschhorn Syndrome. Association of Cytogenetic Technologists, San Antonio, Texas, June 1-4 1995. Applied Cytogenetics 21:115.

49. Zori RT, Boyar FZ, Gray BA, Stalker H, Rimer L, Rasmussen S, Nackashi JA, Williams WN, Williams CA: Palatal defects as mild phenotypic expression of 22q11.2 region microdeletion. American Society of Human Genetics, San Francisco, California, October 1996. Am J Hum Genet, 59A:771.

50. Arn PH, Williams CA, Zori RT Driscoll DJ, Rosenblatt DS: Methylene tetrahydrofolate reductase (MTHFR) deficiency in a patient with phenotypic features of Angelman syndrome. American Society of Human Genetics, San Francisco, California, October 1996. Am J Hum Genet, 59A:463.

51. Graham GE, Brown GK, Robinson BM, Treacy EP, Zori RT, Teebi AS: Features of Ruvalcaba-Myhre-Smith syndrome with oligodactyly of the toes and functional PDM deficiency: Clinical heterogeneity or New syndrome? American Society of Human Genetics, San Francisco, California, October 1996. Am J Hum Genet, 59A:2041.

52. Stalker HJ, Rimer LA, Williams CA, Bent-Williams A, Gray BA, Zori RT: Genetic counseling in 22q11.2 deletions: potential pitfalls. American Society of Human Genetics, San Francisco, California, October 1996. Am J Hum Genet, 59A:972.

53. Boyar FZ, Lossie AC, Gray BA, Filbrandt MM, Burke LW, Zori RT, Driscoll DJ: Molecular characterization of familial cryptic translocation narrows the location of the Angelman gene. American Society of Human Genetics, San Francisco, California, October 1996. Am J Hum Genet, 59A:621.

54. Deng GY, Filbrandt MM, Zori RT, Williams CA, Gray TA, Gabriel JM, Nicholls RD, Driscoll DJ: Analysis of candidates for a major gene in Prader-Willi syndrome. 28th Annual March of Dimes Clinical Genetics Conference and the American College of Medical Genetics 4th Annual Meeting, Ft. Lauderdale, Florida, February 1997.

55. Deng GY, Filbrandt MM, Zori RT, Williams CA, Gray TA, Gabriel JM, Nicholls RD, Driscoll DJ: Analysis of Candidates for a Major Gene in Prader-Willi Syndrome. 12th Annual Prader-Willi Syndrome Association Scientific Conference, Orlando, Florida, July 1997.

56. Lossie AC, Boyar FZ, Filbrandt MM, Zori RT, Williams CA, Driscoll DJ: Analysis of UBE3A in classical and atypical Angelman patients. American Society of Human Genetics, Baltimore, Maryland, October 1997. Am J Hum Genet 61A:1835.

ABSTRACTS (continued):

57. Stalker HJ, Ahmad A, Williams CA, Dietz H, Zori RT: Autosomal recessive syndrome of craniosynostosis, marfanoid habitus, and developmental delay not linked to Fibrillin-1: A new syndrome? American Society of Human Genetics, Baltimore, Maryland, October 1997, Am J Hum Genet 61A:642.

58. Leon BH, Shatz D, Zori RT, Sleasman JW: Sarcoidosis after successful Bone Marrow Transplantation for Omenn Syndrome, American Association of Allergy and Immunology, Washington, DC, 1997.

59. Gray BA, Strauss K, Mitchell D, Zori RT: Cytogenetic evidence of biclonal formation in a case of B-cell CLL suspected of Richter's transformation, American Society of Human Genetics, Denver, Colorado, October 1998. Am J Hum Genet 63A.

60. Boyar FZ, Lossie AC, Filbrandt MM, Gray BA, Keller KL, Stalker HJ, Zori RT, Williams CA, Driscoll DJ: A family with maternally derived interstitial duplication of proximal 15q. American Society of Human Genetics, Denver, Colorado, October 1998. Am J Hum Genet 63A. Available from http://www.ashg.org/genet

61. Lossie AC, Sage BT, Boyar FZ, Filbrandt MM, Nicholls RD, Zori RT, Williams CA, Driscoll DJ: Mutation Analysis of UBE3A in Classical Angelman Syndrome Patient,s American Society of Human Genetics, Denver, Colorado, October 1998. Am J Hum Genet 63A.

62. Lossie AC, Whitney M, Amidon D, Hutson A, Theriaque D, Zori RT, Nicholls RD, Williams CA, Driscoll DJ: Genotype/Phenotype Correlations among the 5 Molecular Classes of Angelman Syndrome. 50th Annual American Society of Human Genetics, Baltimore, Maryland, October 2000. Am J Hum Genet.

63. Lossie AC, Whitney, M, Zori RT, Nicholls RD, Williams CA, Driscoll DJ: Prader-Willi Syndrome and The Role of the Paternal Only Expressed in Genes in 15q11-13: Possible Insights from Angelman Syndrome. PWSA's 15th Annual Scientific Day, Pittsburgh, Pennsylvania, July 2000.

64. Williams CA, Ward AD, Stalker HG, Keller KL, Walker RD, Gray BA, Zori RT. Normal 22q11.2 studies in patients referred for hypospadias. Poster. Annual Clinical Genetic Meeting, American College of Medical Genetics, Miami, FL, March 1-3, 2001.

65. Williams CA, Keller KL, Wharton PW, Paulk MF, Gray BA, Zori RT. Duplication

17p12 involving the Smith-Magenis Region, was not detected in an autism population. Posterj. Annual Clinical Genetic Meeting, American College of Medical Genetics, Miami, FL, March 1-3, 2001.

ABSTRACTS (continued):

66. Gray B, Zori RT, McGuire P, Felton S, Bonde R. Cytogenetic Characterization of the Florida Manatee (Trichechus Manatus Latirostris) By Chromosome Banding Techniques. 14th Biennial Conference on the Biology of Marine Mammals, Vancouver, Canada, September 2001.

67. Zori RT, Gray B, Keller K, Wharton P, Bent-Williams A, Ward A, Stalker H, Paulik M, Williams CA. Cytogenetic studies including FISH studies for 15q11.2/17p12 duplications and subtelomeric rearrangement studies in children with autistic spectrum disorder. 51st Annual American Society of Human Genetics Meeting, San Diego, California, October 2001. Available from http://www.ashg.org/genet

68. Zori RT, Middleton M. Morning Report Presented via the Web. The AAMC Southern Group on Educational Affairs Annual Meeting, Charleston, South Carolina, March 14-17 2002, presented.

69. Zori RT, Middleton M. Morning Report. The 13th International Conference on College Teaching and Learning, Jacksonville, Florida, April 9-13 2002, presented.

70. Berlin MC, Giunta N, Zori RT. A proxy methodology to estimate work RVUs for molecular cytogenetics. Biannual American Cytogenetics Conference, Santa Fe, New Mexico, May 2002, presented.

71. Lapis P, McClune H, Zori RT. Gene Therapy – PKU MICE. Phenyketonuria Present Knowledge and Future Challenges, Elsinore, Denmark, June 27-30 2002, presented.

72. Middleton M, Zori RT. Morning Report Presented via the Web. Annual Irish Pediatric Conference, Dublin, Ireland, Sept. 2002, presented.

73. Laipis PJ Charron CE, Steele HA, Embury JE, Ross K Knapp JM Porvasnik SL, Alexander JJ, Reyes L, Zori RT. Recombinant AAV-based gene therapy of phenylketonuria in the PAH^{enu2} missense mutant mouse. 6th Annual American Society of Gene Therapy Meeting, Washington DC, June 4-8 2003. Mol Ther 2003

74. Marsh DJ, Stalker HJ, Nelson AE, Robinson BG, Zori RT, Howell VM. Molecular diagnosis of Hyperparathyroidism Jaw Tumor Syndrome – germline *HRPT2* mutation in an adolescent presenting with recurrent kidney stones. 53rd Annual

American Society of Human Genetics meeting, Los Angeles, CA, November 2003. Available from http://www.ashg.org/genet

75. Stalker HJ, Wilson RK, Gonzales J, Zori RT. Telehealth in the pediatric genetics clinic: A method of enhancing services to underserved populations.

ABSTRACTS (continued):

Canadian Association of Genetic Counselors Annual Meeting. Edmonton, Canada, October 16-18 2003.

76. Hill-Harfe K, Kaplan L, Stalker HJ, Zori RT, Wallace M. Molecular analysis of a translocation breakpoint in a family with dominant skeletal dysplasia: Toward a novel gene/mechanism. Annual Clinical Genetics meeting, American College of Medical Genetics, Kissimmee, Florida, March 4-7 2004.

77. Williams CA, Zori RT, Gray BA, Stalker HJ. The flip side of the coin: duplications of the Velo-Cardio-Facial syndrome critical region. The Florida Cleft Palate-Craniofacial Association 2004 Winter Symposium. Orlando, Florida, Jan 16-18 2004.

78. Laipis PJ, Charron CE, Embury JE, Perera OP, Porvasnik SL, Fields CR, Knapp JM, Ross K, Zori RT. Correction of Maternal Phenyketonuria Syndrome in the P*ah*^{en2} Missense mouse by r-AAV Mediated Gene Therapy. Poster 878, American Society of Gene Therapy, 7th Annual Meeting, Minneapolis Minnesota, June 2-6, 2004.

79. Stalker H, Zori R, Gray B, Bent-Williams A, Dolwick F, Williams C. High cognitive functioning in Pallister-Killian syndrome. 54rd Annual American Society of Human Genetics meeting, Toronto, Canada, October 26-30, 2004. Available from http://www.ashg.org/genet

80. Williams CA, Wallace MR, Drury KC, Kipersztok S, Edwards RK, Haller MJ, Schatz DA, Silverstein JH, Gray BA, Zori RT. Blood lymphocyte chimerism associated with in vitro fertilization and monochorionic dizygous twinning. 54rd Annual American Society of Human Genetics meeting, Toronto, Canada, October 26-30, 2004. Available from http://www.ashg.org/genet

81. Laipis PJ, Charron CE, Embury JE, Perera OP, Porvasnik SL, Fields CR, Zori RT. Correction of maternal PKU syndrome in the Pahenu2 misssense mouse by r-AAV mediated gene therapy. 54rd Annual American Society of Human Genetics meeting, Toronto, Canada, October 26-30, 2004. Available from http://www.ashg.org/genet

82. Dharmidharka VR, Lin SH, McCune H, Zori RT Gitelman's Syndrome physiology in a boy with argininosuccinic aciduria. American Society of Nephrology Annual Meeting. St Louis , Missouri, October 29-November 1 2004.

83. Stalker HJ, McCune H, Moffett M, Ford E, Gonzalez, J, Wilson R, Zori RT. A multi-tiered approach to provision of regional genetics services. Southeastern Regional Genetics Group (SERGG) 22nd annual meeting, Sandestin, Florida, November 11-13, 2004.

ABSTRACTS (continued):

84. Stalker HJ, McCune H, Wilson R, Zori RT. A multi-tiered approach to provision of regional genetics services. Tenth Annual Meeting & Exposition of the American Telemedicine Association, poster 107 presented in Salt Lake City, Utah, April 2005.

85. Heggestad, AD; Dennis TR; Rodriguez-Jato S; Gray BA; Zori RT; Driscoll DJ; Yang TP. (2005) Spatial organization of the Prader-Willi/Angelman syndrome locus is dependent on the parent of origin. American Society for Human Genetics 55th Annual Meeting, Salt Lake City, UT. (Platform presentation, 249)

86. Heggestad, AD; Dostie J; Dennis TR; Dekker J; Rodriguez-Jato S; Gray BA; Zori RT; Driscoll DJ; Yang TP. Intrachromosomal interactions in the imprinted Angelman/Prader-Willi Syndrome domain. University of Florida College of Medicine Research Day 2006. (Poster)

87. Heggestad, AD; Dostie J; Dennis TR; Dekker J; Rodriguez-Jato S; Gray BA; Zori RT; Driscoll DJ; Yang TP. Intrachromosomal interactions in the imprinted Angelman/Prader-Willi Syndrome domain. Florida Genetics 2005. (Poster)

88. Laipis PJ, Charron C, Ross K, Reyes L, Alexander JJ, Song S, Steele HA, Berns KI, Zori R, Flotte TR 2002 Long-term correction of phenylketonuria in an animal model by recombinant AAV-based gene therapy. J Inherit Metab Dis 25: 615–616

89. McCune HM, Stalker, HS, Zori RT, Interactive website for children with PKU. 3rd Annual Games for Health Conference. Baltimore, MD. Sept 27-28, 2006.

90. Stalker HS, Zori RT. Use of telegenetics consultation in the NICU – an assessment of services. 25th Annual Southeastern Regional Genetics Group (SERGG). New Orleans, LA. July 26-28, 2007.

91. Stalker HS, McCune HM, Porch P, Zori RT. Evaluation of an interactive educational website for children with Phenylketonuria. 25th Annual Southeastern Regional Genetics Group (SERGG). New Orleans, LA. July 26-28, 2007.

92. Dagli A, Zori RT. Homozygous CHRNG Mutation in Twins with Multiple Pterygium Syndrome, Escobar Variant and Schizencephaly. 25th Annual Southeastern Regional Genetics Group (SERGG). New Orleans, LA. July 26-28, 2007.

93. Dagli A, Michalk A, Zori RT, Homozygous CHRNG Mutation in Escobar Variant Multiple Pterygium Syndrome with Schizencephaly. American Society of Human Genetics, 57th Annual meeting. San Diego, CA. October 23-27, 2007

ABSTRACTS (continued):

94. Milunsky JM, Maher TA, Zhao G, Roberts A, Stalker HJ, Zori RT, Clemens M, Smith, R, Mulliken JB, Lin AE. APS*a* Mutations Result in Branchio-Oculo-Facial yndrome (BOFS). American College of Medical Genetics 2007 Annual

    Meeting, Nashville, TN. March 21-27, 2007.

95. Dagli A, Heese B, Driscoll DJ, Zori RT, Edwards P, Stacpoole PW. A Novel and Extreme Case of a Female Patient with Undetectable Pyruvate Dehydrogenase Complex Activity: Early Responsiveness to Diet and Drug Treatment. United Mitochondrial Disease Foundation Meeting. Indianapolis, IL. June 2008

96. ones AG, Li Y, Gray BA, Zori RT, Braylan RC, Al-Quran SZ. The Detection of

    Bone Marrow Hematogones by Multiparameteric Flow Cytometric Analysis in Myelodysplastic Syndromes Predicts the Presence of del 5q. American Society for Clinical Pathology. Baltimore, MD. October 16-19, 2007.

97. Edwards PK, Driscoll DJ, Kerr, DS, McConnell JW, Zori RT, Heese BA, McCune H, Dagli A. Management of PDM Complex Deferring in the Neonate. Second International Nutrition Conference. Atlanta, GA. April 24-26, 2008.

100. Heese B, Driscoll DJ, Zori R, Edwards P, Kerr DS, Stacpoole PW, A Novel and Extreme Case of a Female Patient with Undetectable Pyruvate Dehydrogenase Complex Activity: Early Responsiveness to Diet and Drug Treatment. United Mitochondrial Disease Foundation, Annual Meeting. Indianapolis, IN 2008.

101. Defant JA[1], Stalker HJ[1], Williams CA[1], Knutson N[2], Dagli AI[1], Zori RT[1], 1) A Case of Goltz Syndrome Presenting with a Vascular Type Amniotic Band Sequence Phenotype. American Society of Human Genetics, 58th Annual Meeting, Philadelphia, PA. December 11-15, 2008.

102. Dagli A[1], Zori R[1], McCune, H[1], Ivsic T[3], Maisenbacher M[1], Weinstein, D[2]. Reversal of glycogen storage disease type III related cardiomyopathy with high protein diet. American Society of Human Genetics, 58th Annual Meeting, Philadelphia, PA. December 11-15, 2008

103. Kang S-HL[1], Shchelochkov O[1], Pursley AN[1], Breman AM[1], Sheppard C[2], Bacino CA[1], Dagli A[3], Zori RT[3], Lalani SR[1], Patel A[1], Cheung SW[1]. GATA4 Gene Deletions Identified by Clinical aCGH Analysis in Cases with Congenital Heart Defects Observed on Fetal Ultrasound. American Society of Human Genetics, 58th Annual Meeting, Philadelphia, PA. December 11-15, 2008

104. Milunsky JM[2], Maher T[1], Zhao G[1], Chitayat D[3], Cunningham M[4], Meschino W[5], Megarbane A[6], Stalker H[7], Zori R[7], Lin A[8]. Clinical and Molecular Insights into Branchio-Oculo-Facial Syndrome (BOFS).

ABSTRACTS (continued):

105. Correction of PKU in the Pah^{enu2} mouse by Phenylalanine Ammonium Lyase: Maternal, Biochemical, and Behavioral Studies. P. Laipis[1], W. Zeile[1], J. Embury[1], M. Lewis[2], R. Zori[3], D. Musson[4] , B. Zhao[4] , C. Santamaria[4] , S. Bell[4] A. Cheng[4], L. Tsuruda[4] . American Society of Human Genetics, 59th Annual Meeting. Honolulu, Hawaii. October 24-29, 2009

NON-REFEREED PUBLICATIONS:

1. Zori RT, Angelman Syndrome: Results from a parent questionnaire, Raymond C. Philips Unit Newsletter, February 1991.

2. Zori RT, Ruvalcaba-Myhre-Smith Syndrome, Raymond C. Philips Unit Newsletter, January 1992.

3. Zori RT, Gray BA; Fluorescence In Situ Hybridization, Regional Genetics Newsletter, Spring 1994

4. Zori RT, Cancer Cytogenetics, Regional Genetics Newsletter, Spring 1995.

5. Zori RT, Ketogenic Diet as Therapy for Intractable Epilepsy, Winter 1995.

6. Stalker H, Zori RT; X-linked Mental Retardation, Raymond C. Philips Unit Newsletter, December 1996.

7. Infant Screening Task Force Recommendations and Report (screening, identifying, diagnosing and managing newborns at risk for metabolic, endocrine and genetic disorders and hemoglobinopathies) September 1, 2002 (member of task force)

SEARCH COMMITTEES:

Cytogenetic Laboratory Director, Assistant Professor, Department of Pediatrics, University of Florida, 1990

Genetic Counselor, Department of Pediatrics, University of Florida, 1991, 1994, 1997

Chair, Cytogenetics Laboratory Co-Director, Assistant Professor, Department of Pediatrics, University of Florida, 2002-2003, Dr. Dennis Hired

Chair, Biochemical Geneticist, Assistant Professor, Department of Pediatrics, 2006, Dr. Bryce Heese Hired
Director, Florida State Newborn Screening Laboratory, Jacksonville Florida 2007
Faculty, Department of Pathology, University of Florida, 2008

PART-TIME CLINICAL EMPLOYMENT OR LOCUM TENEMS:

In-house coverage, Neonatal Intensive Care Unit, Baystate Medical

PART-TIME CLINICAL EMPLOYMENT OR LOCUM TENEMS (continued):

Center, Springfield, Massachusetts, July 1981-June 1983
Staff pediatrician, Department of Pediatrics, Skellefteaa, Sweden, June 1986
Physician call (evening and weekends), Sunland Center, Gainesville Department of Health and Rehabilitation Services (Sunland Center is an institution with 1200 mentally retarded residents and a 60 bed licensed hospital), State of Florida, Gainesville, Florida, 1988-1990
Pediatric attending for family practice program, Alachua General Hospital, Department of Community Health and Family Practice, University of Florida, Gainesville, Florida, 1988-1990
Emergency room pediatrician, Shands Teaching Hospital, University of Florida, Gainesville, Florida, 1990
Medical consultant/on-site physician for Alachua County (Florida) Safety Patrol trip to Washington, D.C. (medical responsibility for 1200 children), June 10-15, 1991
Medical consultant/on-site physician for Alachua County (Florida) Safety Patrol trip to Washington, D.C. (medical responsibility for 300 children), June 7-11, 1995
Emergency room coverage, Patrick Air Force Base, Cocoa Beach, Florida, May 1992
Family Practice, Hoopa Indian Reservation Clinic, Hoopa Valley, California, April 1993
"Pediatric After Hours," On-call attending for evenings/weekends ambulatory care service, University of Florida, Gainesville, Florida, 1994-1996
Primary care provider for University of Florida pediatric primary care initiative, Gainesville, Florida, 1994
Pediatrician, Pine Ridge Indian Hospital, Pine Ridge (Lakota) Indian Reservation, South Dakota, July-August 1995; August 1996; March 1997; June 1999
Evening and weekend general pediatric coverage, Jackson Memorial Hospital, Marianna, Florida, 1996
Primary care attending for University of Florida Division of Primary Care, Department of Pediatrics, Gainesville, Florida, 1996-1997