

Curriculum Vitae

EDWARD R.B. McCABE, M.D., Ph.D.

Current Position: Professor and Executive Chair, Department of Pediatrics
Physician-In-Chief, Mattel Children's Hospital at UCLA
Director, UCLA Child Health Research Career Development Award
Director, Human and Molecular Development Training Program
Director, Center for Society, the Individual and Genetics
Member, Molecular Biology Institute
Member, Brain Research Institute
Member, UCLA Mental Retardation Research Center

Address: UCLA Department of Pediatrics
22-412 MDCC
10833 Le Conte Ave.
Los Angeles, California 90095-1752

Education:

1963-1967 Johns Hopkins University, Baltimore, Maryland (BA with General Honors)

1967-1969 University of Maryland School of Medicine, Baltimore, Maryland (Years 1 & 2 of M.D./Ph.D. Program; Biochemistry Department, Dr. Samuel P. Bessman, Advisor)

1969-1972 University of Southern California School of Medicine, Los Angeles, California (Years 3, 4, & 5 of M.D./Ph.D. Program, Ph.D.in Pharmacology, Dr. Samuel P. Bessman, Advisor)

1972-1974 University of Southern California School of Medicine (Years 6 & 7 of M.D./Ph.D. Program, M.D.)

Chronology of Employment:

1974-1976 Internship and Residency, Department of Pediatrics, University of Minnesota Hospitals, Minneapolis, Minnesota

1976-1978 Fellowship, Pediatric Metabolism, University of Colorado School of Medicine, Denver, Colorado

1977-1978 Instructor, Department of Pediatrics, University of Colorado School of Medicine, Denver, Colorado

1978-1982	Assistant Professor, Department of Pediatrics, University of Colorado School of Medicine, Denver, Colorado
1980-1982	Assistant Professor, Dept. of Biochemistry, Biophysics, and Genetics, University of Colorado School of Medicine, Denver, Colorado
1982-1986	Associate Professor, Departments of Pediatrics, and Biochemistry, Biophysics, and Genetics, University of Colorado School of Medicine, Denver, Colorado
1986-1988	Associate Professor, Institute for Molecular Genetics and Department of Pediatrics, Baylor College of Medicine, Houston, Texas
1986-1994	Director, R.J. Kleberg, Jr., Clinical Center, Institute for Molecular Genetics, Baylor College of Medicine, Houston, Texas
1988-1994	Director, Baylor Mental Retardation Research Center, Baylor College of Medicine, Houston, Texas
1988-1994	Director, Baylor Laboratory Training Program, Institute for Molecular Genetics, Baylor College of Medicine, Houston, Texas
1988-1994	Professor, Institute for Molecular Genetics, Baylor College of Medicine, Houston, Texas
1988-1994	Professor, Departments of Pediatrics and Obstetrics and Gynecology, Baylor College of Medicine, Houston, Texas
1990-1994	Director, Baylor Child Health Research Center, Department of Pediatrics, Baylor College of Medicine, Houston, Texas
1992-1993	Chair, Scientific Advisory Committee, Clinical Research Center, Texas Children's Hospital
1992-1994	Vice Chair for Research, Department of Pediatrics, Baylor College of Medicine, Houston, Texas
1993	Acting Director, Institute for Molecular Genetics, Baylor College of Medicine, Houston, Texas
1993-1994	Program Director, Pediatric Clinical Research Center, Texas Children's Hospital, Houston, Texas
1994	Professor and Vice Chair, Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, Texas
1994-Present	Professor and Executive Chair, Department of Pediatrics, University of California, Los Angeles
1994-Present	Director, Pediatric Research, Innovation, and Mentoring Experience (PRIME) Program, Department of Pediatrics
1995-Present	Physician-in-Chief, Mattel Children's Hospital at UCLA
1995-1998	Interim Co-Director, Human Genetics Program, University of California, Los Angeles
1996-Present	Principal Investigator (1996-Present), Program Director (1996-1999) and Core Laboratory Director (1996-Present), UCLA Child Health Research Career Development Award

1998-Present Director, Human and Molecular Development Training Program

2001-Present Director, Center for Society, the Individual and Genetics

Honors:

1961-1969 Research Fellowship, National Institute of Mental Health Neurosciences Training Program, Pediatrics Research Laboratory, University of Maryland School of Medicine, Baltimore, Maryland

1963-1964 Maryland State Scholarship, Johns Hopkins University, Baltimore, Maryland

1963-1967 Kingsville-Perry Hall Lions Club Scholarship, Johns Hopkins University, Baltimore, Maryland

1963-1967 Johns Hopkins University Honorary Scholarship, Johns Hopkins University, Baltimore, Maryland

1967 B.A. with General Honors, Johns Hopkins University, Baltimore, Maryland

1967-1968 Warfield Freshman Merit Scholarship, University of Maryland School of Medicine, Baltimore, Maryland

1968-1969 Dr. Michael Vinceguerra Scholarship, University of Maryland School of Medicine, Baltimore, Maryland

1969-1972 Research Fellowship, National Heart and Lung Institute Training Grant, Department of Pharmacology, University of Southern California School of Medicine, Los Angeles, California

1971 Sigma Xi

1972 American Society of Pharmacology and Experimental Therapeutics Workshop -- Neuropsychopharmacology, Vanderbilt University, Nashville, Tennessee

1972-1973 Aull Scholarship, University of Southern California School of Medicine, Los Angeles, California

1973 Burns-Pine Award for Alcohol Related Research, University of Southern California School of Medicine, Los Angeles, California

1974 United Cerebral Palsy Foundation-J. William Hillman Medical Student Fellowship, Department of Obstetrics and Gynecology, University of Southern California School of Medicine, Los Angeles, California

1974 Alpha Omega Alpha

1974 Phi Kappa Phi

1977-1980 Clinical Associate Physician, Children's Clinical Research Center, University Hospital, Denver, Colorado

1981-1984 Basil O'Connor Starter Research Grantee, University of Colorado School of Medicine, Denver, Colorado

- 1989 Harry A. Waisman Memorial Lectureship, University of Wisconsin, Madison, Wisconsin
- 1991 Bishop John J. Russell and Sister Rita Thomas Lectureship, St. Mary's Hospital, Richmond, Virginia
- 1993 E. Mead Johnson Award, Society for Pediatric Research
- 1995 Ben Kagan Lectureship, Cedars-Sinai Medical Center, Los Angeles, California
- 1995 J.C. Wilt Lectureship, School of Public Health, Winnipeg, Manitoba, Canada
- 1995 NICHD Lectureship, Perinatal Research Society Annual Meeting, Cape Cod, Massachusetts
- 1996 J. Edward Berk Annual Lectureship, University of California, Irvine, California
- 1997 Bilderback Lectureship, Oregon Health Sciences University, Portland, Oregon
- 1997 Harry Gordon Alumni Award for Outstanding Achievement in Academic Medicine, University of Colorado Health Sciences Center, Denver, Colorado
- 1998 Samuel W. Clausen Lectureship, University of Rochester School of Medicine, Rochester, New York
- 2000 Jimmy Simons Lectureship, Wake Forrest University School of Medicine, Winston-Salem, North Carolina
- 2001 Joseph W. St. Geme, Jr., Education Award, Western Society for Pediatric Research, Carmel, California
- 2001 Anna Borun and Harry Borun Foundation Visiting Professor in Cardiology, UCLA Department of Internal Medicine, April 12, 2001, Los Angeles, California
- 2001 Institute of Medicine
- 2002 First Florence Char, M.D., Visiting Professor, Department of Pediatrics, University of Arkansas

Societies:

American Association for the Advancement of Science
 Sigma Xi
 Phi Kappa Phi
 Alpha Omega Alpha
 Society for Inherited Metabolic Disorders-Charter Member
 Western Society for Pediatric Research
 American Society for Human Genetics
 American Academy of Pediatrics - Fellow
 Society for the Study of Inborn Errors Of Metabolism
 American Federation for Clinical Research
 Society for Pediatric Research
 Society for Newborn Screening
 New York Academy of Sciences
 American Pediatric Society
 American Society for Biochemistry and Molecular Biology
 American College of Medical Genetics -- Fellow
 The Endocrine Society

Southwest Pediatric Society
Perinatal Research Society
American Federation for Medical Research
American Diabetes Association

Outside Committees:

1977-1980	Member, Medical Advisory Board, Juvenile Diabetes Foundation, Denver Metro Chapter
1977-1983	Clinic Director, National Collaborative Study of Children Treated for PKU
1977-1986	Member, Medical Advisory Committee, Mountain States Newborn Screening Laboratory for Genetic Disease
1977-Present	National Alumni Schools Committee, Johns Hopkins University, Interviewer for Undergraduate Applicants
1978	Chair, Program Committee, First Scientific Program, Society for Inherited Metabolic Disorders
1978-1980	Medical Staff, Children's Diabetes Foundation at Denver
1978-1981	Board of Directors, Diabetes Mellitus-American Diabetes Association Colorado Affiliate
1979	Member, Task Force on Bioppterin, Subcommittee on Amino Acid Modified Diets, Committee on Nutrition, American Academy of Pediatrics
1980-1981	Consultant, Committee on Genetics, American Academy of Pediatrics
1983-1990	Member, Food and Drug Administration Orphan Drug Review Panel
1984-1986	Member, Food and Drug Administration-American Academy of Pediatrics Task Force on Special Formulas
1984-1986	Clinic Director, Maternal PKU Collaborative Study
1985	Member, National Institute of Child Health and Human Development Special Contract Review Panel
1986-1987	Consultant, Committee on Genetics, American Academy of Pediatrics
1987-Present	Member, Health and Human Services Select Panel on Newborn Screening, Maternal and Child Health Bureau
1987-1991	Chair, Committee on Genetics, American Academy of Pediatrics
1988-1994	Interagency Council for Genetics Services in Texas, Chair (1988-1991); Representative of Private Contractors (1988-1994)
1988-1989	Member, Prevention of Mental Retardation and Related Disabilities in Texas Steering Committee
1988-1994	Member, Texas Sickle Hemoglobinopathy Advisory Committee

1988-1989	Consultant, National Institute of Child Health and Human Development Program Project Grant Review Panel
1988-1996	Maternal and Child Health Genetic Disease Program Ad Hoc Objective Review; Member (1988); Chair (1989, 1990, 1991, 1992, 1993, 1994, 1995, and 1996)
1989-1990	Member, Local Organizing Committee, Fifth International Congress of Inborn Errors of Metabolism
1989	Member, Nutritional Therapy of Inborn Errors of Metabolism, NIH Ad Hoc Review Panel
1989	Ad Hoc Member, NIH Biochemistry Study Section
1989-1992	Mental Retardation Research Committee, National Institute of Child Health and Human Development, Member (1989-1991); Chair (1991-1992)
1989-1994	American Academy of Pediatrics Representative, Newborn Screening Committee, Council of Regional Networks for Genetic Services
1989	Member, Human Genetics Branch Site Visit Committee, National Institute of Child Health and Human Development
1990-1995	Section on Genetics, American Academy of Pediatrics, CoFounder (1990); Member of the Executive Committee and Program CoChair (1990-1993), Chair of the Executive Committee (1993-1995)
1990-1994	Delegate Representing the Texas Genetics Network, Council of Regional Networks for Genetic Services
1991-1994	Member, Advisory Board, Children's PKU Network
1991-1993	Chair, Subcommittee on Biochemical Proficiency Testing, and Member, Genetic Services Committee, American Society of Human Genetics
1991-1994	Member, Advisory Committee, State Governments and the Human Genome Project, The Council of State Governments
1992-1997	Member, Board of Directors, and Rules and Regulations Committee, and Chair, Accreditation Committee (1992-1994); Vice President (1993-1994); President (1995-1996); Member, Recertification Committee (1996-1997); Member, Credentials Committee (1997); American Board of Medical Genetics
1992-1994	American Academy of Pediatrics Representative, Consortium on Rare Diseases, Orphan Products Board, Food and Drug Administration
1992	Ad Hoc Member, Grant Review Panel, March of Dimes Birth Defects Foundation
1992-1994	Chair, Scientific Advisory Committee on Neural Tube Defects, Department of Health, State of Texas
1992-Present	Member, National Institutes of Health Reviewers Reserve
1992-1994	Member, Texas Office of Prevention of Developmental Disabilities Advisory Committee
1993-1994	Chair, Mental Retardation Research Center Directors

1993-1997	Member, Medical Genetics Residency Review Committee, Accreditation Council for Graduate Medical Education
1993-1994	Member, Working Group, NIGMS Human Genetic Mutant Cell Repository
1993-1994	Member, Nominations Committee, American Pediatric Society
1993-1999	Member, Joint Committee on Professional Practices, American College of Medical Genetics
1993-1995	Genetic Screening Subcommittee, Clinical Practice Committee, American College of Medical Genetics; Member (1993-1994); Chair (1994-1995)
1993-1995	Member, Quality Assurance Subcommittee and CAP/ACMG Joint Subcommittee, Laboratory Practice Committee, American College of Medical Genetics
1994-1999	Member (1994-1996) and Chair, (1997-1999), Basil O'Connor Award Committee, and Member (<i>Ex-Officio</i> ; 1997-1999), Scientific Advisory Council, March of Dimes Birth Defects Foundation
1994-1996	Chair, National Institutes of Health Technical Assessment Conference on Gaucher Disease
1995-1996	President, Council of Medical Genetics Organizations
1995-1999	Association of Medical School Pediatric Department Chairmen, Member of the Executive Committee (1995-1998); Chair, Frontiers in Science Program (1995-1999)
1995-1996	Chair, Rapid Action Task Force on Informed Consent for Genetic Research, American Society for Human Genetics
1995-1998	Member (1995-1997) and Chair (1998), Scientific Advisory Committee, Hereditary Disease Foundation
1995-Present	Member, International Healthcare Leadership Council; Member, Health Advisory Board, Starbright
1995-1999	Member, National Advisory Child Health and Human Development Council of the National Institutes of Health; Member (1995-1997) and Chair (1998-1999), Subcommittee on Planning and Policy; Chair (1998-1999), Subcommittee on Center for Research for Mothers and Children
1995-2000	American Pediatric Society Representative to the Council on Pediatric Education, American Academy of Pediatrics; Member, Pediatric Subspecialists of the Future Work Group, Future of Pediatric Education II (FOPE)
1996-2000	Member, Committee on Certification, Subcertification, and Recertification (COCERT), American Board of Medical Specialties
1997-Present	Member, Selection Committee, Pediatric Scientist Development Program
1997-2000	Member, Program Committee, American Society of Human Genetics
1998-Present	Member, Biochemical Genetics Committee, Association of Professors of Human and Medical Genetics

1998-Present	Member, Genetic Testing Workgroup, Centers for Disease Control
1998	American Academy of Pediatrics Representative, Workshop on Ethical, Legal and Social Issues Surrounding Children in Clinical Research, Institute of Medicine
1998-2000	Member, NICHD Planning Committee for the Consensus Development Conference on Screening and Management for Phenylketonuria
1998	Commentator, Fragile X Syndrome Consensus Development Conference, NICHD
1998-2000	Member, Robert Wood Johnson Folic Acid Panel
1999-2004	President-Elect (1999-2000); President (2001-2002); and Immediate Past President (2003-2004), American College of Medical Genetics
1999-Present	Member, Selection Committee, Samuel Rosenthal Foundation Prize for Academic Achievement in Pediatrics
1999-2000	Co-Chair, Newborn Screening Task Force, Maternal and Child Health Bureau and American Academy of Pediatrics
1999-2002	Chair, Secretary's Advisory Committee on Genetic Testing
1999-Present	Member, Advisory Board, Rx Laughter
1999-Present	Member, External Advisory Board, Yale Child Health Research Center
1999-Present	Member, External Advisory Board, Minnesota Child Health Research Center
1999-Present	Member, Sponsor Committee, Glazer Pediatric Clinical Trials Network
2000-Present	Member, American Board of Pediatrics Subspecialties Committee
2001	President-Elect, Western Society for Pediatric Research
2001-2002	Member, Human Cloning Panel, Committee on Science, Engineering and Public Policy, National Academy of Sciences
2002	President, Western Society for Pediatric Research
2002	Co-Chair, Workshop to Develop Newborn Screening Technology for SCID, National Institute of Child Health and Human Development

Other Activities:

University of Southern California School of Medicine

1970-1974	Teaching Assistant, Department of Pharmacology
1972	Participant, American Society of Pharmacology and Experimental Therapeutics Workshop on Neuropsychopharmacology - Fourth Supplementary Training Program in Pharmacology, Vanderbilt University, Nashville, Tennessee

University of Colorado School of Medicine

Committee Activities:

1977-1979	Summer Camp Medical Director, Diabetes Mellitus -- American Diabetes Association, Colorado Affiliate
1978-1982	Alternate for Department of Pediatrics and Member of High-Risk Subcommittee, Human Subjects Committee
1978-1982	Medical Student Advisor, years 3 and 4
1979-1981	Research Coordinator, Diabetes Mellitus -- American Diabetes Association, Colorado Affiliate
1979-1986	By-Laws Committee of the Medical Staff, University Hospital
1980-1981	Chair, Research Committee, Department of Pediatrics
1980-1986	Member, Interdepartmental Genetics Committee
1980-1986	Graduate School Faculty
1981	Health Care Quality Assurance Committee, Consultant for Review of Pediatric Outpatient Management of Juvenile Diabetes
1981-1982	Chair, Curriculum Committee, Interdepartmental Genetics Program
1981-1982	Assistant Director, B. F. Stolinsky Research Laboratories, Department of Pediatrics
1981-1983	Chair, Housestaff Program Committee, Department of Pediatrics
1981-1986	Faculty Incentive Fund Committee, Department of Pediatrics
1981-1986	Affiliate Member, Nutrition Institute, Colorado State University
1982-1983	Member, Human Subjects Committee
1982-1986	Medical Student Advisor, Years 3 and 4, M.D.-Ph.D. Students
1983-1986	Associate Director, B. F. Stolinsky Research Laboratories, Department of Pediatrics
1984-1985	Acting Head, Section of Genetics, Department of Pediatrics
1984-1985	Acting Director, B.F. Stolinsky Research Laboratories, Department of Pediatrics
1985-1986	Member, Steering Committee and Recruiting Subcommittee, Medical Scientist Training Program (MSTP)

Teaching Activities:

1976-1986	Introduction to Clinical Medicine (Medical Students, Year 2) and Practical Instruction (Medical Students, Year 3, Residents, and Fellows), Department of Pediatrics
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- 1979-1986 Topics in Medical Genetics (Masters Students), Supervise Laboratory Rotations (Graduate Students) and Member of Masters and Doctoral Examination Committees, Department of Genetics
- 1981 Tutoring Doctoral Student in Metabolic Regulation for Comprehensive Exams, Department of Genetics
- 1981-1982 Supervise Student Internship, M.A. Program in Applied Early Child Development, Department of Psychology, University of Colorado at Denver
- 1982 Co-Organizer of Interdepartmental Course, "Principles of Gene and Metabolic Regulation: An Aid to Understanding Human Diseases" (Advanced Medical Students and Graduate Students), Departments of Biochemistry, Biophysics, Genetics, Medicine and Pediatrics
- 1982-1984 Clinical Correlation (Medical Students, Year 1), Department of Biochemistry

Clinical Activities:

- 1976-1980 Medical Staff, Pediatric Diabetes Clinic, University Hospital
- 1977-1981 Attending Physician, Pediatric Intensive Care Unit, Denver Children's Hospital
- 1977-1982 Attending Physician, Pediatric Metabolism, Denver Children's Hospital
- 1977-1986 Medical Director, Inherited Metabolic Diseases Clinic, University Hospital
- 1977-1986 Attending Physician, Pediatrics Wards, University Hospital
- 1977-1986 Advisory Committee, Children's Clinical Research Center, University Hospital
- 1980-1986 Medical Staff, Genetics Clinic, University Hospital
- 1982-1986 Attending Physician, Pediatric Genetics, Denver Children's Hospital

Baylor College of Medicine

Committee Activities:

- 1986-1989 Faculty Advisor, Bugher Molecular Cardiology Program
- 1986-1994 Training Faculty, Medical Genetics Research Fellowship Training Program
- 1987-1994 Faculty Advisor, Medical Scientist Training Program
- 1987-1989 Chair, Training Committee, Joseph P. Kennedy, Jr. Mental Retardation Fluid Research Grant
- 1988-1990 Member, Graduate Program Standing Examination Committee, Institute for Molecular Genetics
- 1988-1994 Director, Baylor Mental Retardation Research Center

1988-1994	Director, Baylor Laboratory Training Program
1988-1994	Member, Housestaff Selection Committee, Department of Pediatrics
1989-1994	Member, Clinical Investigation and Publications Committee, Texas Children's Hospital
1989-1994	Member, Department Seminar Committee, Institute for Molecular Genetics
1989-1994	Member, Development and Operations Advisory Committee, Institute for Molecular Genetics
1990	Chair, Liaison Committee on Medical Education Academic Unit Review Team, Department of Biochemistry
1990-1994	Member, Departmental Promotions Committee, Institute for Molecular Genetics
1990-1994	Program Director and Core Laboratory Director, Baylor Child Health Research Center
1990-1994	Member, Baylor Research Advisory Committee
1990-1994	Chair, Clinical Faculty Recruitment Committee, Institute for Molecular Genetics
1992-1994	Member, Executive Committee, Postdoctoral Training Program in Maternal/Infant/Child Nutrition, Baylor Clinical Nutritional Research Center
1992-1994	Member, Advisory Board, Molecular Biology Computer Resource
1992-1994	Chair, Clinical Services Committee, Institute for Molecular Genetics
1992-1994	Chair, Scientific Advisory Committee, Clinical Research Center, Texas Children's Hospital
1992-1994	Member, Medical Scientist Training Program Operating Committee
1993	Member, Search Committee for the Director, Clinical Nutrition Center
1993-1994	Member, Advisory Committee, Molecular Medicine Program
1993-1994	Program Director, Pediatric Clinical Research Center, Texas Children's Hospital, Houston, Texas
1993-1994	Chair, Space Committee, Institute for Molecular Genetics
1993-1994	Member, Department of Molecular Physiology and Biophysics Academic Unit Review Committee
1993-1994	Member, Advisory Committee, Gene Therapy Program
1994	Member, Academic Council, Department of Pediatrics

Teaching Activities

1986-1994	Practical Instruction to Medical Students, Residents and Fellows, Institute for Molecular Genetics
1986-1994	Practical Instruction to Medical Students, Residents and Fellows, Department of Pediatrics

1988-1990 Member, Graduate Student Examinations Committee, Institute for Molecular Genetics

1992-1994 Member, Selection Committee, Medical Genetics Postdoctoral Fellowship Program, Institute for Molecular Genetics

1992-1994 Course Organizer and Lecturer, Academic Pediatrics, Department of Pediatrics

1993-1994 Faculty Member, Graduate Program in Cell and Molecular Biology

Clinical Activities:

1986-1994 Director, R.J.. Kleberg, Jr. Clinical Center, Institute for Molecular Genetics

1986-1994 Medical Staff, Medical Genetics Clinic, Institute for Molecular Genetics

1986-1994 Medical Staff, Metabolic Disease Clinic, Institute for Molecular Genetics

1986-1994 Attending Physician, Texas Children's Hospital

1986-1994 Medical Staff, St. Luke's Episcopal Hospital

1986-1994 Medical Staff, The Methodist Hospital

1986-1994 Medical Staff, Harris County Hospital District

1989-1994 Medical Staff, St. Joseph's Hospital

1991-1994 Medical Staff, Women's Hospital

UCLA School of Medicine

Committee Activities:

1994-1997 Member, Search Committee for Chair, Department of Genetics

1994-Present Editorial Board, UCLA Medicine

1995-1999 Member, Clinical Advisory Council

1995-1996 Chair, Search Committee for Chair, Department of Surgery

1995-Present Member, UCLA Medical Group Board

1995-Present Member, Mental Retardation Research Center Faculty Advisory Committee

1995-Present Member, Medical Staff Executive Committee

1995-1998	Member, Credentials Committee
1995-Present	Member, Clinical Scholars Policy Advisory Committee
1995-1999	Member, Clinical Effectiveness Executive Steering Committee
1996-1999	Member, School of Medicine Frontiers of Medical Science Bridge Award Committee
1996-1997	Member, Rheumatology Search Committee
1996-Present	Member, School of Medicine Lectureship Committee
1996-Present	Member, Cancer Genetics Working Group
1996-1999	Member, Master Plan Steering Committee
1997-1998	Chair, Programs and Partnerships Committee, UCLA Medical Enterprise Master Planning
1997	Member, R-Net Research Community Panel
1997-1999	Member, School of Medicine Research Advisory Committee
1997-1999	Member, Design/Public Areas Planning Committee, UCLA Medical Center Physical Planning and Programming
1997-Present	Member, Human Research Policy Board; Chair, Subcommittee on Genetic Research (1998-Present)
1997-Present	Member, Faculty Advisory Committee, UCLA-DOE Lab of Structural Biology and Molecular Medicine
1998-Present	Member, Dean's Focus Group on Genetics/Genomics
1998-Present	Member, Dean's Focus Group on Developmental Biology
1998	Member, Gordon and Virginia MacDonald Distinguished Chair in Human Genetics Committee
1998-Present	Member, Health Services Research Policy Committee
2000-Present	Member, Clinical Enterprise Executive Committee; Chair, Finance Subcommittee
2000-2002	Chair, Neurology Chair Search
2000-Present	Member, Clinical Pharmacology Core Group
2001-Present	Member, Search Committee for the Division of Digestive Diseases Hepatology Group Faculty
2001-Present	Member, Hospital Board of Advisors
2002	Member, Advisory Group for the Selection of the Executive Vice Chancellor at UCLA

Teaching Activities:

1994-Present	Course Organizer and Lecturer, Academic Pediatrics, Department of Pediatrics
1994-Present	Member, Executive Committee, and Faculty Member, UCLA Intercampus Medical Genetics Training Program
1994-Present	Practical Instruction to Medical Students, Residents and Fellows, Department of Pediatrics
1994-Present	Weekly Chief's Rounds
1995-Present	Faculty, UCLA ACCESS to Graduate Programs in the Molecular and Cellular Life Sciences
1995-Present	Lecturer, UCLA Intercampus Genetics Postdoctoral Training Program
1996 - Present	Member, Advisory Board, Molecular Aspects of Mental Retardation
1996-Present	Co-Organizer (1996-1997), Lecturer (1996-Present) and Course Director (2002-Present), Medical Genetics Course for Medical Students
1996-Present	STAR Program Advisor for the Department of Pediatrics
1997	Co-Organizer and Lecturer, Molecular Genetics 297, Evolution of Sex Determination
1997-Present	Co-Organizer and Lecturer, Leadership Workshop, Department of Pediatrics
1998	Organizer, Spring 1998 Human Genetics Seminars, Molecular Biology Institute
1999	Co-Organizer and Lecturer, Molecular Genetics 297, Molecular Mechanisms of Sex Determination and X-Inactivation
1999-Present	Lecturer, Undergraduate Molecular Genetics Course
2001	Lecturer, Occupational Health Course, School of Nursing

Clinical Activities:

1995-Present	Physician-in-Chief, Mattel Children's Hospital at UCLA
1997-Present	Clinical Director, Spina Bifida Clinic

Editorial Boards:

1979-1987	Biochemical Medicine
1987-1990	Associate Editor, Biochemical Medicine and Metabolic Biology
1989-1994	Associate Editor for Research News, National PKU Newsletter
1990-Present	Editor, Biochemical Medicine and Metabolic Biology (1990-1994); Biochemical and Molecular Medicine (1995-1997); Molecular Genetics and Metabolism (1998-Present)
1991-1994	Advisory Board, Screening

1991-1994	Communicating Editor, Human Mutation
1993-1994	Associate Editor, American Journal on Mental Retardation
1995-1997	Biochemical Genetics Section Editor, American Journal of Medical Genetics
1996	Guest Editor, The Molecular Medicine of Mental Retardation and Developmental Disabilities, Mental Retardation and Developmental Disabilities Reviews

Occasional Consulting Editor:

Science
 Muscle and Nerve
 Pediatric Research
 American Journal of Diseases of Children
 Journal of Pediatrics
 Clinical Chemistry
 American Journal of Human Genetics
 American Journal of Medical Genetics
 Dysmorphology and Clinical Genetics
 Pediatrics
 BioTechniques
 Journal of Genetic Counseling
 Nucleic Acid Research
 Proceedings of the National Academy of Sciences, USA
 Human Genetics
 American Journal of Physiology: Endocrinology and Metabolism
 PCR Methods and Applications
 Biochemical Genetics
 European Journal of Biochemistry
 Genomics
 Prenatal Diagnosis
 Human Mutation
 Human Molecular Genetics
 Journal of Epidemiology
 Journal of the American Medical Association
 Prenatal Diagnosis
 Circulation
 Archives of Medical Research
 Journal of Clinical Investigation
 Journal of Clinical Endocrinology and Metabolism
 Trends in Genetics

Licensure:

State of California G-39458, June 18, 1979

State of Texas H0082, August 23, 1986

Board Certification:

Diplomate, National Board of Medical Examiners (No. 119535, July 1, 1975)

Diplomate, American Board of Pediatrics (No. 22685, April 8, 1979)

Diplomate, American Board of Medical Genetics, Clinical Genetics and Biochemical Genetics (No.1325, March 19, 1982)

Founding Fellow AMA, M.D. of the American College of Medical Genetics (February 1, 1993)

Active Candidate Status, Clinical Molecular Genetics, American Board of Medical Genetics (February 10, 1993)

Laboratory Certification:

Laboratory Director, Genetic Testing, New York State Department of Health (MCCAE1; May 8, 1995)

Research Interests: Developmental Molecular Genetics

Inborn Errors of Metabolism

Newborn Screening

Grants:

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| 1980-1984 | R01 AM26265, Developmental Impact of Glycerol Kinase Deficiency (\$139,474) National Institute of Arthritis, Diabetes, Digestive and Kidney Disease |
| 1981-1984 | 5-332, Basil O'Connor Starter Research Grant, Human Glycerol Kinase: Developmental Biochemistry and Investigations of the Inherited Deficiency State (\$66,874) March of Dimes Birth Defects Foundation |
| 1984-1987 | 2 R01 HD08315, Studies on Glycerol Kinase Deficiency (\$218,490) National Institute of Child Health and Human Development |
| 1986-1987 | RR-052425 (BRSG) Molecular Genetic Studies of Glycerol Kinase Deficiency (\$9,772) National Institutes of Health |
| 1987-1988 | The Joseph P. Kennedy, Jr., Foundation Mental Retardation Fluid Research Grant (\$30,000) The Kennedy Foundation |
| 1987-2004 | 2 R01 HD22563 Molecular Genetic Studies of Glycerol Kinase Deficiency (\$2,175,235) National Institute of Child Health and Human Development; Includes Minority Supplements for Desiree Sylvester-Jackson, Ph.D. and Riki Ohira |
| 1988-1991 | 18-88-18 Expression and Regulation of Hexokinase in HKI Deficiency and Cardiac Development (\$36,000) Predoctoral Graduate Research Training Fellowship to Lisa Griffin, Sponsor: Edward R.B. McCabe, M.D., Ph.D., March of Dimes Birth Defects Foundation |
| 1988-1991 | M CJ-480566, Sickle Cell Anemia: DNA for Newborn Screening Follow-up (\$199,972) Maternal and Child Health and Research Resources |
| 1988-1998 | 2 P30 HD24064, Baylor Mental Retardation Research Center (\$4,538,483) National Institute of Child Health and Human Development |

1988-1994 MCJ-009121, Baylor Laboratory Training Program (\$824,401) Maternal and Child Health Bureau

1989 The Joseph P. Kennedy, Jr. Foundation Mental Retardation Fluid Research Grant (\$10,000), The Kennedy Foundation

1989-1991 NIH NRSA 1 F32 GM13063 Human Hexokinase I Gene and Its Abnormal Variants (\$64,750), Postdoctoral Fellow: Bruce D. Gelb, M.D.; Sponsor: Edward R.B. McCabe, M.D., Ph.D.

1990-1995 1 P30 HD27823 Baylor Child Health Research Center: Molecular Medicine Approaches to Pediatrics (\$1,542,510) National Institute of Child Health and Human Development, Principal Investigator: Ralph D. Feigin, M.D.; Program Director and Core Laboratory Director: Edward R.B. McCabe, M.D., Ph.D.

1990-1992 MCJ-481007 Newborn Screening: Applications of Molecular Genetic Technology (\$144,280) Maternal and Child Health and Research Resources

1990-1995 NIH CIA K08 HL02485 Molecular Linkage and Cloning of X-Linked Cardiomyopathy (\$340,000) Clinical Investigator: Jeffrey A. Towbin, M.D.; Sponsor: Edward R.B. McCabe, M.D., Ph.D.

1991-1994 1 P30 HD00210 Baylor College of Medicine Human Genome Program Center, National Center for Human Genome Research; Principal Investigator: C. Thomas Caskey, M.D.; Molecular Medicine Core Director: Edward R.B. McCabe, M.D., Ph.D. (\$481,759)

1992-1995 MCJ-487G01 DNA/RNA Diagnosis of Thalassemia in Newborn Screening (\$201,006) Maternal and Child Health Bureau

1992-1995 MCJ-487G02 Automated DNA Analysis for Hemoglobin Screening (\$317,544) Maternal and Child Health Bureau

1992-1993 1 R03 RR07841 Automated DNA Testing for Screening and Diagnosis (\$35,000) National Center for Research Resources

1993-1996 NIH NRSA 1 F32 HL03349 Hexokinase in Developing Rat Lung (\$89,700), Postdoctoral Fellow: Susan M. Gray, M.D., Sponsor: Edward R.B. McCabe, M.D., Ph.D.

1993-1994 R13 CCR609182 Public Health Conference Grant - International Conference on Neural Tube Defects (\$20,000) Centers for Disease Control

1993-1995 Brain Hexokinase in Ischemic and Reperfused Piglets (\$100,000) United Cerebral Palsy Research and Educational Foundation, Inc.

1993-1994 International Conferences on Neural Tube Defects (\$10,000) March of Dimes Birth Defects Foundation

1994-1997 Coactivators of the Progesterone Receptor (\$78,000), American Cancer Society, Postdoctoral Fellow: Thomas P. Burris, Ph.D., Sponsor: Edward R.B. McCabe, M.D., Ph.D.

1996-2003 Research Resources Program for Medical Schools, Howard Hughes Medical Institute; Principal Investigator: Lenny Rome, Ph.D.; Disease Mapping Core Director and PRIME Program Director: Edward R.B. McCabe, M.D., Ph.D. (\$1,075,392)

1996-1998 Postdoctoral Research Fellowship (\$36,000), INSERM; Fellow: Eric Vilain, M.D., Ph.D.;

Sponsor: Edward R.B. McCabe, M.D., Ph.D.

- 1996-2001 NHLBI MCSDA Characterization of the EpoR Fusion Partner in TF-1 Cells (\$370,000), Principal Investigator: Lisa Schimmenti, M.D.; Sponsor: Edward R.B. McCabe, M.D., Ph.D.
- 1996-1997 Rapid Diagnosis of Infection and Identification of Pathogenic Bacteria (\$3,100) UCLA Academic Senate Research Award
- 1996-2006 K12 HD34610, NICHD, UCLA Child Health Research Career Development Award (\$3,174,495)
- 1996-1999 K08 HD01103, NICHD, Molecular Genetics of Rett Syndrome (\$281,934), Principal Investigator: N. Carolyn Schanen, M.D., Ph.D.; Sponsor: Edward R.B. McCabe, M.D., Ph.D.
- 1997-1998 Genentech Foundation for Growth and Development (\$45,455); Fellow: Kevin Kaiserman, M.D.; Sponsor: Edward R.B. McCabe, M.D., Ph.D.
- 1997-2002 K08 DK0511, NIDDK, DAX1 Function in Adrenocortical Development (\$392,500), Principal Investigator: Kevin Kaiserman, M.D.; Sponsor: Edward R.B. McCabe, M.D., Ph.D.
- 1997-1998 SBIR, NIAID, Universal PCR for Identification of Emerging Bacteria (\$21,711), Principal Investigator: Sandy Savall, Ph.D.; Subcontractor: Edward R.B. McCabe, M.D., Ph.D.
- 1998-2003 T32 HD07512, Human and Molecular Development Training Program (\$846,192)
- 1999-2000 American Academy of Pediatrics Section on Genetics and Birth Defects 1999 Young Investigator Research Grant, Molecular Mechanisms of Glycerol Kinase Deficiency (\$20,000), Principal Investigator: Katrina Dipple, M.D., Ph.D.; Sponsor: Edward R.B. McCabe, M.D., Ph.D.
- 2000-2001 Bank of America-Giannini Foundation, Molecular Mechanisms of Glycerol Kinase Deficiency (\$30,000); Principal Investigator: Katrina Dipple, M.D., Ph.D.; Sponsor: Edward R.B. McCabe, M.D., Ph.D.
- 2000-2005 R01 HD39233, Molecular Mechanisms of Adrenal Development (\$1,025,000)
- 2000-2002 Maternal and Child Health Bureau, Newborn Screening for the Hemoglobinopathies (\$225,000)
- 2001-2006 K08 DK60055, Molecular Mechanisms of Glycerol Kinase Deficiency (\$563,750), Principal Investigator: Katrina Dipple, M.D., Ph.D.; Mentor: Edward R.B. McCabe, M.D., Ph.D.
- 2001-2004 F32 HD40738, Structural Analysis of DAX1 (\$104,937), Fellow: James K. Phelan, Ph.D.; Mentor: Edward R.B. McCabe, M.D., Ph.D.
- 2001-2005 240-MCHB-01, Consent Process for Newborn Screening (\$1,267,825)
- 2002 American Society of Human Genetics, Oral History of Medical Genetics (\$25,000)
- 2002-2007 National Institute for Biomedical Imaging and Bioengineering, Uropathogen Detectin Using DNA Biosensors, Principal Investigator: Bernard Churchill, M.D.; Probe Development: Edward R.B. McCabe, M.D., Ph.D. (\$350,000)

Formal Private Sector Relationships:

1991-1997	Academic Associate, Nichols Institute, San Juan Capistrano, California
1997-1998	Sub-Contractor, Small Business Innovation Research Grant, Specialty Labs, Santa Monica, California
2002-Present	Consultant, AMGEN
2002-Present	Member, Advisory Council, Genetic Leadership Collaborative, Genzyme

Trainees:

1980-1985	William K. Seltzer, Ph.D., Postdoctoral Fellow, Biochemistry of Glycerol Kinase
1983-1984	Cynthia L. Freehauf, Master's Thesis, Carrier Screening for Phenylketonuria
1985-1986	Catherine M. Walsh Vockley, Master's Thesis, Characterization of Phenylalanine Hydroxylase Restriction Fragment Length Polymorphisms in a Mixed American Population
1986-1989	Jeffrey A. Towbin, M.D., Bugher Molecular Cardiology Fellow, Cloning the Human Glycerol Kinase cDNA
1987-1991	Lisa Griffin, Medical Scientist Training Program Student, Developmental Molecular Genetics of Hexokinase I
1988-1991	Maria Descartes, M.D., Baylor Laboratory Training Program Fellow, Application of Recombinant DNA Techniques to Newborn Screening
1989-1991	Bruce D. Gelb, M.D., Pediatric Cardiology Fellow, Human Hexokinase I Gene and Its Abnormal Variants
1989-1993	Kim Worley, Molecular Genetics Graduate Student, Glycerol Kinase Deficiency and Congenital Adrenal Hypoplasia: Genomic Mapping in Xp21
1990-1992	Volker Adams, Ph.D., Molecular Genetics Postdoctoral Fellow, Enzyme Targeting in Energy Microcompartmentation: Developmental Biology
1991-1994	Katie Coerver, Medical Scientist Training Program Student, Regulation of Expression of Hexokinase Isoenzyme I (HKI)
1991-1992	Michael Levin, M.D., Medical Genetics Postdoctoral Training Program, Automated DNA Screening for MCAD Deficiency, A Treatable Cause of SIDS
1991-1995	Desiree Sylvester-Jackson, Ph.D., Molecular Genetics Postdoctoral Fellow, Molecular Genetic Diagnosis of Sickle Cell Disease
1992-1994	Susan Gray, M.D., Neonatology Fellow, Hexokinase Expression in the Lung
1993-1994	Margaret Sampson, Molecular Genetics Graduate Student, Glycerol Kinase Knockouts: Generation of a Glycerol Kinase Deficient Mouse Model

1993-1995	Sriya Gunawardena, M.D., Hematology/Oncology Fellow, Hexokinase in Tumor Cells
1994-1996	Gary Kaselonis, M.D., Neonatology Fellow, Studies of Hexokinase in Mammary Gland During Lactation
1994-1996	Ghazala Khan, M.D., Critical Care Fellow, Automated DNA Triage for Infectious Disease
1994	Tina Corkran, Molecular Genetics Graduate Student, Structure/Function Relationship of Hexokinase
1994-1998	Weiwen Guo, M.D., Ph.D. Cloning and Characterization of the Gene Responsible for X-Linked Adrenal Hypoplasia Congenita
1994-1996	Thomas P. Burris, Ph.D., X-linked Adrenal Hypoplasia Congenita: Member of the Nuclear Hormone Receptor Superfamily
1995-1998	Kevin Kaiserman, M.D., Mutations Associated with Congenital Adrenal Hypoplasia and Hypogonadotropic Hypogonadism
1996-1998	Eric Vilain, M.D., Ph.D., Gene Therapy for Congenital Adrenal Hypoplasia
1996-2002	Megha Patel, Identification of the DAX1 Gene in the Zebra Finch
1996-Present	Robert Clipsham, D.V.M., Interaction of DAX1 and SF-1
1996-2002	Katrina Dipple, M.D., Ph.D., Channel Function of Porin
1999-2001	Koji Nagano, M.D., Ph.D., Gene Therapy for Glycerol Kinase Deficiency
1999-Present	Riki Ohira, Mutations in Glycerol Kinase Deficiency
1999-Present	Phoebe Dewing, DAX1 and Other Genes in Adrenal Development
1999-Present	Jim Phelan, Ph.D., Crystal Structure of DAX1
2000-Present	Krzysztof Stanczak, DAX1 and Other Genes in Adrenal Hypoplasia Congenita
2001-Present	Kathy Niakan, Functional Breakpoints in Glycerol Kinase Deficiency Mutations
2001-Present	John Ho, Sequence Structure for Xp21
2001-Present	Julian Martinez, M.D., Ph.D., Glycerol Kinase Deficiency in Drosophila
2001-Present	Michelle Lewis, J.D., M.D., Informed Consent for Newborn Screening
2002-Present	Anita Iyer, Identification of Genes Interacting with DAX1 and Their Interaction Motifs
2002-Present	Yan Zhao, Adrenal Development in the Zebrafish

Biobibliography

Refereed Publications:

1. **E.R.B. McCabe**, E.C. Layne, D.F. Saylor, N. Slusher and S.P. Bessman: Synergy of ethanol and a natural soporific--gamma-hydroxybutyrate. *Science* 171:404-406, 1971.
2. S.P. Bessman and **E.R.B. McCabe**: 1,4-Butanediol: A substrate for rat liver and horse liver alcohol dehydrogenases. *Biochemical Pharmacology* 21:1135-1142, 1972.
3. S.P. Bessman, P.G. Geiger, T.-C. Lu and **E.R.B. McCabe**: Separation and automated analysis of phosphorylated metabolic intermediates. *Analytical Biochemistry* 59:533-546, 1974.
4. **E.R.B. McCabe**, P.V. Fennessey, M.A. Guggenheim, B.S. Miles, W.W. Bullen, D.J. Sceats and S.I. Goodman: Human glycerol kinase deficiency with hyperglycerolemia and glyceroluria. *Biochemical and Biophysical Research Communications* 78:1327-1333, 1977.

5. S.I. Goodman, **E.R.B. McCabe**, P.V. Fennessey, B.S. Miles, J.W. Mace and E. Jellum: Methylmalonic/beta-hydroxy-n-valeric aciduria due to methylmalonyl-CoA mutase deficiency. *Clinica Chimica Acta* 87:441-449,1978.
6. R.O. Fisch, **E.R.B. McCabe**, D. Doeden, L.J. Koep, J.G. Kohlhoff, A. Silverman and T.E. Starzl: Homotransplantation of the liver in a patient with hepatoma in hereditary tyrosinemia. *Journal of Pediatrics* 93:592-596,1978.
7. M.E. Pichichero and **E.R.B. McCabe**: Recurrent Reye's syndrome. *American Journal of Diseases of Childhood* 132:1097-1099,1978.
8. **E.R.B. McCabe**, J.H. Kersey, P.R. Vandersteen and G.J. Vosika: Reverse transcriptase in a patient with scleroderma. *Biochemical Medicine* 21:47-54,1979.
9. D.A. Stumpf, **E.R.B. McCabe**, J.K. Parks, W.W. Bullen and S. Schiff: Loosely coupled oxidative phosphorylation induced by protoporphyrin *Biochemical Medicine* 21:182-189, 1979.
10. S.I. Goodman, **E.R.B. McCabe**, P.V. Fennessey and J.W. Mace: Multiple acyl-CoA dehydrogenase deficiency (glutaric aciduria type II) with transient hypersarcosinemia and sarcosinuria; possible inherited deficiency of an electron transport flavoprotein. *Pediatric Research* 14:12-17, 1980.
11. R.L. Leibel, V.E. Shih, S.I. Goodman, M.L. Bauman, **E.R.B. McCabe**, R.G. Zwerdling, I. Bergman and C. Costello: Glutaric acidemia: A metabolic disorder causing progressive choreoathetosis. *Neurology* 30:1163--1168, 1980.
12. M.A. Guggenheim, **E.R.B. McCabe**, M. Roig, S.I. Goodman, G.M. Lum, W.W. Bullen and S. Ringel: Glycerol kinase deficiency with neuromuscular, skeletal and adrenal abnormalities. *Annals of Neurology* 7: 441-449, 1980.
13. **E.R.B. McCabe**, T.R. Melvin, D. O'Brien, R.R. Montgomery, W.A. Robinson, C. Bhaskar and B.I. Brown: Neutropenia in a patient with type IB glycogen storage disease: *In vitro* response to lithium chloride. *Journal of Pediatrics* 97:944-946, 1980.
14. P.B. Acosta, P.M. Fernhoff, H.S. Warshaw, K.M. Hambidge, A. Ernest, **E.R.B. McCabe** and L.J. Elsas: Zinc and copper status of treated children with phenylketonuria. *Journal of Parenteral and Enteral Nutrition* 5:406-409, 1981.
15. P.B. Acosta, P.M. Fernhoff, H.S. Warshaw, L.J. Elsas, K.M. Hambidge, A. Ernest, and **E.R.B. McCabe**: Zinc status and growth of children undergoing treatment for phenylketonuria. *Journal of Inherited Metabolic Disease* 5:107-110, 1982.
16. **E.R.B. McCabe**, D. Sadava, W.W. Bullen, W.K. Seltzer, H.A. McKelvey and C.I. Rose: Human glycerol kinase deficiency: Enzyme kinetics and fibroblast hybridization. *Journal of Inherited Metabolic Disease* 5: 177-182, 1982.
17. S.I. Goodman, D.O. Stene, **E.R.B. McCabe**, M.D. Norenberg, R.H. Shikes, D.A. Stumpf and G.K. Blackburn: Glutaric aciduria type II: Clinical, biochemical and morphologic considerations. *Journal of Pediatrics* 100:946-950, 1982.
18. **E.R.B. McCabe**, S.I. Goodman, P.V. Fennessey, B.S. Miles, M. Wall and A. Silverman: Glutaric, 3-hydroxypropionic, and lactic aciduria with metabolic acidemia, following extensive small bowel resection. *Biochemical Medicine* 28:229-236, 1982.
19. J.A. Bartley, D.K. Miller, J.T. Hayford and **E.R.B. McCabe**: The concordance of X-linked glycerol kinase deficiency with X-linked adrenal hypoplasia in two families. *Lancet* ii:733-736, 1982.

20. W.K. Seltzer, W.W. Bullen and **E.R.B. McCabe**: Human glycerol kinase: Comparison of properties from fibroblasts and liver. *Life Sciences* 32:1721-1726, 1983.
21. **E.R.B. McCabe**: Glycerol kinase deficiency: An inborn error of compartmental metabolism. *Biochemical Medicine*, 30:215-230, 1983.
22. **E.R.B. McCabe**, L. McCabe, G.A. Mosher, R.J. Allen and J.L. Berman: Newborn screening for phenylketonuria: Predictive validity as a function of age. *Pediatrics* 72:390-398, 1983.
23. W.K. Seltzer and **E.R.B. McCabe**: Human and rat adrenal glycerol kinase: Subcellular distribution and bisubstrate kinetics. *Molecular and Cellular Biochemistry* 62:43-50, 1984.
24. C.L. Freehauf, D. Lezotte, S.I. Goodman and **E.R.B. McCabe**: Carrier screening for phenylketonuria: Comparison of two discriminant analysis procedures. *American Journal of Human Genetics* 36:1180-1189, 1984.
25. E.I. Ginns, J.A. Barranger, S.W. McClean, E. Schaefer, R.O. Brady, R Young, S.I. Goodman and **E.R.B. McCabe**: Juvenile form of glycerol kinase deficiency with episodic vomiting acidemia and stupor. *Journal of Pediatrics* 104:736-739, 1984.
26. W.K. Seltzer and **E.R.B. McCabe**: Subcellular distribution and kinetic properties of soluble and particulate-associated bovine adrenal glycerol kinase. *Molecular and Cellular Biochemistry* 64:51-61, 1984.
27. M.C. Reardon, K.B. Hammond, F.J. Accurso, C.D. Fisher, **E.R.B. McCabe**, E.K. Cotton and C.M. Bowman: Nutritional deficits exist before two months of age in some infants with cystic fibrosis identified by screening test. *Journal of Pediatrics* 105:271-274, 1984.
28. D.R Ambruso, **E.R.B. McCabe**, et al. (18 additional coauthors): Infectious and bleeding complications in patients with glycogenosis Ib: Relationship to neutrophil and platelet function. *American Journal of Diseases of Children* 139:691-697, 1985.
29. B.F. Pennington, W.J. van Doorninck, L. McCabe and **E.R.B. McCabe**: Neuropsychological deficits in early treated phenylketonurics. *American Journal of Mental Deficiency* 89:467-474, 1985.
30. W.K. Seltzer, H. Firminger, J. Klein, A. Pike, P. Fennessey, and **E.R.B. McCabe**: Adrenal dysfunction in glycerol kinase deficiency. *Biochemical Medicine* 33:189-199, 1985.
31. S.-Z. Huang, M.L. Law, W.K. Seltzer and **E.R.B. McCabe**: DNA microextraction from dried blood spots on filter paper blotters, and its application in the identification of sex. *Shanghai Medical Journal* 2:346-348, 1986.
32. D.R. Ambruso, B. Hawkins, D.L. Johnson, A.R. Fritzberg, W.C. Klingensmith and **E.R.B. McCabe**: Measurement of adenosine triphosphate and 2,3-diphosphoglycerate in stored blood with ³¹p nuclear magnetic resonance spectroscopy. *Biochemical Medicine and Metabolic Biology* 35:376-383, 1986.
33. L.C. Stork, D.R Ambruso, S.F. Wallner, J.E. Sambrano, L.C. Moscinski, H.L. Wilson and **E.R.B. McCabe**: The pancytopenia of propionic acidemia: Hematologic evaluation and studies of hematopoiesis *in vitro*. *Pediatric Research* 20:783-788, 1986.
34. W.K. Seltzer, G. Dhariwal, H.A. McKelvey and **E.R.B. McCabe**: 1- Thioglycerol: Inhibitor of glycerol kinase activity *in vitro* and *in situ*. *Life Sciences* 39:1417-1424, 1986.
35. **E.R.B. McCabe**, S.-Z. Huang, W.K. Seltzer and M.L. Law: DNA microextraction from dried blood spots on filter paper blotters: Potential applications to newborn screening. *Human Genetics* 75:213-216, 1987.

36. J.E. Wise, R Matalon, A.M. Morgan, and **E.R.B. McCabe**: Phenotypic features of patients with congenital adrenal hypoplasia and glycerol kinase deficiency. *American Journal of Disease of Children* 141:744-747, 1987.
37. A. Kohlschutter, R.P. Willig, D. Schlamp, K. Kruse, **E.R.B. McCabe**, H.J. Schafer, G. Beckenkamp and R. Rohkamm: Infantile glycerol kinase deficiency - A condition requiring prompt identification - Clinical, biochemical and morphological findings in two cases. *European Journal of Pediatrics* 146:575-581, 1987.
38. U. Francke, J.F. Harper, B.T. Darras, J.M. Cowan, **E.R.B. McCabe**, A. Kohlschutter, W.K. Seltzer, F. Saito, J. Goto, J.-P. Harpey, and J.E. Wise: Congenital adrenal hypoplasia, myopathy and glycerol kinase deficiency: Molecular genetic evidence for deletions. *American Journal of Human Genetics* 40:212-227, 1987.
39. D. Sadava, M. Depper, M. Gilbert, B. Bernard and **E.R.B. McCabe**: Development of enzymes of glycerol metabolism in human fetal liver. *Biology of the Neonate* 52:26-32, 1987.
40. **E.R.B. McCabe**, A. M. Nord, A. Ernest and L. McCabe: Evaluation of a phenylalanine-free product for treatment of phenylketonuria. *American Journal of Diseases of Children* 141:1327-1329, 1987.
41. D.H. Mahoney, D.R Ambruso, **E.R.B. McCabe**, D. Anderson, J.V. Leonard and D.B. Dunger. Lack of effect of lithium carbonate in patients with glycogenosis Ib. *American Journal of Diseases of Children* 141: 985-986, 1987.
42. K. Peterson, R Slover, S. Gass, W.K. Seltzer, L. McCabe and **E.R.B. McCabe**: Blood phenylalanine estimation for the patient with phenylketonuria using a portable device. *Biochemical Medicine and Metabolic Biology* 39:98-104, 1988.
43. M.P. Whyte, J.D. Mahuren, K.N. Fedde, F.S. Cole, **E.R.B. McCabe**, and S.P. Coburn: Perinatal hypophosphatasia: Tissue levels of vitamin B6 are unremarkable despite markedly increased circulating concentrations of pyridoxal-5'-phosphate (Evidence for an ectoenzyme role for tissue nonspecific alkaline phosphatase.) *Journal of Clinical Investigation* 81:1234-1239, 1988.
44. B.A. Burke, M. Wick, R King, T. Thompson, J. Hansen, B.T. Darras, U. Francke, W.K. Seltzer, **E.R.B. McCabe** and B. Scheithauer: Congenital adrenal hypoplasia and selective absence of pituitary luteinizing hormone - a new autosomal recessive disorder. *American Journal of Medical Genetics* 31:75-97, 1988.
45. B. Kirshon, N. Wasserstrum, R Willis, G.E. Herman and **E.R.B. McCabe**: Teratogenic effects of first trimester cyclophosphamide. *Obstetrics and Gynecology* 72:462-464, 1988.
46. A.M. Nord, L. McCabe and **E.R.B. McCabe**: Biochemical and nutritional status of children with hyperphenylalaninemia. *Journal of Inherited Metabolic Disease* 11:431-432, 1988.
47. **E.R.B. McCabe**, J. Towbin, J. Chamberlain, L. Baumbach, J. Witkowski, G.J.B. van Ommen, M. Koenig, L.M. Kunkel and W.K. Seltzer: cDNA probes for the Duchenne muscular dystrophy locus demonstrate a previously undetectable deletion in a patient with dystrophic myopathy, glycerol kinase deficiency and congenital adrenal hypoplasia. *The Journal of Clinical Investigation* 83:95-99, 1989.
48. R.J. Sokol, **E.R.B. McCabe**, A.M. Kotzer, and S.I. Langendoerfer: Pitfalls in diagnosing galactosemia: False negative newborn screen following red blood cell transfusion. *Journal of Pediatric Gastroenterology and Nutrition* 8:266-268, 1989.
49. D.C. Jinks, M. Minter, D.A. Tarver, M. Vanderford, J.F. Hejtmancik and **E.R.B. McCabe**: Molecular genetic diagnosis of sickle cell disease using dried blood specimens from newborn screening blotters. *Human Genetics* 81:363-366, 1989.
50. M. Minter, J. Towbin, J. Harter, and **E.R.B. McCabe**: Enzyme product blot for nondestructive enzymatic assay of proteins in polyacrylamide gels. *Analytical Biochemistry* 178:22-26, 1989.

51. W.K. Seltzer, C. Angelini, G. Dhariwal, S.P. Ringel, and **E.R.B. McCabe**: Muscle glycerol kinase in Duchenne dystrophy and glycerol kinase deficiency. *Muscle and Nerve* 12:307-313, 1989.
52. L.D. Griffin, G.R. MacGregor, D.M. Muzny, J. Harter, R.G. Cook, and **E.R.B. McCabe**: Synthesis and characterization of a bovine hexokinase 1 cDNA probe by mixed oligonucleotide primed amplification of cDNA using high complexity primer mixtures. *Biochemical Medicine and Metabolic Biology* 41:125-131, 1989.
53. F. Greenberg, W. Wasiewski, and **E.R.B. McCabe**: Weaver Syndrome: The changing phenotype with age. *American Journal of Medical Genetics* 33:127-129, 1989.
54. J.A. Towbin, D. Wu, J. Chamberlain, P. Larsen, W.K. Seltzer, and **E.R.B. McCabe**: Characterization of patients with glycerol kinase deficiency (GKD) utilizing cDNA probes for the Duchenne muscular dystrophy (DMD) locus. *Human Genetics* 83:122-126, 1989.
55. J.A. Towbin, M. Minter, D. Brdiczka, V. Adams, V. De Pinto, F. Palmieri, and **E.R.B. McCabe**: Demonstration and characterization of human cardiac porin: A voltage-dependent channel involved in adenine nucleotide movement across the outer mitochondrial membrane. *Biochemical Medicine and Metabolic Biology* 42:161-169, 1989.
56. L. McCabe, A.E. Ernest, M.R. Neifert, S. Yannicelli, A.M. Nord, P.J. Garry and **E.R.B. McCabe**: The management of breast feeding among infants with phenylketonuria. *Journal of Inherited Metabolic Disease* 12:467-474, 1989.
57. B. Perelmuter, S.I. Goodman, and **E.R.B. McCabe**: Galactosemia with cerebral edema and tyrosinemia. *Journal of Inherited Metabolic Disease* 12:489-490, 1989.
58. M.A. Kay, W. O'Brien, B. Kessler, R. McVie, S. Ursin, K. Dietrich, and **E.R.B. McCabe**: Transient organic aciduria and methemoglobinemia with acute gastroenteritis. *Pediatrics* 85:589-592, 1990.
59. J.A. Towbin, J.S. Chamberlain, D. Wu, D.M. Pillers, W.K. Seltzer, and **E.R.B. McCabe**: DXS28 (C7) maps centromeric to DXS68 (L 1-4) and DXS67 (B24) by deletion analysis. *Genomics* 7:442-444, 1990.
60. L. McCabe, L.D. Griffin, A. Kinzer, M. Chandler, J.B. Beckwith, and **E.R.B. McCabe**: Overo lethal white foal syndrome: Equine model of aganglionic megacolon (Hirschsprung Disease). *American Journal of Medical Genetics* 36:336-340, 1990.
61. M.A. Kay and **E.R.B. McCabe**: *E. coli* sepsis and prolonged hypophosphatemia following exertional heat stroke. *Pediatrics* 86:307-309, 1990.
62. J.R. Lupski, Y.H. Zhang, M. Rieger, M. Minter, B. Hsu, B.G. Ooi, T. Koeuth, and **E.R.B. McCabe**: Mutational analysis of the *E. coli* glpFK region with Tn5 mutagenesis and the polymerase chain reaction (PCR). *Journal of Bacteriology* 172:6129-6134, 1990.
63. D.M. Pillers, J.A. Towbin, J.S. Chamberlain, D. Wu, J. Ranier, B.R. Powell, and **E.R.B. McCabe**: Deletion mapping of Aland Island Eye Disease to Xp21 between DXS67 (B24) and Duchenne Muscular Dystrophy. *American Journal of Human Genetics* 47:795-801, 1990.
64. M.C. Welsh, B.F. Pennington, S. Ozonoff, B. Rouse, and **E.R.B. McCabe**: Neuropsychology of early-treated phenylketonuria: Specific executive function deficits. *Child Development* 61:1697-1713, 1990.
65. C.G. Azen, R. Koch, E.G. Friedman, S. Berlow, J. Coldwell, W. Krause, R. Matalon, **E.R.B. McCabe**, M. O'Flynn, R. Peterson, B. Rouse, C.R. Scott, B. Sigman, D. Valle, and R. Warner: Intellectual development in 12-year-old children treated for phenylketonuria. *American Journal of Diseases of Children* 145:35-39, 1991.

66. V. Adams, L. Griffin, J. Towbin, B. Gelb, K. Worley, and **E.R.B. McCabe**: Porin interaction with hexokinase and glycerol kinase: Metabolic microcompartmentation at the outer mitochondrial membrane. *Biochemical Medicine and Metabolic Biology*. 45:271-291, 1991.
67. V. Adams, L.D. Griffin, B.D. Gelb, and **E.R.B. McCabe**: Protein kinase activity of rat brain hexokinase. *Biochemical and Biophysical Research Communications*. 177:1101-1106, 1991.
68. W.K. Seltzer, F. Accurso, M.Z. Fall, A.J. Van Riper, M. Descartes, Y. Huang, and **E.R.B. McCabe**: Screening for cystic fibrosis: Feasibility of molecular genetic analysis of dried blood specimens. *Biochemical Medicine and Metabolic Biology*. 46:105-109, 1991.
69. Y. Matsubara, K. Narisawa, K. Tada, H. Ikeda, Y. Ye-Qi, D.M. Danks, A. Green, and **E.R.B. McCabe**: Prevalence of K329E mutation in medium-chain acyl-CoA dehydrogenase gene determined from Guthrie cards. *Lancet*. 338:552-553, 1991.
70. J. Versalovic, T. Koeuth, **E.R.B. McCabe**, and J.R. Lupski: Use of the polymerase chain reaction for physical mapping of *Escherichia coli* genes. *Journal of Bacteriology* 173:5253-5255, 1991.
71. A.L. Pettigrew, **E.R.B. McCabe**, F.F.B. Elder, and D.H. Ledbetter: Isodicentric X chromosome in a patient with Turner syndrome-implications for localization of the X-inactivation center. *Human Genetics* 87:498-502, 1991.
72. B.D. Gelb, J.A. Towbin, **E.R.B. McCabe**, and E. Sujansky: The San Luis Valley Recombinant Chromosome 8 and Tetralogy of Fallot: A review of Chromosome 8 anomalies and congenital heart disease. *American Journal of Medical Genetics* 40:471-476, 1991.
73. **E.R.B. McCabe**: Utility of PCR for DNA analysis from dried blood spots on filter paper blotters. *PCR Methods and Applications*. 1:99-106, 1991.
74. L.D. Griffin, B.D. Gelb, D.A. Wheeler, D. Davison, V. Adams, and **E.R.B. McCabe**: Mammalian hexokinase 1: Evolutionary conservation and structure to function analysis. *Genomics* 11:1014-1024, 1991.
75. B.D. Gelb, V. Adams, S.N. Jones, L.D. Griffin, G.R. MacGregor, and **E.R.B. McCabe**: Targeting of hexokinase 1 to liver and hepatoma mitochondria. *Proceedings of the National Academy of Sciences USA* 89:202-206, 1992.
76. P.S. Subramanian, J. Versalovic, **E.R.B. McCabe**, and J.R. Lupski: Rapid mapping of *Escherichia coli*:Tn5 insertion mutations by REP-Tn5 PCR. *PCR Methods and Applications* 1:187-194, 1992.
77. L.D. Griffin, B.D. Gelb, V. Adams, and **E.R.B. McCabe**: Developmental expression of hexokinase 1 in the rat. *Biochimica et Biophysica Acta* 1129:309-317, 1992.
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183. J. Santiprabhob, E.P. Smith, **E.R.B. McCabe**, and D.R. Repaske: Late onset of adrenal insufficiency in adrenal hypoplasia congenita caused by a frameshift mutation of the *DAX1* gene. Presented to the Endocrine Society Meeting, June 15-18, 2000, Toronto, Ontario, Canada.
184. M.A. Suchard, R.E. Weiss, K.S. Dorman, M. Patel, **E.R.B. McCabe**, and J.S. Sinsheimer: Similarity in evolutionary patterns: Bayesian extensions and the introduction of a scaled evolutionary tree metric. Presented to the Joint Statistical Meeting, August, 2000.
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195. R.C. Clipsham, Y.-H. Zhang, B.-L. Huang, and **E.R.B. McCabe**: *AHCH* expression in murine embryonic stem cells: A model system for investigating the role of *AHCH* in early ontogeny. Accepted for presentation to The Endocrine Society Annual Meeting, June 20-23, 2001, Denver. Colorado.
196. M.V. Patel, G.B. Golding, A.P. Arnold, J.S. Sinsheimer, and **E.R.B. McCabe**: *DAX1/SHP* family of the nuclear hormone receptor superfamily appears predisposed to sequence variability. Accepted for presentation to The Endocrine Society Annual Meeting, June 20-23, 2001, Denver. Colorado.
197. J.K. Phelan, Y.-H. Zhang, B.-L. Huang, K.M. Dipple, L.L. McCabe, E. Vilain, and **E.R.B. McCabe**: Eighty *NROB1* intragenic mutations in patients with adrenal hypoplasia congenita and distribution of these mutations in the DAX1 protein. Accepted for presentation to The Endocrine Society Annual Meeting, June 20-23, 2001, Denver. Colorado.
198. **E.R.B. McCabe** and E. Vilain: Mammalian sex determination: From gonads to brain. Submitted for presentation to Pediatric Endocrinology Montreal 2001 LWPES/ESPE 6 Joint Meeting, July 6-10, 2001, Montreal, Quebec, Canada.
199. R.C. Clipsham and **E.R.B. McCabe**: Single tube gene-specific expression analysis by high primer density multiplex reverse transcription. *The American Journal of Human Genetics*. 69:959A, 2001.
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204. S.V. Stepanian, K.M. Dipple, and **E.R.B. McCabe**: Characterization of the human glycerol kinase promoter. The American Journal of Human Genetics. 69:1750A, 2001.
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207. M.L. Lewis, L.L. McCabe and **E.R.B. McCabe**: Informed decision-making in newborn screening: Highly variable regulatory language. Journal of Investigative Medicine. 50:20A, 2002.
208. R.C. Clipsham, Y.-H. Zhang, B.-L. Huang, and **E.R.B. McCabe**: Transcriptional network analysis for identification of optimal steroidogenic axis model cell lines. Journal of Investigative Medicine. 50:87A, 2002.
209. L.L. McCabe and **E.R.B. McCabe**: Genomic medicine: Population screening. Accepted for presentation to the American Association for the Advancement of Science Meeting, February 14-19, 2002, Boston, Massachusetts.
210. M.H. Lewis, L.L. McCabe and **E.R.B. McCabe**: Informed decision-making in newborn screening: Highly variable regulatory language. Pediatric Research. 51:42A, 2002.
211. T. Huang, A. Shalci, D. Ho, Y.-H. Zhang, **E.R.B. McCabe**, and C.-M. Ho: Rapid bacterial diagnosis: MEMS-based DNA detection. Pediatric Research. 51:276A-277A. 2002.
212. U. Bhardwaj, Y.-H. Zhang, D. Sylvester-Jackson, G.R. Buchanan, B. Therrell, and **E.R.B. McCabe**: DNA confirmation of β -globin cluster deletion in newborn screening follow-up. Pediatric Research. 51:240A, 2002.
213. K.M. Dipple, Y.-H. Zhang, J. Van Hove, and **E.R.B. McCabe**: Gestational diabetes associated with a novel mutation (378-379insTT) in the glycerol kinase gene. Presented to the Endocrine Society Meeting, June 17-22, 2002, San Francisco, California.
214. S.V. Stepanian, S.T. Huyn, **E.R.B. McCabe**, and K.M. Dipple: HNF-4 alpha binds the human glycerol kinase promoter in the 5' untranslated region important for increased expression of glycerol kinase. Presented to the Endocrine Society Meeting, June 17-22, 2002, San Francisco, California.
215. R.C. Clipsham, Y.-H. Zhang, B.-L. Huang, and **E.R.B. McCabe**: Transcriptional network analysis in steroidogenic axis-derived model cell lines. Presented to the Endocrine Society Meeting, June 17-22, 2002, San Francisco, California.
216. M. Patel, J.S. Sinsheimer, and **E.R.B. McCabe**: The *NROB* subfamily members DAX1 and SHP exhibit sequence variability C-terminal to the LXXLL domain. Presented to the Endocrine Society Meeting, June 17-22, 2002, San Francisco, California.
217. Y.-H. Zhang, U. Bhardwaj, B.-L. Huang, M. Karpodinis, and **E.R.B. McCabe**: *NROB-1* intragenic inversion

causes adrenal hypoplasia congenita (AHC). Accepted for presentation to the American Society of Human Genetics. October 16-19, 2002, Baltimore, Maryland.

218. U. Bhardwaj, Y.-H. Zhang, L.L. McCabe, W. Blackburn, and **E.R.B. McCabe**: Rapid confirmation of Southeast Asian ($--^{SEA}$) and Filipino ($--^{FIL}$) α -thalassemia genotypes from newborn screening samples. Accepted for presentation to the American Society of Human Genetics. October 16-19, 2002, Baltimore, Maryland.
219. A. Maestrejuan, M. Meldrum, and **E.R.B. McCabe**: Oral history of human genetics project. Accepted for presentation to the American Society of Human Genetics. October 16-19, 2002, Baltimore, Maryland
220. M.H. Lewis, L.L. McCabe and **E.R.B. McCabe**: Informed decision-making in newborn screening: Highly variable regulatory language. Accepted for presentation to the American Society of Human Genetics. October 16-19, 2002, Baltimore, Maryland.
221. S.V. Stepanian, S.T. Huyn, **E.R.B. McCabe** and K.M. Dipple: HNF-4 α enhances expression of human glycerol kinase in a human hepatocellular carcinoma cell line. Accepted for presentation to the American Society of Human Genetics. October 16-19, 2002, Baltimore, Maryland.
222. R.H. Ohira, K.M. Dipple, and **E.R.B. McCabe**: Human glycerol kinase (GK): Tissue-specific expression patterns of two alternatively spliced mRNAs from the Xp21 GK gene. Accepted for presentation to the American Society of Human Genetics. October 16-19, 2002, Baltimore, Maryland.

Invited Presentations

Clinical Subtypes of Glycerol Kinase Deficiency, Their Diagnosis and Management, National Institutes of Health, June 1984, Washington, D.C.

Glycerol Kinase Deficiency and Its Involvement with the Porin Microcompartmentation System, Department of Biology, University of Konstanz, September 1986, Konstanz, West Germany

Newborn Screening and the Diagnosis and Management of Inherited Metabolic Disease, Shanghai Children's Hospital, Xin-Hua Children's Hospital, First Medical College of Shanghai, and Genetics Society of Shanghai, June 1987, Shanghai, China

Molecular Genetic Approaches to Newborn Screening for Sickle Cell Disease and Other Disorders, Center for Disease Control, April 4-6, 1988, Atlanta, Georgia

Glycerol Kinase and Hexokinase: Biochemical and Molecular Genetic Investigations of Compartmented Enzymes, Department of Pharmacology, University of Southern California School of Medicine, May 20, 1988, Los Angeles, California

The Top Ten Problems in Screening Programs, 6th National Neonatal Screening Symposium, May 22-25, 1988, Portland, Oregon

Newborn Screening for Sickle Cell Disease: Molecular Genetic Strategy, 6th National Neonatal Screening Symposium, May 22-25, 1988, Portland, Oregon

Newborn Screening: New Diseases and New Directions, American Academy of Pediatrics Annual Meeting, October 20, 1988, San Francisco, California

Glycerol Kinase and Hexokinase: Genetics and Biology of Two Channel-Binding Enzymes, Oregon Health Sciences Center, May 25, 1989, Portland, Oregon

Newborn Screens for Genetic Diseases, Pediatric Grand Rounds, Wesley Medical Center, June 30, 1989, Wichita, Kansas

Glycerol Kinase Deficiency: A Contiguous Gene Syndrome Involving This Channel Binding Enzyme, University of Alabama School of Medicine, July 18, 1989, Birmingham, Alabama

Metabolic Emergencies, John A. Burns School of Medicine, University of Hawaii at Manoa, September 22, 1989, Honolulu, Hawaii

Glycerol Kinase Deficiency and Related Xp21 Loci, John A. Burns School of Medicine, University of Hawaii at Manoa, September 22, 1989, Honolulu, Hawaii

Recombinant DNA Diagnosis Using Blood Spots from Newborn Screening Blotters, John A. Burns School of Medicine, University of Hawaii at Manoa, September 22, 1989, Honolulu, Hawaii

Molecular Genetic Investigations of Mental Retardation, 17th Harry A. Waisman Memorial Lecture, Harry A. Waisman Center on Mental Retardation and Human Development, University of Wisconsin, October 5, 1989, Madison., Wisconsin

Molecular Genetic Analysis of Dried Blood Specimens on Filter Paper Blotters: Evolving Applications for Newborn Screening and Other Programs, 7th National Neonatal Screening Symposium, November 16-18, 1989, New Orleans, Louisiana

Glycerol Kinase Deficiency: A Contiguous Gene Syndrome Involving This Channel Binding Enzyme, Department of Pharmacology, University of Southern California School of Medicine, February 23, 1990, Los Angeles, California

Molecular Genetic Investigations of Mental Retardation, Sparks Center for Mental Retardation, University of Alabama School of Medicine, March 13, 1990, Birmingham, Alabama

Genetic Screening for the Next Decade: Application of Present and New Technologies, Keynote Address, New Technologies for Genetic and Newborn Screening, Yale University School of Medicine, April 23, 1990, New Haven, Connecticut

Molecular Genetic Applications to Newborn Screening, Southern Genetics Group, July 19-21, 1990, Destin, Florida

DNA Analysis on Dried Blood Spots: The New Genetics Comes to Newborn Screening, Nichols Institute, July 31, 1990, San Juan Capistrano, California

DNA from Dried Blood Specimens: Molecular Genetics Comes to Newborn Screening. Mountain States Regional Genetic Services Network Meeting, August 23-25, 1990, Vail, Colorado

DNA - Coming Soon to a Lab Near You, Special Genes Meeting/New York State Newborn Screening Program 25th Anniversary Celebration, September 14, 1990, Albany, New York

DNA Analysis in Newborn Screening, Pacific Southwest Regional Genetics Network, September 28, 1990, Berkeley, California

DNA Forensics: Effective Application to Pediatric Practice, American Academy of Pediatrics Annual Meeting, October 10, 1990, Boston, Massachusetts

Direct DNA Amplification without Microextraction Facilitates Molecular Genetic Analysis of Neonatal Screening Specimens, 8th National Neonatal Screening Symposium, January 31, 1991, Saratoga Springs, New York

DNA Analysis Using Newborn Screening Specimens: Evolving Applications for Dried Blood Spot Technology, DNA-Based Diagnosis: From Research to Application, The Banbury Center, Cold Spring Harbor Laboratory, March 31-April 3, 1991, Cold Spring Harbor, New York

DNA Forensics: Rape, Murder, Paternity, Bishop John J. Russell and Sister Rita Thomas Lectures, St. Mary's Hospital, April 3, 1991, Richmond, Virginia

The Impact of Genetic Information on the Practice of Pediatrics, Biotechnology and the Diagnosis of Genetic Disease, Georgetown University Medical Center, April 19, 1991, Arlington, Virginia

Molecular Genetics of Mental Retardation: Gene Mapping, Function, and Therapy, Minnesota Center for Research in Developmental Disabilities, University of Minnesota, May 14, 1991, Minneapolis, Minnesota

Congenital Adrenal Hypoplasia: Molecular Genetic Analysis and Diagnosis, The Nichols Institute, June 4, 1991, San Juan Capistrano, California

Developmental Expression, Evolution, and Structure to Function Analysis of a Cloned HKI cDNA, March of Dimes Birth Defects Foundation Pre/Postdoctoral Research Fellows Symposium, June 11, 1991, Hilton Head, South Carolina

Prenatal Genetics Laboratory Techniques, The Perinatal Scientist in the 1990's, 3rd Annual NICHD Aspen Conference, August 23, 1991, Aspen, Colorado

Basic Research in Sickle Cell Diagnosis: DNA Analysis, Sickle Cell: Twenty Years of Progress, September 14, 1991, Houston, Texas

Molecular Medicine and Biotechnology: New Directions in Health Care Delivery, The Claremont Colleges, October 28, 1991, Claremont, California

Recognition and Management of Inborn Errors of Metabolism, American Academy of Pediatrics Annual Meeting, October 30, 1991, New Orleans, Louisiana

Biotechnology and Molecular Medicine: New Directions in Pediatric Practice, Pediatric Grand Rounds, Harbor UCLA Medical Center, October 31, 1991, Torrance, California

What's New in DNA/RNA?; Problems in Newborn Screening Programs in Developed Countries, Eighth International Neonatal Screening Symposium, November 12-15, 1991, Leura, Australia

Glycerol Kinase and Hexokinase: Molecular Genetics and Interactions with Porin, November 19, 1991, University of Konstanz, Konstanz, Germany

DNA Techniques for Screening of Inborn Errors of Metabolism, Selective Screening for Inborn Errors of Metabolism. November 20-22, 1991, Fulda, Germany

Glycerol Kinase and Hexokinase: Molecular Genetics and Mitochondrial Binding, Jefferson Medical College, March 12, 1992, Philadelphia, Pennsylvania

Molecular Genetic Investigations of Suspected Mitochondrial Disease, Role of Mitochondrial Genetics in Mental Retardation, NICHD Workshop, March 30-April 1, 1992, Bethesda, Maryland

Molecular Genetics of Glycerol Kinase Deficiency and Hexokinase Binding to Porin, Department of Pathology and Laboratory Medicine, University of Texas Health Science Center at Houston, April 2, 1992, Houston, Texas

DNA Fingerprinting for Quality Control of the Organisms Used in Bacterial Inhibition Assays, Ninth National Newborn Screening Symposium, April 7-11, 1992, Raleigh, North Carolina

Integration of DNA and RNA Methods into a Neonatal Hemoglobinopathy Screening Program; Ninth National Newborn Screening Symposium, April 7-11, 1992, Raleigh, North Carolina

Contiguous Gene Syndromes: Role in the Etiology of Mental Retardation, American Academy of Pediatrics Spring Session, April 11-16, 1992, New York, New York

Introduction to DNA Diagnosis and Gene Therapy, American Academy of Pediatrics Spring Session, April 11-16, 1992, New York, New York

Complex Glycerol Kinase Deficiency: An Xp21 Contiguous Gene Syndrome, Human Genetics Seminar Series, The Mount Sinai Medical Center, April 14, 1992, New York, New York

Glycerol Kinase and Hexokinase: Molecular Genetics and Mitochondrial Targeting, Division of Pediatric Biochemistry, University of Maryland School of Medicine, May 4, 1992, Baltimore, Maryland

RNA and Primer Mismatch DNA Amplification from Initial Specimens Facilitate Neonatal Hemoglobinopathy Screening Confirmation, Society for Pediatric Research Annual Meeting, May 4-7, 1992, Baltimore, Maryland

Guthrie Cards as DNA Source, DNA Databanks and Repositories, Armed Forces Institute of Pathology, May 15-16, 1992, Bethesda, Maryland

Glycerol Kinase and Hexokinase: Molecular Genetics and Mitochondrial Binding, Department of Pediatrics, University of Washington, June 17, 1992, Seattle, Washington

American Academy of Pediatrics Policy Statements and Issues Related to DNA and RNA Microextraction from Newborn Screening Blood Spots, Workshop on Genetic Diagnosis, Testing and Screening Services: Newborn Screening, Committee on Assessing Genetic Risks: Issues and Implications for Health, Institute of Medicine, June 27-29, 1992, Irvine, California

Role of Porin-Kinase Interactions in Disease, NATO ARW, Molecular Biology of Mitochondrial Transport Systems, September 17-21, 1992, Pisa, Italy

Introduction to Genetics, The Shape of Things to Come: Genetics for Clinic, Classroom and Community, Scott and White, October 2, 1992, Temple, Texas

Indications for Referral, The Shape of Things to Come: Genetics for Clinic, Classroom and Community, Scott and White, October 2, 1992, Temple, Texas

New Technologies/The Human Genome Project, The Shape of Things to Come: Genetics for Clinic, Classroom and Community, Scott and White, October 2, 1992, Temple, Texas

Regulation, Certification and Public Health, The Future of DNA-Based Diagnosis, The Banbury Center, Cold Spring Harbor Laboratory, January 13, 1993, Cold Spring Harbor, New York

Molecular Genetics Follow-up, Newborn Screening Hemoglobinopathy Program Annual Meeting, February 22, 1993, Austin, Texas

Identification of the Glycerol Kinase Gene Using Genomic Scanning and Patient Deletions, Society of Inherited Metabolic Diseases Annual Meeting, March 15, 1993, Asilomar, California

DNA Diagnosis in Newborn Screening, Association of Maternal and Child Health Programs Annual Meeting, March 22, 1993, Washington, D.C.

Role of Biotechnology in Pediatrics, American Academy of Pediatrics Annual Meeting, March 23, 1993, Chicago, Illinois

Newborn Screening: Principles, Practice and Prudence, Pediatric Postgraduate Symposium, April 17, 1993, Houston, Texas

Rapid Genomic Scanning for Expressed Sequences: Application to the Glycerol Kinase Region of Xp21. Society for Pediatric Research Annual Meeting, May 4, 1993, Washington, D.C.

Molecular Pediatrics: From Rare Diseases to Public Health, E. Mead Johnson Award Presentation, Society for Pediatric Research Annual Meeting, May 5, 1993, Washington, D.C.

Epidemiology of Genetic Disorders, Primary Prevention of Developmental Disabilities, Vanderbilt University, May 17, 1993, Nashville, Tennessee

Progress Highlights of the Baylor Mental Retardation Research Center, American Association on Mental Retardation Annual Meeting, June 3, 1993, Washington, D.C.

DNA Diagnosis: Future Trends, Newborn Screening for Sickle Cell Disease Conference, June 11, 1993, Washington, D.C.

Cloning the Complete Coding Sequence for the Glycerol Kinase Gene Using a New Genomic Scanning Approach, Department of Genetics, University of Leiden, June 24, 1993, Leiden, The Netherlands

Cloning and Expression of the Human Glycerol Kinase cDNA, University of Konstanz, June 28, 1993, Konstanz, Germany

Cloning the Complete Coding Sequence for the Glycerol Kinase Gene Using a New Genomic Scanning Approach, Institute for Clinical Chemistry, City Hospital, June 30, 1993, Munich, Germany

Cloning and Expression of the Human Glycerol Kinase cDNA, Institute for Cell Biology, ETH Honggerberg, July 2, 1993, Institute for Cell Biology, ETH Honggerberg, Zurich, Switzerland

Inborn Errors of Metabolism: Emerging Opportunities and Potential Problems in Gene Therapy, Gene Therapy for Mental Retardation and Developmental Disabilities: Prospects and Strategies, NICHD Conference, July 8, 1993, Bethesda, Maryland

Newborn Screening: PKU, Galactosemia, and the Hemoglobinopathies, A Practical Approach to Incorporating Genetics into Primary Health Care, Texas Medical Association, August 27, 1993, Austin, Texas

Automation of Molecular Genetic Screening, 9th International Screening Symposium, September 16, 1993, Lille, France

Evaluation of the Dysmorphic Child, Genetics Training Program for Nurses, Northwest Memorial Hospital, October 2, 1993, Houston, Texas

Microcompartmentation of Energy Metabolism at the Outer Mitochondrial Membrane: Role in Diabetes and Other Disorders, Energy Genes and Mitochondrial Disease Symposium, American Society for Human Genetics Annual Meeting, October 6, 1993, New Orleans, Louisiana

Automation of Molecular Genetic Screening, Annual Meeting of the Northeastern Section of the American Association for Clinical Chemistry, October 16, 1993, Albany, New York

Molecular Pediatrics: Role of Biotechnology in Clinical Practice, 4th Annual Pediatric Research Symposium, Department of Pediatrics, University of Michigan, October 22, 1993, Ann Arbor, Michigan

The Future of Molecular Genetics Testing in Newborn Screening, Annual American Public Health Association Meeting, October 26, 1993, San Francisco, California

Newborn Screening: What Do Those Blood Spots Tell Us?, Genetics at a Turning Point, October 30, 1993, Houston, Texas

Molecular Pediatrics from Rare Disease to Public Health, Pediatric Grand Rounds, University of Texas Health Science Center, November 2, 1993, Houston, Texas

Cloning of the Glycerol Kinase Gene by CAIGES: A New Genomic Scanning Approach, University of Kentucky School of Medicine, December 7-8, 1993, Lexington, Kentucky

Phenylketonuria: Newborn Screening and Gene Therapy, University of Kentucky School of Medicine, December 7-8, 1993, Lexington, Kentucky

Molecular Genetic Diagnosis and Treatment: The Future is Upon Us, Department of Genetics, Yale University School of Medicine, December 14, 1993, New Haven, Connecticut

New Directions in Newborn Screening, Mental Retardation Research Center Seminar, The Children's Hospital of Philadelphia, February 8, 1994, Philadelphia, Pennsylvania

DNA Analysis in Sickle Cell Disease, Southeastern Regional Genetics Group, February 25-27, 1994, Atlanta, Georgia

DNA Techniques for Population-Based Newborn Screening, Southeastern Regional Genetics Group, February 25-27, 1994, Atlanta, Georgia

Molecular Pediatrics: The Role of Biotechnology in Clinical Practice, Pediatric Grand Rounds, University of Minnesota Medical Center, March 9, 1994, Minneapolis, Minnesota

Complex Glycerol Kinase Deficiency: Genomic and Functional Insights from a Contiguous Gene Syndrome, Institute of Human Genetics, University of Minnesota Medical Center, March 10, 1994, Minneapolis, Minnesota

Genetic Disease Diagnosis, Advanced Biotechnology, Food and Drug Administration, April 12, 1994, Washington, D.C.

Metabolic Emergencies, California Association for Medical Laboratory Technicians Seminar, April 30, 1994, Walnut Creek, California

Molecular Genetic Screening: Technological Advances and Ethical Dilemmas; Society for Pediatric Research Annual Meeting, May 2, 1994, Seattle, Washington

Automated DNA Screening and Diagnosis, Society for Pediatric Research Annual Meeting, May 2, 1994, Seattle, Washington

Gene Therapies: Future Therapy for Mental Retardation, American Association for Mental Retardation Annual Meeting, June 3, 1994, Boston, Massachusetts

Automation of Molecular Genetic Screening, 10th National Neonatal Screening Symposium, June 10, 1994, Seattle, Washington

Newborn Screening: Sample Collection and Storage for Future Genetic Studies, Informed Consent for Genetic Studies Using Stored Tissue Samples, NIH/CDC Conference, July 7-8, 1994, Bethesda, Maryland

Frequency of Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency Among California Children Dying of Sudden Infant Death Syndrome (SIDS) or Liver Disease: Two-tiered Analysis of Newborn Screening Specimens. Annual Meeting of the American Society of Human Genetics, October 22, 1994, Montreal, Quebec, Canada

Gene Therapy; Ethical Issues in Genetic Screening, Annual Meeting of the American Academy of Pediatrics, October 25, 1994, Dallas, Texas

Xp21 Deletions and Duplications: Simple Explanations Emerge for Complex Phenotypes. UCLA Intercampus Medical Genetics Program Seminar, November 11, 1994, Los Angeles, California

Xp21 Contiguous Gene Syndrome Includes Multiple Loci for Mental Retardation, UCLA Mental Retardation Research Center Retreat, January 12, 1995, Los Angeles, California

Excursions Along the X Chromosome: Exploring a Contiguous Gene Syndrome, The Claremont Colleges, February 22, 1995, Claremont, California

Excursions Along the X Chromosome: Exploring a Contiguous Gene Syndrome, The Claremont Colleges, February 22, 1995, Claremont, California

Xp21 Disorders: Potential for Gene Therapy, Gene Therapy Program Seminar, Los Angeles, Children's Hospital, March 16, 1995, Los Angeles, California

Strategies to Cope with Early Discharge: Molecular Genetic Laboratory Paradigms for Newborn Screening, Impact of Early Discharge on Newborn Screening Conference, April 1, 1995, Washington, D.C.

Molecular Pediatrics: From Rare Disease to Public Health, Pediatric Grand Rounds, Ventura County Medical Center, April 25, 1995, Ventura, California

Impact of Biotechnology on Pediatrics, Pediatric Grand Rounds, Santa Monica Hospital, May 24, 1995, Santa Monica, California

Adrenal Cortical Development and Its Disorders, Ben Kagan Lecture, Cedars-Sinai Medical Center, June 8, 1995, Los Angeles, California

Excursions on the X Chromosome: Molecular Pediatrics and the Human Genome Initiative, Pediatric Grand Rounds, Health Sciences Center, June 15, 1995, Winnipeg, Manitoba, Canada

Genetic Screening and Diagnosis: Technological and Ethical Challenges, J.C. Wilt Lecture, School of Public Health, Health Sciences Center, June 15, 1995, Winnipeg, Manitoba, Canada

Impact of Biotechnology on the Daily Practice of Pediatrics, Pediatric Grand Rounds, Children's Hospital of Los Angeles, June 23, 1995, Los Angeles, California

Progress and Opportunities in Prevention and Treatment, Intellectual Disability: Programs, Policies, and Planning for the Future, United Nations, June 30, 1995, New York, New York

Molecular Basis of Adrenal Hypoplasia Congenita, Hypogonadotropic Hypogonadism and Sex Reversal, Recent Progress in Hormone Research, July 30, 1995, Stevenson, Washington

Molecular Genetic Screening for Inherited and Infectious Diseases, NICHD Lectureship, Perinatal Research Society Annual Meeting, September 18, 1995, Cape Cod, Massachusetts

Polymerase Chain Reaction for Bacterial Identification and Screening for Childhood Infections, Pediatric Grand Rounds, Huntington Memorial Hospital, September 20, 1995, Pasadena, California

Molecular Basis of Adrenal Hypoplasia Congenita, Hypogonadotropic Hypogonadism, and Sex Reversal, Combined Endocrine Conference, UCLA School of Medicine, September 27, 1995, Los Angeles, California

Recognition and Management of Inborn Errors of Metabolism, American Academy of Pediatrics Annual Meeting, October 17, 1995, San Francisco, California

Evaluating the Dysmorphic Child, Pediatric Grand Rounds, UCLA School of Medicine, November 3, 1995, Los Angeles, California

Excursions on the X-Chromosome: Positional Cloning in an Xp21 Contiguous Gene Syndrome, Genetics Research Seminar, University of Southern California School of Medicine, November 6, 1995, Los Angeles, California

Positional Cloning of Genes in Xp21 Using a Novel Genomic Scanning Approach, Molecular Biology Institute Faculty Research Seminar, UCLA School of Medicine, November 7, 1995, Los Angeles, California

Molecular Genetic Strategies to Detect Genetic Defects in Energy Metabolism, Mead Johnson Perinatology Symposium, November 17, 1995, Marco Island, Florida

Energy Metabolism at the Outer Mitochondrial Membrane, Mead Johnson Perinatology Symposium, November 17, 1995, Marco Island, Florida

Molecular Basis of Adrenal Hypoplasia Congenita, Hypogonadotropic Hypogonadism and Sex Reversal, Combined Endocrine Conference, Harbor-UCLA Medical Center, December 5, 1995, Torrance, California

Excursions on the X-Chromosome: Positional Cloning in a Contiguous Gene Syndrome, Human and Molecular Development Research Seminar, UCLA School of Medicine, January 18, 1996, Los Angeles, California

Molecular Genetics: From Rare Disorders to Public Health, Pediatric Grand Rounds, Encino-Tarzana Medical Center, January 30, 1996, Tarzana, California

Metabolic Emergencies, Pediatric Grand Rounds, Olive View-UCLA Medical Center, February 28, 1996, Sylmar, California

Recognition and Management of Inborn Errors of Metabolism, Pediatric Grand Rounds, Memorial Miller Children's Hospital, March 1, 1996, Long Beach, California

Genetics for the Generalist, Harvard-UCLA Lecture Series, March 31, 1996, Long Beach, California

Impact of Biotechnology on Pediatrics, Pediatric Grand Rounds, University of Southern California School of Medicine, April 9, 1996, Los Angeles, California

Is It Bacterial or Viral? Rapid Diagnosis with DNA, American Academy of Pediatrics Annual Meeting, April 13-16, 1996, Chicago, Illinois

Genetics for the Practicing Pediatrician, American Academy of Pediatrics Annual Meeting, April 13-16, 1996, Chicago, Illinois

New Genetics for the Practicing Physician, Clinical Volunteer Faculty Association, University of California, Irvine, May 28, 1996, Irvine, California

Syndrome of Muscle, Adrenal, and CNS Abnormalities: 20 Year Interplay Between the Bedside and the Bench, J. Edward Berk Annual Lecture, Pediatric Grand Rounds, University of California, Irvine, May 29, 1996, Orange, California

Mutations in DAX1 Identified by Sequencing of Genomic DNA from Patients with Adrenal Hypoplasia Congenita (AHC), 10th International Congress of Endocrinology, June 14, 1996, San Francisco, California

Molecular Genetic Testing for Bacterial Infection: Methodology for Rapid Diagnosis and Speciation of Bacterial Infection, Nichols Institute, June 27, 1996, San Juan Capistrano, California

Worldwide Progress and Opportunities in Prevention of Mental Retardation, International Association for the Scientific Study of Intellectual Disability 10th World Congress, July 10, 1996, Helsinki, Finland

Excursions on the X Chromosome: Molecular Basis of a Contiguous Gene Syndrome, Department of Human Molecular Genetics, National Public Health Institute, July 12, 1996, Helsinki, Finland

Molecular Triage of Bacterial Infection, Specialty Laboratories, August 9, 1996, Santa Monica, California

Harnessing Molecular Medicine for Improved Prevention, Diagnosis, and Management of Mental Retardation and Developmental Disabilities, President's Committee on Mental Retardation, September 27, 1996, Washington, D.C.

Becoming a Scientist: Let Nature Ask the Questions, Junior Science and Humanities Symposium, Museum of Science and Industry, November 20, 1996, Los Angeles, California

Genetic Screening: Controversy and Opportunity, Southwest Pediatric Society, November 20, 1996, Los Angeles, California

Channel-Kinase Interactions at the Outer Mitochondrial Membrane, Anesthesia Research Seminar, UCLA School of Medicine, December 2, 1996, Los Angeles, California

Newborn Screening Blood Blotters as DNA Databases, The Joint Conference on DNA Databanks and Repositories, December 12, 1996, Tallahassee, Florida

Newborn Screening Informed Consent Issues, Newborn Screening for Cystic Fibrosis: A Paradigm for Public Health Genetics Policy Development, Centers for Disease Control, January 13, 1997, Atlanta, Georgia

Dried Blood Specimens: Diagnostic Opportunities, Epidemiologic Advantages and Ethical Implications, American College of Medical Genetics Annual Meeting, March 2, 1997, Ft. Lauderdale, Florida

Genetics Testing: Meeting the Ethical Challenges in the 21st Century, Harvard-UCLA Pri-Med Conference, April 5, 1997, Long Beach, California

Molecular Genetic Diagnosis of Infectious Diseases, Human and Molecular Development Research Seminar, UCLA, April 17, 1997, Los Angeles, California

Basis of an Xp21 Contiguous Gene Syndrome with Muscle, CNS and Adrenal Abnormalities, Human Genetics Research Seminar, Oregon Health Sciences University, April 23, 1997, Portland, Oregon

Impact of the Human Genome Project on the Practice of Pediatrics, Bilderback Lecture, Oregon Health Sciences University, April 24, 1997, Portland, Oregon

Rapid DNA Diagnosis of Infectious Disease: Molecular Triage of Sepsis, Portland Academy of Pediatrics, April 24, 1997, Portland Oregon

Evaluating the Dysmorphic Child, Pediatric Grand Rounds, Legacy Emanuel Hospital, April 25, 1997, Portland, Oregon

Preparation and Presentation of Abstracts at Scientific Meetings, Pediatric Academic Societies' Annual Meeting, May 2, 1997, Washington, D.C.

What Is Required to Increase Children's Clinical and Health Services Research?, Public Policy Plenary, Pediatric Academic Societies' Annual Meeting, May 4, 1997, Washington, D.C.

Genetics and Cancer: What is New?, American Academy of Pediatrics Annual Meeting, May 10, 1997, San Diego, California

Dysmorphology for the General Pediatrician: Diagnosis and Office Management, American Academy of Pediatrics Annual Meeting, May 10, 1997, San Diego, California

Impact of the Human Genome Project on our Understanding of Mental Retardation, Genetic Advances in Understanding Mental Retardation (Pauline Wilson Horner Genetics Symposium), Case Western Reserve University, June 4, 1997, Cleveland, Ohio

Molecular Triage for Rapid Diagnosis of Infectious Disease, Pediatric Grand Rounds, Case Western Reserve University School of Medicine, June 5, 1997, Cleveland, Ohio

Molecular Genetic Approaches to the Diagnosis of Infectious Disease, OB/GYN Grand Rounds, UCLA School of Medicine, June 6, 1997, Los Angeles, California

From Newborn Screening to Infectious Disease Diagnosis: Molecular Genetic Technology Bridges Public Health Disciplines, Centers for Disease Control, June 20, 1997, Atlanta, Georgia

What's New in Genetics for the Practicing Pediatrician, 40th Annual Pediatric Program, University of Colorado School of Medicine, June 27, 1997, Denver, Colorado

Excursions on the X Chromosome: Twenty Year Interplay Between the Bedside and the Bench, Harry Gordon Award for Outstanding Achievement in Academic Medicine, University of Colorado School of Medicine, June 27, 1997, Denver, Colorado

Is It Viral or Bacterial? Rapid Diagnosis of Infectious Disease, 40th Annual Pediatric Program, University of Colorado School of Medicine, June 28, 1997, Denver, Colorado

Impact of the Human Genome Project on Our Understanding of Mental Retardation, Keynote Address, International Conference on Mental Retardation: Genes, Brain and Behavior, July 10, 1997, Staten Island, New York

Rapid Diagnosis of Infection, NICHD Perinatology Conference, August 22, 1997, Aspen, Colorado

Molecular Pediatrics: Impact of the New Biology on Clinical Practice, Grand Rounds, British Columbia Children's Hospital, October 3, 1997, Vancouver, British Columbia, Canada

Excursions on the X Chromosome: The Molecular Basis of a Contiguous Gene Syndrome with Muscle, Adrenal and CNS Abnormalities, Medical Genetics Departmental Seminar, University of British Columbia School of Medicine, October 3, 1997, Vancouver, British Columbia, Canada

DNA Testing and New Technologies, California Newborn Screening Program Annual Meeting, October 8, 1997, Los Angeles, California

Molecular Genetics for the Practicing Physician, Internal Medicine Grand Rounds, White Memorial Hospital, October 16, 1997, Los Angeles, California

Impact of Early Discharge on Newborn Screening, American Academy of Pediatrics Annual Meeting, November 5, 1997, New Orleans, Louisiana

New Genetics for the Practicing Pediatrician; Genetic Testing: Ethical Challenges for the 21st Century; and Introduction to and Visual Diagnosis of Dysmorphology/Genetic Syndromes; 39th Annual Southern California Permanente Medical Group Pediatric Symposium, November 7-8, 1997, San Diego, California

Hexokinase: Role in Diabetes and Cancer, Combined Endocrine Conference, UCLA Medical Center, November 26, 1997, Los Angeles, California

New Genetics for the Practicing Pediatrician, Grand Rounds, Department of Pediatrics, Brown University Medical Center, December 7, 1997, Providence, Rhode Island

Excursions Along the X Chromosome: Molecular Basis of a Contiguous Gene Syndrome, Genetics Seminar, Brown University Medical Center, December 7, 1997, Providence, Rhode Island

Advances in the Diagnosis of Genetic Disease, Grand Rounds, Department of Pediatrics, King/Drew Medical Center, February 10, 1998, Los Angeles, California

Integrating Newborn Screening into Health Care Delivery, American College of Medical Genetics Annual Meeting, March 1, 1998, Los Angeles, California

Criteria for Expansion of Newborn Screening, 13th National Neonatal Screening Symposium, March 2, 1998, San Diego, California

Balancing Personal and Professional Lives, Pediatrician Scientist Development Program Annual Meeting, March 5, 1998, Bal Harbour, Florida

Integration of the Basic Sciences with Clinical Medicine, Council on Medical Student Education in Pediatrics Annual Meeting, March 8, 1998, Bal Harbour, Florida

NIH Funding for Research in Pediatric Departments; and Developing Research-Oriented Faculty: The Child Health Research Center, Association of Medical School Pediatric Department Chairs Annual Meeting, March 9, 1998, Bal Harbour, Florida

Newborn Screening, Genetic Medicine and the Practicing Physician, March 14, 1998, New Orleans, Louisiana

Hexokinase: Role in Diabetes Mellitus and Cancer, Basic Science Lecture, Department of Medicine, Harbor-UCLA Medical Center, April 21, 1998, Torrance, California

Preparation and Presentations of Abstracts at Scientific Meetings, American Pediatric Society/Society for Pediatrics Research Workshop, May 1, 1998, New Orleans, Louisiana

Developmental Molecular Genetics of the Adrenal Cortex, State of the Art Plenary, American Pediatric Society/Society for Pediatric Research/Lawson Wilkins Pediatric Endocrine Society, May 3, 1998, New Orleans, Louisiana

Genetic Testing: Meeting the Ethical Challenges of the 21st Century, Pediatric Grand Rounds, Rochester General Hospital, May 12, 1998, Rochester, New York

Rapid Diagnosis of Infectious Disease: The Molecular Revolution, Department of Pediatrics, University of Rochester Medical Center, May 12, 1998, Rochester, New York

Impact of the Human Genome Project on the Practice of Pediatrics, Samuel W. Clausen Lecture, Children's Hospital at Strong, May 13, 1998, Rochester, New York

Screening for Genetic Disease, Pediatric Grand Rounds, Ventura County Medical Center, May 26, 1998, Ventura, California

Genetic Discrimination and Confidentiality of Genetic Testing, Animal Cloning and Other Gene Manipulations: Ethical, Medical and Public Policy Issues Symposium, June 9, 1998, Beverly Hills, California

Mental Retardation Associated with X-Linked Adrenal Hypoplasia Congenita: Identification of CNS-Expressed Genes in This Region, Endocrine Society, June 25, 1998, New Orleans, Louisiana

Genes that Function in Sexual Differentiation, Determinants of Sexual Differentiation Symposium, Endocrine Society, June 26, 1998, New Orleans, Louisiana

Academic Pediatrics, Department of Pediatrics, University of Hawaii, July 6-10, 1998, Honolulu, Hawaii

Sexual Differentiation and Sexual Development, Genetics Seminar, University of Hawaii, July 8, 1998, Honolulu, Hawaii

Genetic Testing: Meeting the Ethical Challenges of the 21st Century, UCLA Department of Medicine Grand Rounds, August 12, 1998, Los Angeles, California

Balancing Personal and Professional Lives, NICHD Perinatal Conference, August 27, 1998, Aspen, Colorado

Gene Therapy, NICHD Perinatal Conference, August 28, 1998, Aspen, Colorado

Linkages with Young Investigators, National Association of Children's Hospitals and Related Institutions Annual Meeting, October 14, 1998, Houston, Texas

Impact of the Human Genome Project on the Practice of Pediatrics, Grand Rounds, UCLA Department of Pediatrics, October 16, 1998, Los Angeles, California

Balancing Your Life: Academics vs Family, Career Development in Pediatrics Symposium, Western Society for Pediatric Research, January 27, 1999, Carmel, California

Dried Blood Spot Technology, Newborn Screening for Diabetes Risk, Centers for Disease Control, February 16, 1999, Atlanta Georgia

Molecular Genetic Diagnosis of Bacterial Infection, Grand Rounds, UCLA Department of Pediatrics, February 19, 1999, Los Angeles, California

You and Your Mentor: How to Succeed in a Basic Science Laboratory, Pediatric Scientist Development Program Annual Meeting, March 4, 1999, Tampa, Florida

Why Evaluate Faculty: The Good, the Bad and the Ugly, New Chairs' Meeting, American Medical School Pediatrics Department Chairs Annual Meeting, March 4, 1999, Tampa, Florida

Genetics and Cancer: What's New?, American Academy of Pediatrics Annual Meeting, April 20, 1999, Chicago, Illinois

Excursions on the X Chromosome: Molecular Basis of a Syndrome Affecting Muscle, Adrenal and Brain, Seminars in Biology and Chemistry, Children's Memorial Institute for Education and Research, Northwestern University, May 20, 1999, Chicago, Illinois

State of the Art DNA Technology in Neonatal Screening, International Society for Neonatal Screening Meeting, June 14, 1999, Stockholm, Sweden

Xp21 Contiguous Gene Syndrome Including Duchenne Muscular Dystrophy, Glycerol Kinase Deficiency, and Adrenal Hypoplasia Congenita, Department of Genetics, Hopital Enfants Malades, June 21, 1999, Paris, France

Financing, Planning and Balancing a Research Career, NICHD Perinatal Conference, August 26, 1999, Aspen, Colorado

Gene Therapy, NICHD Perinatal Conference, August 27, 1999, Aspen, Colorado

What's New in Newborn Screening?, American Academy of Pediatrics Annual Meeting, October 9, 1999, Washington, D.C.

Presymptomatic Genetic Screening in Childhood, American Academy of Pediatrics Annual Meeting, October 12, 1999, Washington, D.C.

The Human Genome Project: How Does This Project Offer a New Paradigm for Health Promotion?, Incorporating Genetic Medicine and Technology Into Practice and Service, Maternal and Child Health Bureau Meeting, November 1, 1999, Arlington, Virginia

Preparing a Curriculum Vitae and Applying for Fellowships, UCLA Pediatric Resident Retreat, November 2, 1999, Lake Arrowhead, California

Dysmorphology for the General Pediatrician: Diagnosis and Office Management; Genetic Testing: Ethical Challenges for the 21st Century; Rapid Diagnosis of Infectious Disease: The Molecular Revolution; and What's New in Genetics for the Practicing Physician, Tenth Annual Pediatric Symposium, Joe DiMaggio Children's Hospital, November 13-14, 1999, Hollywood, Florida

Human Genome Project: Impact on Health, LIFE Society, University of California, Riverside, November 16, 1999, Riverside, California

How to Apply for a Fellowship, Life After Residency, January 14, 2000, Marina Del Rey, California

Solving a Clinical Puzzle at the Bench: Odyssey on the X Chromosome, Medical Scientist Training Program, UCLA, January 31, 2000, Los Angeles, California

Presymptomatic Genetic Testing: The Future of Genetic Medicine, Western Society for Pediatric Research, February 11, 2000, Carmel, California

Molecular Genetics of Glycerol Kinase Deficiency, UCLA ACCESS Graduate Program Meet the Professor Lunch, February 16, 2000, Los Angeles, California

The Importance of Collaboration, American Association of Administrators of Pediatrics, February 17, 2000, Universal City, California

New Pathogenesis of Glycerol Kinase Deficiency: Functional Genomics, Basic Science Lecture, Harbor/UCLA Medical Center, February 22, 2000, Torrance, California

Implications of Genetic Research and Biotechnology, UCLA Anderson School of Business Graduate Seminar, February 28, 2000, Los Angeles, California

Ethics and Newborn Screening, High School Students Day, American College of Medical Genetics Annual Meeting, March 9, 2000, Palm Springs, California

What's New in Newborn Screening?, Genetic Screening Workshop, American College of Medical Genetics Annual Meeting, March 11, 2000, Palm Springs, California

Role of DAX1 in Sex Determination and Adrenal Function, Ligand, March 17, 2000, La Jolla, California

Human Genome Project: New Principles and Technologies for Screening and Diagnosis, Gatlinberg Conference, March 18, 2000, San Diego, California

Presymptomatic Genetic Testing and Predictive Medicine, Pediatric Grand Rounds, UCLA, April 14, 2000, Los Angeles, California

Mapping Human Genes for Diagnosis and Treatment: Exciting Possibilities and Ethical Dilemmas, Center for Research and Training in Humane and Ethical Medical Care, Santa Monica-UCLA Medical Center, April 18, 2000, Santa Monica, California

Phenotypes of Patients with “Simple” Mendelian Disorders Are Complex Traits: Thresholds, Modifiers and Systems Dynamics, UCLA Department of Human Genetics Research Seminar, April 25, 2000, Los Angeles, California

Respondent to Playing God With Our Genes? The Cloning and Stem Cell Controversies, UCLA Center for the Study of Religion, May 1, 2000, Los Angeles, California

Genetic Testing: Technological Advances and Public Perspectives, Basic Science Seminar Series, University of Utah School of Medicine, May 9, 2000, Salt Lake City, Utah

Career Paths in Academic Medicine: Clinical/Fellows, Pediatric Academic Societies and American Academy of Pediatrics Joint Meeting, May 12, 2000, Boston, Massachusetts

Newborn Screening: Continuities and Change, Pediatric Academic Societies and American Academy of Pediatrics Joint Meeting, May 16, 2000, Boston, Massachusetts

Post-Genomic Medicine: Presymptomatic Testing for Prediction and Prevention, Pediatric Grand Rounds, Harbor-UCLA Medical Center, June 8, 2000, Torrance, California

Ethical Issues in Genetic Testing, The Endocrine Society Annual Meeting, June 24, 2000, Toronto, Ontario, Canada

Leadership in Medicine, National Youth Leadership Forum, June 26, 2000, Los Angeles, California

What’s Your Specialty?, National Youth Leadership Forum, June 29, 2000, Los Angeles, California

Genetics: Impact on Public Policy, NICHD Conference on Perinatal Medicine, August, 26, 2000, Aspen, Colorado

Human Genome Project: Interface of Technology and Public Policy, Jimmy Simon, M.D., Lecture, Wake Forrester University School of Medicine, September 27, 2000, Winston-Salem, North Carolina

Dysmorphology for General Pediatricians, Department of Pediatrics, Wake Forest University School of Medicine, September 27, 2000, Winston-Salem North Carolina

Rapid Diagnosis of Infectious Disease: The Molecular Revolution, Kaiser Permanente, September 29, 2000, Panorama City, California

Secretary’s Advisory Committee for Genetic Testing, Overcoming Regulatory and/or Reimbursement Barriers to Introducing New Genetic Tests for Clinical Use, American Society of Human Genetics Annual Meeting, October 4, 2000, Philadelphia, Pennsylvania

Post Genomic Medicine: The Future of the Public’s Health, Centers for Disease Control, October 10, 2000, Atlanta, Georgia

Academic Medicine, Academic Medicine Interest Group, First and Second Year Medical Students, UCLA School of Medicine, October 23, 2000, Los Angeles, California

Systematic Screening and Referral Process, The American Association for Home-Based Early Interventionists, October 26, 2000, Chicago, Illinois

Syndromes: Outcomes and Interventions, The American Association for Home-Based Early Interventionists, October 26, 2000, Chicago, Illinois

The Human Genome Project, American Academy of Pediatrics Board of Directors' Meeting, October 27, 2000, Chicago Illinois

Genetic Testing: Is It Ready for Primetime?, American Academy of Pediatrics Annual Meeting, October 28, 2000, Chicago, Illinois

Genetic Unknowns, American Academy of Pediatrics Annual Meeting, October 29, 2000, Chicago, Illinois

Applying for Fellowships and Establishing a Research Career, UCLA Pediatric Residents Annual Retreat, November 8, 2000, Lake Arrowhead, California

The Secretary's Advisory Committee on Genetic Testing, Association for Molecular Pathology Annual Meeting, November 12, 2000, Denver, Colorado

After the Genome Project: Predictive Medicine, Southwestern Pediatric Society, November 15, 2000, Los Angeles, California

Compassion, Advocacy and Health Disparities: Recent Policy Changes in Pediatrics and Genetics, Grand Rounds, UCLA Department of Pediatrics, January 12, 2001, Los Angeles, California

How to Apply for a Fellowship, Life After Residency, January 12, 2001, Marina Del Rey, California

Faculty Development Workshop, Women in Academic Medicine, University of Utah, School of Medicine, January 25, 2001, Salt Lake City, Utah

Mentoring in Difficult Times, Department of Pediatrics Senior Faculty, University of Utah, School of Medicine, January 26, 2001, Salt Lake City, Utah

How to Succeed in Academics, Department of Pediatrics, University of Utah, School of Medicine, January 27, 2001, Salt Lake City, Utah

Perinatal Genomic Medicine: Screening, Pharmacogenomics, and Infectious Disease, Western Perinatal Club, Western Regional Meeting, February 9, 2001, Carmel, California

Genetic Testing: Issues in Newborn Screening and Oversight, American Association for the Advancement of Science Annual Meeting, February 17, 2001, San Francisco, California

Clinical Genetics: Compassion, Access, Science and Advocacy, Presidential Address, American College of Medical Genetics Annual Meeting, March 1, 2001, Miami, Florida

Modifier Genes and Metabolic Disease Phenotypes, American College of Medical Genetics/Society for Inherited Metabolic Diseases Annual Meeting, March 4, 2001, Miami, Florida

You and Your Mentor: How to Succeed in a Basic Science Laboratory, Pediatric Scientist Development Program Fellows Workshop, March 8, 2001, San Diego, California

Phenotypes of "Simple" Mendelian Disorders are Complex Traits, Molecular Development Seminar, California Institute of Technology, March 21, 2001, Pasadena, California

Genetic Privacy, DNA Sciences, March 23, 2001, DNA.com

Role of Mentoring in Career Development, Pediatric Emergency Medicine National Fellow's Conference, March 25, 2001, Marina del Rey, California

The Human Genome Project: Interface of Technology and Public Policy, The 4th Annual Harvey Genetics Colloquium, Greater Baltimore Medical Center, March 28, 2001, Baltimore, Maryland

What's New in Newborn Screening?, American Academy of Pediatrics Workshop on Perinatal Practice Strategies, March 31, 2001, Scottsdale, Arizona

Genomic Medicine: Technology and Public Policy, Beyond the Human Genome Project Lecture Series, Harvey Mudd College, April 4, 2001, Claremont, California

Orientation to Academic Medicine, UCLA Department of Pediatrics, April 5, 2001, Los Angeles, California

Screening for Hemochromatosis, Anna Borun and Harry Borun Foundation Visiting Professor in Cardiology, UCLA Department of Internal Medicine, April 12, 2001, Los Angeles, California

Glucose Metabolism and Mitochondrial Compartmentation, UCLA Cardiovascular Research Laboratory Seminar, April 16, 2001, Los Angeles, California

Genetic Screening, Ethical and Policy Challenges in the Genetic Revolution, University of California San Diego, April 17, 2001, La Jolla, California

Complexity of Single Gene Disorders, Pediatric Academic Societies Meeting, April 28, 2001, Baltimore, Maryland

Advocacy, Compassion, Science and Health Disparities: Current Policy Issues in Pediatrics and Genetics, Pediatric Grand Rounds, Cedars Sinai/UCLA, May 10, 2001, Los Angeles, California

Mentoring: The Key to Academic Success, Neurology Grand Rounds, June 15, 2001, West Los Angeles Veterans' Administration Hospital, Los Angeles, California

How to Succeed in Academics, Department of Internal Medicine, Mayo Clinic, July 11, 2001, Rochester, Minnesota

Mentoring: The Key to Academic Success, Internal Medicine Grand Rounds, Mayo Clinic, July 11, 2001, Rochester, Minnesota

Update of the Secretary's Advisory Committee on Genetic Testing (SACGT), Ontario Advisory Committee on New Predictive Genetic Technology Horizon Scanning Session, September 19, 2001, Toronto, Ontario, Canada

Human Genome Project: Ethical, Legal and Social Issues, UCLA School of Law Seminar, September 24, 2001, Los Angeles, California

Consequences of Complexity within Proteomic Networks; Robustness and Health, or Vulnerability and Disease, UCLA Medical Scientist Training Program Seminar, September 24, 2001, Los Angeles, California

Impact of the Human Genome Project on Predictive Medicine, Pediatric Grand Rounds, Huntington Hospital, September 28, 2001, Pasadena, California

Selecting a Subspecialty and Applying for Fellowships, Pediatric Residents Retreat, October 3, 2001, Lake Arrowhead, California

What's New in Newborn Screening?, The Sick Newborn: Diagnosis and Treatment, Citrus Valley Medical Center, October 5, 2001, West Covina, California

What's New in Newborn Screening?, International Society of Nurses in Genetics Annual Meeting, October 12, 2001, San Diego, California

Modifier Genes and Metabolic Disease Phenotypes, American Society of Human Genetics Annual Meeting, October 15, 2001, San Diego, California

What's New in Newborn Screening?, American Academy of Pediatrics Annual Meeting, October 24, 2001, San Francisco, California

What's New in Newborn Screening?, Grand Rounds, Childrens Hospital Los Angeles, October 26, 2001, Los Angeles, California

Translational Genomics: Dissection of an Xp21 Contiguous Gene Syndrome, Research Seminar Series, Children's Hospital Oakland Research Institute, October 30, 2001, Oakland, California

Genetic Privacy, Law, Technology and the Human Genome, California Institute of Technology, November 10, 2001, Pasadena, California

Consequences of Complexity within Proteomic Networks: Robustness and Health or Vulnerability and Disease, UCLA Human and Molecular Development Research Seminar, December 20, 2001, Los Angeles, California

After the Human Genome: Predictive Medicine, First Florence Char, M.D. Visiting Lectureship, Department of Pediatrics, University of Arkansas, January 7, 2002, Little Rock, Arkansas

On Mentoring, Department of Pediatrics, University of Arkansas, January 7, 2002, Little Rock, Arkansas

After the Human Genome Project: Predictive Medicine, Genetics Policy Forum, January 12, 2002, Scottsdale, Arizona

Mentorship, Is There Life After Pediatric Residency?, January 18, 2002, Marina del Rey, California

What's New in Newborn Screening?, Preventing Birth Defects and Infant Mortality, January 25, 2002, Long Beach, California

How to Succeed in Academics, Western Regional Meeting, February 8 and 9, 2002, Carmel, California

Genomic Medicine: Population Screening, American Association for the Advancement of Science Annual Meeting, February 15, 2002, Boston, Massachusetts

Dissecting the Complexity of a Simple Mendelian Disorder, Tribute to Lew Barnes, University of South Florida, February 16, 2002, Tampa, Florida

How to Succeed in Academics, University of California San Francisco, February 26, 2002, San Francisco, California

Translational Genomics in Medical Genetics, Presidential Address, American College of Medical Genetics Annual Meeting, March 14, 2002, New Orleans, Louisiana

What's New in Newborn Screening? Pediatric Grand Rounds, White Memorial Hospital, April 11, 2002, Los Angeles, California

Ethical Issues in Human Genetics, Hadassah Sabra Group, April 15, 2002, Beverly Hills, California

Mentoring: The Key to Academic Success, and Developing Teaching Skills; University of Michigan, May 1, 2002, Ann Arbor, Michigan

Dissecting Complexity in Patients with an Xp21 Contiguous Gene Syndrome, Department of Human Genetics Seminar, University of Michigan, May 1, 2002, Ann Arbor, Michigan

Dissecting Complexity in Patients with Mental Retardation and an Xp21 Contiguous Gene Syndrome, Department of Human Genetics Seminar, Mt Sinai School of Medicine, May 29, 2002, New York, New York

How to Succeed in Academics, Mt. Sinai School of Medicine Pediatrics Research Day, Mt. Sinai School of Medicine, May 30, 2002, New York, New York

Impact of the Human Genome Project on Medicine and Genetic Research, Keynote Address, Genetic Privacy and Discrimination Symposium, University of Rochester, June 14, 2002, Rochester, New York

Molecular Genetics of Adrenal Hypoplasia Congenita, Xth International Adrenal Cortex Conference, June 17, 2002, San Francisco, California

Introduction to Newborn Screening Programs and Overview of Current Technology, Workshop to Develop Newborn Screening Technology for SCID, National Institute of Child Health and Human Development, July 25, 2002, Bethesda, Maryland

Human Genome Project: Impact on Genetic Privacy and Discrimination, Internal Medicine Grand Rounds, Mayo Clinic, September 4, 2002, Rochester, Minnesota