Curriculum Vitae Date: 1/6/17

A.Identifying data:

Name:	Gregory Mark Enns, M.B., Ch.B.
Ethnicity:	Caucasian
Citizenship:	United States of America
Licensure:	California #A52095 Hawaii #MD12592 United Kingdom #3469202

B.Academic History:

Undergraduate:	B.A., Biology: May, 1984 Pomona College Pomona, CA August, 1980–May, 1984
Graduate:	Diploma, Medical Science: June, 1987 University of St. Andrews St. Andrews, Scotland September, 1985–June, 1987 M.B., Ch.B.: June, 1990 University of Glasgow Glasgow, Scotland September, 1987–June, 1990
Honors and Awards:	Kellogg Foundation Scholarship, UC Davis, 1984 P.T. Herring Memorial Prize in Histology, University of St. Andrews, 1986 First Class Merit Certificates: Anatomy, Physiology, Biochemistry, General Pathology, Microbiology, and Pharmacology, University of St. Andrews, 1985-87 Neil Arnott Memorial Prize in Clinical Physics, University of Glasgow, 1988 University of Glasgow Clinical Travel Grant, for clinical studies in China and Nepal, 1988 C.H.L.A. Board of Directors Award for outstanding service as Pediatric Chief Resident, 1995 U.C.S.F. Liver Center Pilot/Feasibility Grant for Hepatic Gene Therapy Research, 1998 Pete and Arline Harman Scholarship, 2005

Stanford University School of Medicine Honor Roll for Teaching, 2005-2006
Distinguished Service Citation, American Academy of Pediatrics, 2011
Outstanding Service Citation, United Mitochondrial Disease Foundation, 2011
Stanford University School of Medicine Excellence in Teaching Citation, 2011–2012
PCARES Rose Award, Lucile Packard Children's Hospital, 2015

Post-Graduate Training: Junior House Officer, Pediatric Surgery
Royal Hospital for Sick Children, Yorkhill
Glasgow, Scotland
August, 1990–January, 1991

Junior House Officer, General Medicine
Glasgow Royal Infirmary
Glasgow, Scotland
January, 1991–June, 1991

Intern and Resident, Pediatrics
Children's Hospital of Los Angeles
Los Angeles, California
June, 1991–June, 1994

Chief Resident, Pediatrics
Children's Hospital of Los Angeles
Los Angeles, California
June, 1994–June, 1995

Fellow, Medical Genetics
University of California, San Francisco
San Francisco, California
June, 1995 – June, 1998

Board Certification: American Board of Pediatrics (10/12/94, 6/14/01, 11/06/08)
American Board of Medical Genetics, Diplomate, Clinical Genetics (9/1/99, 1/1/10)
American Board of Medical Genetics, Diplomate, Clinical Biochemical Genetics (9/1/99, 1/1/10)
ECFMG 0-447-053-0

Research/Contract Support:

RP103-MITO-001 (Enns) 10/08/14–09/30/16 3.18 CM
Raptor Pharmaceuticals $254,794
An Open-label, Dose-Escalating Study to Assess the Safety, Tolerability and Efficacy of Cysteamin Bitratrate Delayed-release capsules (RP103) for the Treatment of Children with Inherited Mitochondrial Disease
This is an open-label study focusing on the safety and preliminary efficacy of cysteamine bitatartrate to treat patients with Leigh syndrome and other inherited mitochondrial disorders.

RP103-MITO-002 (Enns) 07/20/15–05/31/17 1.66 CM
Raptor Pharmaceuticals $359,162
A Long-Term Open-Label Extension Study of RP103-MITO-001 to Assess the Safety, Tolerability and Efficacy of Cysteamine Bitratrate Delayed-release capsules (RP103) for the Treatment of Children with Inherited Mitochondrial Disease
This is an extension study for the RP103 protocol, a study using cysteamine bitartrate to treat patients with Leigh syndrome and other inherited mitochondrial disorders.

IND#107,401 (Enns) 12/09/13 – 11/3016 0.56 CM
Edison Pharmaceuticals $224,765
Long-Term Safety and Efficacy Evaluation of EPI-743 in Children with Leigh Syndrome
This is a long-term follow-up study to the Phase 2B randomized clinical trial in Leigh syndrome, the first randomized trial using a novel redox-modulating agent to treat mitochondrial disease.

EPI743 (Enns) 08/20/10–04/14/16 0.12 CM
Emergency Use Protocol for EPI-743 in Acutely Ill Patients with Inherited Mitochondrial Respiratory Chain Disease Within 90 Days of End-of-life Care
This is an emergency treatment protocol for mitochondrial disease patients using a novel redox-modulating agent.

SPO117002 (Enns) 04/01/15 – 03/31/16 0.24 CM
National Institutes of Health
Lumina Diagnostics, Inc.
Breath Ammonia Monitoring Device for Children with Urea Cycle Disorders
The goal of this project is to develop a novel breath sensor to measure ammonia in patients with urea cycle disorders.

1R01DK1028201A1 (Peltz) 01/01/15–12/31/17 0.60 CM
National Institutes of Health $382,029
Stem Cell-Based In vivo Models of Human Genetic Liver Diseases
The goal of this grant is to generate murine models of genetic liver diseases, including polymerase gamma deficiency and Alagille syndrome, using stem cell techniques to generate "humanized" liver tissue.

Data and Website (Enns) 07/01/09–06/30/16 0.12 CM
Genzyme Corporation $63,885
Gaucher, Fabry, MPS 1 and other Genetically Based Metabolic Disorders
This is a longitudinal registry program for lysosomal storage disorders.

LAL-CL02 (Enns) 05/31/13–03/31/17 0.60 CM
Synageva BioPharma Corp. $406,690
A Multicenter, Randomized, Placebo-Controlled Study of SBC-102 in Patients with Lysosomal Acid Lipase Deficiency.
This is a clinical trial using a novel enzyme replacement therapy to treat LAL deficiency.

GOS (Enns) 09/16/11–08/31/18 0.12 CM
Shire Human Genetics Therapies, Inc. $126,201
Gaucher Disease Outcome Survey (GOS)
This is a longitudinal registry program for Gaucher Disease.

NIH 14-GG006326 (Enns) 09/01/13–08/31/18 0.01 CM
North American Mitochondrial Disease Consortium $580
This is a registry program for mitochondrial disease patients.

Foundation Grant (Enns) 06/01/14–03/31/16 0.12 CM
Austin Memorial Foundation $45,873
A cell-based method for screening mitochondrial disease therapies.
This study focuses on the use of spectromicroscopy and tandem mass spectrometry to study fibroblasts obtained from mitochondrial disease patients.

C. **Employment History:**

Academic Appointments: Clinical Instructor, Pediatrics
University of Southern California
June, 1994–June, 1995

Clinical Instructor, Pediatrics
University of California, San Francisco
July, 1998–present

Assistant Professor of Pediatrics
Stanford University
October, 1998–October, 2006

Associate Professor of Pediatrics
Stanford University
November, 2006–May, 2015

Professor of Pediatrics
Stanford University
June, 2015–present

Administrative Appointments:

Director, Biochemical Genetics Program
Stanford University School of Medicine
October, 1998–present

Scientific Advisory Board, Genotyping Core
Stanford University School of Medicine
May, 2000–June, 2001

Co–Director, UCSF/Stanford Lysosomal Disease Center
Stanford University Medical Center
July, 2000–present

Director, Metabolic Special Care Center
Lucile Salter Packard Children's Hospital
June, 2001–present

Medical Consultant, Newborn Screening Area Service Center
Stanford University Medical Center
June 2003–present

Program Director, Medical Genetics Residency Training Program
Stanford University School of Medicine
September, 2007–April, 2013

Associate Program Director, Medical Genetics Residency Training Program
Stanford University School of Medicine
May, 2013–April, 2014

Hospital/Clinic Privileges:

Division of Emergency Medicine, C.H.L.A., CA, 1995
Queen's Medical Center, Honolulu, HI, 2003–2011
University of California, San Francisco, CA, 1998–present
Stanford University Hospital, CA, 1998–present
Lucile Salter Packard Children's Hospital, CA, 1998–present
El Camino Hospital, Mountain View, CA, 2002–present
Kapiolani Children's Hospital, Honolulu, HI, 2003–present

D. Public and Professional Service:

Committees:

Children's Hospital of Los Angeles –
Residency Restructuring Committee, 1994–1995
Healthcare Economics Committee, 1994–1995
Intern Selection Committee, 1994–1995
Ethics Committee, 1994–1995
Medical Records Committee, 1994–1995

Lucile Salter Packard Children's Hospital –
Peer Review Committee, 5/01–2/04
Care Improvement Committee, 6/01–9/10
Chair, Medical Specialties Peer Review Committee, 2/04–9/10
Pediatric Research Center Steering Committee, 1/05–6/05
Child Health Research Program Subcommittee on Mentoring and Tutoring, 7/05–10/07
Pediatric Research Fund Grant Review Committee – 11/29/10, 3/25/15, 5/11/15

Stanford University Hospital –
Biochemical Laboratory Director Search Committee, 6/01–1/02
Pediatric Laboratory Oversight Committee, 2/02–7/06
Institutional Review Board, Panel 4 10/02–9/06
OB/GYN Maternal Fetal Medicine UTL Search Committee, 12/03–2/05
Molecular Pathology Education Committee, 2/05–present

State of California –
Advisory Board, Health Resources and Services Administration, California Tandem Mass Spectrometry Research Project, California Department of Health Services, 5/02–7/05

Consultant, Expanded Newborn Screening Using MS/MS Financial, Ethical, Legal and Social Issues (FELSI) Project, California Department of Health Services, 1/04–7/06
Newborn Screening Metabolic Disorders Guidelines Committee, California Department of Health Services, 7/06–present
Chair, Newborn Screening Metabolic Disorders Guidelines Committee, California Department of Health Services, 3/09–present

State of Hawaii –
Steering Committee, Hawaii Department of Health Regional Genetics Collaborative Network Project in Newborn Screening, 1/04–present

National Organizations –
U.S. Department of Health and Human Services, Interfacing MS/MS Outcome Information into Newborn Screening Systems panel meeting, Baltimore, MD 6/18/01
United Mitochondrial Disease Foundation Registry Design Team 9/01/02–8/30/04
Rare Disease Steering Committee (RDSC) of the National Disease Research Interchange (NDRI), 12/02–12/04
NIH National Institute of Child Health and Human Development Mental Retardation Research Subcommittee 6/16/03–6/15/04
American College of Medical Genetics Committee for the Development of Clinical Practice Guidelines for the Evaluation of Hypotonia 3/01/04–2/28/05
NIH National Institute of Child Health and Human Development Special Emphasis Panel: Disorders of Mitochondrial Energy Metabolism 7/29/04
March of Dimes, Expanded Newborn Screening Educational Outreach Taskforce 10/5/04–7/15/05
Fellow, American Academy of Pediatrics 4/1/05–present
American Academy of Pediatrics Committee on Genetics 7/1/05–6/30/11
Rare Diseases Clinical Research Network (RDCRN) Protocol Review Committee, Department of Health & Human Services, NIH 3/10/06–3/09/07
Society for Inherited Metabolic Disorders North American Metabolic Academy, 3/26/07–9/27/08
United Mitochondrial Disease Foundation Grant Review Committee, 8/07–6/11
NIH Therapeutic Approaches to Genetic Diseases Study Section (TAG) 5/1/2010
NIH National Institute of Neurological Disorders and Stroke Protocol Review Committee 7/9/10–7/8/11

NIH National Institute of Child Health and Human Development Best Pharmaceuticals for Children Act Program Evaluator 7/1/10–6/30/11
United Mitochondrial Disease Foundation Research Policy Review Committee, 10/14/10–present
Association of Public Health Laboratories Newborn Screening Meeting Planning Committee, 11/30/10–10/15/11
United Mitochondrial Disease Foundation "Mitochondrial Champion", 3/10/11–present
United Mitochondrial Disease Foundation Symposium Steering Committee, 11/30/11–present
United Mitochondrial Disease Foundation Clinical Research Committee, 1/4/13–present
Mitochondrial Medicine Society, Program Chair, 5/29/12–6/13/13
National Organization for Rare Disorders (NORD) Scientific and Medical Advisory Committee, 7/18/12–present
NIH National Human Genome Research Institute Special Emphasis Panel ZHG1 HGR-M (J1), 11/27/13
American Society of Human Genetics Program Committee, 12/18/13–10/22/16
American College of Medical Genetics ClinGen Workgroup, 6/24/14 – present

Board of Directors –
Society for Inherited Metabolic Disorders, 3/27/07–3/09/14

Data Safety Monitoring Board Membership –
BioMarin Pharmaceutical Inc.: PKU 003, "A Phase 3, Randomized, Double-Blind, Placebo-Controlled Study to Evaluate the Safety and Efficacy of Phenoptin in Subjects with Phenylketonuria." 12/16/05–2/15/06

University of Florida: "Phase 3 trial of coenzyme Q_{10} in mitochondrial diseases." 11/6/06–7/30/10

BioMarin Pharmaceutical Inc.: PAL 001, "A Phase 1, Open-Label, Dose-Escalation Study to Evaluate the Safety, Tolerability, and Pharmacokinetics of Single, Subcutaneous Doses of rAvPAL-PEG in Subjects with Phenylketonuria."2/20/08–3/31/09

BioMarin Pharmaceutical Inc.: MOR-004, "A Phase 3, Randomized, Double Blind, Placebo-Controlled, Multinational Clinical Study to Evaluate the Efficacy and Safety of 2.0 mg/kg/week and 2.0 mg/kg/every other week BMN 11 in Patients with

Mucopolysaccharidosis IVA (Morquio A syndrome)." 12/17/10–1/25/13

BioMarin Pharmaceutical Inc.: A Phase 3 Switchover Study of the Efficacy and Safety of BMN 701 (GILT-tagged Recombinant Human GAA) and Long-Term Study for Extended Treatment in rhBAA Exposed Subjects with Late-Onset Pompe Disease. 4/9/14–present

Data Safety Monitoring Board Chair –

BioMarin Pharmaceutical Inc.: PAL-002, "A Phase 2, Open-Label, Dose-Finding Study to Evaluate the Safety, Efficacy, and Tolerability of Multiple Subcutaneous Doses of rAvPAL-PEG in Subjects with PKU." 10/29/10–9/28/11

BioMarin Pharmaceutical Inc.: PAL 003, "Long-term Extension of a Phase 2, Open-Label, Dose-Finding Study to Evaluate the Safety, Efficacy, and Tolerability of Multiple Subcutaneous Doses of rAvPAL-PEG in Subjects with PKU."10/29/10–9/28/11

BioMarin Pharmaceutical Inc.: PAL 004, "A Phase 2, Open-Label Study to Evaluate the Safety, Tolerability, and Efficacy of 4 Subcutaneous Dose Levels of rAvPAL-PEG Administered Daily in Subjects with Phenylketonuria."10/29/10–9/28/11

Amicus Therapeutics: AT2220-010, An Open-Label, Multi-Center Study to Investigate Drug-Drug Interactions Between AT2220 (duvoglustat hydrochloride) and Alglucosidase Alfa in Patients with Pompe Disease. 11/23/11–3/21/13

BioMarin Pharmaceutical Inc.: 165-301,302 A Three-Part, Randomized, Double Blind, Placebo Controlled, Four-Arm, Discontinuation Study to Evaluate the Efficacy and Safety of Subcutaneous Injections of BMN 165 Self-Administered by Adults with Phenylketonuria. 7/12/13–present

Ultragenyx Pharmaceutical: UX007-CL201 Phase 2 Study of Triheptanoin in Fatty Acid Oxidation Disorders. 3/24/14–present

Ultragenyx Pharmaceutical: UX003-CL201 An Open-Label Phase 1/2 Study to Assess the Safety, Efficacy and Dose of UX003 rhGUS Enzyme Replacement Therapy in Patients with MPS 7. 4/30/14–1/6/17

Scientific Advisory Board Membership–
Familial Dysautonomia Hope Foundation 4/05–present
United Mitochondrial Disease Foundation 3/07–6/11
Hyperion Therapeutics 9/07–6/15
Medical & Scientific Advisory Committee of the SSADH Association, 12/12–present

Organizations Formed: The Stanford University Mitochondrial Interest Group ("MitoGroup"), 2/04
The Bay Area Mitochondria Association, 6/04 (established in conjunction with Bertram Lubin, MD and Bruce Ames, MD, Children's Hospital Oakland Research Institute, CA)

Other Public Service: Muscular Dystrophy Association, Website "Mitochondrial Chat" session 8/23/04

Journal peer review:
(*editorial board)* – Molecular Genetics and Metabolism (10/05–12/13)
Journal of Inherited Metabolic Disease (8/11–present)
Molecular Genetics and Metabolism Reports (12/13–present)
Journal of Inborn Errors of Metabolism and Screening (1/15 – present)

(*ad hoc*) – Acta Paediatrica
American Journal of Hematology
American Journal of Medical Genetics
American Journal of Medicine
Application of Clinical Genetics
Archives of Disease of Childhood
Biochimica et Biophysica Acta
Biochimica et Biophysica Acta – Molecular Cell Research
BMC Pediatrics
Brain
Clinical Genetics
Clinical Nutrition
Developmental Medicine & Child Neurology
EMBO Molecular Medicine
European Journal of Obstetrics & Gynecology and Reproductive Biology
Expert Opinion on Orphan Drugs
Expert Opinion on Pharmacotherapy
FASEB Journal
Future Medicine
Gene
Genetics in Medicine

Human Gene Therapy
Human Genetics
Human Mutation
JAMA Neurology
Journal of Adolescent Health
Journal of Genetic Counseling
Journal of Hepatology
Journal of Inherited Metabolic Disease
Journal of Inherited Metabolic Disease Reports
Journal of Medical Genetics
Journal of Neurodevelopmental Disorders
Journal of Neurology
Journal of Neuroscience Research
Journal of Pediatric Gastroenterology and Nutrition
Journal of Pediatrics
Lancet Neurology
Mitochondrion
Molecular Genetics and Metabolism
Molecular Genetics and Metabolism Reports
Muscle & Nerve
Nanotechnology
Nature Clinical Practice Neurology
Nature Communications
Neonatology
NeoReviews
Neurology
Neuropediatrics
Neurotherapeutics
New England Journal of Medicine
Orphanet Journal of Rare Diseases
Pediatric Pulmonology
Pediatric Research
Pediatric Transplantation
Pediatrics
PLoS Genetics
PLoS ONE
Therapeutics and Clinical Risk Management
Transplantation
Ultrastructural Pathology

Other peer review:
(*ad hoc*) –

GeneReviews at GeneTests
National Organization for Rare Disorders
Oxford University Press

E.Memberships in Professional Associations and Learned Societies:

General Medical Council, U.K., 1991–present
American Academy of Pediatrics, 1994–2016
American Society of Human Genetics, 1997–present
Western Society for Pediatric Research, 1998–present
United Mitochondrial Disease Foundation, 1999–present
The Mitochondrial Interest Group, 2000–present
The Mitochondrial Medicine Society, 2000–present
Society for the Study of Inborn Errors of Metabolism, 2000–present
Society for Inherited Metabolic Disorders, 2000–present
American Association for the Advancement of Science, 2000–present
American Academy of Pediatrics, Section on Genetics and Birth Defects, 2005–2011
Bay Area Mitochondrial Association, 2004–present

F. Scholarly Publications:

Peer-Reviewed Articles (90):

1. Warburton D, Buckley S, Cosico S, **Enns GM**, Saluna T. Combined effects of corticosteroids, thyroid hormones, and beta–agonist receptor binding in fetal lamb lung. Pediatr Res 24:166–170, 1988.

2. **Enns GM,** Cox VA, Goldstein RB, Gibbs DL, Harrison MR, Golabi M. Congenital diaphragmatic defects and associated syndromes, malformations, and chromosome anomalies: A retrospective study of 60 patients and literature review. Am J Med Genet 79:215–222, 1998.

3. Ewart-Toland A, **Enns GM**, Cox VA, Chandra Mohun G, Golabi M. Severe congenital anomalies requiring transplantation in children with Kabuki syndrome. Am J Med Genet 80:362–367, 1998.

4. **Enns GM**, Barkovich AJ, Weisiger K, Ohnstad C, Packman S. Progressive neurological deterioration and MRI changes in *cblC* methylmalonic acidaemia treated with hydroxocobalamin. J Inherit Metab Dis 22:599–607, 1999.

5. **Enns GM**, Roeder E, Chan RT, Ali-Catts Z, Cox V, Golabi M. Cyclophosphamide (Cytoxan) embryopathy: A distinct phenotype? Am J Med Genet 86:237–241, 1999.

6. **Enns GM**, Martinez DR, Kuzmin AI, Koch R, Wakeem C, Woo SLC, Eisensmith RC, Packman S. Molecular correlations in phenylketonuria: Mutation patterns and

corresponding biochemical and clinical phenotypes in a heterogeneous California population. Pediatr Res 46:594–602, 1999.

7. **Enns GM**, Bennett MJ, Hoppel C, Weisiger C, Ohnstad C, Golabi M, Packman S. Mitochondrial respiratory chain complex I deficiency presenting with clinical and biochemical features of long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency. J Pediatr 136(2):251–254, 2000.

8. Westphal V, **Enns GM**, McCracken M, Freeze H. Functional analysis of two new mutations in a congenital disorder of glycosylation (CDG) Ia patient with mixed Asian ancestry. Mol Genet Metab 73(1):71–76, 2001.

9. **Enns GM**, Seppala R, Musci TJ, Weisiger K, Ferrell LD, Wenger DA, Gahl WA, Packman S. Clinical course and biochemistry of sialuria. J Inherit Metab Dis 24(3):328–336, 2001.

10. Hintz S, Matern D, Strauss A, Bennett MJ, Hoyme HE, Schelley S, Kobori J, Colby C, Lehman NL, **Enns GM**. Early neonatal presentation of long-chain 3-hydroxyacyl-CoA dehydrogenase and mitochondrial trifunctional protein deficiencies. Mol Genet Metab 75(2):120–127, 2002.

11. **Enns GM**, Steiner RD, Buist N, Cowan C, Leppig KA, McCracken MF, Westphal V, Freeze HH, O'Brien J, Jaeken J, Matthijs G, Behera S, Hudgins L. Clinical and molecular features of congenital disorder of glycosylation in patients with diverse ethnic origins and a type 1 sialotransferrin pattern. J Pediatr 141(5):695–700, 2002.

12. Newell JW, Seo N-S, **Enns GM**, McCracken M, Mantovani JF, Freeze HH. Congenital diosrder of glycosylation Ic in patients of Indian origin. Mol Genet Metab 79(3):221– 228, 2003.

13. Adam MP, Manning MA, Beck AE, Kwan A, **Enns GM**, Clericuzio C, Hoyme HE. Methotrexate/misoprostol embryopathy: report of four cases resulting from failed medical abortion. Am J Med Genet 123A(1):72–78, 2003

14. Wong L-J C, Perng C-L, Hsu C-H, Bai R-K, Schelley S, Vladutiu GD, Vogel H, **Enns GM**. Compensatory amplification of mitochondrial DNA in a mitochondrial DNA deletion syndrome with a novel deletion/duplication and high mutant load. J Med Genet 40(11):e125, 2003.

15. **Enns GM**, Barkovich AJ, van Kuilenberg ABP, Manning M, Sanger T, Witt D, van Gennip AH. Head imaging abnormalities in dihydropyrimidine dehydrogenase deficiency. J Inherit Metab Dis 27(4):513–22, 2004.

16. Chen KC, Cherry AM, Hahn JS, **Enns GM**. Mild developmental delay in terminal chromosome 6p deletion. Am J Med Genet 129A(2):201–5, 2004.

17. Manning MA, Cassidy SB, Clericuzio C, Cherry AM, Schwartz S, Hudgins L, **Enns GM**, Hoyme HE. Terminal 22q deletion syndrome: a newly recognized cause of speech and language disability in the autistic spectrum. Pediatrics 114(2):451–7, 2004.

18. Currier SC, Lee CK, Chang BS, Bodell AL, Pai GS, Job L, lagae LG, Al-Gazali LI, Eyaid WM, **Enns GM**, Dobyns WB, Walsh CA. Mutations in *POMT1* are found in a minority of patients with Walker–Warburg syndrome. Am J Med Genet 133A(1):53–57, 2005.

19. Nagarajan S, **Enns GM**, Millan MT, Winter S, Sarwal MM. Management of methylmalonic acidemia by combined liver–kidney transplantation. J Inherit Metab Dis 28(4):517–524, 2005.

20. **Enns GM**, O'Brien WE, Kobayashi K, Shinzawa H, Pelligrino JE. Postpartum "psychosis" in mild argininosuccinate synthetase deficiency. Obstet Gynecol 105(5):1244–1246, 2005.

21. van Kuilenberg ABP, Meinsma R, Beke E, Bobba B, Boffi P, **Enns GM**, Witt DR, Dobritzsch D. Identification of three novel mutations in the dihydropyrimidine dehydrogenase gene associated with altered pre–mRNA splicing or protein function. Biol Chem 386(4):319–324, 2005.

22. **Enns GM**, Hoppel CL, DeArmond S, Schelley S, Bass N, Weisiger K, Horoupian D, and Packman S. The relationship of mitochondrial dysfunction to the pathogenesis of fiber type abnormalities in skeletal muscle. Clin Genet 68(4):337-348, 2005.

23. Gallagher RC, Cowan TM, Goodman SI, **Enns GM**. Glutaryl-CoA dehydrogenase deficiency and newborn screening: retrospective analysis of a low excretor provides further evidence that some cases may be missed. Mol Genet Metab 86(3):417–420, 2005.

24. **Enns GM**, Bai RK, Beck AE, Wong LJ. Molecular–clinical correlations in a family with variable tissue mitochondrial DNA T8993G mutant load. Mol Genet Metab 88(4):364–371, 2006.

25. Abdul-Rahman OA, Trang HL, Kwan A, Schlaubitz S, Barsh GS, **Enns GM**, Hudgins L. Genitopatellar syndrome: expanding the phenotype and excluding mutations in *LMX1B* and *TBX4*. Am J Med Genet 140(14):1567–1572, 2006.

26. Shieh JTC, Swidler P, Martignetti JA, Ramirez MCM, Balboni I, Kaplan J, Kennedy J, Abdul-Rahman O, **Enns GM**, Sandborg C, Hoyme HEH. Systemic hyalinosis: a distinctive early–onset childhood disorder characterized by mutations in the anthrax receptor 2 gene (*ANTRX2*). Pediatrics 118(5):e1485–92, 2006.

27. Veeravagu A, Hou LC, Hsu AR, **Enns GM**, Huhn SL. Glutaric acidemia type I: a neurosurgical perspective. J Neurosurg 107(2):167-172, 2007.

28. **Enns GM**, Berry SA, Berry GT, Rhead WT, Brusilow SW, Hamosh A. Survival after treatment with phenylacetate and benzoate for urea–cycle disorders. New Engl J Med 356(22):2282–92, 2007.

29. Dimmock DP, Zhang Q, Dionisi-Vicci C, Carrozzo R, Shieh J, Tanag LY, Truong C, Schmitt E, Sifry-Platt M, Lucioli S, Santorelli FM, Ficicioglu CH, Rodriguez M, Wierenga K, **Enns GM**, Longo N, Lipson MH, Vallance H, Craigen WJ, Scaglia F, Wong LJ. Clinical and molecular features of mitochondrial DNA depletion due to mutations in deoxyguanosine kinase. Hum Mutat – Mutation in Brief #990 (2007) Online.

30. Cheyette BNR, Cheyette SNR, Cusmano-Ozog, K **Enns GM**. Dopa–responsive dystonia presenting as delayed and awkward gait. Pediatr Neurol 38(4):273–275, 2008.

31. Longo N, Schrijver I, Vogel H, Pique LM, Cowan TM, Pasquali M, Hedlund GL, Ernst SL, Gallagher RC, **Enns GM**. Progressive cerebral vascular degeneration with mitochondrial encephalomyopathy. Am J Med Genet Part A 146A:361–7, 2008.

32. Yuan N, El-Sayed YY, Ruoss SJ, Riley E, **Enns GM**, Robinson TE. Successful pregnancy and Cesarean delivery via non–invasive positive pressure ventilation in a patient with mitochondrial thymidine kinase 2 deficiency. J Perinatol 29(2):166-167, 2009.

33. A novel homozygous *SCO2* mutation, G193S, causing fatal infantile cardio-encephalomyopathy. Mobley BC, **Enns GM**, Wong LJ, Vogel H. Clin Neuropathol 28(2):143-149, 2009.

34. Miousse IR, Watkins D, Lavallée J, Coelho D, Clarke JTR, Crombez EA, Vilain E, Cederbaum S, Bernstein JA, Cowan T, **Enns GM**, Fowler B, Rosenblatt DS. Clinical and genetic findings in patients with the *cblD* inborn error of cobalamin metabolism. J Pediatr 154(4):551–556, 2009.

35. Scharfe C, Lu HH-S, Neuenberg JK, Allen E, Klopstock T, Cowan TM, **Enns GM**, Davis RW. Mapping gene associations in human mitochondria using clinical disease phenotypes. PLoS Comput Biol 5(4):e1000374, 2009.

36. Atkuri KR, Cowan TM, Kwan T, Ng A, Herzenberg LA, Herzenberg LA, **Enns GM**. Inherited disorders affecting mitochondrial function are associated with glutathione deficiency and hypocitrullinemia. Proc Natl Acad Sci USA 106(10):3941–3945, 2009.

37. Shieh JTC, Berquist WE, Zhang Q, Chou PC, Wong LJC, **Enns GM**. Novel deoxyguanosine kinase gene mutations and viral infection predispose apparently healthy children to fulminant liver failure. J Pediatr Gastroenterol Nut 49(1):130–132, 2009.

38. Abdul–Rahman OA, Edghill EL, Kwan A, **Enns GM**, Hattersley AT. Hypoplastic glomerulocystic kidney disease and hepatoblastoma: a potential association not caused by mutations in hepatocyte nuclear factor 1ß. J Pediatr Hematol Oncol 31(7):527–529, 2009.

39. Stevenson T, Millan MT, Berquist WE, Wayman K, Sarwal M, Esquivel CO, **Enns GM**. Long–term outcome following pediatric liver transplantation for metabolic disorders. Pediatr Transpl 14(2):268–275, 2010.

40. **Enns GM**, Koch R, Brumm V, Blakely E, Suter R, Jurecki E. Suboptimal outcomes in patients with PKU treated early with diet alone: revisiting the evidence. Mol Genet Metab 101(2-3):99–109, 2010.

41. Casper RC, Gilles A, Fleisher BE, Baran J, DeBattista A, **Enns GM**, Lazzeroni LC. Length of prenatal exposure to selective serotonin reuptake inhibitors (SSRI) Antidepressants: effects on neonatal adaptation and psychomotor development. Psychopharmacology (Berl) 217(2):211-9, 2011.

42. Shen P, Wang W, Krishnakumar S, Palm C, Chi AK, **Enns GM**, Davis RW, Speed TP, Mindrinos M, Scharfe C. High quality DNA sequence capture of 524 disease candidate genes. Proc Natl Acad Sci USA 108(16):6549–54, 2011.

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100. Kim IK, Niemi AK, **Enns GM**, Esquivel C. Pediatric liver transplantation as definitive therapy for urea cycle disorders. Transplantation 94(10S):29, 2012. Poster presentation American Transplant Society Annual Meeting, Boston, MA, 2012. Platform presentation The Transplant Society Annual Meeting, Berlin, Germany, July, 2012.

101. Hendriksz C, Wraith E, Dhawan A, Whitley C, Banikazemi M, Bialer M, Guardamagna O, Jones S, Raiman J, Cederbaum S, DiRocco M, Domm J, **Enns GM**, Finegold D, Simonds A, Eckert S, Schneider E, Quinn A. Natural history study of early onset lysosomal acid lipase (LAL) deficiency (Wolman disease) confirms a severe and rapid disease course. Poster presentation Society for the Study of Inborn Errors of Metabolism Annual Meeting, October, 2012.

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103. Jones S, **Enns GM**, Balwani M, Breen C, Sharma R, Deegan P, Malinova V, Honzik T, Valayannopoulos V, Schneider E, Burg J, Quinn AG. Enzyme replacement with recombinant human lysosomal acid lipase (rhLAL) in patients with cholesteryl ester storage disease, the late onset form of LAL deficiency, produces sustained decreases in transaminases and reduction in liver fat content. Submitted to American Association for the Study of Liver Diseases Annual Meeting.

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105. Balwani M, Malinova V, Sharma R, Valayannopoulos V, Stock EO, Boyadjiev SA, Kessler B, Deegan P, **Enns GM**, Breen C, Kane JP, Schneider E, Quinn AG. Long term safety and clinical activity of SBC-102, a recombinant human lysosomal acid lipase (rhLAL), in patients with late onset LAL deficiency. Platform presentation Lysosomal Disease Network WORLD Conference, Orlando, FL, 2013.

106. Gallant NM, Wilnai Y, Lee C, Lorey F, Feuchtbaum L, Tang H, Leydiker K, **Enns GM**, Packman S, Lin HJ, Wilcox W, Cederbaum SD, Abdenur JE. Informing the debate: newborn screening for systemic primary carnitine deficiency. Platform presentation SSIEM 2013.

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M, Domm J, **Enns GM**, Finegold D, Gargus J, Zaki O, Eckert S, Schneider E, Quinn AG. Severe and rapid disease course in the natural history of early onset lysosomal acid lipase (LAL) deficiency (Wolman Disease). Submitted to North American Society of Pediatric Gastroenterology Hepatology and Nutrition, Chicago, IL, 2013.

108. Atwal PS, Schelley S, **Enns GM**. Mutations in the complex III assembly factor tetratricopeptide 19 gene *TTC19* are a rare cause of Leigh syndrome. Poster presentation United Mitochondrial Disease Foundation Annual Meeting, Newport Beach, CA, 2013.

109. Bai R, Haverfield E, Higgs J, Suchy SF, Arjona D, Rettner K, Smaoui N, Richard G, Bale S, Kendall FD, Parikh S, Gropman A, Haas R, Goldstein A, Panzer JA, Yum SW, Falk MJ, Saneto RP, **Enns GM**, Chung WK. First-line genetic testing for mitochondrial disorders in the next-generation sequencing era: comprehensive known disease gene panel or exome sequencing? Poster presentation United Mitochondrial Disease Foundation Annual Meeting, Newport Beach, CA, 2013.

110. Wilnai Y, Cox R, Bai R, **Enns GM**. Severe multi-systemic presentation of *COX10* deficiency. Poster presentation United Mitochondrial Disease Foundation Annual Meeting, Newport Beach, CA, 2013.

111. Khouzam A, Platt J, Cox R, **Enns GM**. Novel c.316G>A mutation in *PDHA1* causing PDH deficiency. Poster presentation United Mitochondrial Disease Foundation Annual Meeting, Newport Beach, CA, 2013.

112. Platt J, Cox R, **Enns GM**. Mitochondrial NGS panels: is more better? Poster presentation United Mitochondrial Disease Foundation Annual Meeting, Newport Beach, CA, 2013.

113. Atwal PS, Le AT, Moore T, Cowan T, **Enns GM**. Biomarkers for evidence of mitochondrial dysfunction in cobalamin C disease. Poster presentation Wellcome Trust Scientific Conference – Mitochondrial disease: translating biology into new treatments, Cambridge, UK, 2013.

114. Holman E, **Enns GM**, Blankenberg FG. HMPAO uptake assay with t-BHP or irradiation of primary patient SURF-1 fibroblast cultures. Poster presentation Society of Nuclear Medicine and Molecular Imaging Annual Meeting, 2014. J Nuc Med Meeting Abstracts, May 2014;55:1805.

115. Blankenberg FG, **Enns GM**. Monitoring mitochondrial disease related nephropathy and subclinical redox stress of the brain in the kd/kd murine model. Poster presentation Society of Nuclear Medicine and Molecular Imaging Annual Meeting, 2014. J Nuc Med Meeting Abstracts, May 2014;55:1806.

116. **Enns GM**, Shashi V, Zahir F, Gambello MJ, Bast T, Zoghbi HY, Platt J, Cox R, Bernstein J, Crimian R, Schoch K, Scavina M, Walter RS, Need AC, Oviedo A, Langlois S, Gibbs R, Bainbridge M, Schaaf C, Snyder M, Boyle S, Butte AJ, FORGE Canada Consortium, He P, Freeze HH, Goldstein DB. Mutations in *NGLY1* cause an inherited disorder of the endoplasmic reticulum-associated degradation (ERAD) pathway. Mol Genet Metab 111(3):236, 2014. Oral presentation Society for Inherited Metabolic Disorders Annual Meeting, Carmel, CA 2014.

117. **Enns GM**, Moore T, Le A, Atkuri K, Shah MK, Cusmano-Ozog K, Niemi AK, Cowan TM. Glutathione deficiency and redox imbalance in mitochondrial disorders. Poster presentation Society for Inherited Metabolic Disorders Annual Meeting, Carmel, CA 2014.

118. Frazier DM, Van Calcar SC, **Enns GM**, McGuire PJ. Need for evidence-based research to assess benefits of nutrition treatments for rare inborn errors of metabolism. Mol Genet Metab 111(3):295, 2014. Poster presentation Society for Inherited Metabolic Disorders Annual Meeting, Carmel, CA 2014.

119. Atwal AT, Le T, Moore T, Cowan T, **Enns GM**. Biomarkers for evidence of mitochondrial dysfunction in cobalamin C disease. Mol Genet Metab 111(3):247, 2014. Poster presentation Society for Inherited Metabolic Disorders Annual Meeting, Carmel, CA 2014.

120. Wilnai Y, Alcorn D, Benitz W, Berquist W, Bernstein JA, Blumenfeld YJ, Castillo R, Concepcion W, Palma J, Summar ML, Schelley S, Vishnu PA, **Enns GM**. Prenatal treatment of ornithine transcarbamylase deficiency. Mol Genet Metab 111(3):248, 2014. Poster presentation Society for Inherited Metabolic Disorders Annual Meeting, Carmel, CA 2014.

121. Niemi AK, Cox R, Platt J, Wayman K, Mark J, **Enns GM**. Mitochondrial complex care clinic model – empowering families as part of the care team. Mol Genet Metab 111(3):254, 2014. Poster presentation Society for Inherited Metabolic Disorders Annual Meeting, Carmel, CA 2014.

122. **Enns GM**, Moore T, Le A, Atkuri K, Shah MK, Cusmano-Ozog K, Niemi AK, Cowan TM. Correlation of glutathione redox potential with mitochondrial disease etiology and clinical severity. Mol Genet Metab 111(3):268, 2014. Poster presentation Society for Inherited Metabolic Disorders Annual Meeting, Carmel, CA 2014.

123. Soler-Alfonso C, **Enns GM**, Koenig MK, Saavedra H, Bonfante-Megia E, Northrup H. Identification of *HIBCH* gene mutations causing autosomal-recessive Leigh syndrome: a gene involved in valine metabolism. Poster presentation American College of Medical Genetics Annual Meeting, Nashville, TN 2014.

124. Cox R, Platt J, van Haren K, **Enns GM**. A treatable mimic of Leigh syndrome: biotin- and thiamine-responsive basal ganglia disease. Poster presentation United Mitochondrial Disease Foundation Annual Meeting, Pittsburgh, PA, 2014.

125. Splinter BA, Niemi AK, Cox R, Platt J, Shah M, **Enns GM**, Kasahara M, Bernstein JA. Parents report impaired health-related quality of life in children with methylmalonic acidemia with improvements in development and health following liver transplantation. Poster presentation National Society of Genetic Counselors Conference Annual Meeting, 2014.

126. **Enns GM**, Moore T, Le A, Shah MK, Cusmano-Ozog K, Niemi AK, Cowan TM. Evidence of secondary mitochondrial dysfunction in patients with organic acidemias. Poster presentation American Society of Human Genetics Annual Meeting, San Diego, CA 2014.

127. Balwani M, Burton B, Baric I, Bialer M, Burrow TA, Camarena Grande C, Coker M, Consuelo Sanchez A, Deegan P, Di Rocco M, **Enns GM**, Erbe RW, Ezgu FS, Feillet F, Ficicioglu C, Furuya KN, Guelberg NB, Kane J, Kostyleva M, Laukaitis CM, Valinova V, Mengel E, Murphy E, Neilan EG, Nightingale S, Rosado JP, Peters H, Rahman Y, Scarpa M, Schwab KO, Smolka V, Taybert J, Valayannopoulos V, Wood M, Zeniya M, Yang Y, Eckert S, Rojas-Caro S, Quinn AG. Results of a global phase 3, randomized, double-blind,placebo-controlled trial evaluating the efficacy and safety of sebelipase alfa as an enzyme replacement therapy in children and adults with lysosomal acid lipase deficiency. Oral presentation American Association for the Study of Liver Diseases Annual Meeting, Boston, MA 2014.

128. Leduc MS, Yang Y, Scaglia F, He W, Brown CW, Elsea SH, Cox R, **Enns GM**, Ward PA, Braxton AA, Vaughn TR, Eng CM, Gibbs RA, Beaudet AL. Medical benefit of individual genome sequencing: examples from BCM clinical whole exome sequencing. Submitted to American College of Medical Genetics and Genomics Annual Meeting.

129. Gomez-Ospina N, **Enns GM**. Neonatal cholestasis due to mutations in the farnesoid X receptor. Oral presentation Western Society for Pediatric Research, Carmel, CA 2015.

130. Bai R, Balog A, Higgs J, Retterer K, Arjona D, Juusola J, Vitazka P, **Enns GM**, Goldstein A, Tarnopolsky M, Parikh S, Copeland W, Niyazov D, Falk M, Chung W, Bale S, Richard G. Whole exome sequencing and whole mitochondrial genome sequencing for the molecular diagnosis of mitochondrial disorders: Lesson from 865 cases. Poster presentation UMDF Annual Meeting, Herndon, VA 2015.

131. Zarkoob H, Shen P, **Enns GM**, Sabatti C, Scharfe C. Characterizing the human mitochondrial proteome using a genome-wide functional linkage network. Mitochondrion 24:S26, 2015. Poster presentation UMDF Annual Meeting, Herndon, VA 2015.

132. Vockley J, Charrow J, Ganesh J, Eswara M, Diaz GA, **Enns GM**, Marsden DL. Response to compassionate use of triheptanoin in infants with cardiomyopathy due to long chain fatty acid oxidation defects (LC-FAODs). Poster presentation SSIEM, Lyon, France.

133. Bai R, Balog, A, Higgs J, Retterer K, Arjona D, Juusoal J, Ali Abu Q, Suchy S, **Enns GM**, Haas R, Goldstein A, Tarnopolsky M, Parikh S, Copeland WC, Niyazov D, Falk MJ, Bale SJ, Chung WK, Richard G. Whole exome sequencing and whole mitochondrial genome sequencing for the molecular diagnosis of mitochondrial disorders. Poster presentation ASHG Annual Meeting, Baltimore, MD 2015.

134. Myers A, Goyal N, Vagelos R, **Enns GM**. OPA-1-plus syndromes may include cardiomyopathy – expanding the phenotype. Poster presentation SIMD, FL 2016.

135. Scott AI, Cusmano-Ozog K, **Enns GM**, Cowan TM. Rate of leucine clearance in MSUD. Poster presentation SIMD, FL 2016.

136. Cohen BH, **Enns GM**, Haas R, Longo N, Scaglia F, Lang W, Sile S. An open-label, dose-escalating study to assess the safety, tolerability, efficacy, pharmacokinetics and pharmacodynamics, of cysteamine bitartrate delayed-release capsules (RP103) for treatment of children with inherited mitochondrial disease. Poster presentation UMDF, Seattle, WA 2016.

137. Scott AI, Myers A, **Enns GM**, Cowan T. Aicardi-Goutieres syndrome and the ADAR1 dominant negative mutation: a case report. Poster presentation ASHG Annual Meeting, Vancouver, Canada 2016.

138. Kohler J, Fisk D, Grove ME, Zastrow DB, Fernandez L, Zornio P, Dries A, Schelley S, **Enns GM**, Taylor R, Olahova M, Members of the Undiagnosed Diseases Network, Ashley EA, Fisher P, Bernstein JA, Wheeler M. Novel mutation in *ATP5D* as a cause of mitochondrial ATP synthase deficiency. Poster presentation ASHG Annual Meeting, Vancouver, Canada 2016.

Books (1):

1. Hudgins L, Toriello HV, **Enns GM**, Hoyme HE, Eds. Signs and Symptoms of Genetic Conditions: A Handbook. Oxford University Press, New York, 2014.

Book Chapters (28):

1. **Enns GM**. Inborn errors of metabolism with features of hypoxic–ischemic encephalopathy. In *Fetal and Neonatal Brain Injury: Mechanisms, Management and*

the Risks of Practice, 3rd edition. Stevenson DK, Benitz WE, Sunshine P eds. Cambridge University Press, Cambridge, UK, 2002, pp.392–408.

2. **Enns GM**, Packman S. Metabolic Disorders: Phenylketonuria and biopterin disorders. In *Gellis and Kagan's Current Pediatric Therapy 17.* Burg FD, Ingelfinger JR, Wald ER, Polin RA eds. Saunders, Philadelphia, 2002, pp.731–736.

3. **Enns GM**. Multiple pterygium syndrome. In *The NORD Guide to Rare Disorders.* Gruson ES, ed. Lippincott, Williams and Wilkins, Philadelphia, 2003, p.241.

4. **Enns GM**. Simpson dysmorphia syndrome. In *The NORD Guide to Rare Disorders.* Gruson ES, ed. Lippincott, Williams and Wilkins, Philadelphia, 2003, pp.251–252.

5. **Enns GM**. Fryns syndrome. In *The NORD Guide to Rare Disorders.* Gruson ES, ed. Lippincott, Williams and Wilkins, Philadelphia, 2003, pp.715–716.

6. **Enns GM**. Inborn errors of metabolism: Mitochondrial disease. In *Forfar and Arneil's Textbook of Pediatrics, 6th edition.* McIntosh N, Helms P, Smyth R eds. Churchill Livingstone, London, 2003, pp.1219–1231.

7. **Enns GM**, Gibson M. Inborn errors of metabolism: Congenital disorders of glycosylation. In *Forfar and Arneil's Textbook of Pediatrics, 6th edition.* McIntosh N, Helms P, Smyth R eds. Churchill Livingstone, London, 2003, pp.1247–1249.

8. **Enns GM**. Oxidative Metabolism. In *Encyclopedia of the Neurological Sciences*. Aminoff, M, and Daroff, R, eds. Academic Press, San Diego, 2003, Vol.3, pp.708–713.

9. **Enns GM**. Respiratory Chain Disorders. In *Encyclopedia of the Neurological Sciences*. Aminoff, M, and Daroff, R. eds. Academic Press, San Diego, 2003, Vol.4, pp.154–160.

10. Kuppermann N, **Enns GM**, Kuppermann N. Metabolic disease. In *APLS: The Pediatric Emergency Medicine Resource, 4th edition.* Fuchs S, Gausche-Hill M, Yamamoto L eds. Jones and Bartlett, Massachusetts, 2004, pp.186–207.

11. **Enns GM**, Steiner RL. Lysosomal storage disorders. In *Pediatrics*. Osborn L, DeWitt T, First L, Zenel J eds. Elsevier Mosby, Philadelphia, 2004, pp.1007–1012.

12. **Enns GM**, Steiner RL. Diagnosis and treatment of children with suspected metabolic disease. In *Pediatrics*. Osborn L, DeWitt T, First L, Zenel J eds. Elsevier Mosby, Philadelphia, 2004, pp.1866–1875.

13. **Enns GM**, Cowan TM, Klein O, Packman S. Aminoacidemias and organic acidemias. In *Pediatric Neurology: Principles & Practice, 4th edition.* Swaiman KF, Ashwal S, Ferriero DM, Eds. Mosby Elsevier, 2006, pp.567-602.

14. Glaser N, **Enns GM**, Kuppermann N. Hypoglycemia. In *Baren: Pediatric Emergency Medicine.* Rothrock S, Ed. Elsevier., 2007, pp.765-770.

15. **Enns GM**, Buist NRM. Inborn errors of metabolism: Mitochondrial disease. In *Forfar and Arneil's Textbook of Pediatrics, 7th edition.* McIntosh N, Helms P, Smyth R eds. Churchill Livingstone, London, 2008, pp.1081-8

16. **Enns GM**, Gibson M. Inborn errors of metabolism: Congenital disorders of glycosylation. In *Forfar and Arneil's Textbook of Pediatrics, 7th edition.* McIntosh N, Helms P, Smyth R eds. Churchill Livingstone, London, 2008, pp.1119-21

17. Cowan TM, Steiner RL, **Enns GM**. Lysosomal disorders. In *Essential Pediatric Endocrinology and Inborn Errors of Metabolism.* Sarafoglou K, Ed. McGraw-Hill, 2009, pp.721–755.

18. **Enns GM**. Inborn errors of metabolism with features of hypoxic–ischemic encephalopathy. In *Fetal and Neonatal Brain Injury: Mechanisms, Management and the Risks of Practice, 4th edition.* Stevenson DK, Benitz WE, Sunshine P, Hintz SR, Druzin ML Eds. Cambridge University Press, Cambridge, UK, 2009, pp.389–401.

19. **Enns GM**. Metabolic Disease. In *Encyclopaedia Britannica: Online Academic Edition*. Encyclopaedia Britannica Corporation. http://www.britannica.com/ EBchecked/ topic/377311/metabolic-disease, 2012.

20. **Enns GM**, Cowan TM, Klein O, Packman S. Aminoacidemias and organic acidemias. In *Pediatric Neurology: Principles & Practice, 5th edition.* Swaiman KF, Ashwal S, Ferriero DM, Schor NF Eds. Mosby Elsevier, 2012, pp.328-356.

21. **Enns GM**, Hudgins L, Cowan TM. Genetic Testing. In *Signs and Symptoms of Genetic Conditions: A Handbook.* Hoyme HE, Hudgins L, Enns GM, Toriello H, Eds. Oxford University Press, New York, 2014, pp.1–8.

22. Sherr EH, **Enns GM**. Hypotonia. In *Signs and Symptoms of Genetic Conditions: A Handbook.* Hoyme HE, Hudgins L, Enns GM, Toriello H, Eds. Oxford University Press. New York, 2014, pp.145–163.

23. Wang CH, **Enns GM**. Ataxia. In *Signs and Symptoms of Genetic Conditions: A Handbook.* Hoyme HE, Hudgins L, Enns GM, Toriello H, Eds. Oxford University Press. New York, 2014, pp.190–208.

24. Cowan TM, **Enns GM**. Metabolic acidosis. In *Signs and Symptoms of Genetic Conditions: A Handbook.* Hoyme HE, Hudgins L, Enns GM, Toriello H, Eds. Oxford University Press. New York, 2014, pp.235–247.

25. **Enns GM**, Cowan TM. Hyperammonemia. In *Signs and Symptoms of Genetic Conditions: A Handbook*. Hoyme HE, Hudgins L, Enns GM, Toreillo H, Eds. Oxford University Press, New York, 2014, pp.261–279.

26. **Enns GM**. Inborn errors of metabolism and newborn screening. In *Newborn Medicine – Practical Approaches to Newborn Care: A Global Perspective in the Age of Information*. Misra S, Govindaswami B, Eds. Jaypee Brothers Medical Publishers, New Delhi, India. (in press)

27. **Enns GM**. Inborn Errors of Metabolism and single gene disorders with features of neonatal encephalopathy. In *Fetal and Neonatal Brain Injury: Mechanisms, Management and the Risks of Practice, 5th edition*. Stevenson DK, Benitz WE, Sunshine P, Hintz SR, Druzin ML eds. Cambridge University Press, Cambridge, UK. (in press)

28. Gallagher R, **Enns GM**, Cowan TM, Mendelsohn B, Lee C, Packman S. Aminoacidemias and organic acidemias. In *Pediatric Neurology: Principles & Practice, 6th edition*. Swaiman KF, Ashwal S, Ferriero DM, Eds. Mosby Elsevier (in press)

Book Reviews (1):

1. **Enns GM.** *Review of:* Mitochondrial Disorders: from pathophysiology to acquired defects: Desnuelle C, DiMauro S, Eds. Springer, France 2002. Doody Publishing.

Electronic Publications (5):

1. **Enns GM**. Galactosemia. In *Pocket Medicine/Internal Medicine: Energy and Metabolism*. Scharschmidt B, Kraemer FB, Hoffman A eds. PocketMedicine.com, Inc., 2002.

2. **Enns GM**. Homocystinuria. In *Pocket Medicine/Internal Medicine: Energy and Metabolism*. Scharschmidt B, Kraemer FB, Hoffman A eds. PocketMedicine.com, Inc., 2002.

3. **Enns GM**. Hyperoxaluria. In *Pocket Medicine/Internal Medicine: Energy and Metabolism*. Scharschmidt B, Kraemer FB, Hoffman A eds. PocketMedicine.com, Inc., 2002.

4. **Enns GM**. Mucopolysaccharidoses. In *Pocket Medicine/Internal Medicine: Energy and Metabolism*. Scharschmidt B, Kraemer FB, Hoffman A eds. PocketMedicine.com, Inc., 2002.

5. **Enns GM**. Purine and pyrimidine metabolic disorders. In *Pocket Medicine/Internal Medicine: Energy and Metabolism*. Scharschmidt B, Kraemer FB, Hoffman A eds. PocketMedicine.com, Inc., 2002.

G.**Invited presentations (238):**

1. Grand Rounds, Children's Hospital of Los Angeles. "Case presentations." 12/16/94.

2. Grand Rounds, Children's Hospital of Los Angeles. "Case presentations." 2/24/95.

3. Medical Genetics Journal Club, University of California, San Francisco. "Velocardiofacial syndrome." 12/15/95.

4. School of Genetic Counseling, University of California, Berkeley. "Mucopolysaccharidoses and peroxisomal disorders." 4/25/96.

5. Medical Genetics Journal Club, University of California, San Francisco. "Adenoviral gene therapy in tyrosinemia." 6/20/96.

6. Pediatric Resident Conference, University of California, San Francisco. "Arginase deficiency." 2/25/97.

7. Monterey County Department of Public Health, Community Genetics Teaching Conference. "Genetics and autism." 3/8/97.

8. School of Genetic Counseling, University of California, Berkeley. "An introduction to the principles of gene therapy for inborn errors of metabolism." 3/20/97.

9. Department of Optometry, University of California, Berkeley. "Systemic manifestations of congenital developmental disorders." 4/11/97.

10. School of Genetic Counseling, University of California, Berkeley. "Mitochondrial disease." 4/17/97.

11. Pediatric Resident Conference, University of California, San Francisco. "Long-chain 3-hydroxyacylCoA dehydrogenase deficiency." 4/22/97.

12. School of Genetic Counseling, University of California, Berkeley. "An introduction to inborn errors of metabolism." 9/18/97.

13. Pediatric Genetics Conference, Lucile Packard Children's Hospital. "Genotype-phenotype correlations in phenylketonuria." 12/12/97.

14. School of Optometry, University of California, Berkeley. "Systemic manifestations of congenital developmental disorders." 2/13/98

15. Department of Pediatrics, Pediatric Genetics Conference, Oregon Health Sciences University. "Genotype-phenotype correlations in phenylketonuria." 8/11/98.

16. Grand Rounds, Department of Pediatrics, University of California, San Francisco. "Gene therapy for inborn errors of metabolism." 12/17/98.

17. Genetics Board Review Course, University of California, San Francisco. "Approach to the diagnosis of inborn errors of metabolism." 2/10/99.

18. Genetics Board Review Course, University of California, San Francisco. "Mitochondrial disorders." 2/17/99.

19. Genetics Board Review Course, University of California, San Francisco. "Fatty acid oxidation disorders." 2/17/99.

20. Genetics Board Review Course, Stanford University. "Approach to the diagnosis of inborn errors of metabolism." 2/19/99.

21. Human Genetics Journal Club, Stanford University. "Treatments in X-linked Adrenoleukodystrophy." 3/9/99

22. Pediatric Noon Conference, Lucile Packard Children's Hosptial. "Primer of inborn errors of metabolism." 5/13/99.

23. Department of Pediatrics, Pediatric Genetics Conference, Lucile Packard Children's Hospital. "Clinical and biochemical diagnosis of mitochondrial disorders." 7/16/99

24. Pediatric Quality of Patient Care Conference, Lucile Packard Children's Hospital. "Metabolic evaluation of acute liver failure." 8/3/99

25. University of California San Francisco Liver Center Annual Meeting. "Treatment of hepatic inborn errors of metabolism with adeno-associated viral (AAV) vectors." 10/2/99

26. Pediatric Quality of Patient Care Conference, Lucile Packard Children's Hospital. "Approach to neonatal hyperammonemia." 10/5/99

27. Pediatric CME Program Lecture, El Camino Hospital, Mountain View, CA. "New topics in inborn errors of metabolism." 11/22/99

28. Grand Rounds, Department of Pediatrics, Lucile Packard Children's Hospital. "Clinical, biochemical, and molecular features of mitochondrial diseases in children." 12/10/99

29. Human Genetics Journal Club, Stanford University. "Do mitochondrial mutations dim the fire of life?" 1/25/2000

30. Grand Rounds, Department of Dermatology, Stanford University. "Metabolic causes of angiokeratomata." 2/8/2000

31. Grand Rounds, Department of Neurology, University of California, San Francisco. "Muscle fiber-type disproportion associated with mitochondrial respiratory chain disorders." 4/19/2000

32. Pediatric Noon Conference, Department of Pediatrics, Lucile Packard Children's Hospital. "An approach to the diagnosis of inborn errors of metabolism." 5/17/2000

33. Northern California Clinical Genetics Exchange, Spring Meeting, Stanford University. "Mitochondrial DNA depletion syndrome." 5/18/2000

34. Pediatric Intensive Care Unit Conference, Department of Pediatrics, Lucile Packard Children's Hospital. "Fatty acid oxidation defects and the acute management of inborn errors of metabolism." 5/25/2000

35. Medical Genetics Grand Rounds, Department of Pediatrics, Lucile Packard Children's Hospital. "Carbohydrate deficient glycoprotein syndrome." 5/26/2000

36. Pediatric Endocrinology Division Meeting, Lucile Packard Children's Hospital. "The biochemical genetics of hypoglycemia in infancy and childhood." 10/31/2000

37. Human Genetics Journal Club, Stanford University. "Gene therapy for mitochondrial disorders." 10/31/2000

38. American Academy for the Advancement of Science Annual Meeting Symposium: Screening for Inborn diseases – Trouble in the Postgenome Days, San Francisco, CA. "Screening for metabolic and genetic disorders: Today and the future." 2/17/2001

39. Grand Rounds, Department of Pediatrics, Lucile Salter Packard Children's Hospital. "The future of newborn screening in California." 2/23/2001

40. School of Optometry, University of California, Berkeley. "Systemic manifestations of congenital developmental disorders." 2/23/2001

41. Grand Rounds, Mad River Community Hospital, Arcata, California. Humbolt-Del Norte Consortium Continuing Medical Education Program. "Screening for metabolic and genetic disorders: Today and the future." 3/15/2001

42. Grand Rounds, St. Joseph Hospital, Eureka, California. Humbolt-Del Norte Consortium Continuing Medical Education Program. "Screening for metabolic and genetic disorders: Today and the future." 3/15/2001

43. Medical Genetics Grand Rounds, Stanford University School of Medicine. "Inborn errors of metabolism with features of hypoxic-ischemic encephalopathy." 4/13/2001

44. Northern California Clinical Genetics Exchange, Spring Meeting, Valley Children's Hospital, Madera, CA. "Clinical and molecular features in North American congenital disorders of glycosylation type I patients with diverse ethnic origins." 5/4/2001

45. Pediatric Noon Conference, Department of Pediatrics, Lucile Packard Children's Hospital. "An approach to the diagnosis of inborn errors of metabolism." 5/9/2001

46. Technological Advances Symposium, 34th Annual Advances and Controversies in Clinical Pediatrics, UCSF School of Medicine, San Francisco, CA. "Tandem mass spectrometry: Should it supplement current newborn testing?" 5/12/2001

47. Neonatology Morbidity and Mortality Conference, O'Connor Hospital, San Jose, CA. "The treatment of a neonate with methylmalonic acidemia." 6/8/2001

48. Ninth Annual Pediatric Update Seminar, Lucile Packard Children's Hospital, Stanford, CA. "What's new in newborn screening?" 7/20/2001

49. Neonatology Morbidity and Mortality Conference, O'Connor Hospital, San Jose, CA. "The evaluation of a neonate with a suspected inborn error of metabolism." 9/7/2001

50. Neonatology Board Review Course, Lucile Packard Children's Hosptial, Stanford, CA. "Inborn errors of metabolism in the neonate." 9/7/2001

51. Jewish Community Federation Maimonides Society Symposium, Palo Alto, CA. "Jewish genetics in the 21st century." 11/8/2001

52. Pediatric Quality of Patient Care Conference, Lucile Packard Children's Hospital. "Reye syndrome." 11/13/2001

53. Genetics: Impact on the Future Symposium, Center for Education and Professional Development, Division of Patient Care Services, Lucile Packard Children's Hospital. "Population screeing: New techniques and new diseases." 11/14/2001

54. Human Genetics Journal Club, Stanford University. "A tale of 2 genomes: Nuclear mutations and the mitochondrial dNTP pool in the mtDNA depletion syndrome." 01/08/2002

55. Medical Genetics Grand Rounds, Stanford University School of Medicine. "Genetics of autism." 2/15/2002

56. Neonatology Conference, Lucile Packard Children's Hospital. "Newborn screening by tandem mass spectrometry." 2/25/2002

57. Pediatric Residents Noon Conference, Lucile Packard Children's Hospital. "Care of the patient with an inborn error of metabolism in the Emergency Room." 2/27/2002

58. Pediatric CME Program Lecture, El Camino Hospital. "What's new in newbron screening?" 3/18/2002

59. Neurology Residents Conference, Stanford University Hospital. "Neurometabolic testing in children." 3/27/2002

60. Northern California Clinical Genetics Exchange, Spring Meeting, Stanford University School of Medicine. "Postpartum psychosis in mild late-onset argininosuccinic acid synthetase deficiency." 5/3/2002

61. Pediatric Residents Noon Conference, Lucile Packard Children's Hospital. "A diagnostic approach to the evaluation of inborn errors of metabolism." 5/15/2002

62. Medical Genetics Board Review Course, University of California San Francisco. "Clinical, biochemical, and molecular features of mitochondrial disorders." 5/20/2002

63. Department of Emergency Medicine Meeting, Stanford University. "Inborn errors of metabolism in the emergency room." 5/22/2002

64. California State Newborn Screening Conference, University of California Extension Building, Berkeley. "A review of inborn errors of metabolism: aminoacidemias, fatty acid oxidation defects, and organic acidemias." 5/22/2002

65. Department of Emergency Medicine In-service, Stanford University. "Inborn errors of metabolism in the emergency room." 6/13/2002

66. Department of Pediatrics, Gastroenterology Division Meeting, Stanford University. "Metabolic liver disease." 10/11/2002

67. Genzyme Corporation, Framingham, MA. "Congenital disorders of glycosylation: potential therapies." 10/21/2002

68. Advanced Postgraduate Program in Clinical Investigation Seminar, Department of Biochemistry and Molecular Biology, University of Florida, Gainesville, FL. "New advances in the diagnosis and therapy of mitochondrial disorders." 11/14/2002

69. Human Genetics Journal Club, Stanford University. "New diagnostic techniques for mitochondrial disorders." 01/07/2003

70. Medical Genetics Grand Rounds, Stanford University School of Medicine. "Congenital muscular dystrophies." 1/10/2003

71. School of Optometry, University of California, Berkeley. "Systemic manifestations of congenital developmental disorders." 2/21/2003

72. Palo Alto Medical Foundation, Palo Alto, CA. "Newborn screening by tandem-mass (MS/MS) spectrometry." 3/25/2003

73. Advances in the Management of MPS I Disease: Setting New Standards of Care with Enzyme Replacement Therapy Training Meeting, San Francisco, CA. "Lysosomal storage diseases and enzyme replacement therapy." 5/3/2003

74. Pediatric Academic Societies Topic Symposium, Washington State Convention and Trade Center, Seattle, WA. "The contribution of mitochondrial genetics to common complex disorders." 5/6/2003

75. Pediatric Residents Noon Conference, Lucile Packard Children's Hospital. "A review of inborn errors of metabolism." 5/29/2003

76. Northern California Clinical Genetics Exchange, Spring Meeting, Stanford University School of Medicine. "Congenital disorder of glycosylation type Ib: the 2nd North American patient." 6/6/2003

77. University of Hawaii, Kapi'olani Medical Center for Women and Children, Honolulu, HI. "Metabolic emergencies." 6/27/2003

78. Symposia Medicus, 16th Advances in Perinatal & Pediatric Nutrition, Stanford, CA. "New therapies and controversies in galactosemia and phenylketonuria." 7/14/2003

79. Morning Report, Lucile Packard Children's Hospital, Stanford, CA. "Metabolic emergencies." 7/15/2003

80. Department of Pediatrics, Gastroenterology Division Meeting, Stanford University. "Metabolic liver disease." 10/10/2003

81. Department of Pediatrics, Neonatology Division Meeting, Stanford University, "Metabolic catastrophes in the nursery: what's a neonatologist to do?" 10/17/2003

82. American Academy of Pediatrics 2003 National Conference, New Orleans, LA. "Plenary Session: Enzyme Replacement Therapy for Genetic Diseases: A Treatment Whose Time Has Come?" 11/01/2003

83. American Academy of Pediatrics 2003 National Conference, New Orleans, LA. "Audience Response Case Discussion: Genetic Unknowns." 11/01/2003

84. American Academy of Pediatrics 2003 National Conference, New Orleans, LA. "Selected Short Subject: Metabolic Catastrophes in the Nursery: What's a Pediatrician to Do?" 11/02/2003

85. Stanford University School of Medicine, Stanford, CA. "Practice of Medicine: Ornithine transcarbamylase deficiency." 11/24/2003

86. Human Genetics Journal Club, Stanford University. "Is cancer a mitochondrial disorder?" 11/25/2003

87. Department of Emergency Medicine, Stanford University. "Genetic emergencies." 11/26/2003

88. Grand Rounds, Department of Pediatrics, Lucile Packard Children's Hospital. "Enzyme replacement therapy for inborn errors of metabolism." 12/05/2003

89. Medical Genetics Group Conference, Division of Medical Genetics, University of California San Francisco, CA. "Proteomics in the study of mitochondriopathies." 12/05/2003

90. Mid-Coastal California Perinatal Outreach Program 24th Annual Meeting, Perinatal Potpourri: Reaching Out and Looking Forward, Monterey, CA. "Demystifying neonatal metabolic disease." 01/08/2004

91. Mid-Coastal California Perinatal Outreach Program 24th Annual Meeting, Perinatal Potpourri: Reaching Out and Looking Forward, Monterey, CA. "Metabolic disease *v.* hypoxic-ischemic encephalopathy: making the diagnosis." 01/08/2004

92. Seminar, Children's Hospital Oakland Research Institute, Oakland, CA. "New therapies and diagnostics for mitochondrial disorders." 2/17/2004

93. School of Optometry, University of California, Berkeley. "Systemic manifestations of congenital developmental disorders." 2/20/2004

94. Seminar, Late Treated PKU Program, Agnews Developmental Center, San Jose, CA. "Variations of PKU and mutation analysis." 2/25/2004

95. Department of Pediatrics, Neonatology Division Meeting, Stanford University, "Metabolic disease *v.* hypoxic-ischemic encephalopathy: making the diagnosis." 2/27/2004

96. California Policymakers' Symposium on Issues in Newborn Genetic Screening, State Capitol, Sacramento, California. "Introduction to newborn screening." 3/11/2004

97. Pacific Southwest Regional Genetics Network Meeting, Lucile Packard Children's Hospital, Stanford, CA. "New therapies and diagnositics for mitochondrial disorders." 3/20/2004

98. General Pediatrics Board Review, Lucile Packard Children's Hospital, Stanford, CA. "An approach to the evaluation of a child with a suspected inborn error of metabolism." 5/5/2004

99. Medical Genetics Grand Rounds, Stanford University, Stanford, CA. "Metabolic disease *v.* hypoxic–ischemic encephalopathy: Making the diagnosis." 5/7/2004

100. Pediatric Noon Conference, Department of Pediatrics, Lucile Packard Children's Hospital. "Metabolic emergencies." 7/22/2004

101. University of Hawaii, Kapi'olani Medical Center for Women and Children, Honolulu, HI. "Mitochodrial disease." 8/11/2003

102. PREP The Course: An Intensive Review of Pediatrics, American Academy of Pediatrics, Costa Mesa, CA. "Genetics/Dysmorphology Review." 9/14/2004

103. PREP The Course: An Intensive Review of Pediatrics, American Academy of Pediatrics, Costa Mesa, CA. "Genetics/Dysmorphology Cases." 9/14/2004, 9/15/2004

104. American College of Healthcare Executives Conference: Keeping Pace with the Times - The Emergence of Public Health Preparedness and Genomics, University of Berkeley School of Public Health, Berkeley, CA. "Impact of genomics on clinical practice." 9/27/2004

105. Grand Rounds, Department of Pediatrics, Lucile Packard Children's Hospital. "Expanded newborn screening in California: what pediatricians should know." 10/01/2004

106. American Academy of Pediatrics, 2004 National Conference and Exhibition, San Francisco, CA. "Genetic unknowns." 10/9/2004

107. American Academy of Pediatrics Satellite Symposium: Early Diagnosis of Developmental Disabilities – Case Studies in Autism, ADHD, and Lysosomal Storage

Disorders, San Francisco, CA. "Diagnosing and treating mucopolysaccharidosis I (MPS I)." 10/9/2004

108. PKU Parents Conference, Roseville, CA. "Expanded newborn screening in California." 10/23/2004

109. Human Genetics Journal Club, Stanford University. "Homoplasmic mitochondrial DNA mutations in disease: syndrome X and beyond." 11/23/2004

110. Pediatric Rounds, Community Hospital of Monterey Peninsula, Monterey, CA. "Expanded screening by tandem mass spectrometry: what pediatricians need to know." 1/4/2005

111. Medical Genetics Grand Rounds, Stanford University. "L-γ-Glutamyl-L-cysteinyl-glycine (glutathione)." 1/14/2005

112. School of Optometry, University of California, Berkeley. "Systemic manifestations of congenital developmental disorders." 2/11/2005

113. California Association of Neonatologists and AAP District IX Section on Perinatal Pediatrics 11th Annual Conference, Current Topics and Controversies in Perinatal and Neonatal Medicine, Coronado, CA. "Metabolic disease for the neonatologist." 3/5/2005

114. University of California San Francisco School of Medicine 4th Annual Developmental Disabilities Conference, An Update for Health Professionals, San Francisco, CA. "Expanded newborn screening for metabolic disorders is coming: what primary care providers need to know." 3/11/2005

115. State of California Department of Health Services, Tandem Mass Spectrometry Training Course, Richmond, CA. "Organic acidemias." 3/16/2005

116. Seminar, Current Concepts and Dilemmas in Genetic Testing, Stanford University School of Medicine, Stanford, CA. "Tandem mass spectrometry expanded newborn screening." 4/13/2005

117. Perinatal Morbidity and Mortality Conference, El Camino Hospital, Mountain View, CA. "Tandem mass spectrometry expanded newborn screening." 4/15/2005

118. Pediatric Noon Conference, Department of Pediatrics, Lucile Packard Children's Hospital. "Metabolic Unknowns." 5/26/2005

119. Department of Developmental Services, Phenylketonuria Seminar, Regional Center of the East Bay, Oakland, CA. "Newborn screening, Diagnosis, Variations of PKU, Mutation Analysis, Early Detection: Where have we gone from there." 6/3/2005

120. Seminar, Far Northern Regional Center, Eureka, CA. "Expanded newborn screening for metabolic disorders is here: what primary care providers need to know." 8/11/2005

121. Seminar, Biomarin Pharmaceuticals, Inc., Novato, CA. "The mucopolysaccharidoses: an overview." 8/16/2005

122. Neurology Grand Rounds, Department of Neurology, Stanford University, CA. "Contemporary diagnosis of mitochondrial diseases." 9/9/05

123. NICU CME Lecture Series, Salinas Valley Memorial Hospital, Salinas, CA. "Expanded newborn screening for metabolic disorders is here: what primary care providers need to know." 10/7/05

124. Human Genetics Journal Club, Stanford University. "Alternative approaches to the derivation of human embryonic stem cells." 11/22/2005

125. Grand Rounds, California Pacific Medical Center, San Francisco, CA. "Expanded newborn screening for metabolic disorders is here: what primary care providers need to know." 12/9/05

126. Medical Genetics Grand Rounds, Stanford University. "Pompe disease." 1/6/06

127. 6th Annual March of Dimes Health Professional Conference, Long Beach, CA. "Expanded newborn screening for metabolic disorders is here: the California experience." 1/20/06

128. School of Optometry, University of California, Berkeley. "Systemic manifestations of congenital developmental disorders." 2/17/2006

129. Seminar, Department of Gastroenterology, Stanford University. "Metabolic disease and liver transplantation." 3/31/06

130. UP 1204-003 Investigator Meeting, San Francisco, CA. "Introduction to urea cycle disorders." 4/27/06

131. UCSF/Stanford Lysosomal Disease Center Meeting – Hematological Manifestations of Gaucher Disease, San Francisco, CA. "Gaucher disease." 4/28/06

132. Hawaii Community Genetics, Honolulu, HI. "Disorders of carnitine transport." 6/5/06

133. Hawaii Community Genetics, Honolulu, HI. "Oxidative metabolism." 6/606

134. Bay Area CCS Medical Consultants Meeting, California Department of Health Services, Oakland, CA. "Mitochondrial disease." 6/21/06

135. Medicine Grand Rounds, Fairchild Auditorium, Stanford University, CA. "Metabolism happens: Adult presentations of genetic diseases." 7/27/06

136. Medical Genetics Grand Rounds, B200, Stanford University, CA. "Metabolic emergencies in the NICU." 8/11/06

137. NICU Seminar, LPCH, Stanford, CA. "Urea cycle disorders." 8/11/06

138. Seminar, Far Northern Regional Center, Eureka, CA. "An overview of metabolic emergencies." 8/24/2006

139. Grand Rounds, Sutter Medical Center, Santa Rosa, CA. "What every primary care provider needs to know about the new metabolic screen." 9/15/06

140. Grand Rounds, Department of Pediatrics, Lucile Packard Children's Hospital. "Enzyme replacement therapy for lysosomal disorders: a treatment whose time has come? 10/6/06

141. Seminar, Children's Hospital Oakland, Oakland, CA. "Mitochondrial disease and redox imbalance: implications for therapy." 11/10/06

142. Symposium – Genetic disorders in the Ashkenazi Jewish population, California State University Northridge, USU San Fernando Valley Hall, Northridge, CA. "Founder effect in Ashkenazim and diagnosis, management, and treatment of Ashkenazi Jewish disorders. 12/9/06

143. School of Optometry, University of California, Berkeley. "Systemic manifestations of congenital developmental disorders." 2/9/2007

144. Seminar, Department of Neurology, Stanford University, H3150. "Metabolic disorders of childhood." 2/28/07

145. Society for Inherited Metabolic Disorders Annual Meeting, Nashville, TN. "Survival of patients with urea cycle disorders following treatment of hyperammonemia with intravenous sodium phenylacetate and sodium benzoate." 3/28/07

146. Pediatric Grand Rounds – Aaron Michael Graham Lectureship, Childrens Hospital Los Angeles, Los Angeles, CA. "Enzyme replacement therapy for metabolic disorders – a therapy whose time has come?" 5/11/07

147. UCSF 40th Annual Advances and Controversies in Clinical Pediatrics, San Francisco, CA. "Expanded newborn screening: early results, surprises, and the future." 5/31/07

148. Seminar, Methylmalonic Acidemia Meeting, National Institutes of Health, Bethesda, MD. "Combined liver–kidney transplantation in methylmalonic acidemia." 6/18/07

149. Seminar, "Glutathione – a biomarker of redox imbalance." Edison Pharmaceutical, San Jose, CA. 10/11/07

150. Seminar, Hyperion Therapeutics, San Francisco, CA. "Urea cycle disorders – an overview." 10/15/07

151. Seminar, Hyperion Therapeutics San Francisco, CA. "Urea cycle disorders – diagnosis and laboratory evaluation." 10/16/07

152. Seminar, Hyperion Therapeutics San Francisco, CA. "Urea cycle disorders – therapy and outcome." 10/16/07

153. American Academy of Pediatrics, 2007 National Conference and Exhibition, San Francisco, CA. "Genetic unknowns." 10/28/2007

154. American Academy of Pediatrics, 2007 National Conference and Exhibition, San Francisco, CA. "Testing for inborn errors of metabolism: who to test, what tests to send, and how to interpret the results." 10/28/2007

155. Human Genetics Journal Club, Department of Genetics, Stanford University, Stanford, CA. "Break on through to the other side – therapeutics that cross the blood-brain barrier." 12/11/07

156. Medical Genetics Grand Rounds, Department of Pediatrics, Division of Medical Genetics, Stanford University, CA. "Lysosomal disorders – pain." 1/4/08

157. Pediatric Grand Rounds, California Pacific Medical Center, San Francisco, CA. "Testing for inborn errors of metabolism: who to test, what tests to send, and how to interpret the results." 1/11/08

158. School of Optometry, University of California, Berkeley. "Systemic manifestations of congenital developmental disorders." 2/8/2008

159. Kuvan Regional Advisory Board Meeting, San Diego, CA. "Kuvan efficacy and safety data." 2/21/08

160. Kuvan Regional Advisory Board Meeting, San Diego, CA. "Initiation of Kuvan therapy and patient management." 2/21/08

161. Symposium, American College of Medical Genetics Annual Meeting, Phoenix, AZ. "Urea cycle disorders: Best practices and new developments." 3/13/08

162. Symposium, Genetic Metabolic Dietician International Conference, Atlanta, GA. "Medical criteria for transplantation in organic acidemias and urea cycle disorders." 4/26/04

163. Seminar, Department of Neurology, Stanford University, H3150. "Initial approach to the child with a suspected metabolic disorder." 4/30/08

164. Seminar, Department of Genetics, MIND Institute, Sacramento, CA. "Novel therapies for phenylketonuria." 7/3/08

165. North American Metabolic Academy, Warrenton, VA. "Mitochondrial disorders." 9/23/08

166. North American Metabolic Academy, Warrenton, VA. "Metal disorders." 9/24/08

167. North American Metabolic Academy, Warrenton, VA. "Neurological manifestations of lysosomal storage disorders." 9/25/08

168. North American Metabolic Academy, Warrenton, VA. "Pain in lysosomal storage disorders." 9/25/08

169. Patient Town Hall Meeting, San Francisco, CA. "Overview and discussion of Kuvan." 10/11/08

170. Grand Rounds, University of California, Davis, CA. "Fabry disease in the pediatric patient." 11/14/08

171. Human Genetics Journal Club, Department of Genetics, Stanford University, Stanford, CA. "Lysine-tryptophan degradation in the Amish: a glimpse into a mechanism of brain degeneration." 11/18/08

172. Gastroenterology Noon Conference, Department of Pediatrics, Stanford University, Stanford, CA. "Liver inborn errors of metabolism: cholestasis." 12/5/08

173. NICU Seminar, LPCH, Stanford, CA. "Urea cycle disorders: best practices and new developments." 1/30/09

174. University of California San Francisco School of Medicine 8th Annual Developmental Disabilities Conference, San Francisco, CA. "Autism, metabolic diseases, and immunizations." 3/6/2009

175. Children's Hospital of Orange County's 3rd Annual Symposium, Stem Cell Therapies for Pediatric Diseases and Injuries, Orange County, CA. "Clinical trial of hNSCs in lysosomal storage disorders." 3/12/09

176. Medical Genetics Grand Rounds, Division of Medical Genetics, Stanford University, Stanford, CA. "Neurocognitive, behavioral and social outcomes with phenylketonuria (PKU)." 3/20/09

177. Platform Session, Mitochondrial Medicine 2009: Capitol Hill, United Mitochondrial Disease Foundation, Washington, DC. "Mitochondrial disease biomarkers." 6/27/09

178. International Symposium – Improving Neurological Outcomes in Urea Cycle Disorders, University College London, Institute of Child Health, London, UK. "Urea cycle disorders: current treatment in the US and potential improvements." 7/8/09

179. Medical Genetics Grand Rounds, Division of Medical Genetics, Stanford University, Stanford, CA. "An introduction to inborn errors of metabolism and newborn screening." 7/17/09

180. 3rd International Satellite on Urea Cycle Disorders, Overcoming Barriers: New Developments and Future Directions for Urea Cycle Disorders. La Jolla, CA. "Nitrogen sparing therapy revisited 2009." 8/29/09

181. Pediatric Grand Rounds, Children's Hospital Oakland, Oakland, CA. "Inborn errors of metabolism in children: who to test, what tests to send, and how to interpret the results." 9/15/09

182. Seminar, Hawaii Community Genetics, Honolulu, HI. "Nitrogen sparing therapy revisited 2009." 10/26/09

183. Seminar, Hawaii Community Genetics, Honolulu, HI. "Mitochondrial disease biomarkers." 10/27/09

184. Leadership Forum, Arrillaga Alumni Center, Stanford University, Stanford, CA. "Update on newborn screening." 11/6/09

185. Human Genetics Journal Club, Department of Genetics, Stanford University, Stanford, CA. "Parkinson disease: genetics meets the environment." 1/19/10

186. Metabolic University Satellite Symposium, Denver, CO. "Neurocognitive and behavioral outcomes in PKU." 2/5/10

187. 7th Annual Advanced Practice neonatal Nurses Conference, San Francisco, CA. "Neonatal metabolic disorders and newborn screening." 3/18/10

188. Lucile Salter Packard Society Holiday Tea, Allied Arts Guild, Menlo Park, CA. "Where there's biochemical smoke, there's fire – a mitochondrial approach to medicine in the 21st century." 12/7/10

189. Human Genetics Journal Club, Department of Genetics, Stanford University, Stanford, CA. "Mitochondrial sirtuins – novel therapeutic targets for redox-related disorders?" 1/4/11

190. Scientific Symposium – New Clinical Approaches to NAGS Deficiency: From Diagnosis to Treatment, Asilomar, CA. "Clinical overview of NAGS deficiency." 3/1/11

191. Immunology Journal Club, Department of Pediatrics, Stanford University, Stanford, CA. "Immune response and enzyme replacement therapy for lysosomal storage disorders." 4/21/11

192. Pediatric Grand Rounds, CHOC Children's Hospital, Orange County, CA. "Mitochondrial medicine in the 21st century – from bench to bedside." 5/11/11

193. Special Symposium Session, United Mitochondrial Disease Foundation Annual Meeting, Chicago, IL. "Preliminary report on initial subjects diagnosed with genetically confirmed mitochondrial disease at end-of-life treated with EPI-743 under FDA-approved expanded access protocol EPI-2009-1." 6/17/11

194. Visting Professor, Sanford Black Hills Pediatric Symposium, Deadwood, SD. "Where there's biochemical smoke, there's fire – a mitochondrial approach to medicine in the 21st century." 7/15/11

195. Visting Professor, Sanford Black Hills Pediatric Symposium, Deadwood, SD. "Urea cycle disorders: best practices and new developments." 7/15/11, 7/16/11

196. Seminar, California Department of Health Services, Genetic Disease Branch, Stanford, CA. "Aminoacidemias and organic acidemias." 7/19/11

197. Platform Presentation, VIIIth Latin American Congress of Inborn Errors of Metabolism and Neonatal Screening, Cusco, Peru. "Treatment alternatives in mitochondrial diseases." 9/21/11

198. Seminar, SRI International Biosciences Division, Menlo Park, CA. "Mitochondrial diseases: is treatment possible?" 11/11/11

199. Human Genetics Journal Club, Department of Genetics, Stanford University, Stanford, CA. "Exercise and eat dark chocolate – the Willy Wonka approach to improving mitochondrial function." 1/10/12

200. Grand Rounds, Children's Hospital Oakland, Oakland, CA. "Mitochondrial disease in the 21st century: is treatment possible?" 1/17/12

201. Grand Rounds, Lucile Packard Children's Hospital, Stanford, CA. "Lysosomal storage disorders – clinical clues and new therapies." 3/16/12

202. Seminar, Society for Inherited Metabolic Disorders 35th Annual Meeting, Charlotte, NC. "New therapies in mitochondrial disease." 4/2/12

203. Symposium, Genetic Metabolic Dietitians International Annual Meeting, New Orleans, LA. "Urea cycle disorders: diagnosis to transplant." 4/20/12

204. Featured Symposium: Pediatrics – Long Term Outcomes and Recurrent Disease, International Liver Transplantation Society 18th Annual International Congress, San Francisco, CA. "Inborn errors of metabolism – To transplant or not to transplant?" 5/19/12

205. Mitochondrial Medicine 2012: Capitol Hill, United Mitochondrial Disease Foundation Annual Meeting, Bethesda, MD. "Redox biomarkers in mitochondrial disease." 6/14/12

206. Human Genetics Journal Club, Department of Genetics, Stanford University, Stanford, CA. "Brain mitochondrial glutathione transport – a link to neurodegeneration." 1/16/13

207. Pediatric Critical Care Conference, Department of Pediatrics, Stanford University, Stanford, CA. "Metabolic emergencies." 2/19/13

208. Neuro-NICU Training Course, Department of Pediatrics, Stanford University, Stanford, CA. "Inborn errors of metabolism masquerading as hypoxic-ischemic encephalopathy." 2/21/13

209. Neurology Seminar, Department of Pediatrics, Stanford University, Stanford, CA. "Biochemical Genetics: neurological aspects and therapy – Part 1." 4/3/13

210. Neurology Seminar, Department of Pediatrics, Stanford University, Stanford, CA. "Biochemical Genetics: neurological aspects and therapy – Part 2." 4/10/13

211. Fourth Annual Pediatrics Research Retreat, Stanford University, Stanford, CA. "Mitochondrial disease biomarkers." 4/26/13

212. Mitochondrial Medicine 2013: United Mitochondrial Disease Foundation Annual Meeting, Newport Beach, CA. "Treatment of mitochondrial disease." 6/12/13

213. Mitochondrial Medicine Society Clinical Directors Workshop, Newport Beach, CA. "Planning clinical trials, choosing outcome measures and more." 6/13/13

214. Neurobiology of Disease in Children Symposium: Mitochondrial Disease, Session III – Translational Science and Clinical Frontiers, Child Neurology Society 42nd Annual Meeting, Austin, TX. "Clinical trials: Edison Pharmaceuticals, EPI-743." 10/30/13

215. Newborn Screening Meeeting, California Department of Public Health, Richmond, CA. "Newborn screening for mitochondrial disorders." 11/22/13

216. Human Genetics Journal Club, Department of Genetics, Stanford University, Stanford, CA. "mTOR: a new pathway for mitochondrial disease therapy." 1/8/14

217. Gastroenterology Seminar, Department of Pediatrics, Stanford University, Stanford, CA. "Inborn errors of metabolism and liver disease." 1/17/14

218. MitoAction Mitochondrial Disease Clinical Conference, Los Angeles, CA. "Current approaches to mitochondrial disease treatment." 2/8/14

219. Medical Genetics Grand Rounds, Department of Pediatrics, Stanford University, Stanford, CA. "Mitochondrial bioenergetics." 2/14/14

220. 5th Annual Rare Disease Day Symposium, Sanford-Burnham Medical Research Institute, San Diego, CA. "The clinical phenotype of *NGLY1* deficiency." 2/28/14

221. Annual Association of Auxiliaries for Children Meeting, Stanford, CA. "Mitochondria, energy and life – a bioenergetics approach to medicine." 3/3/14

222. CME/CNE Regional Roundtable Meeting Series, San Francisco, CA. "Inborn errors of metabolism: advances in the management of nitrogen in patients with urea cycle disorders." 6/3/14

223. International Symposium: Challenging Urea Cycle Disorder Treatment Targets for Optimal Neurological Outcomes – 5 Years On, What has Changes?, Stirling, Scotland. "Treating urea cycle disorders 5 years on – a view from California." 6/18/14

224. CME/CNE Regional Roundtable Meeting Series, Houston, TX. "Inborn errors of metabolism: advances in the management of nitrogen in patients with urea cycle disorders." 6/24/14

225. The Mitochondrial Conundrum: Novel Presentations and Treatments of Mitochondrial Diseases Conference, Bell Harbor International Conference Center, Seattle, WA. "New Therapeutics; EPI-743." 9/12/14

226. Nutritional Interventions in Primary Mitochondrial Disorders Workshop, NIH Neurosciences Center, Rockville, MD. "Mendelian mitochondrial disorders." 12/2/14

227. Seminar, Department of Anesthesia, Stanford, CA. "Anesthesia use in mitochondrial disorders." 1/22/15

228. Human Genetics Journal Club, Department of Genetics, Stanford University, Stanford, CA. "NGLY1 deficiency – new insights into the pathogenesis of a novel disorder of the endoplasmic reticulum-associated degradation (ERAD) pathway." 1/28/15

229. Seminars in Genetics, Kaiser-Permanente Northern California Regional Genetics Department 2015 Webinar Series. "Treatment of Mitochondrial Disorders." 2/12/15

230. Pediatric Critical Care Conference, Department of Pediatrics, Stanford, CA. "Metabolic emergencies." 3/30/15

231. Association of Health Care Journalists Annual Meeting, Santa Clara, CA. "A genetics primer for journalists." 4/23/15

232. United Mitochondrial Disease Foundation, Mitochondrial Medicine: 2015, Herndon, VA. "Mitochondrial disease – the basics." 6/19/15

233. United Mitochondrial Disease Foundation, Mitochondrial Medicine: 2015, Herndon, VA. "Trial-ready drugs: what is in the pipeline for mitochondrial diseases?" 6/20/15

234. Neurology Grand Rounds, Department of Neurology, Stanford University, Stanford, CA. "Update on diagnostic and therapeutic issues in mitochondrial diseases." 8/14/15

235. Medscape Education Seminar, San Francisco, CA. "Lysosomal acid lipase deficiency: presentation, diagnosis, and treatment." 11/15/15

236. Rare Diseases Clinical Research Network (RDCRN) Certificate Program webinar. "The role of industry in clinical research." 2/2/16

237. Johns Hopkins School of Medicine Gaucher Regional Meeting, San Francisco, CA. "Comprehensive care for patients with Gaucher disease: emerging concepts for improved outcomes." 7/28/16

238. Johns Hopkins School of Medicine Gaucher Regional Meeting, Los Angeles, CA. "Comprehensive care for patients with Gaucher disease: emerging concepts for improved outcomes." 8/4/16

H.**Scientific and Professional Meetings Attended:**

American Academy of Pediatrics, 1995, 2003 (3 presentations), 2004
American Academy of Pediatrics, PREP The Course, 2004
American College of Medical Genetics, 2000, 2008 (1 poster)
American Society of Gene Therapy, 1998
American Society of Human Genetics, 1995, 1996, 1997 (2 posters), 1998 (1 poster), 1999 (1 poster), 2000 (1 poster), 2002 (1 poster), 2003 (1 poster), 2005 (4 posters), 2006 (6 posters), 2007 (1 platform, 1 poster), 2009 (4 posters), 2010 (1 poster), 2014 (1 poster), 2015 (1 poster), 2016 (2 posters)
D.W. Smith Workshop on Malformations and Morphogenesis 1996 (1 platform, 2 posters)
International Congress on Inborn Errors of Metabolism, 2009 (1 platform, 3 posters)
Latin American Congress of Inborn Errors of Metabolism and Neonatal Screening, 2011 (1 platform)
Lysosomal Storage Disease Registries Meeting, 2005
The Mitochondrial Society, 1998
MS/MS Program Implementation Meeting, 2003
Mucopolysaccharidosis I Annual Meeting, 2005
Pediatric Academic Society, 1999 (1 poster), 2003 (1 presentation)
San Francisco Bay Area Lysosomal Disease Registry Working Group, 2009
Society of Inherited Metabolic Disorders, 1998 (2 posters), 1999, 2001 (1 poster), 2002 (1 poster), 2007 (2 platforms, 1 poster), 2008 (1 platforms), 2011 (2 posters), 2012 (1 platform, 1 poster), 2014 (1 platform, 5 posters)
Western Society for Pediatric Research, 1997 (platform, 1 poster), 1998 (Platform presentation, 1 poster), 1999, 2000, 2001 (platform), 2002 (2 platforms), 2003 (platform), 2004 (3 platforms), 2005 (1 platform), 2008 (1 platform), 2010 (4 platforms)
UCSF-Stanford Lysosomal Disease Center/Fabry Support and Information Group, 1/18/05
UCSF-Stanford Lysosomal Disease Center/National Gaucher Foundation, 1/18/05
United Mitochondrial Disease Foundation, 2000 (1 poster), 2005 (1 platform), 2008 (1 platform), 2009 (1 platform, 1 poster), 2010, 2011 (1 platform, 1 poster), 2012 (1 platform, 1 poster), 2013 (1 platform, 5 posters), 2015 (2 platform, 1 poster)

I. Scientific and Professional Sessions Organized/Moderated (25):

1. Western Society for Pediatric Research, Genetics Session, Carpenter Hall, Sunset Center, Carmel, CA. 1/31/2003
2. Pediatric Academic Societies' Annual Meeting, Genetics/Inborn Errors of Metabolism Session, Washington State Convention Center, Room 4C2, Seattle, WA. 5/5/2003
3. American Society of Human Genetics Annual Meeting, Session 43. Inborn Errors of Metabolism, and Biochemical Genetics II, Los Angeles Convention Center, Los Angeles, CA. 11/7/2003
4. Bay Area Mitochondrial Association, Stanford University, Stanford, CA. 2/23/2005
5. 7th Annual Northern California Genetics Exchange, Stanford University, Stanford,

CA. 6/2/2006

6. Western Society for Pediatric Research, Genetics Session, Rehearsal Room, Sunset Center, Carmel, CA. 2/2/2007
7. Kuvan Regional Advisory Board Meeting, San Diego, CA. 2/21/08
8. Ask the Experts!, Session 7, Society for Inherited Metabolic Disorders Annual Meeting, Asilomar, CA. 3/4/08
9. Kuvan Regional Advisory Board Meeting, Baltimore, MD. 6/5/09
10. Mitochondrial Medicine 2009: Capitol Hill, Washington, DC. 6/24/09–6/27/09
11. 11th International Congress of Inborn Errors of Metabolism, Society for the Study of Inborn Errors of Metabolism, San Diego, CA. 8/29/09–9/02/09
12. Session 42: Therapies for Genetic Disorders: 59th Annual Meeting, American Society of Human Genetics, Honolulu, HI. 10/20/09–10/24/09
13. Kuvan Regional Advisory Board Meeting, Chicago, IL. 11/13/09
14. Mitochondrial Medicine 2011: Chicago, IL. 6/15/11–6/17/11
15. Session 23: Therapy for Genetic Disorders: 61st Annual Meeting, American Society of Human Genetics, Montreal, Canada. 10/11/11–10/15/11
16. 2011 Newborn Screening & Genetic Testing Symposium, Association of Public Health Laboratories, San Diego, CA. 11/7/11–11/10/11
17. Session 23: Therapy for Genetic Disorders: 12th International Congress on Human Genetics (ICHG)/61st American Society of Human Genetics Annual Meeting, Montreal, Canada. 10/11/11–10/15/11
18. Co-Chair of Research Strategies session: Nutrition and Dietary Supplement Interventions for Inborn Errors of Metabolism Stakeholder Workshop, NIH, Rockville, MD. 12/6/11–12/7/11
19. Co-Chair of Session 4: Product Development Activities of the Industry – Lessons Learned, Translational Research in Primary Mitochondrial Diseases: Challenges and Opportunities Workshop, NHGRI Conference Area, Rockville, MD. 3/8/12–3/9/12
20. Program Chair, United Mitochondrial Disease Foundation Annual Meeting: Mitochondrial Therapeutics – From Bench to Bedside, Newport Beach, CA. 6/12/13–6/15/13
21. Session Co-Director and Moderator Neurobiology of Disease in Children Symposium: Mitochondrial Disease, Session III – Translational Science and Clinical Frontiers, Child Neurology Society 42nd Annual Meeting, Austin, TX. 10/30/13
22. Planning Committee, MitoAction Mitochondrial Disease Clinical Conference, Los Angeles, CA. 2/8/14
23. Course Director, Inborn Errors of Metabolism: Advances in the Management of Nitrogen in Patients with Urea Cycle Disorders – CME/CE Activity, Palo Alto, CA. 2/18/14
24. Rare Disease Symposium, ERAD defects in the cytoplasm: the *NGLY1* story, Sanford-Burnham Medical Research Institute, San Diego, CA. 2/28/14
25. Planning Committee, NIH Office of Dietary Supplements, "Nutritional Inverventions in Primary Mitochondrial Disorders: Developing an Evidence Base" Workshop. NIH Neuroscience Center, Rockville, MD. Moderator Session 4: "Challenges and Barriers to Dietary Supplement Use in PMD." 12/2/14–12/3/14

J. Other teaching:

1. Teaching Attending, Lucile Salter Packard Children's Hospital, Ward 3W. 11/6/00–12/3/00

2. Lecturer, Medical Biochemistry 204, Stanford University School of Medicine. 1/23/01

3. Lecturer, Medical Biochemistry 204, Stanford University School of Medicine. 2/16/01

4. Lecturer, Medical Biochemistry 204, Stanford University School of Medicine. 4/26/01

5. Qualifying Exam Committee, Genetics Ph.D. Program, Stanford University School of Medicine. Joylette L. Portlock 2/11/02

6. Lecturer, Genetics 201, Stanford University School of Medicine. 2/21/02

7. Teaching Attending, Lucile Salter Packard Children's Hospital, General Pediatrics Wards. 3/26/02–4/19/02

8. Lecturer, Genetics 201, Stanford University School of Medicine. 4/22/02, 4/25/02

9. American College of Medical Genetics Exhibit Vendor, The Endocrine Society's 84th Annual Meeting, San Francisco, CA. 6/19–6/21/02

10. American Academy of Pediatrics, Pediatric UPDATE program. "Newborn screening." Boston, MA. 10/20/02

11. Lecturer, Medical Biochemistry 204, Stanford University School of Medicine. 1/27/03

12. Teaching Attending, Lucile Salter Packard Children's Hospital, General Pediatrics Wards. 2/3/03–2/28/03

13. Lecturer, Medical Biochemistry 202, Stanford University School of Medicine. 2/14/03, 2/21/03

14. Teaching Attending, Lucile Salter Packard Children's Hospital, General Pediatrics Wards. 2/3/03–2/28/03

15. Lecturer, Genetics 201, Stanford University School of Medicine. 4/21/03, 4/24/03

16. Newborn Screening Area Service Center Training Course. 5–6/03.

17. Lecturer, Genetics 202, Stanford University School of Medicine. 10/21/03, 10/23/03.

18. Lecturer, Practice of Medicine, Clinical Correlations, Stanford University School of Medicine. 11/24/03

19. Teaching Attending, Lucile Salter Packard Children's Hospital, General Pediatrics Wards. 2/3/04–3/2/04

20. Faculty Mentor, Practice of Medicine, Stanford University School of Medicine. 3/29/04

21. Lecturer, Medical Biochemistry 202, Stanford University School of Medicine. 10/26/04, 10/28/04

22. Teaching Attending, Lucile Salter Packard Children's Hospital, General Pediatrics Wards. 2/1/05–2/24/05

23. Lecturer, Genetics 238, Stanford University School of Medicine. 4/13/05

24. Lecturer, Genetics 202, Stanford University School of Medicine. 11/1/05, 11/3/05

25. Lecturer, Genetics 202, Stanford University School of Medicine. 10/31/06, 11/2/06

26. Lecturer, Genetics 202, Stanford University School of Medicine. 10/30/07, 11/1/07

27. Faculty, North American Metabolic Academy. 9/21/08–9/26/08

28. Lecturer, Genetics 202, Stanford University School of Medicine. 11/04/08, 11/18/08

29. Lecturer, Genetics 285b, Stanford University Genetic Counseling Program. 2/5/09

30. Lecturer, Genetics 202, Stanford University School of Medicine. 11/10/09, 11/12/09

31. Lecturer, Genetics 202, Stanford University School of Medicine. 10/19/10, 10/28/10

32. Lecturer, Genetics 202, Stanford University School of Medicine. 10/25/11, 10/27/11

33. Lecturer, Pediatric Board Review, Lucile Packard Children's Hospital. 5/10/12

34. Lecturer, Genetics 202, Stanford University School of Medicine. 10/23/12, 10/25/12

35. Lecturer, Genetics 202, Stanford University School of Medicine. 11/5/13, 11/7/13

36. Lecturer, Genetics 202, Stanford University School of Medicine. 11/7/14, 11/11/14, 11/13/14

37. Lecturer, Genetics 280, Stanford University School of Medicine. 1/8/15

38. Lecturer, Pediatric Board Review, Lucile Packard Children's Hospital. 9/28/15

39. Lecturer, Genetics 202, Stanford University School of Medicine. 10/22/15, 10/26/15, 10/27/15