**Curriculum Vitae** September 2019

**Ellen Roy Elias, M.D., FAAP, FACMG**

**Current Position:**

Professor with Tenure, Departments of Pediatrics and Genetics

Director, Special Care Clinic for Primary and Consultative Services

Children’s Hospital Colorado

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**Personal Information:**

Husband: Anthony D. Elias, MD –

Martha Cannon Dear Professor of Medicine

University of CO School of Medicine

Children: Benjamin DOB: 12/27/83

 Daniel DOB: 4/18/86

 Sarah DOB: 10/26/88

**EDUCATION**

1976 A.B. (cum laude in chemistry), Princeton University, Princeton, NJ

1980 M.D. New York University School of Medicine, New York, NY

**POSTDOCTORAL TRAINING**

1980-1981 Internship, Department of Pediatrics, Johns Hopkins Hospital, Baltimore, MD

1981-1983 Residency, Department of Pediatrics, Johns Hopkins Hospital, Baltimore, MD

1983-1985 Fellow, Developmental Evaluation Clinic, Children's Hospital, Boston, MA

**ACADEMIC APPOINTMENTS**

1986-1988 Instructor in Pediatrics, Tufts University School of Medicine, Boston, MA

1988- 1996 Assistant Professor in Pediatrics, Tufts University School of Medicine,

 Boston, MA

1997- 2000 Assistant Professor in Pediatrics, Harvard Medical School, Boston, MA

1. Visiting Associate Professor in Pediatrics, Harvard Medical School, Boston, MA
2. Associate Professor of Pediatrics, University of Colorado School of Medicine

2009 Professor of Pediatrics, University of Colorado School of Medicine

2016 Awarded Tenure

**HOSPITAL APPOINTMENTS**

1985-1986 Pediatrician, Kennedy Memorial Hospital for Handicapped Children,

 Boston, MA

1985-1986 Clinical Fellow, PKU-lnborn Errors of Metabolism, Children's Hospital,

 Boston, MA

1986-1996 Assistant Pediatrician, Clinical Genetics, New England Medical Center

 Hospitals, Boston, MA

1988-1996 Medical Director, Myelodysplasia Clinic, New England Medical Center

 Hospitals, Boston, MA

* 1. Director, Coordinated Care Services, Children’s Hospital, Boston
	2. Assistant in Medicine, Department of Medicine, Children’s Hospital Boston

1998-2000 Associate in Medicine, Department of Medicine, Children’s Hospital Boston

2001-present Director, Special Care Clinic, Children’s Hospital Colorado

2016-present Director, Osteogenesis Imperfecta Multidisciplinary Clinic

**AWARDS AND HONORS**

1987 Society for Developmental Pediatrics

1992 Advocate Award, 2nd Annual Hearing & Language Center Awards, Department

 of Speech, Hearing and Language, New England Medical Center Hospitals

1994 Travel Award from SIMD, to attend Vl International Congress of Inborn Errors

 of Metabolism, Milan, Italy

1997 Book of the Year Award in 1998 from Doody’s Health Science Book Review Journal awarded to chapter on Beckwith-Wiedemann Syndrome, which appeared in Jameson: Principles of Molecular Medicine, Humana Press

2002 Fellow, American Academy of Pediatrics

2003 Elected to the Executive Committee, Council of Children with Disabilities, AAP

2005 Editor, Developmental-Behavioral Pediatrics, 4th Edition, Elsevier, Phila, PA

2010 Editor for Special Edition of Journal of Behavioral Pediatrics devoted to Genetic Issues in Children with Disabilities

2011 Nominated for Best Role Model at the Golden Stethoscope Award night, sponsored by CU School of Medicine, Foundations of Doctoring Curriculum

2011 Received Teaching Award from the CU School of Medicine Class of 2014 for Favorite Clinical Correlate for Down Syndrome and Prader-Willi lectures

2011 Received Citation from AAP in recognition of distinguished service rendered as a member of the Executive Committee of the Council on Children with Disabilities 2003-2010

2012 Abstract on SLOS research project received “Best in Show Award” at the AAPOS meeting, March 2012

2012 Nominated for Best Pediatric Preceptor at the Golden Stethoscope Award night, sponsored by CU School of Medicine, Foundations of Doctoring Curriculum

2013 Received the Golden Stethoscope Award from CU School of Medicine, Foundations of Doctoring Program for Best Specialty Preceptor, April 2013

2013 Visiting Professor at the University of Wisconsin, Madison, Feb 2013. Gave lectures and led a grad student seminar

2009-present TOP DOC in 5280 Magazine in Neurodevelopmental disabilities

2016-present America’s Best Physicians

2017 Keynote Speaker for Nutrition Conference at CHCO

 Co-Chair of Faculty Promotions Committee since Nov 2017

2018 Career Teaching Scholar Award from the Dept of Pediatrics Nov 30, 2018

**PROFESSIONAL SOCIETIES:**

NATIONAL

1985 Member, Society of Developmental Pediatrics

1987 Member, American Academy of Cerebral Palsy & Developmental Medicine

1. Member, American Society of Human Genetics

2002 Fellow, American Academy of Pediatrics

2003-2010 Member, Executive Committee, Section on Children with Disabilities, AAP

2008 Fellow, American College of Medical Genetics

2010-2017 Member, Autism Subcommittee of the AAP Council on Children with Disabilities

**INSTITUTIONAL COMMITTEES**

DEPARTMENTAL

1998-2000 Committee on Post-graduate Education, Children’s Hospital Boston

1997-2000 Mentorship committee, Children’s Hospital Boston

2012-2016 Member, Promotions Committee, Department of Pediatrics, University of CO

2014-2017 CHA-CARE project participant

2017- 2019 SIM project participant for both working group and QI group

2016-present Transition project of patients with medical complexity to adulthood

MEDICAL SCHOOL

1993-1996 Admissions Committee, Tufts University Medical School

2002-2016 SOM Admissions Committee, University of Colorado

2016-present Member, Promotions Committee, University of CO School of Medicine

2017-present Co-Chair, Faculty Promotions Committee, University of CO School of Medicine

HOSPITAL

2003-present Interview incoming residents

2013-2014 Member of Medication Reconciliation Work Group

2014-2017 Member CHA-CARE Project Advisory Group

2014-present Member Care Coordination Implementation Team

2014-2017 Member Readmissions Work Group

2017-2019 member of SIM work group and QI group

UNIVERSITY

2004-2015 Member, Scientific Advisory Review Committee (SARC) of the Pediatric General Clinical Research Center

2008-present Member, Intellectual and Developmental Disabilities Research Center (IDDRC)

2008-present Member, Working Group on Down syndrome and related chromosome disorders

NATIONAL

2003-2010 Elected member of the Executive Committee of the AAP Council on Children with Disabilities (COCWD)

2010-2017 Elected member of the Autism Sub-committee of the AAP COCWD

**COMMUNITY SERVICE RELATED TO PROFESSIONAL WORK:**

1988-2000 Medical Advisory committee for the Cotting School (a private school for children with special health care needs and developmental disabilities in MA)

1989-2000 Understanding our Differences, a disability awareness program,

 Coordinator for Chronic Medical Conditions, Physical Disabilities,

 Learning Disabilities and Mental Retardation Units,

 Underwood School, Newton

1998-current Understanding our Differences, a disability awareness program,

 Medical Advisory Board, Newton city-wide program, Newton, MA

2004 CATCH grant committee on transition of young adults with disabilities

2011-current Member of Advisory Board, Smith-Lemli-Opitz Parent Support Group

2011-current Presented lecture and information support regarding testing for Ashkenazi Jewish Genetic Diseases (AJGD) at synagogues in Denver and Boulder, invited speaker at breakfast for Women Rabbis, June 2015;

2015 Speaker and Program Director for planned educational sessions in Nov 2015 for Obstetricians and Jewish Clergy and community members regarding updates in recommendations for screening for AJGD, sponsored by the Ashkenazi Jewish Genetic Diseases Consortium based in NY, and the Victor Center for the Prevention of Jewish Genetic Diseases, Phila, PA

2016-present Invited speaker on Jewish Genetic Disorders for AP Biology class at local HS

2017 Interviewed by Intermountain Jewish News with subsequent article published regarding recommendations for Jewish Genetic testing

**LICENSURE**

1983 Massachusetts State Medical License

2001-present Colorado State Medical License

**CERTIFICATION**

1981 National Board of Medical Examiners

1985 American Academy of Pediatrics

1. American Board of Medical Genetics/ Recertification 2003/ Recertification 2005/ Recertification 2007/ Recertification 2009-2019

2001 Neurodevelopmental disabilities 2001/ Recertification 2011-2021

**REVIEW AND REFEREE WORK**

2000-present Ad hoc reviewer for Journal of Pediatrics, American Journal of Medical Genetics, the Journal of Pediatric Gastroenterology and Nutrition, Journal of Developmental/Behavioral Pediatrics, Journal of Inherited Metabolic Disease, and Pediatrics

2004-2015 Scientific Advisory Committee for General Clinical Research Center at Children’s Hospital Colorado

2004-2010 As Executive Committee member of COCWD, I reviewed many policy statements, clinical reviews related to genetic disorders and developmental disabilities for AAP, published in Pediatrics

2010 Editor for Special Edition of Journal of Behavioral Pediatrics devoted to Genetic Issues in Children with Disabilities

2010-2017 As a member of Autism Subcommittee of the AAP, I reviewed policy statements and clinical reviews related to autism and associated genetic disorders, which are to be published in Pediatrics

2011-current Member of Advisory Board, Smith-Lemli-Opitz Parent Support Group, I review research proposals related to SLOS submitted for funding

**Invited Presentations:**

LOCAL

2002: 1. “Children should be seen and heard: Finding their voice in healthcare decision making, at *Ethics Conference at TCH, Denver*, CO April 2003

 2. “Recent advances and Current Controversies” at *Perinatal Conference at TCH, Denver*, CO, May 2003

2003: 3. “Smith-Lemli-Opitz Syndrome, Prenatal Diagnosis, Introduction and overview” and “Medical Management of Infants and Children”, at the ***5th Scientific Symposium on SLOS,*** *Denver, Co*, June 2003

 4. “New Medical Techniques and Technology in Spina Bifida”, at *Spina Bifida Conference run by Rehab Dept at TCH,* Sept, 2003

2005 5.**Grand Rounds**: Oct 21, 2005 at *The Children’s Hospital, Denver*; “Smith-Lemli-Opitz Syndrome (SLOS): An update on a fascinating disorder”

2008: 6. “Feeding Modalities for Children with Special Health Care Needs”, **Hospitalist Lecture series** *at The Children’s Hospital,* May 6, 2008

7. **Grand Rounds** for *Dept of Pediatrics* on “Smith-Lemli-Opitz Syndrome, the Tip of the Iceberg and Below”, at *the Marshfield Clinic, Marshfield, WI*, May 23, 2008

2009 8. “The Genetics of Autism, **Grand Rounds at The Children’s Hospital,** Denver, CO, Nov 2009

2011 9. **Grand Rounds** for *Dept of Pediatrics* on “Smith-Lemli-Opitz Syndrome: SLOS on the Range; where are we roaming in 2011” presented September 2011 at Children’s Hospital Colorado

2015: 10. Panelist for the Grand Rounds Panel for the Ethical Issues in Genetics: A Family Perspective Symposium March 2015

2016: Talk to Maternal Fetal Medicine on prenatal diagnosis of Smith-Lemli-Opitz syndrome, September 2016

 Talk to Maternal Fetal Medicine on prenatal diagnosis of Osteogenesis Imperfecta

2017: Invited speaker on “The Genetic Evaluation of the Child with Intellectual Disabilites” for the 33rd Annual Community and School health Pediatric Conference, June 8, 2017, Children’s Hospital CO

 Invited Speaker for the 6th Annual Updates on “Integration of Medical and Nutritional Care for Children with Medical Complexity” for the Clinical Nutrition Conference, Nov 3, 2017 Children’s Hospital CO

REGIONAL

2008: 1. “Respiratory Problems in Patients with Skeletal Dysplasias”, at the **Regional Conference of the Little People of America***, Westminster, CO,* April 26, 2008

2009: 2. “The Genetic Evaluation of the Child with Intellectual Disabilities and Autism”, at **Grand Rounds at Denver Health**, April 2009

2010: 3. “The Genetic causes of Autism in Down Syndrome: presented at the **Down Syndrome and Autism Symposium**, University of CO, Oct 8, 2010

2011: 4. “Antioxidant Treatment in SLOS” presented at the **SLOS Scientific Conference** held in Denver, CO July 2011

2015: 5. Speaker during EES Awareness month for the local chapter of the Ehlers-Danlos Support Group on “Updated Medical and Genetic Issues regarding the diagnosis and management of patients with EDS, May 28, 2015

2016: Speaker on Ashkenazi Jewish Genetic Disorders to Biology students at DAT, Dec, 2016

2017: “The Genetic Evaluation of the Child with Intellectual Disabilities” presented at the 33rd Annual Community and School Health Pediatric conference, June 8, 2017 at Children’s Hospital Colorado

 **Lyn Stevenson Pediatric Nutrition Lectureship:** “Children with Special Health Care Needs: A Medical and Nutritional Overview” at the 6th Annual Updates in Clinical Nutrition Conference Nov 3, 2017, Children’s Hospital Colorado

2019: Speaker on Ashkenazi Jewish Genetic Disorders to Biology students at DAT, Jan 2019

 Psychiatry Grand Rounds on Transition of patients with Medical Complexity to Adult care Sept 2019

NATIONAL

2000: 1.“Smith-Lemli-Opitz Syndrome: The Paradigm of a Metabolic Disorder as

Cause of Dysmorphic Features, Multiple Congenital Anomalies, and

Mental Retardation.” ***123rd Meeting of the American Association for***

***Mental Retardation***, Washington DC, June 2000

 2002 2.“Retinal dysfunction in patients with the Smith-Lemli-Opitz Syndrome”, ***American Society of Human Genetics Mtg***, Philadelphia, PA, October,2000

 3. “Connecting the Docs”, at the ***National Smith-Magenis Conference****, Colorado,* July 2002

 4. “Medical Issues including Sleep Apnea” at the ***National Down Syndrome Congress****, Denver, CO* August, 2002

2004: 5. Children and Adolescents with Disabilities: How and when do we address issues of Puberty and Sexuality; ***Instructional course*** *given Oct 1 at the annual* ***American Academy of Cerebral Palsy and Developmental Medicine Mtg*** in LA, CA Oct 2004

2005: 6. “The Genetics of Autism” presented at the Council on Children with Disabilities program Oct 8, 2005, part of the **AAP National Conference and Exhibition**, Washington, DC

2007: 7. Smith-Lemli-Opitz Syndrome - **Grand Rounds** for *Genetics Dept at the Marshfield Clinic, Marshfield, WI*, May 2007

8. “Caring for Children with Special Needs in the Hospital” **–** 1 hr session presented at the **AAP National Conference and Exhibition***, San Francisco ,* October, 2007

2008 9. “The Child who has developmental regression”,presented Oct 2008 at the **AAP National Conference and Exhibition***, Boston, MA*

10. “Children with Multiple Congenital Anomalies” at the **National SOFT Conference**, Denver, CO July 2008

2010: 11. “Beyond Developmental Screening: Laboratory testing of the child with Delayed Development” presented Oct 2010 at the **AAP National Conference and Exhibition***, San Francisco, CA*

2012 12. “Down Syndrome: Growing up in the Medical Home” presented Oct 23, 2012 in New Orleans at the **AAP National Conference and Exhibition**

2012 13. “Beyond Cholesterol: Antioxidant treatment for patients with SLOS” presented at the **ASHG annual meeting** in San Francisco, Nov 8, 2012

2013 Was invited as a **Visiting Professor at the University of Wisconsin,** Madison, Gave lectures and led a grad student seminar

14. “Beyond Cholesterol: Antioxidant Treatment for patients with the Smith-Lemli-Opitz Syndrome (SLOS)”

15. “The Genetics of Autism Spectrum Disorder”

2013 16. “Genetics and the Dual Diagnosis of Down syndrome and Autism Spectrum Disorder (DS/ASD)”, presented in Denver, CO at the **National Down Syndrome Congress, July 2013**

2015: 17. “Why is medical complexity so common in neurodevelopmental disorders?”**,** Part of the 37th **Annual Spectrum of Developmental Disabilities, a Symposium on the Intersection of Chronic Disease and Neurodevelopmental Disabilities**, Mar 23, 2015, Johns Hopkins Hospital and Kennedy Krieger Institute, Baltimore, MD

18. “Complex Care Update of the Child with a Genetic Syndrome: Carefully Coordinating Co-Morbidities and Co-Existing Conditions.”To be presented at the **Joint Program of the AAP’s Council on Children with Disabilities and Section on Developmental and Behavioral Pediatrics at the 2015 AAP NCE Meeting** in Washington, DC on October 24, 2015.

19. **Ashkenazi Jewish Genetic Disorders program: sponsored by the AJGD Consortium, held in Nov 2015:**

* **Grand Rounds at University Hospital Dept of OB/GYN**
* **Grand Rounds at Rose Hospital Dept of OB/GYN**
* Spoke to Rabbinic Counsel of Denver

2016: I gave a Telemedicine Course on the Genetic Evaluation of children with Developmental Disabilities as part of the ECHO project , in July 2016

2018: I planned and spoke at the 1st conference on Ehlers-Danlos Syndromes held in Colorado titled: **Ehlers-Danlos Syndromes and Associated Conditions: Improving the Diagnosis and Treatment for All Ages.** This two day conference, Mar 2nd for medical providers and Mar 3rd for patients and families, included invited speakers from all over the country, and speaker panels on which I participated.

2019: Invited Speaker at Smith-Lemli-Opitz Conference at the NIH, Bethesda, MD, June 27-28

Invited Speaker at National Jewish Hospital symposium on Neuroimmunological Disorders, talking about Ehlers-Danlos Syndrome and associated morbidities

Keynote Speaker at Ehlers-Danlos Conference held in Kansas City, MI, Sept 6-7

Speaker at AAP NCE on Sexuality in patients with Disabilities, Oct 28-29

INTERNATIONAL

2001: 1. “Smith-Lemli-Opitz Syndrome: update on results of cholesterol treatment” ***4th International SLOS mtg*,** Detroit, MI June 2001

2005: 2. Smith-Lemli-Opitz June, 2005 **6th International SLOS Conference** *in Baltimore*; presented at both conference for researchers and parent support group meeting

2007 3. Retinal Dysfunction in patients with SLOS presented at the **7th International Smith-Lemli-Opitz Conference** *in Portland, Oregon*, June 2007.

4. Management of Pubertal Issues in patients with SLOS, presented at **7th International Smith-Lemli-Opitz Conference** *in Portland, Oregon,* June, 2007

2009: 5. “SLOS on the Range” at the **8th International Smith-Lemli-Opitz Conference** in Boston, MA, June 2009

2011: 6. “Antioxidant Treatment in SLOS”, at the **9th** **International Smith-Lemli-Opitz Conference** in Denver, Co, 2011

2017 **4 Talks at the** **11th International Smithy-Lemli-Opitz Conference** in Cincinatti, including: Retinal and Audiologic Changes in Patients with Smith-Lemli-Opitz Syndrome presented at the Scientific Session and “Forms of Cholesterol Supplementation”, Antioxidant Studies: Retinal Disease in patients with SLOS”, and “Updated Research Plans for future Cholic Acid Studies” presented at the Family Session

2018 I presented 2 invited talks at the Ehlers-Danlos Society held in Baltimore, MD Aug 5, 2018. I presented a talk on the Pediatric presentation of Ehlers-Danlos for the **Ehlers-Danlos Society Medical Professionals Day**, and a talk entitled “Life Planning for Children with EDS” for the **Rarer Types Day of the Ehlers-Danlos Society International Conference**

**Teaching RECORD**

1a. *Medical student teaching:*

 1998- 2000 Patient-Doctor II Course to Harvard Medical School Second Year Students

2001-present Univ of Colorado SOM students who care for pediatric patients during pediatrics rotation

2005-present Molecules to Medicine Course: Clinical presentations on Down Syndrome and on Prader-Willi Syndrome

2009-2012 I mentored a Medical Student, Kirk Vance Mitchell MS 2012 in his research project regarding how well physicians are taught during medical school and residency to care for patients with Down Syndrome. This study was funded by a grant from the Special Olympics.

2010-2014 I taught two medical students, Alia Broman and Charles Johnson, who were doing their Foundations of Doctoring Curriculum with me in the Special Care Clinic. Dr Broman also worked with me on her research project studying the genetic causes of Ehlers-Danlos syndrome.

2013-present I mentored a former medical student Tiffany Pointon who did a research project on Audiologic problems in patients with Smith-Lemli-Opitz Syndrome which she presented at a national meeting and is submitting for publication. I am currently mentioning Daniele Martinez on her research project studying the effects of bisphosphonates on nephrocalcinosis. I have a new Foundations student, Sam Russell who started in 2016.

2016-present Molecules to Medicine – in addition to teaching courses on Down syndrome and Prader-Willi/Angelman syndrome, I have been giving a 3rd lecture on Imprinting

 b. *Graduate courses/Seminars:*

2001-present Attend genetics conferences at CHCO and Colorado Genetics Lab, discussing cases with Genetics Fellows

2001-present Attend Morning Report daily with the pediatric residents to review interesting cases

2005 Genetics of Autism presented to housestaff, Sept 2005

2006 Update on Smith-Lemli-Opitz Syndrome, presented at Faculty Genetics conference Jan 2006

2006-present Lecture to Neonatology Fellows on Common Genetic Disorders

2007 Lecture on Genetics of Neural Tube Defects to Rehab Fellows and Attendings

2013 **I developed a new Graduate Student Course along with Dr Katheleen Gardiner, and presented 3 lectures in Jan 2013** during the new course on Intellectual Disabilities and underlying Genetic causes. Plans are to repeat this course in Jan 2015

2015 Myelomeningocele – talk on genetics and medical complications to Genetic Counseling Graduate students

2015-present I have been working with Dr Austin Larsen on developing and teaching in the Genetics Academic Half Day course. I have taught at the Genetics Half Day course every year since, including in Oct 2017 and 2018, and May 2019.

2019 I continue to lecture to Developmental Fellows on Genetic Causes of Intellectual Disabilities, and Care of Patients with Medical Complexity, May and June 2019

1. *CME Courses:*

2000 Update on Medical Management in Children with Mental Retardation and Myelodysplasia, CME course for pediatricians at the Massachusetts Medical Society, Waltham, MA

2002: “Children should be seen and heard: Finding their voice in healthcare decision making, at Ethics Conference at CHCO Denver, CO April 2003

 “Recent advances and Current Controversies” at Perinatal Conference at TCH, Denver, CO, May 2003

2003: I organized the 5th Scientific Symposium on SLOS, Denver, Co, June 2003, a CME course attended by SLOS researches from around the country, as well as Denver area physicians, including pediatricians, Ob/GYN, neurologists, psychiatrists and geneticists. I also presented the following 2 lectures: “Smith-Lemli-Opitz Syndrome, Prenatal Diagnosis, Introduction and overview and “Medical Management of Infants and Children”, at the conference

2004: Children and Adolescents with Disabilities: How and When do we address issues of Puberty and Sexuality; Instructional course given Oct 1 at the annual American Academy of Cerebral Palsy and Developmental Medicine Mtg in LA, CA Oct 2004

2005: Smith-Lemli-Opitz June, 2005 Baltimore Conference; presented at both conference for researches and parent support group meeting

 “The Genetics of Autism” presented at the Council on Children with Disabilities program Oct 8, 2005, part of the AAP National Conference and Exhibition, Washington, DC

 Grand Rounds: Oct 21, 2005 at The Children’s Hospital, Denver; “Smith-Lemli-Opitz Syndrome (SLOS): An update on a fascinating disorder”

2007: Smith-Lemli-Opitz Syndrome: Update on a fascinating disorder, Grand Rounds for Genetics Dept at the Marshfield Clinic, Marshfield, WI, May 2007

 Invited Speaker on several topics at the Smith-Lemli-Opitz Conference in Portland, Oregon, June 2007. Topics include Retinal Dysfunction and Pubertal Issues

 “Caring for the Child with Special Health Care Needs in the Hospital,” presented at the Section on Hospitalists program at the AAP National Conference and Exhibition, Oct 2007, San Francisco

 Evaluation of the child with Birth defects – given as a Telemedicine conference for Grand Rounds in Wyoming, Oct 2007

2008: “Feeding Modalities for Children with Special Health Care Needs”, *Hospitalist Lecture Series at The Children’s Hospital,* May 6, 2008

**Grand Rounds** for *Dept of Pediatrics* on “Smith-Lemli-Opitz Syndrome, the Tip of the Iceberg and Below”, at *the Marshfield Clinic, Marshfield, WI*, May 23, 2008

“The Child who has developmental regression”,presented Oct 2008 at the *AAP National Conference and Exhibition, Boston, MA*

2010: I spoke at the NCE in October, 2010 on the genetic evaluation of children with developmental disabilities

2011: Antioxidant Treatment of patients with SLOS at the SLOS Scientific Conference held in Denver, July 2011

 **Grand Rounds** for *Dept of Pediatrics* on Smith-Lemli-Opitz Syndrome titled “SLOS on the range – where are we roaming in 2011”

2012: “Down Syndrome: Growing up in the Medical Home” presented Oct 23, 2012 in New Orleans at the *AAP National Conference and Exhibition*

“Beyond Cholesterol: Antioxidant treatment for patients with SLOS” presented at the ASHG annual meeting in San Francisco, Nov 8, 2012

2013: **Visiting Professor in Genetics at the Univ of WI, Madison, WI**. February 2013; Presented the following two lectures:

“Beyond Cholesterol: Antioxidant Treatment for patients with the Smith-Lemli-Opitz Syndrome (SLOS)”

 “The Genetics of Autism Spectrum Disorder”

2013 “Genetics and the Dual Diagnosis of Down syndrome and Autism Spectrum Disorder (DS/ASD)”, presented in Denver, CO at the National Down Syndrome Congress, July 2013

2017 “The Genetic Evaluation of the Child with Intellectual Disabilities” presented at the 33rd Annual Community and School Health Pediatric conference, June 8, 2017 at Children’s Hospital Colorado conference CTR

“Retinal and Audiologic Changes in Patients with Smith-Lemli-Opitz Syndrome” presented at the Scientific Session of the 11th International Smithy-Lemli-Opitz Conference in Cincinatti

 **Lyn Stevenson Pediatric Nutrition Lectureship:** “Children with Special Health Care Needs: A Medical and Nutritional Overview” at the 6th Annual Updates in Clinical Nutrition Conference Nov 3, 2017, Children’s Hospital Colorado

2018 I directed, planned the program and spoke at the 1st Conference on **Ehlers-Danlos Syndrome** held at Children’s Hospital Colorado on March 2, 2018.

2019 I was a speaker at the upcoming Smith-Lemli-Opitz conference in June 2019 at the NIH on various clinical topics

I was an invited speaker on management of complex patients with EDS on July 20, 2019 at National Jewish hospital

 I was the Keynote speaker for the Ehlers-Danlos Conference in Kansas City on Sept 7, 2019.

I spoke on Sexuality in patients with developmental disabilities at the AAP NCE in New Orleans, Oct 2019

*Resident/FellowTeaching:*

1996-2000 At Children’s Hospital, Boston, I designed the schedule and format for resident rotation (mandatory) on the Coordinated Care Service (CCS), an inpatient and outpatient exposure to medical management of children with special health care needs (CSHCN). I helped develop the core curriculum, and taught many of the didactic sessions.

2001-2006 At Children’s Hospital, Denver, I was the inpatient attending for children admitted to the Special Care Clinic service approximately one third of the year. In this role, I supervised and taught the residents how to manage CSHCN.

I gave didactic conferences for the residents addressing issues of medical complexity and common genetic disorders, both on the wards and during the Noon conference series.

2007 With the change (since January 2007) to the use of hospitalists as attendings on the inpatient service, I refocused my role. I continue to teach pediatric residents the care and management of Children with Medical Complexity, during the times that I am ward attending, during Morning Report which I attend daily when possible

2008-2015 I give lectures to the inpatient residents on topics related to the care of children with Medical Complexity.

2001-2014 I gave Noon conferences, about 3-6 per year, to the house staff on various topics related to Developmental Disabilities and Genetic disorders.

2010-present I mentored a faculty member in the Child Development Unit, Dr Nicole Tartaglia, who is continuing her research on the developmental profile of children with sex chromosome abnormalities

2010-2013 I supervised two residents doing their Continuity Clinic in the Special Care Clinic, Dr Shanlee Davis, and Dr Austin Larson. I also shared in supervision of a third Resident, Dr Krista Eschbach. Dr Davis is currently an Attending Endocrinologist at CHCO, and Dr Larson is a Genetics and Metabolism Attending at CHCO. Dr Eschbach is a Pediatric Neurologist at CHCO. I was also the supervisor/mentor for Dr Davis’ research project on the auto-immune endocrine disorders in patients with Down syndrome. Dr Davis presented this at 3 national meetings during her 3rd year of residency in the spring of 2013. I have continued to be involved in her research using Testosterone for patients with Sex chromosome abnormalities.

2013-2015 I supervised two pediatric residents doing their Continuity Clinic in the Special Care Clinic, Dr Ben Apple and Dr Kristin Wigby. Both of these residents were been accepted to Fellowships in Clinical Genetics. Dr Wigby has a successful career in Genetics in San Diego.

2015-present I am the Faculty Advisor for and gave lectures on several genetic topics in Jan 2015 -2019, to the residents as part of the Genetics Academic Half-Day program. I most recently did this on march 2019.

2015-present I supervised Maricarmen Shields, a pediatric resident, who did her Continuity Clinic in Special Care Clinic on a weekly basis and graduated in 2018. Since 2017, I had a second pediatrics resident, Emily Shelkowitz whom I precepted on a weekly basis, who graduated in 2019 and started her Fellowship in Genetics at CHCO.

2016-present I have accepted a Foundations of Doctoring medical student, Sam Russell

2017 Talk to Psychiatry Faculty and Fellows on Behavioral disorders in patients with Smith-Lemli-Opitz Syndrome Oct 2017

2019 I have a new resident, Dr Amanda Appel, who started her Residency in June 2019. She is in the Physiatry program.

**Administrative Responsibilities:**

* 1. I was the Director of the Coordinated Care Service at Children’s Hospital, Boston. As Director of CCS, I supervised a busy inpatient and outpatient program which cared for more than 1,000 children per year with complex medical issues and developmental disabilities. I helped develop a curriculum to teach residents about the care of children with special needs. While at Children’s Hospital, I was the Principal Investigator of a complicated research protocol to treat children with Smith-Lemli-Opitz syndrome. I was one of two attendings in the Down Syndrome Program, along with the late Dr Allen Crocker, a nationally recognized DS expert.

2001-present In 2001, I moved to Denver, and assumed the directorship of the Special Care Clinic at Children’s Hospital Colorado. The Special Care Clinic currently cares for over 4800 children with special health care needs and genetic disorders. We provide both primary and consultative care to children from CO, as well as all surrounding states.

2007-present A diagnosis-based clinic was started in November, 2007. The clinic is devoted to the care of children with Skeletal Dysplasias, and is a multidisciplinary clinic with myself, Leah Rowe, a genetic counselor, Dr Nancy Miller of Orthopedics, and Dr Joyce Olesyk of Rehabilitation Medicine. It is held once a month, and is the only clinic for individuals with Skeletal Dysplasia in this part of the country. I am the senior geneticist and complex care expert in this clinic.

2016- present Medical Director of the Osteogenesis Imperfecta Clinic, a Multi-disciplinary Clinic for patients with OI which includes providers from Genetics (me), Endocrinology (Dr Maggie Chan), Orthopedics, Rehabilitation Medicine, Social Work and nursing.

2019 through a generous donation from a philanthropist, I will be starting a new Center of Excellence for patients with Ehlers-Danlos syndrome, which will begin in the fall of 2019.

**GRANT SUPPORT**

I am the Principal Investigator for a study entitled “Treatment of the Cholesterol Defect in Children with the Smith-Lemli-Opitz Syndrome”, which has been supported by MOI-RR00069, the Pediatric Clinical Translational Research Center (CTRC) at Children’s Hospital Colorado. The CTRC paid for the investigational antioxidant medication dispensed to the patients, as well as the some of the laboratory studies of the patients in the past thru 2013. Currently insurance pays for lab work and studies under anesthesia.

I was accepted as a PI into the NICHD-supported National Consortium which studies sterol disorders, called STAIR, as of June 2016. I have written a new protocol to be funded by STAIR to use Cholic acid in addition to cholesterol and antioxidant treatment, which will start in 2019. The protocol has approved by NIH reviewers and is in the final stages of planning.

Between the fall of 2007 and June 2013, I participated in a CDC funded protocol called CADDRE, developed to assess the underlying genetic contributions to autism. This was a multi-institutional study. This Project paid for 5-10% of my salary through June 2013. This project is currently in the analytic phase, and I have co-authored several abstracts presented at genetics meetings regarding this project. The actual paper being written regarding the results of this study, for which I am a co-author, have been recently published.

I was awarded a $100,000 grant by the Co-Pilot Project of the CCTSI to study the Genetic Cause of Autism in Patients with Down syndrome in 2010. The funding for this project was extended through 2012. This project is continuing under Dr Tamim Shaik and I continue as a co-PI.

I continue to be involved in two grants funded by the Linda Crnic Center since 2013-2014:

* “Investigating the Genetic Etiology of Autism in Patients with Down Syndrome”, COMIRB 10-0276. This is a continuation of the study funded by the CCTSI in 2010, but using new genetic technologies to perform more sophisticated testing including whole exome sequencing. The PI for this project is Dr Tamim Shaikh.
* “Investigating the Genetic Etiology of Autoimmune Disorders in Patients with Down Syndrome”, COMIRB 13-0397. This is a study using sophisticated genetic testing to evaluate genes associated with an increased risk of multiple autoimmune diseases such as Thyroid, Diabetes and Celiac Diseases in patients with DS. The PI for this project is Dr Richard Spritz. This project is winding down due to the imminent retirement of Dr Spritz.

I am a co-investigator with Dr Dennis Roop and his team at the Gates center for Regenerative Medicine, looking at pluripotent stem cell studies in patients with Ehlers-Danlos Syndrome.

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**Other Products of Scholarship:**

**Autism Toolkit**, published by the AAP initially in 2007 and revised in 2012, a comprehensive package of information for the pediatrician with information and recommendations regarding care of the child with an Autism Spectrum Disorder. This packet includes the two Clinical Reports published in 2007 and reaffirmed subsequently (lead authors Scott Myers and Chris Johnson). I participated in editing and partly writing the sections on recommendations for genetic testing and descriptions of disorders associated with autism

**Letters to the Editor:**

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