**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.

**Ellen Roy Elias, MD Bibliography**

**Papers Published in Peer-Reviewed Journals**

1. Stamberg J, Jabs E, and **Elias ER**. Terminal deletion of (4) (q33) in a white male infant. Clin Genet 1982; 21:125-129.

2. Kaplan LC, Osbourne P, **Elias ER**. The diagnosis of muscular dystrophy in patients referred for evaluation of language delay. J Child Psychol Psychiat 1986; 27:545-549.

3. Kaplan LC, Wharton R, **Elias ER**, Mandell F, Donlon T, Latt SA. Clinical heterogeneity associated with deletions in the long arm of chromosome 15: report of three new cases and their possible genetic significance. Am J Med Genet 1987; 28: 19-27.

4. McCauley RG, Beckwith JB, **Elias ER** et al. Benign hemorrhagic adrenocortical macrocysts in Beckwith-Wiedemann syndrome. Am J Roent 1991; 157:549-552.

5. Dobyns WB, **Elias ER**, Newlin AC, Pagon RA, Ledbetter DH. Causal heterogeneity in isolated lissencephaly. Neurology 1992; 42: 1375-1388.

6. **Elias ER**, Sadeghi-Nejad A. Precocious puberty in girls with myelodysplasia. Pediatrics 1994; 92:521-522.

7. Irons M, **Elias ER**, Salen G, Tint GS, Batta AK. Defective cholesterol biosynthesis in Smith-Lemli-Opitz syndrome. Lancet 1993; 341:1414.

8. Tint GS, Irons M, **Elias ER**, Batta AK, Friedan R, Chen TS, Salen G. Defective cholesterol biosynthesis associated with the Smith-Lemli-Opitz syndrome. New Eng J Med 1994; 330:107-113.

9. Irons, M, **Elias ER**, Tint GS, Salen G, Friedan R, Buie TM, Ampola A. Abnormal cholesterol metabolism in the Smith-Lemli-Opitz syndrome. Report on clinical and biochemical findings in 4 patients and treatment in 1 patient. Am J Med Genet 1994; 50:347-352.

10. Batta AK, Tint GS, Salem G, Shefer S, Irons M, **Elias ER**. Identification of 7-dehydrocholesterol and related sterols in patients with Smith-Lemli-Opitz syndrome. Am J Med Genet 1994; 50: 334.

11. Shefer S, Salen G, Batta AK, Tint GS, Irons M, **Elias ER**. Reduced 7-dehydrocholesterol--reductase activity in Smith-Lemli-Opitz syndrome. Am J Med Genet 1994;50:336.

12. Moser A, Rasmussen M, Naidu S, Watkins P, Mcguinness M, Hajra A, Chen C, Raymond G, Liu A, Gordon D, Carnass K, Walton D, Skjeldal, Guggenheim M, Jackson L, **Elias ER**, Moser H. Phenotype of 173 peroxisome disorder patients subdivided into 16 complementation groups. J Peds 1995;127:13-22.

13. Irons M, **Elias E**, Abuelo D, Tint GS, Salen G. Clinical features of the Smith-Lemli-Opitz syndrome and treatment of the cholesterol metabolic defect. Int'l Peds 1995;10:28-32.

14. Tint, GS, Salen G, Batta, AG, Shefer S, Irons, M, **Elias ER**, Abuelo DN, Johnson VP, Lambert M, Lutz R. Correlation of outcome with plasma sterol in Smith-Lemli-Opitz Syndrome. J Peds 1995; 127:82-87.

15 Salen G, Tint GS, Xu G, Batta AK, Irons M, **Elias ER**. Abnormal cholesterol biosynthesis in the Smith-Lemli-Opitz Syndrome. Ital J Gastroenterol 1995, 27:506-508

16 Shefer S, Salen G, Batta AK,Honda A, Tint GS, Irons M, **Elias ER**, Holick MF. Markedly inhibited 7-DHC 7 reductase activity in liver microsomes from Smith-Lemli-Opitz heterozygotes. J Clin Invest 1995;96:1779-1785

17 DS Theodoropoulos, JM Cowan, **ER Elias**, C Cole, Physical Findings in 21q22 Deletion Suggest Critical Region for 21q- Phenotype in q22, Amer J Med Genet 1995; 59:161-163 .

18. Tint GS, Seller M, Hughes-Benzie R, Batta AK, Shefer S, Genest D, Irons M, **Elias ER.** Markedly increased tissue concentrations of 7-dehydrocholesterol combined with low levels of cholesterol are characteristic of the Smith-Lemli-Opitz Syndrome. J Lipid Res 1995 ; 36:89-95

19. Salen G, Shefer S, Batta AK, Tint GS, Xu G, Honda A, Irons M, **Elias ER**. Abnormal cholesterol biosynthesis in the Smith-Lemli-Opitz Syndrome. J Lipid Res 1996 37:1169-1180

20. JM Milunsky, HE Wyandt, X Huang, X Kang, **ER Elias**, A Milunsky, Trisomy 15 Mosaicism and Uniparental Disomy (UPD) in a Liveborn Infant, Amer J Med Genet 1996; 61:269-273 .

21. **ER Elias** et al., Clinical Effects of Cholesterol Supplementation in Six Patients with the Smith-Lemli-Opitz Syndrome (SLOS) - Amer J Med Gen, 1997; 68:305-310

22. Irons M, **Elias ER**, Abuelo D, Bull MJ, Greene CL, Johnson VP, Keppen L, Schanen C, Tint GS, Salen G. Treatment of Smith-Lemli-Opitz Syndrome: Results of a multicenter trial. Amer J Med Genet, 1997;68:311-314

23. ML Huie et al. Glycogen Storage Disease Type II - Identification of 4 novel missense mutations and 2 insertions/deletions in the acid -glucosidase locus of patients of differing phenotype. Biochem and Biophys Research Communications, 1998; 244: 921-927.

24. Giampietro PF, Auerbach AD, **Elias ER** et al. A new recessive syndrome characterized by increased chromosomal breakage and several features which overlap with Fanconi Anemia. Amer J Med Genet, 1998; 78:70-75.

26. Tint GS, Abuelo D, Till M, Cordier MP, Batta AK, Shefer S, Honda A, Honda M, Xu G, Irons M, **Elias ER**, Salen G. Fetal Smith-Lemli-Opitz Syndrome can be detected accurately and reliably by measuring amniotic fluid dehydrocholesterols. Prenatl Diagn; 1998, 18:651-658

27. **Elias ER**, Mobasseleh M, Hajra AK, Moser AM. Developmental delay and growth failure caused by deficiency of dihydroxyacetonephosphate acyl transferase (DHAP-AT): a new peroxisomal disorder. Amer J Med Genet 1998; 80 (#3): 223-226

28. Huang T, **Elias ER**, Mulliken JB, Kirse DJ, Holmes LB. A New Syndrome: Heart Defects, Laryngeal Anomalies, Preaxial Polydactyly and Colonic Aganglionosis in Sibs. Genetics in Medicine 1999; 1 (#3):223-226

29. Borer JG, Kaefer M, Barnewolt CE, **Elias ER**, Hobbs N, Retik AB, Peters CA. Renal Findings on Radiological Followup of Patients with Beckwith-Wiedemann Syndrome. J of Urology 1999;161:235

30. Metzl JD, **Elias ER**, Berul CI. A Case of Infant Sudden Death: severe hypertrophic cardiomyopathy in Pompe’s disease. Pacing and Clinical Electrophysiology 1999; 22:821-22

31. Wheeler PG, Sadeghi-Nejad A, **Elias ER**. The 3C Syndrome: evolution of the phenotype and growth hormone deficiency. Amer J Med Genet 1999; 87: 61-62.

32. Toriello HV, Carey JC, Addor M-C, Allen W, Burke L, Chun N, Dobyns W, **Elias E**, et al. Toriello-Carey Sydnrome: Delineation and Review Amer J Med Genet 2003; 123A:84-90

33. **Elias ER**, Hansen RM, Irons M, Quinn NB, Fulton, AB. Rod Photoreceptor responses in Children with Smith-Lemli-Opitz Syndrome. Arch Ophthalmol 2003; 121:1738-1743

34.Caruso PA, Poussaint TY, Tzika AA, Zurakowski D, Astrakas LG, **Elias ER**, Irons MB.

Neuroradiology MRI and (1) MRI findings in Smith-Lemli-Opitz syndrome 2004; 46:3-14

36 Pauliks LB, Chan K-C, Lorts A, **Elias ER**, Cayre RO, Valdes-Cruz LM. Ultrasound in Medicine Sphrintzen-Goldberg syndrome with tetralogy of Fallot and subvalvar aortic stenosis – case report and review of the literature. 2005 J Ultrasound May;24(5):703-6

37 Berman S, Rannie M, Moore L, **Elias E,** Dryer LJ, Jones MD. Utilization and costs for children who have special health care needs and are enrolled in a hospital-based comprehensive primary care clinic. Pediatrics. 2005 Jun;115(6):e637-42

38 Pickler, L, **Elias, ER,** “The Genetic Evaluation of the Child with Autism Spectrum Disorders”, Pediatric Annals, in edition titled Autism Spectrum Disorders: What’s New and What’s to Do?, Edited by C. Johnson and S. Myers Pediatric Annals 2009 38(1): 26-29

39 Garry D, Hansen RM, Moskowitz A, **Elias ER,** Irons M, Fulton AB. “ Cone ERG Responses in patients with Smith-Lemli-Opitz Syndrome (SLOS)”. Doc Ophthalmol 2010 121(2):85-91

43 Stein, MT, **Elias, ER,** Saenz, M, Pickler, L and Reynolds, A**. “**Autistic Spectrum Disorder in a 9 yr old with Macrocephaly”. Journal of Developmental and Behavioral Pediatrics, volume 31, pages 632-634 (2010)

44 Kaplan FS, Shore EM, Pignolo RJ (eds), **Elias, ER**, and The International Clinical Consortium on FOP.  The medical management of fibrodysplasia ossificans progressiva: current treatment considerations. **Clin Proc Intl Clin Consort FOP** 4:1-100, 2011].

45 Liu, W, Xu, L Lamberson, CR, Merkins, LS, Steiner, RD, **Elias, ER**, Haas, D and Porter, NA. Assays of Plasma Dehydrocholesteryl Esters and Oxysterols from Smith-Lemli-Opitz Patients, L Lipid Research, 2013; 54:244-253

46 Larson, A, LeRoux, J, **Elias, ER,** Exonic deletion of *OPHN1* resulting in seizures, intellectual disability, and brain malformations, Advances in Genomics and Genetics, 2014; 4, 1-4

47 Yu HC, Coughlin CR, Geiger EA, Salvador BJ, Elias ER, Cavanaugh JL, Chatfield KC, Miyamoto SD, Shaikh TH. Discovery of a potentially deleterious variant in TMEM87B in a patient with a hemizygous 2q13 microdeletion suggests a recessive condition characterized by congenital heart disease and restrictive cardiomyopathy. Cold Spring Harb Mol Case Stud. 2016 May;2(3):a000844. doi: 10.1101/mcs.a000844.

48 Rachubinski, AL, Hepburn, S, **Elias**, **ER** ; Katheleen Gardiner, Tamim H. Shaikh. The co-occurrence of Down syndrome and autism spectrum disorder: is it due to additional genetic variations? Prenatal Diagnosis, 2017 Jan 37(1):31-36

49 Pointon T, **Elias ER**. Auditory findings in Smith-Lemli-Opitz Syndrome. Clin of Otorhinolaryngology (2017) 2:1 017

50 Shapira, SK, Tian LH, Aylsworth, AS, **Elias, ER,** Hoover-Fong, JE, Meeks, NJ, Souders, MC, Tsai, Anne C-H, Zackai, EH, Alexander, AA, Yeargin-Allsopp, M, Shieve, LA. A Novel Approach to Dysmorphology to Enhance the Phenotypic Classification of Autism Spectrum Disorder in the Study to Explore Early Development. Journal of Autism and Developmental Disorders, (2019) 49:2184-2202

51 Shelkowitz, E, Singh, JK, Larson, A, **Elias, ER,** IRF2BPL Gene Mutation: Expanding on Neurologic Phenotypes, AJMG Part A (2019): 1-9

**Books and Monographs:**

Developmental-Behavioral Pediatrics, 4th Edition, Editors William B Carey, Allen C Crocker, William L Coleman, Heidi M Feldman, **Ellen R** **Elias,** Saunders/ Elsevier, Philadelphia, PA ; 2009

**Book Chapters, Invited Articles and Reviews**

1. **Elias ER**, Bauer S. Urologic disorders, in Rubin IL and Crocker AC (eds): Developmental Disabilities: Delivery of medical care for children and adults. Philadelphia, Lea & Febiger; 1989, p.282.

2. **Elias, ER**, Irons,M; Abnormal Cholesterol Metabolism in Smith-Lemli-Opitz Syndrome, in Korf B (ed): Current Opinion in Pediatrics: Genetics section, Philadelphia, Current Science; 1995, 7:710-714.

3. **Elias, ER**, DeBaun MR, Feinberg AP: Beckwith-Wiedemann Syndrome, in Jameson: Principles of Molecular Medicine, Genetic Basis of Congenital Malformations, Ethylin Wang Jabs, Ed., Totowa, NJ, Humana Press, 1998 Chap 116: p 1047-1052

4. **Elias ER**, Hobbs N. Spina Bifida. Contemporary Pediatrics, 1998; 15 vol 4: 156-171

1. Irons MB, **Elias ER**. Glycogen Storage Diseases, in The Molecular Genetics of Cardiac Electrophysiology, Berul CI and Towbin JA, eds Klower Acad Publishers 1999
2. Glader, L, **Elias, ER**, Madsen, J. Myelodysplasia in Manual of the Intensive Care of the Surgical Newborn, Hansen and Puder, Ed’s, Hamilton, Ontario, BC Decker Inc, 2003 Chap 10 Part II, pg 394-405
3. **Elias ER**, Tsai A H-C, Manchester D, Genetics and Dysmorphology, in Current Pediatric Diagnosis and Management, 16th ed, The McGraw-Hill Companies, 2003 Chapter 33, Genetics and Dysmorphology; pg 1009-1050
4. **Elias ER**. Genetic Evaluation in the Newborn. NeoReviews 2003 Vol 4 No 10:e277-282
5. **Elias ER**, Tsai A H-C, Manchester D, Genetics and Dysmorphology, in Current Pediatric Diagnosis and Management, 17th ed, The McGraw-Hill Companies, NY, NY 2005 Chapter 33, Genetics and Dysmorphology; pg 1039-1079
6. **Elias ER**, Tsai A H-C, Manchester D, Genetics and Dysmorphology, in Current Pediatric Diagnosis and Management, 18th ed, The McGraw-Hill Companies, NY, NY 2005 Chapter 33, Genetics and Dysmorphology; pg 1011-1048
7. **Elias ER**, Tsai A H-C, Manchester D, Genetics and Dysmorphology, in Current Pediatric Diagnosis and Management, 19th ed, The McGraw-Hill Companies, NY, NY 2008 Chapter 33, Genetics and Dysmorphology;
8. Glader, L, **Elias, ER**, Madsen, J. Chapter 9 Neurological Disorders, Part 2

Myelodysplasia in Manual of the Intensive Care of the Surgical Newborn 2nd Ed, Hansen and Puder, Ed’s, People’s Medical Publishing House, Shelton, CT 2009 Pg 459-472

1. **Elias ER**, Chapter 22 The Biomedical Basis of Development and Behavior in Developmental-Behavioral Pediatrics, 4th Edition, Saunders/Elsevier, Philadelphia, PA, 2009 pg 209
2. **Elias ER**, Chapter 74 The Medical Management of the Child with Multiple Disabilities in Developmental-Behavioral Pediatrics, 4th Edition, Saunders/Elsevier, Philadelphia, PA, 2009 pg 737
3. **Elias ER,** Chapter 68 Intellectual Disabilities (formerly called Mental Retardation) (coauthoring with Dr Allen Crocker of Boston Children’s Hospital ), in Developmental-Behavioral Pediatrics, 4th Edition, Saunders/Elsevier, Philadelphia, PA, 2009, pg 663
4. **Elias, ER,** Chapter 26 Genetic Syndromes and Dysmorphology, (coauthoring with Proud, V), in Developmental-Behavioral Pediatrics, 4th Edition, Saunders/Elsevier, Philadelphia, PA, 2009, pg 246
5. **Elias, ER** Final Chapter, The Right to Be Different (coauthoring with Dr’s Carey, W, Coleman, W, Crocker, A and Feldman, H) in Developmental-Behavioral Pediatrics, 4th Edition, Saunders/Elsevier, Philadelphia, PA, 2009, pg 989
6. Tsai, C-H, Manchester, D and **Elias ER.** Genetics and Dysmorphology, in Current Pediatric Diagnosis and Treatment 20th Edition, 2010 McGraw Hill Pg 1020
7. David Fox, MD, Karen Kelminson, MD, and **Ellen Roy Elias**, MD Overview of Primary Care for the Child with Special Health Care Needs in Berman's Pediatric Decision Making, 5th Edition 2011
8. Karen Kelminson, MD, **Ellen Roy Elias, MD** and Edward Goldson, MD. Down Syndrome in Berman's Pediatric Decision Making, 5th Edition 2011
9. Tsai, C-H, Manchester, D and **Elias ER.** Genetics and Dysmorphology, in Current Pediatric Diagnosis and Treatment 21st Edition, 2012 McGraw Hill
10. Rosenblatt, AI, Carbone PS Autism Spectrum Disorders, What Every Parent Needs to Know, American Academy of Pediatrics, 2013 (contributed to Chapter 2, What Causes Autism Spectrum Disorders)
11. **Ellen Roy Elias, MD, FAAP, FACMG** The Genetics of Down Syndrome and Autism Spectrum Disorder in When Down Syndrome and Autism Intersect, a Guide to DS-ASD for Parents and Professionals, 2013, Woodbine House
12. Saenz, Margarita, Tsai, C-H, Manchester, D and **Elias ER.** Genetics and Dysmorphology, in Current Pediatric Diagnosis and Treatment 22nd Edition, 2013 McGraw Hill
13. Saenz, Margarita, Meeks, Naomi, Tsai, C-H, and **Elias ER.** Genetics and Dysmorphology, in Current Pediatric Diagnosis and Treatment 23rd Edition, 2015 McGraw Hill
14. Larson, AL, and **Elias ER,** Chap 4,Biological Influences on Child Development and Behavior and Medical Evaluation of Children with Developmental-Behavioral Delays/Disorders, published in Developmental and Behavioral Pediatrics*, 2nd edition,* edited by Dr*.* Robert Voigt, Dr. Michelle Macias, Dr. Scott Myers, and Dr. Carl Tapia. American Academy of Pediatrics 2018
15. **Ellen R Elias**, “The Trials of Transition – how well are we doing, and how can we do better?”Requested Commentary in the Journal of Graduate Medical Education, published in April 2017
16. Meeks, Naomi, Saenz, Margarita, Tsai, C-H, and **Elias ER.** Genetics and Dysmorphology, in Current Pediatric Diagnosis and Treatment 24th Edition, 2018 McGraw Hill
17. Jarrell J.A., **Elias E.R**., Kang T.I. (2018) Palliative Care in Adolescents and Young Adults with Special Healthcare Needs. In: Hergenroeder A., Wiemann C. (eds) Health Care Transition. Springer, Cham

**Other Publications including Policy Papers**

(The first 5 citations are peer-reviewed Clinical Reports published in Pediatrics that provide national guidelines to pediatricians regarding the care of children with disabilities)

1. N Murphy, **ER Elias**. and the Executive Committee on Children with Disabilities of the AAP, Sexuality of Children and Adolescents with Developmental Disabilities; in Pediatrics 2006; Vol 118(1), 398-40
2. Myers, S, Johnson, CP and the Executive Committee on Children with Disabilities of the AAP, Management of Children with Autism Spectrum Disorders, Pediatrics Vol. 120 No. 5 November 1, 2007, pp. 1162 -1182
3. Johnson, CP, Myers, SM and the Executive Committee on Children with Disabilities of the AAP, Identification and Evaluation of Children with Autism Spectrum Disorder Pediatrics Vol. 120 No. 5 November 1, 2007, pp 1183-1215
4. **Elias, ER**, Nancy Murphy and the Executive Committee on Children with Disabilities of the AAP, Home Care of Children and Youth with Complex Health care Needs and Technology Dependencies. Pediatrics published online: April 30, 2012 (doi: peds.2012.0606)
5. Adams, R, **Elias, ER** and the Executive Committee on Children with Disabilities of the AAP, Nonoral Feeding for Children and Youth with Developmental or Acquired Disabilities, Pediatrics published online: November 24, 2014 (doi: peds.2014.2829)
6. **Genetic Testing for Developmental Disabilities, Intellectual Disability, and Autism Spectrum Disorder,** AHRQ Publication No. 15-EHC024-EF June 2015 Technical Brief #23; I served as one of the Key informants for this extensive work geared towards making evidence-based recommendations for genetic testing.

**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:**

1. Elias AD, **Elias ER**. Matching of couples in the NRMP. New Eng J Med 1980; 302: 1425-1426.

2. **Elias ER**. Response to “Cholesterol Supplementation in Smith-Lemli-Opitz Syndrome. Amer J Med Gen, 1998, 78:379-380

**ABSTRACTS:** *(all of the abstracts listed below were competitive and presented as posters unless otherwise noted)*

1. **Elias ER**, Rubin IL. Urinary tract infections in patients with cerebral palsy. 38th Annual Meeting of the American Academy of Cerebral Palsy and Developmental Medicine, October 1984.

2. Kaplan, LC, **Elias ER**. The diagnosis of muscular dystrophy in patients referred for evaluation of language delay. 39th Annual Meeting of the American Academy of Cerebral Palsy and Developmental Medicine, October, 1985.

3. **Elias ER**, Rubin IL. The longterm sequelae of the congenital rubella syndrome. Dev Med Child Neurol 1986; 28 (Suppl 53):39.

4. **Elias ER**, Siber M. Tethered cord syndrome. Dev Med Child Neurol 1987; 30 (Suppl 57):26.

5. Kellogg C, **Elias E**, Nielsen H, Sadeghi-Nejad A. Familial primary adrenal hypoplasia (FPAH) and short-limbed dwarfism in male and female half-siblings. Pediatr Res 1990; 27: 133A.

6. Nass S, Mehttretter S, Eanuzzo J, Michaud L, Ward BE, **Elias ER** and Gersen SL. Chromosome abnormality in a child with a novel peroxisomal defect. Cytogenetics Meetings, April 1992.

7. **Elias ER**, Mobassaleh M, Moser A, Hajra AK. A unique variant of plasmalogen synthesis. SIMD Annual Meeting, March 1993.

8. Irons M, **Elias ER**, Tint GS, et al. A new disorder of cholesterol biosynthesis in three patients with the Smith-Lemli-Opitz syndrome. American Society of Human Genetics, New Orleans, October 1993.

9. **Elias ER**, Irons M, Tint S, Salen G. Cholesterol biosynthesis defect in the Smith-Lemli-Opitz syndrome. 1st Annual Meeting, American College of Medical Genetics, Orlando, March 1994.

10. **Elias ER**, Mobassaleh M, Moser A, Hajra AK. A unique variant of plasmalogen synthesis. Vl International Congress of Inborn Errors of Metabolism, Milan, Italy, May 1994.

11. **Elias ER**, Irons M, Tint S, Salen G. Cholesterol biosynthesis defect in the Smith-Lemli-Opitz syndrome. Vl International Congress of Inborn Errors of Metabolism, Milan, Italy, May 1994.

12. **Elias ER**, Irons M, Tint S, Salen G. Treatment of the Cholesterol Defect in the Smith-Lemli-Opitz syndrome. American Society of Human Genetics, Montreal Canada, October 1994.

13. **Elias ER**, Sadeghi-Nejad A. Precocious Puberty in Boys with Myelomeningocele. 2nd Annual meeting of the American College of Medical Genetics, Los Angeles, CA, March, 1995.

14. Irons M, **Elias ER**, Nwokoro NA, Mulvihill JJ, Bull MJ, Keppen L. Fat-soluble vitamins in the Smith-Lemli-Opitz Syndrome. 2nd Annual meeting of the American College of Medical Genetics, Los Angeles, CA, March, 1995.

15. **Elias ER**, Irons M, Tint S, Salen G. Treating the Cholesterol defect in Smith-Lemli-Opitz syndrome: effects on multiple organ systems. 2nd Annual meeting of the American College of Medical Genetics, Los Angeles, CA, March, 1995.

16. **Elias ER**, Irons, M, Hurley A, et al. Neuropsychological and behavioral effects of Cholesterol Therapy in Children with the Smith-Lemli-Opitz (SLO) Syndrome. The American Society of Human Genetics, 45th Annual Mtg., Minneapolis, MN, Oct 1995.

17. Irons M, **Elias ER**, Abuelo D, Bull MJ, Greene CL, Johnson VP, Keppen L, Schanen C, Tint GS, Salen G, Treatment of Smith-Lemli-Opitz syndrome (SLOS): Results of a Multicenter Trial. The American Society of Human Genetics, 45th Annual Mtg., Minneapolis, MN Oct 1995.

18. Cowan JM, **Elias ER**, Trisomy 12 mosaicism in three tissues in a live born infant. The American Society of Human Genetics, 45th Annual Mtg., Minneapolis, MN Oct 1995.

19. Berlin BM, Shephard BA, **Elias ER**, Lazar EC, Bianchi DW, Abnormal serum triple screen and cri-du-chat syndrome. The American Society of Human Genetics, 45th Annual Mtg., Minneapolis, MN Oct 1995.

20. **Elias ER**, Laio Y, Irons M, Clinical factors which predict an abnormal sterol profile in patients with the Smith-Lemli-Opitz Syndrome (SLOS). 3rd Annual Mtg, Amer College of Med Genetics, San Antonio, TX, Mar 1996

21. **Elias ER**, Ampola M, Kaye E, Sege R, Shih V. Treatment of Molybdenum Cofactor Deficiency (MoCD) with betaine and special diet. 3rd Annual Mtg, Amer College of Med Gentics, San Antonio, TX, Mar 1996

22. **Elias ER**, Irons M, Hurley A, Tint GS, Salen G. Behavioral and Neuropsychological effects of cholesterol therapy in children with the Smith-Lemli-Opitz Syndrome (SLOS). 3rd Annual Mtg, Amer College of Med Genetics, San Antonio, TX, Mar 1996

23. Hobbs, NM, **Elias, ER**. Report of two siblings with severe Osteogenesis Imperfecta (OI), one with nephrocalcinosis. 4th Annual Mtg, Amer College of Med Genetics, Ft Lauderdale, FLA, Mar 1997

24. **Elias ER**, Irons, M, Klempner, M. Neutrophil function in patients with the Smith-Lemli-Opitz Syndrome (SLOS), 4th Annual Mtg, Amer College of Med Genetics, Ft Lauderdale, FLA, Mar 1997

25. **Elias ER** Irons, M, Wolfe, LC. Hematologic abnormalities in children with the Smith-Lemli-Opitz syndrome (SLOS). 4th Annual Mtg, Amer College of Med Genetics, Ft Lauderdale, FLA, Mar 1997

26. Irons M, **Elias ER** et al. Lipid profiles in Smith-Lemli-Opitz syndrome, SPR, 1997

27. Sarneso CA, et al. Prenatal diagnosis and cholesterol supplementation in a pregnancy affected by SLOS, ASHG mtg, Baltimore, MD, Oct 97, AJMG 61:A162;1997

28. **Elias ER**, et al. Effect of cholesterol deficiency on erythrocytes and neutrophils in children with SLOS. ASHG mtg, Baltimore, MD, Oct 97, AJMG 61:A250; 1997

29. **Elias ER**, Ware J, Choy YS, Tint GS. Exploring a new frontier: the mild end of the spectrum of the Smith-Lemli-Opitz Syndrome (SLOS). ASHG mtg Denver CO, Oct 1998, AJHG 63#4:A103;1998

30. **Elias ER**, Irons M, Tint GS, Salen, G. Five Year Experience in Treating the Smith-Lemli-Opitz Syndrome; What have we learned and where are we going? 6th Annual Mtg, Amer College of Med Genetics, Miami, FL Mar 99

1. **Elias ER**, Irons M, Fulton A, Tint GS, Salen S. Cholesterol supplementation for patients with the Smith-Lemli-Opitz Syndrome: a review of five years’ experience and avenues for future research. ASHG mtg, San Francisco, Oct 99
2. **Elias ER**, Fulton, A, Mayer DL, Hansen, RM. Retinal dysfunction in patients with the Smith-Lemli-Opitz Syndrome (SLOS). ASHG mtg, Philadelphia, PA Oct. 2000
3. Washington KA, Hoffenberg EJ, Tsai C-H, **Elias, ER**. Unusual GI Pathology in a patient with Rett Syndrome (RS). ASHG mtg San Diego, CA Oct 2001
4. **Elias ER**, Kochevar IE, Taylor CR. Photosensitivity in patients with the Smith-Lemli-Opitz Syndrome (SLOS). ASHG mtg, San Diego, CA Oct 2001
5. Irons M, **Elias ER**, Bay C, Pober B, Tint GS, Salen G. Improvement in sterol levels with cholesterol treatment in Smith-Lemli-Opitz Syndrome (SLOS). ASHG mtg, San Diego, CA Oct 2001
6. **Elias ER**, Tsai C-H, Zemel S. Pamidronate use in infants with Osteogenesis Imperfecta. ASHG mtg, Baltimore, MD Oct 2002
7. Manchester DK, **Elias ER**. Arthrogryposis, gastroschisis, and spinal cord atresia with dysmorphic features: a variant of dyssegmental dysplasia or a new syndrome? ASHG mtg, Baltimore, MD October 2002
8. **Elias ER**, Maleki, A, McGavran L. A complex and unique chromosomal abnormality. ASHG mtg Los Angeles, CA, Nov 2003
9. **Elias ER**. A child with multiple congenital anomalies, dysmorphic features and a rare skeletal dysplasia. ACMG mtg., Orlando FLA, Mar 2004
10. Morrisey MC, Bateman JB, Durairaj VD, Kelley PE, **Elias ER**. Delineation of the phenotype of trisomy 6p/monosomy 12p (unbalanced translocation) including novel nasal and ophthalmologic features. ACMG mtg., Orlando FLA, Mar 2004
11. Morrisey MC, Mandava N, **Elias ER**, Bateman, JB. Report of ocular features including retinal photoreceptor degeneration associated with a novel unbalanced chromosomal translocation (6:12)(p21.1;p13) in an infant, ARVO April, 2004, Ft Lauderdale, FLA
12. Berman, S, Rannie, M, **Elias ER**, Jones, MD. Ambulatory and inpatient utilization and costs for children with special health care needs enrolled or not enrolled in a hospital-based comprehensive primary care clinic. SPR, May 2004
13. **Elias ER**, Zeitler, P. Hypercalciuria and nephrocalcinosis in Infants with Osteogenesis Imperfecta (OI). ASHG, Oct 2004, Toronto, CA
14. **Elias ER**, Giampietro, P. Autism may be caused by Smith-Lemli-Opitz Syndrome (SLOS). ACMG mtg, March 2005, Dallas, TX
15. **Elias ER**, Pickler L, Bieschel LS, Johnson JP. Atypical Phenotype of Angelman Syndrome (AS) in two patients mosaic for imprinting mutations. ACMG mtg, March 2006 San Diego, CA
16. **Elias ER,** Garrington, T, Schaeffer M, Swisshelm K, Geiersbach K. Ring Chromosome 6 with Wilms Tumor and Tetralogy of Fallot. ACMG mtg, March 2007, Nashville, TN
17. **Elias ER.** Bannayan-Riley-Ruvalcaba Syndrome, a cause of autism not to miss. ACMG mtg, March 2008, Pheonix, AZ
18. Swisshelm K, Lunt B, Meltesen L, **Elias ER.** Interstitial 1q deletion detected by array comparative genomic hybridization (aCGH). March 2008, Phoenix, AZ
19. Braverman RS, Curtis T, Drack A, Bateman JB, Mandava N, **Elias ER.** Smith-Lemli-Opitz Syndrome – retinal pigment and epithelial abnormalities and photoreceptor function determined by electroretinogram. Amer Assoc of Ped Ophthal and Strabismus. April 2009, San Francisco, CA
20. **Elias, ER**, Braverman RS, Tong, S, A Novel Treatment for Retinal Dysfunction in patients with the Smith-Lemli-Opitz Syndrome. American College of Medical Genetics. Albuquerque, NM, March 2010
21. **Elias, ER**, Moen A, Lichtenfels J, Enzenauer R, Gessner J, McGavran, L. A Child with Peter’s anomaly and a deletion of 6p25.3 involving FOXC1. American College of Medical Genetics, Vancouver, BC March 2011
22. Eroglu Y, Nguyen-Driver M, Freeman K, Merkens L, Merkens M< Roullet J-B, **Elias E,** Sarphare G, Porter FD, Tierney E, Steiner R. Smith-Lemli-Opitz Syndrome with Normal IQ. PAS/ASPR Denver, CO May 2011
23. Zakerani, N, Nguyen-Driver, M, Eroglu, Y, Freeman, K, Roullet, JB, **Elias, ER**, Sarphare, G, Porter, F, Tierney, E, & Steiner, R. Cognitive Functioning in Children with Smith-Lemli-Opitz Syndrome: a Descriptive Case Series, Society for Pediatric Psychology, San Antonio, TX, April 2011.
24. Braverman, RS, **Elias ER,** Treatment of Smith-Lemli-Opitz syndrome and retina function determined by electroretinography**.** AAPOS meeting March 2012
25. **Elias, ER**, Sharer, Gunter, Gardiner, Katheleen. Genetic Etiologies of Autism Spectrum Disorder in patients with Down Syndrome ACMG March 2012
26. Alia Broman, **Elias, ER**, A Novel cause of Ehlers-Danlos Syndrome associated with a 2q duplication. ACMG March 2012
27. **Elias, ER.** Beyond Cholesterol: Antioxidant Treatment for patients with Smith-Lemli-Opitz Syndrome. ASHG Nov 2012 – platform presentation
28. **Elias, ER.** Autism associated with an Xq12 deletion involving the gene OPHN1 – importance of pursuing a genetic etiology for an Autism Spectrum Disorder (ASD) ASHG Oct 2013
29. **Elias ER,** Swisshelm, K. Thirty-month-old boy with feeding problems, severe sleep disorder, mild developmental disabilities and a maternally inherited deletion of 13q14.11-q14.2 involving *HTR2A* ACMG March 2014
30. S.K. Shapira, L.H. Tian, A.S. Aylsworth, **E.R. Elias**, J.E. Hoover-Fong, N.J. Meeks, M.C. Souders, A.C.-H. Tsai, E. H. Zackai, A.A. Alexander and L.A. Schieve; Development of a Novel Protocol for Characterizing Dysmorphology to Enhance the Phenotypic Classification of Autism Spectrum Disorder, presented at IMFAR, May 2014
31. **Elias ER**, Stille C. Genetic Disorders in Children with Medical Complexity, ACMG, Salt Lake City, Utah, March 2015
32. Apple, B,Bellus, G, March, J, Oleszek, J, Miller, N**, ER Elias.**A child with a novel mutation in WNT5A causing Robinow syndrome. ACMG, Salt Lake City, Utah, March 2015
33. Wigby, K, D’Epagnier, C, CordeiroL, Howell, S, **Elias, ER,** Tartaglia, N. Expanding the phenotype of triple X syndrome: A comparison of prenatal vs postnatal diagnosis. ACMG, Salt Lake City, Utah, March 2015 - platform
34. Angulo, AS, **Elias ER,** Developmental Delay and Skeletal Dysplasia associated with a Maternally Inherited Deletion of 3p26.3 with a phenotypically normal mother, ASHG, Baltimore, MD, Oct 2015
35. Pointon, T, Hayes, D, **Elias ER**, Auditory pathway findings in Smith-Lemli-Opitz syndrome (SLOS), ASHG, Baltimore, MD, Oct 2015
36. Hung-Chun Yu, Curtis R. Coughlin II, Elizabeth A. Geiger, **Ellen R. Elias**, Jean L. Cavanaugh, Shelley D. Miyamoto, Tamim H. Shaikh.Discovery of a deleterious variant in *TMEM87B* in a patient with a hemizygous 2q13 microdeletion, uncovers a recessive condition characterized by severe congenital heart defects, ASHG, Baltimore, Oct 2015
37. **Ellen R. Elias,** Bethany Tucker, Severe Osteogenesis (OI) in 3 Vietnamese Siblings with a Homozygous Mutation in Prolyl 3-Hydroxylase 1 (P3H1), also known as LEPRE 1, ASHG, Vancouver, BC, Oct 2016
38. Karen Swisshelm, Susan Toomey, Jamie LeRoux, Rachel Rowe, Kieu Ha, Billie Carsten and **Ellen R Elias,** Co-existence of a complex three-way translocation with and a 4.6 Mb deletion in 8q22.3-8q23.1. ASHG, Vancouver, BC, Oct 2016
39. **Ellen R Elias,** Phil Zeitler, Michael Wang, Value of updated genetic testing: a case report, ACMG, Phoenix, AZ, Mar 2017