

**The Faculty of Medicine of Harvard University
Curriculum Vitae**

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Education

1999	B.S. summa cum laude	Biological Anthropology and Anatomy	Duke University
2006	M.D.	Medicine	Harvard Medical School
2006	M.P.P.	Public Policy	Harvard Kennedy School

Postdoctoral Training

07/06-06/07	Internship	Pediatrics	Boston Combined Residency Program in Pediatrics (Children's Hospital Boston & Boston Medical Center)
07/07-06/09	Residency	Pediatrics	Boston Combined Residency Program in Pediatrics (Children's Hospital Boston & Boston Medical Center)
07/09-06/12	Fellowship	Medical Genetics	Harvard Medical School American Board of Medical Genetics Training Program, Genetics (Children's Hospital Boston, Massachusetts General Hospital, Brigham & Women's Hospital, Dana Farber Cancer Institute)

Faculty Academic Appointments

2011-2013	Instructor	Pediatrics	Harvard Medical School
2013-2017	Assistant Professor	Pediatrics	Harvard Medical School
2018-	Associate Professor	Pediatrics	Harvard Medical School

Appointments at Hospitals/Affiliated Institutions

07/06-06/09	Clinical Fellow	Pediatrics	Children's Hospital Boston
07/06-06/09	Teaching Fellow	Pediatrics	Boston University School of Medicine
07/09-06/12	Clinical Fellow	Genetics	Children's Hospital Boston, Brigham & Women's Hospital, Massachusetts General Hospital, Dana Farber Cancer Institute
07/12-12/16	Assistant in Pediatrics	Pediatrics	Massachusetts General Hospital
12/16-	Assistant Pediatrician	Pediatrics	Massachusetts General Hospital
04/13-	Affiliate Faculty Member	Center for Human Genetics Research	Massachusetts General Hospital
10/13-	Member	Leukemia Program	Dana-Farber/Harvard Cancer Center
08/16-	Associate Member	Ariadne Labs	Harvard Medical School/Brigham & Women's Hospital
10/18-	Emma Campbell Endowed Chair on Down Syndrome	Pediatrics	Massachusetts General Hospital

Other Professional Positions

2003-2006	Program Coordinator, More to Life Than Genes	Harvard Medical School
1999-2004	National Chair, Disability Task Force	American Red Cross

2001-2015	Board of Directors (Chair 2009-2011)	Massachusetts Down Syndrome Congress
2001-2008	Board of Directors	Massachusetts Developmental Disabilities Council
2004-2007	National Board of Governors	American Red Cross
2005-2007	Clinical Director, Health Promotion	Special Olympics
2007-2013	Professional Advisory Council	National Down Syndrome Congress
2007-2017	Board of Directors	Band of Angels Foundation
2007-2012	Secretary, Board of Directors	National Down Syndrome Society
2007-2012	Chair, Clinical Advisory Board	National Down Syndrome Society
2010-2011	Medical Advisor, Board of Directors	Lettercase
2015-	Chair, Medical and Science Advisory Council	Massachusetts Down Syndrome Congress

Major Administrative Leadership Positions

Local

2012-2017	Clinical Co-Director, Down Syndrome Program	Massachusetts General Hospital
2018-	Clinical Director, Down Syndrome Program	Massachusetts General Hospital

Committee Service

Local

2007-2009	Advocacy in Clinical Training (ACT) Committee	Children's Hospital Boston
2012-	T32 Steering Committee	Harvard Medical School American Board of Medical Genetics Training Program
2018	Pilot Feasibility Studies Selection Committee	Massachusetts General Hospital <i>for</i> Children

National

2017-	Council Member, Down Syndrome and Alzheimer's Disease Professional Interest	Alzheimer's Association International Society to Advance Alzheimer's Research
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Area

and Treatment (ISTAART)

Professional Societies

2006-2009	American Academy of Pediatrics	Member
2009-	American College of Medical Genetics and Genomics (ACMG)	Member
2012 and 2016		Member, Working Group for ACMG Consensus statement on Noninvasive Prenatal Screening
2009-	American Society of Human Genetics (ASHG)	Member
2012-	Down Syndrome Medical Interest Group	Member
2015-	Society for Pediatric Research	Member
2015-	T21 Research Society	Member
2017-		Member, Down Syndrome Developmental Clinical Committee

Editorial Activities

Ad hoc Reviewer

Advances in Therapy

American Journal of Bioethics – Primary Research

American Journal on Intellectual and Developmental Disabilities

American Journal of Medical Genetics, Part A

American Journal of Medical Genetics, Part B

American Journal of Medical Genetics, Part C

American Journal of Obstetrics and Gynecology

Archives of Diseases in Childhood

BMC Health Services Research

British Journal of Obstetrics & Gynecology

Child: Care, Health & Development

CNS Drugs

European Journal of Human Genetics

Frontiers Psychiatry

Genetics in Medicine

Journal of Alzheimer's Disease

Journal of Genetic Counseling

Journal of Intellectual Disability Research

Journal of Perinatology
Journal of Psychology and Psychotherapy
Journal on Developmental Disabilities
Neuropsychiatric Disease and Treatment
New England Journal of Medicine
Obstetrics & Gynecology
Pediatrics
Pediatric Drugs
Pediatric Quality and Safety
Perspectives in Biology and Medicine
PLOS ONE
Research in Developmental Disabilities
The CRISPR Journal
The Journal of Pediatrics
The Lancet Psychiatry
Therapeutics and Clinical Risk Management
Universal Access in the Information Society
Vaccine

Honors and Prizes

1997-2001	Angier B. Duke Memorial Scholarship	Duke University	Merit scholarship for top students
1999	Lord Rothermere Scholarship	Duke University/Oxford University	Merit scholarship for international study
2001	All American Academic First Team	USA Today	Top college students in nation
2001	Faculty Scholar	Duke University	Top merit award to graduating seniors
2004-2005	Jack Kent Cooke Scholarship	Jack Kent Cooke Foundation	Top merit award to graduating seniors
2004	Knowles Fellow	Harvard University	Top merit award
2004	Margaret Schneider Austin Scholarship	Harvard Kennedy School	Top merit award
2006	Presidential Scholars Public Service Initiative Award	Harvard Medical School	Recognizing impressive public service
2008	Dean's Community Service Award	Harvard Medical School	Recognizing leadership through service

2010-2014	NIH Pediatric Loan Repayment Award	National Institutes of Health	Pediatric research
2012	Good Catch Award	Children's Hospital Boston	Excellence in clinical safety
2015	William K. Bowes Jr. Award in Medical Genetics	Partners Healthcare Personalized Medicine	Recognizing an outstanding physician who has recently completed training in medical genetics

Report of Funded and Unfunded Projects

Funding Information

Past

1997-2006	Literacy Research in Children with Rett Syndrome Duke University institutional grant Co-P.I. (\$5,000) Investigated the communicative use of storybooks in children with Rett syndrome		
1997-2008	Memory Formation in Amnesic Patient, H.M. American Foundation of Aging Research P.I. (\$8,000) Studied the preoperative and postoperative semantic memories in amnesic patient, H.M.		
1999-2001	Neuroanatomical Research on the Mammalian Tongue Howard Hughes Research Fellowship P.I. Analyzed the neuromuscular development of the mammalian tongue, using mouse models for Down syndrome		
2001-2005	Prenatal and Postnatal Support Services for Mothers of Children with Down Syndrome in Spain Mental Health & Disability Scholarship, Children's Hospital Boston; Rosemary F. Dybwad International Fellowship P.I. Investigated maternal perceptions of receiving a prenatal or postnatal diagnosis of Down syndrome in Spain		
2001-2005	Prenatal and Postnatal Support Services for Mothers of Children with Down Syndrome in U.S. Tim White Foundation, Children's Hospital Boston P.I. (\$15,000) Investigated maternal perceptions of receiving a prenatal or postnatal diagnosis of Down		

syndrome in U.S.

- 2006-2009 Family Attitudes Toward Persons with Down Syndrome
Fred Lovejoy Housestaff Research and Education Fund, Joel and Barbara Alpert
Scholarship, Tim White Foundation
P.I. (\$51,000)
Surveyed mothers, fathers, siblings, and people with Down syndrome across the country
about family life when someone has Down syndrome
- 2010-2012 Predicting Obstructive Sleep Apnea in Down Syndrome
NIH/T32; Sircar/Dynan Fund, Division of Genetics, Children's Hospital Boston
P.I. (\$85,000)
Developing a screening tool that could be used by physicians to predict obstructive sleep
apnea in Down syndrome
- 2011-2013 Predicting Obstructive Sleep Apnea in Down Syndrome
NICHD/F32 HD068101-01A1
P.I. (\$117,432)
Developing a screening tool that could be used by physicians to predict obstructive sleep
apnea in Down syndrome
- 2013-2014 A Screening Protocol to Assess Adult and Adolescent Individuals with Down syndrome
for Eligibility to Participate in an Upcoming Study to Evaluate the efficacy, safety and
tolerability of RO5186582 (Study BP28947)
F. Hoffmann-La Roche Ltd.
Site P.I. (\$36,844)
Industry-initiated Screening Protocol for Phase 2 clinical drug trial on a pharmaceutical
that might enhance cognition, memory, and language in adults with Down syndrome
- 2013-2015 A 4-Week Randomized, Double-Blind, Placebo-Controlled, Phase 2a Safety and
Pharmacokinetic Study of Oral ELND005 in Young Adults With Down Syndrome Without
Dementia
Transitions Therapeutics
Site P.I. (\$201,455)
Industry-initiated Phase 2a clinical drug trial on a pharmaceutical that might enhance
cognition, memory, and language in adults with Down syndrome and stave off
Alzheimer's disease related dementia
- 2015-2016 A Randomized, Double-blind, Placebo-controlled, Parallel Group 26-week Dose-
Investigating Study to Explore the Pharmacokinetics, Pharmacodynamic Effects, Efficacy,
Safety and Tolerability of RO5186582 in Children with Down Syndrome Aged 6-11 Years
(Study WP28760)
F. Hoffmann-La Roche Ltd.
Site P.I. (\$150,282)
Industry-initiated Phase 2 clinical drug trial on a pharmaceutical that might enhance
cognition, memory, and language in children with Down syndrome
- 2015-2016 A Multicenter, Longitudinal, Non-drug Study to Assess the Suitability of Neurocognitive

Tests and Functioning Scales for the Measurement of Cognitive and Functioning Changes in Children with Down Syndrome (Study BP29589)
F. Hoffmann-La Roche Ltd.
Site P.I. (\$169,011)
Industry-initiated Phase 2 clinical drug trial looking at appropriate neurocognitive measures in children with Down syndrome

2014-2016 A Multi-Center, Randomized, Double-Blind, Placebo-Controlled Phase 2 Study of the Efficacy, Safety and Tolerability of RO5186582 in Adults and Adolescents with Down Syndrome (Study BP27832)
F. Hoffmann-La Roche Ltd.
Site P.I. (\$789,910)
Industry-initiated Phase 2 clinical drug trial on a pharmaceutical that might enhance cognition, memory, and language in adults with Down syndrome

2012-2016 Predicting Obstructive Sleep Apnea in Down Syndrome
HRSA/R40MC25322-01-00
P.I. (\$935,540)
Developing a screening tool that could be used by physicians to predict obstructive sleep apnea in Down syndrome

2016-2019 Virtual Evidence-based Healthcare for Underserved Patients with Down Syndrome
Patient-Centered Outcomes Research Institute (PCORI)/AD-1507-31567
P.I. (\$2,100,000)
Through a randomized-control national trial, we will be creating and testing a direct-to-public, always-available, online service yielding comprehensive yet personalized recommendations for patients with Down syndrome who don't have access to specialty care.

Current

2016-2019 Making Activity and Nutritional Goals Obtainable (MANGO)
Internal award, Massachusetts General Hospital & Research Grant from Google, Inc.
P.I. (\$95,000)
Through use of focus groups and design workshops, we are creating of a mobile app for patients with Down syndrome so that they can improve their nutrition and exercise habits.

2017-2020 A Phase 1b Multi-Center, Double-Blind, Randomized, Placebo-Controlled Dose Escalation Study of the Safety, Tolerability, and Immunogenicity of ACI-24 in Adults with Down Syndrome
NIH/1R01AG047922-01
Site P.I.
NIH-funded Phase 1b clinical drug trial on a vaccine that might prevent Alzheimer's disease in patients with Down syndrome.

2019-2021 Longitudinal Investigation For the Enhancement of Down Syndrome Research (LIFE-DSR)
LuMind IDSC Foundation
Co-I (\$64,636)

The LIFE-DSR study is an observational investigation with the goal to better understand the cognitive and behavioral changes along with the health issues found in adults with Down syndrome as they progress toward Alzheimer's disease.

Report of Local Teaching and Training

Teaching of Students in Courses

2015-2016	Human Genetics Course 1 st year MD and PhD students in Health, Sciences, & Technology Program	Harvard Medical School and Massachusetts Institutes of Technology Presented 90-minute lecture on prenatal testing for Down syndrome and how best to deliver prenatal diagnoses to expectant parents
2015-2018	PhD Program in Biological and Biomedical Sciences 1 st year PhD students	Harvard Medical School Organized and presented 3-hour course on Down syndrome
2018	AISC-Human Genetics course 1 st year MD students	Harvard Medical School Preceptor
2019-	The Developing Physician, The Practice of Medicine 1 st year MD students	Harvard Medical School Course instructor

Formal Teaching of Residents, Clinical Fellows and Research Fellows (post-docs)

2009-current	Introduction to Metabolic Disorders 1 st year medical genetics fellows	Children's Hospital Boston, Massachusetts General Hospital, Brigham & Women's Hospital, Dana Farber Cancer Institute One-hour lecture
2009	A Challenging Dynamic: Presenting a Diagnosis of Down Syndrome to New and Expectant Parents Med-Peds Residency Program	Massachusetts General Hospital One-hour lecture
2013	Healthcare Updates for Patients with Down Syndrome Pediatrics residents (all years)	Massachusetts General Hospital One-hour lecture
2014	Newborn Care for Patients with Down Syndrome Pediatrics residents (all years)	Massachusetts General Hospital One-hour lecture

2016	A Challenging Dynamic: Presenting a Diagnosis of Down Syndrome to New and Expectant Parents Obstetric Residents and Fellows	Brigham & Women's Hospital One-hour lecture
2019	Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional Genetic counseling graduate students	Massachusetts General Hospital Institutes of Healthcare Professions One-hour lecture

Clinical and Supervisory Training Responsibilities

2012-current	Down Syndrome Program Clinic Preceptor / Massachusetts General Hospital	One half session per week
2012-current	Medical Genetics Clinic Preceptor / Massachusetts General Hospital	One half session per week
2012-current	Medical Genetics and Metabolism Inpatient Services Consultation Supervising Attending / Massachusetts General Hospital	2 hours/day for one week every 2 months

Laboratory and Other Research Supervisory and Training Responsibilities

2011-2012	Supervision of Research Assistant for Division of Developmental and Behavioral Pediatrics, Children's Hospital Boston	5-10 hours/week for 12 months
2012-current	Supervision of Research Assistant and Clinical Research Coordinator for the MassGeneral Hospital Down Syndrome Program	5-10 hours/week supervision
2016-current	Supervision of Program Manager for the MassGeneral Hospital Down Syndrome Research Program	5 hours/week supervision

Formally Supervised Trainees

2011-2012	Laura Kline / Genetic Counseling Student, Brandeis University Thesis approved; abstract presentation at National Society of Genetic Counselors
2011-2013	Timothy Ridge / Clinical Psychology, Massachusetts Professional School of Psychology Thesis approved
2014-2015	Devon Haynes / Genetic Counseling Student, University of South Carolina School of Medicine Thesis approved

- 2015-2016 Alex Ilacqua / Genetic Counseling Student, The Ohio State University
Thesis approved
- 2015-2017 Alexandra Garza-Flores, M.D. / Medical Genetics Resident, Harvard Medical School
American Board of Medical Genetics Training Program
NIH F32 submitted; oversaw the implementation of her clinical research project
- 2016; 2017
(summer) Sierra Weiss / Undergraduate student, Emory University
Created multiple patient and family handouts for our Massachusetts General Hospital
Down Syndrome Program
- 2016-2017 Ibrahim Elsharkawi, MB, BCH / Research Fellow, Massachusetts General Hospital
Multiple manuscripts published
- 2016-2018 Maggie Salinger / MPP Candidate at Harvard Kennedy School and MD Candidate at
Emory University School of Medicine
Developing business analysis of the financial sustainability for our Massachusetts General
Hospital Down Syndrome Program
- 2017
(summer) Sophia Campbell / Undergraduate student, Harvard University
Created multiple patient and family handouts for our Massachusetts General Hospital
Down Syndrome Program
- 2017
(summer) Elaina Ramos / MD Candidate at Boston University Medical Center
Created multiple patient and family handouts for our Massachusetts General Hospital
Down Syndrome Program
- 2018 Courtney Verscaj / MD Candidate at Boston University Medical Center
Created an online CME-course through Harvard Medical School for primary care
physicians to learn about medical care for patients with Down syndrome
- 2018 Daniel Kats / MD Candidate at Case Western University Medical Center
Developed a survey tool to measure human rights for people with Down syndrome across
the world; also conducted structured phone interviews with families whose children have
Down syndrome and infantile spasms

Formal Teaching of Peers (e.g., CME and other continuing education courses)

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| 2010- | Down Syndrome: Healthcare Updates for the Primary
Care Pediatrician
Harvard Medical School | Single presentation

Online CME course |
| 2010- | What the Other Children Without Down Syndrome
Are Thinking: Sibling Issues for the Pediatrician | Single presentation |

	Nemours/Alfred I. duPont Hospital for Children	Online CME course
2013-	Prenatal Diagnoses of Down Syndrome: Delivering Results in Our New Age of Genetic Testing Massachusetts General Hospital Academy	Single presentation Online CME course
2013-2016	Keeping Children and Adolescents with Down Syndrome Healthy: Medical Updates for the Primary Care Physician Harvard Medical School 35 th Annual Primary Care Pediatrics Conference	Single presentation CME course
2015	Prenatal Diagnoses of Down Syndrome: Delivering Results in Our New Age of Genetic Testing Harvard Center for Bioethics Conference: Families Matter Ethically, Legally, and Clinically	Two presentations CME course

Local Invited Presentations

No presentations below were sponsored by outside entities

2007	Prenatal and Postnatal Medical Support: Capturing the Sentiments of Parents Who Have Children with Down Syndrome / Lecture Division of Genetics, Massachusetts General Hospital
2008	A Challenging Dynamic: Delivering and Prenatal and Postnatal Diagnosis of Down Syndrome to New and Expectant Parents / Lecture Department of Newborn Medicine, Children's Hospital Boston
2009	Prenatal Diagnosis of Down Syndrome: New Advances and Updates for Healthcare Professionals / Lecture Division of Genetics, Massachusetts General Hospital
2009	Fasten Your Seatbelt: A Crash Course on Down Syndrome for Brothers and Sisters / Lecture Department of Pediatrics, Children's Hospital Boston
2009	Multiple Options on a Changing Menu / Walk Rounds Division of Genetics, Children's Hospital Boston
2009	A Dip in Time / Walk Rounds Division of Genetics, Children's Hospital Boston
2009	A Challenging Dynamic: Presenting a Diagnosis of Down Syndrome to New and Expectant Parents / Lunchtime Talk Pediatric Advanced Care Team, Children's Hospital Boston

- 2009 Sharing the Diagnosis of Neurofibromatosis / Seminar
Harvard Medical School Center for Neurofibromatosis and Allied Disorders
- 2009 Prenatal Diagnosis of Down Syndrome: New Advances and Updates for Healthcare Professionals / Grand Rounds
Department of OB/GYN, Mount Auburn Hospital, Cambridge, MA
- 2010 Making a Diagnosis of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Department of OB/GYN, Massachusetts General Hospital
- 2010 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Grand Rounds
Department of OB/GYN, Beth Israel Deaconess Medical Center
- 2010 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Grand Rounds
Department of Pediatrics, Mt. Auburn Hospital, Cambridge, MA
- 2010 A Second Look: Jacobsen syndrome / Walk Rounds
Division of Genetics, Children's Hospital Boston
- 2010 Down Syndrome / Lecture
Partners Healthcare Center for Personalized Genetic Medicine
- 2010 What your other children without Down syndrome are thinking: Sibling Issues for Parents / Allen C. Crocker Lecture Series
Division of Developmental and Behavioral Pediatrics, Children's Hospital Boston
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Grand Rounds
Department of OB/GYN, Cambridge Hospital
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Children's Hospital Boston Community Outreach Conference
- 2011 Down Syndrome Healthcare Updates for Older Children / Lecture
Division of Developmental and Behavioral Pediatrics, Children's Hospital Boston
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Department of Neonatology, Children's Hospital Boston
- 2011 Down syndrome / Lecture
Partners Healthcare Center for Personalized Genetic Medicine
- 2011 Community Partnerships / Panel Discussion

Harvard Medical School Center for Neurofibromatosis and Allied Disorders

- 2011 Keeping Children and Adolescents with Down Syndrome Healthy / Lecture
Division of Nutrition, Children's Hospital Boston
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates
for the Healthcare Professional / Lecture
Department of Maternal Fetal Medicine, Brigham and Women's Hospital
- 2011 Predicting Obstructive Sleep Apnea in Down Syndrome / Lecture
Division of Genetics, Children's Hospital Boston
- 2012 Predicting Obstructive Sleep Apnea in Down Syndrome / Lecture
Division of Genetics, Massachusetts General Hospital
- 2012 Predicting Obstructive Sleep Apnea in Down Syndrome / Lecture
Division of Sleep Medicine, Children's Hospital Boston
- 2012 Down Syndrome / Lecture
Partners Healthcare Center for Personalized Genetic Medicine
- 2012 Behavioral Concerns in Children and Adolescents with Down Syndrome: A
comprehensive approach / Lecture
MassGeneral Hospital Psychology Assessment Center
- 2012 Keeping Children and Adolescents with Down Syndrome Healthy: All the Updates that
Healthcare Professionals Need to Know / Grand Rounds
Department of Pediatrics, Massachusetts General Hospital
- 2012 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates
for the Healthcare Professional / Grand Rounds
Department of Pediatrics, Newton-Wellesley Hospital
- 2013 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates
for the Healthcare Professional / Grand Rounds
Department of Perinatal Services / Massachusetts General Hospital
- 2013 Keeping Children and Adolescents with Down Syndrome Healthy: All the Updates that
Healthcare Professionals Need to Know / Grand Rounds
Department of Audiology / Massachusetts Eye and Ear Infirmary
- 2013 Prenatal Diagnoses of Down Syndrome, Delivering Results in our New Age of Genetic
Testing / Grand Rounds
Pediatrics Department, Cambridge Hospital, Cambridge, MA
- 2014 Keeping Children and Adolescents with Down Syndrome Healthy: Medical Updates for
the Primary Care Physician / Grand Rounds
Division of Genetics, Massachusetts General Hospital, Boston, MA

- 2014 Keeping Children and Adolescents with Down Syndrome Healthy: Medical Updates for the Primary Care Physician / Grand Rounds
North Shore Medical Center, Salem, MA
- 2015 Representations of Down Syndrome in Art / Grand Rounds
Division of Genetics, Massachusetts General Hospital, Boston, MA
- 2015 Prenatal Diagnoses of Down Syndrome, Delivering Results in our New Age of Genetic Testing / Lecture
Division of Newborn Medicine, Brigham & Women's Hospital, Boston, MA
- 2015 Prenatal Diagnoses of Down Syndrome, Delivering Results in our New Age of Genetic Testing / Lecture
Division of Neurology, Department of Pediatrics, Massachusetts General Hospital, Boston, MA
- 2016 Keeping Children and Adolescents with Down Syndrome Healthy: Medical Updates for the Primary Care Physician / Grand Rounds
Division of Endocrinology, Department of Pediatrics, Massachusetts General Hospital, Boston, MA
- 2016 Prenatal Diagnoses of Down Syndrome, Delivering Results in our New Age of Genetic Testing / Lecture
Division of Neurology, Department of Pediatrics, Massachusetts General Hospital, Boston, MA
- 2017 Creating a Fulfilling Career in Academic Medicine: Four Decision Points / Lecture
Pediatric Intensive Care Unit, Department of Pediatrics, Massachusetts General Hospital, Boston, MA
- 2017 Sharing a diagnosis: An initial discussion of strategies and lessons learned / Grand Rounds
Division of Medical Genetics, Department of Pediatrics, Massachusetts General Hospital, Boston, MA
- 2018 Down Syndrome Across the Lifespan / Grand Rounds, co-presented with Clo Cottrell
Department of Social Work, Massachusetts General Hospital, Boston, MA
- 2019 Keeping Children and Adolescents with Down Syndrome Healthy: Medical Updates for the Primary Care Physician / Grand Rounds
Division of Endocrinology, Department of Pediatrics, Massachusetts General Hospital, Boston, MA
- 2019 Down Syndrome Clinic to You (DSC2U): An Online Personalized Care Plan for Caregivers and Primary Care Providers / Lecture
Ariadne Labs, Brigham & Women's Hospital, Boston, MA
- 2019 Down Syndrome Clinic to You (DSC2U): An Online Personalized Care Plan for

Caregivers and Primary Care Providers / Lecture, MGHfC Research Day
Department of Pediatrics, Massachusetts General Hospital, Boston, MA

2019 Strategic Planning for the Future
Cystic Fibrosis Program, Department of Pediatric Pulmonology, Massachusetts General Hospital, Boston, MA

Report of Regional, National and International Invited Teaching and Presentations

Presentations sponsored by outside entities indicated in brackets after talk.

Invited Presentations and Courses

Regional

- 2007 Prenatal and Postnatal Medical Support: Capturing the Sentiments of Parents Who Have Children with Down Syndrome / Grand Rounds
Department of OB/GYN, Boston University Medical Center, Boston, MA
- 2008 A Challenging Dynamic: Delivering a Diagnosis of Down Syndrome to New and Expectant Parents / Grand Rounds
Hartford Hospital & University of Connecticut Medical Center, Hartford, CT
- 2009 A Challenging Dynamic: Delivering a Diagnosis of Down Syndrome to New and Expectant Parents / Grand Rounds
Hasbro Children's Hospital, Providence, RI
- 2009 Brothers and Sisters Who Have Siblings with Down Syndrome: Questions and Answers / Lecture
Department of Pediatrics, Boston University Medical Center, Boston, MA
- 2009 A Challenging Dynamic: Delivering and Prenatal and Postnatal Diagnosis of Down Syndrome to New and Expectant Parents / Grand Rounds
Department of OB/GYN, South Shore Hospital, South Weymouth, MA
- 2009 Prenatal Diagnosis of Down Syndrome: New Advances and Updates for Healthcare Professionals / Grand Rounds
Department of Pediatrics, Tufts Medical Center, Boston, MA
- 2010 Best Practices in Healthcare Communication and Relationships / Lecture
Department of Pediatrics, Tufts Medical Center, Boston, MA
- 2010 New Prenatal Tests for Down Syndrome: Everything You Need to Know and Should be Doing / Grand Rounds
Department of Family Medicine and Community Health, UMass Memorial, Boston, MA
- 2010 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates

- for the Healthcare Professional / Lecture
Department of Genetic Counseling, Boston Medical Center, Boston, MA
- 2010 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Grand Rounds
Genetic Counseling Department, Brandeis University, Boston, MA
- 2010 New Prenatal Testing for Down Syndrome: Everything You Need to Know and Should be Doing / Grand Rounds
Department of OB/GYN, Tufts Medical Center, Boston, MA
- 2010 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Grand Rounds
Department of OB/GYN, Boston University Medical Center, Boston, MA
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates / Lecture
Brown University, Providence, RI
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Lecture
Women and Infant's Hospital, Providence, RI
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Grand Rounds
Department of OB/GYN, South Shore Hospital, South Weymouth, MA
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Grand Rounds
Department of OB/GYN, Winchester Hospital, Winchester, MA.
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Division of Genetic Counseling, Brandeis University, Waltham, MA
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates / Lecture
Boston College, Boston, MA
- 2011 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Physicians Need to Know / Lecture
Department of Pediatrics, BayState Health, Springfield, MA
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Grand Rounds
Department of Pediatrics, BayState Health, Springfield, MA
- 2012 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates

for Today's Physicians / Grand Rounds
OB/GYN Department, Lowell General Hospital, Lowell, MA

- 2012 Best Practices in Healthcare Communication and Relationships / Lecture
Tufts Medical School, Boston, MA
- 2012 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates
for the Healthcare Professional / Lecture
Boston University School of Medicine, Boston, MA
- 2012 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Physicians Need to Know / Lecture
University of New England College of Osteopathic Medicine, Biddeford, ME
- 2013 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates
for the Healthcare Professional / Lecture
Boston University School of Medicine, Boston, MA
- 2013 Prenatal Diagnoses of Down Syndrome, Delivering Results in our New Age of Genetic
Testing / Grand Rounds
OB/GYN & Pediatrics Departments, MetroWest Medical Center, Framingham, MA
- 2013 Prenatal Diagnoses of Down Syndrome, Delivering Results in our New Age of Genetic
Testing / Lecture
Center for Biotechnology and Biomedical Sciences, Merrimack College, North Andover,
MA
- 2014 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates
for the Healthcare Professional / Lecture
Boston University School of Medicine, Boston, MA
- 2014 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates
for the Healthcare Professional / Lecture
South Shore Hospital, South Weymouth, MA
- 2015 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Physicians Need to Know / Lecture
Bridgewater State University, Bridgewater, MA
- 2015 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates
for the Healthcare Professional / Lecture
Boston University School of Medicine, Boston, MA
- 2015 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Physicians Need to Know / Lecture
Tufts University Medical Center, Boston, MA
- 2015 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical

Updates that Clinicians Need to Know
Boston Medical Center Primary Care Conference, Boston, MA

- 2015 Prenatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Grand Rounds
Pediatrics Department, Boston Medical Center, Boston, MA
- 2015 Prenatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Grand Rounds
Department of OB/GYN, South Shore Hospital, South Weymouth, MA
- 2016 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Physicians Need to Know / Lecture
Tufts University Dental School, Boston, MA
- 2016 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Boston University School of Medicine, Boston, MA
- 2017 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Boston University School of Medicine, Boston, MA
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Physicians Need to Know / Lecture
Tufts University Dental School, Boston, MA
- 2017 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Boston University School of Medicine, Boston, MA
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Physicians Need to Know / Lecture
Edmund Ervin Pediatric Center, MaineGeneral Medical Center, Waterville, ME
- 2017 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows
Edmund Ervin Pediatric Center, MaineGeneral Medical Center, Waterville, ME
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Physicians Need to Know / Lecture
Bridgewater State University, Bridgewater, MA
- 2018 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Boston University School of Medicine, Boston, MA
- 2018 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Grand Rounds

OB/GYN Department, St. Elizabeth's Medical Center, Brighton, MA

- 2018 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Boston University School of Medicine, Boston, MA
- 2019 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Physicians Need to Know / Lecture
Tufts University Dental School, Boston, MA
- 2019 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Boston University School of Medicine, Boston, MA
- 2019 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Physicians Need to Know / Lecture
Franciscan Children's Hospital, Brighton, MA
- 2019 Down Syndrome Clinic to You (DSC2U): A New Virtual Clinic for All Families
Massachusetts Institutes of Technology, Boston, MA

National

- 2008 Compassionate Delivery of a New Diagnosis to New and Expectant Parents of Children with Down Syndrome / Lecture
Vanderbilt Kennedy Center & Vanderbilt Medical School, Nashville, TN
- 2008 Brothers and Sisters Who Have Siblings with Down Syndrome: Questions and Answers / Lecture
Department of Pediatrics, Vanderbilt Medical Center, Nashville, TN
- 2008 Prenatal and Postnatal Medical Support: Capturing the Sentiments of Parents Who Have Children with Down Syndrome / Grand Rounds
Department of Obstetrics and Gynecology, Vanderbilt Medical Center, Nashville, TN
- 2008 Prenatal and Postnatal Medical Support: Capturing the Sentiments of Parents Who Have Children with Down Syndrome / Lecture
Riley Hospital for Children, Indianapolis, IN
- 2009 A Challenging Dynamic: Delivering a Diagnosis of Down Syndrome to New and Expectant Parents / Grand Rounds
Children's Mercy Hospital, Kansas City, MO
- 2009 Brothers and sisters who have siblings with Down syndrome: Questions and Answers / Lecture
Children's Mercy Hospital, Kansas City, MO
- 2009 A Challenging Dynamic: Delivering a Diagnosis of Down Syndrome to New and

- Expectant Parents / Lecture
St. Louis University Hospital, St. Louis, MO
- 2009 A Challenging Dynamic: Delivering a Diagnosis of Down Syndrome to New and Expectant Parents / Lecture
St. John's Mercy Hospital, St. Louis, MO
- 2009 A Challenging Dynamic: Delivering a Diagnosis of Down Syndrome to New and Expectant Parents / Grand Rounds
Department of Pediatrics, Christiana Hospital, Wilmington, DE
- 2009 What the Other Children Without Down Syndrome are Thinking: Sibling Issues for the Pediatrician / Lecture
Al DuPont Hospital, Wilmington, DE
- 2009 Making a Diagnosis of Down Syndrome: New Advances and Updates for Healthcare Professionals / Lecture
OB/GYN Department, University Hospitals, Cleveland, OH
- 2009 Making a Diagnosis of Down Syndrome: New Advances and Updates for Healthcare Professionals / Lecture
Pediatrics Department, University Hospitals, Cleveland, OH
- 2009 Accessing Quality Healthcare for your Brother or Sister: New Advances and Updates / Lecture
The Ohio State University, Columbus, OH
- 2009 Prenatal and Postnatal Diagnoses of Down Syndrome / Lecture
U.S. Congress, Washington, D.C.
- 2010 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Grand Rounds
Gerber Center for Infant Development and Nutrition at Helen DeVos Children's Hospital, Grand Rapids, MI
- 2010 Making a Diagnosis of Down Syndrome: New Advances and Updates for Healthcare Professionals / Grand Rounds
Pediatrics Department, Helen DeVos Children's Hospital, Grand Rounds, MI
- 2010 Delivering a Sensitive Diagnosis / Lecture
Helen DeVos Children's Hospital, Grand Rapids, MI
- 2010 Making a Diagnosis of Down Syndrome: New Advances and Updates for Healthcare Professionals / Grand Rounds
Texas Children's Hospital, Houston, TX
- 2010 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Grand Rounds

Department of Pediatrics, Children's Hospital of Wisconsin, Milwaukee, WI

- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: All of the Things that Residents Need to Know / Lecture
The Children's Hospital, Oklahoma University Medical Center, Oklahoma City, OK
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Grand Rounds
Department of OB/GYN, Holy Cross Hospital, Silver Spring, MD
- 2011 What Your Other Children Without Down Syndrome are Thinking: Sibling Issues / Lecture
The Children's Hospital of Philadelphia, Philadelphia, PA
- 2011 Celebrating Life with Down Syndrome / Keynote
The Children's Hospital of Philadelphia, Philadelphia, PA
- 2011 Keeping Children and Adolescents with Down Syndrome Healthy: All of the Medical Updates that Parents Need to Know / Lecture
The Children's Hospital of Philadelphia, Philadelphia, PA
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Lecture
Bergan Mercy Hospital, Omaha, NE
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Lecture
Methodist Women's Hospital, Omaha, NE
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Lecture
Division of Genetics, Duke University Medical Center, Durham, NC
- 2011 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Grand Rounds
Department of Pediatrics, Children's Hospital of Los Angeles, Los Angeles, CA
- 2012 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Lecture
Bergan Mercy Medical Center, Omaha, NE
- 2012 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Grand Rounds
Children's Hospital & Medical Center, Omaha, NE
- 2012 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Cold Spring Harbor Laboratory, Cold Spring Harbor, NY

- 2012 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings with Down Syndrome / Lecture
United Nations, New York, NY
- 2012 Genetics and Obstetrics: New Down Syndrome Testing / Lecture
Nurse Practitioners in Women's Health conference, Orlando, FL
- 2012 Does NextGen sequencing diminish the value of individuals with Down syndrome? / Oral platform
National Society of Genetic Counseling, Boston, MA
- 2012 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Physicians Need to Know / Lecture
East Tennessee Children's Hospital, Knoxville, TN
- 2013 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Grand Rounds
Carolinas Medical Center, Charlotte, NC
- 2013 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Physicians and Parents Need to Know / Lecture
University of Arkansas for Medical Sciences, Little Rock, AR
- 2013 Celebrating Life with Down Syndrome / Keynote
University of Arkansas for Medical Sciences, Little Rock, AR
- 2013 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Lecture
Duke University, Durham, NC
- 2014 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Grand Rounds
Department of Pediatrics, Medical University of South Carolina (MUSC), Charleston, SC
- 2014 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Lecture
College of Charleston, Charleston, SC
- 2014 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Grand Rounds
Department of OB/GYN, Medical College of Wisconsin, Milwaukee, WI
- 2014 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Department of Neonatology, Novant Health Presbyterian Medical Center, Charlotte, NC
- 2014 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic

Testing / Grand Rounds
Department of Pediatrics, University of Wisconsin School of Medicine and Public Health,
Madison, WI

- 2015 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Grand Rounds
Gundersen Healthcare, La Crosse, WI
- 2015 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Gundersen Healthcare, La Crosse, WI
- 2015 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Grand Rounds
Marshfield Clinic, Marshfield, WI
- 2015 Launching a New Clinical Program / Concurrent Session
American College of Medical Genetics and Genomics Annual Conference, Salt Lake City, UT
- 2015 Down syndrome: Caring for Patients Across a Lifespan / Concurrent Session
American College of Medical Genetics and Genomics Annual Conference, Salt Lake City, UT
- 2015 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Grand Rounds
OB/GYN Department, University of Nebraska Medical Center, Omaha, NE
- 2015 Medical Work-up for Regression in Patients with Down Syndrome
Down Syndrome Medical Interest Group National Meeting, Phoenix, AZ
- 2015 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Grand Rounds
OB/GYN Department, Bellin Health Care, Green Bay, WI
- 2015 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Grand Rounds
Division of Medical Genetics, Baylor Medical Center, Houston, TX
- 2016 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Grand Rounds
OB/GYN Department, Christiana Healthcare, Wilmington, DE
- 2016 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Family Medicine Department, St. Francis Healthcare, Wilmington, DE
- 2016 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical

- Updates that Parents Need to Know / Lecture
Department of Pediatrics, Nemours, Wilmington, DE
- 2016 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Family Medicine Department, Christiana Healthcare, Wilmington, DE
- 2016 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Grand Rounds
Department of Pediatrics, Christiana Healthcare, Wilmington, DE
- 2016 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Grand Rounds
Department of Pediatrics, East Tennessee Children's Hospital, Knoxville, TN
- 2016 Building a National Down Syndrome Biobank
Down Syndrome Medical Interest Group national meeting, Orlando, FL
- 2016 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Grand Rounds
Department of Pediatrics, Maimonides Medical Center, Brooklyn, NY
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Cumberland Pediatric Foundation, Nashville, TN
- 2017 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Grand Rounds
OB/GYN Department, Vanderbilt University Medical Center, Nashville, TN
- 2017 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Grand Rounds
OB/GYN Department, University of Rochester Medical Center, Rochester, NY
- 2017 Urinary Biomarkers and Obstructive Sleep Apnea in Patients with Down Syndrome / Lecture
Trisomy 21 Research Society, Northwestern Medical Center, Chicago, IL
- 2017 Down Syndrome & Unexplained Regression
Down Syndrome Medical Interest Group, Sacramento, CA
- 2017 Predicting Obstructive Sleep Apnea in Patients with Down Syndrome
Down Syndrome Medical Interest Group, Sacramento, CA
- 2017 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Lecture
St. Luke's Hospital of Kansas City, Kansas City, MO

- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Grand Rounds
Children's Mercy Hospital, Kansas City, MO
- 2017 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / McBride Lecture
Simpson College, Indianola, IA
- 2017 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing / Grand Rounds
Division of Genetics, University Hospitals, Cleveland, OH
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Grand Rounds
Department of Pediatrics, Cleveland Clinics, Cleveland, OH
- 2017 Delivering a Prenatal Diagnosis of Down Syndrome: Lessons Learned from Evidence-Based Literature / Lecture
5th annual Advances in Prenatal Molecular Diagnostics conference, Boston MA
- 2018 The Latest and Greatest Research on Down Syndrome / Lecture
Department of Pediatrics, Nationwide Children's Hospital, Columbus, OH
- 2018 The Latest and Greatest Research on Down Syndrome / Lecture
Quillen College of Medicine, East Tennessee State University, Johnson City, TN
- 2018 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Clinicians Need to Know / Lecture
Quillen College of Medicine, East Tennessee State University, Johnson City, TN
- 2018 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows / Grand Rounds
Department of Pediatrics, Children's Hospitals and Clinics of Minnesota – Minneapolis Campus, Minneapolis, MN
- 2018 Down Syndrome Clinic to You (DSC2U): An Online Personalized Care Plan for Caregivers and Primary Care Providers / Lecture
Down Syndrome Medical Interest Group Annual Meeting, Dallas, TX
- 2018 Two Years Later: The Implementation of ACMG's NIPS Guidelines / Lecture
6th annual Advances in Prenatal Molecular Diagnostics conference, Boston MA
- 2018 Prenatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers / Lecture
Combined Society Meeting of Obstetricians and Pediatricians, Buffalo, NY
- 2018 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Healthcare Professionals Need to Know / Grand Rounds

Department of Pediatrics, Oishei Children's Hospital, Buffalo, NY

- 2018 Prenatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers / Lecture
Jacobs School of Medicine and Biological Sciences, Buffalo, NY
- 2019 Down Syndrome Clinic to You (DSC2U): An Online Automated Clinic for Patients and Families
American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting, Seattle, WA
- 2019 What does it mean to live with Down syndrome?
AC Immune Key Opinion Leaders meeting, New York City, NY
[sponsored by AC Immune]
- 2019 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Grand Rounds
Department of Obstetrics and Gynecology, Medical College of Wisconsin, Milwaukee, WI
- 2019 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Physicians Need to Know / Lecture
Department of Pediatrics, Mayo Clinic Health System, La Crosse, WI
- 2019 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Department of Pediatrics, University of Wisconsin Health University Hospital, Madison, WI
- 2019 Brain Train: SMART Actions that Might Boost Cognition and Prevent Alzheimer's Disease in Patients with Down Syndrome / Lecture
Waisman Center at University of Wisconsin-Madison, Madison, WI
- 2019 Brain Train: SMART Actions that Might Boost Cognition and Prevent Alzheimer's Disease in Patients with Down Syndrome / Lecture
Department of Pediatrics, Gundersen Healthcare, La Crosse, WI
- 2019 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows / Lecture
Northwell Health 2019 Perinatal Neonatal Nursing Conference, New Hyde Park, NY
- 2019 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Department of Family Medicine, Saint Joseph Hospital, Ft. Wayne, IN
- 2019 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for the Healthcare Professional / Lecture
Central New York Regional Perinatal Program, Crouse Hospital, Syracuse, NY

International

- 2011 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Albert Einstein Hospital, Sao Paulo, Brazil
- 2012 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers
Japan Society of Human Genetics, Tokyo, Japan
- 2012 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers
Japan Association for Bioethics, Kyoto, Japan
- 2013 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers
KK Women's & Children's Hospital, Singapore
- 2013 Celebrating Life with Down Syndrome
Karolinska University Hospital, Stockholm, Sweden
- 2013 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers
Karolinska University Hospital, Stockholm, Sweden
- 2013 Let's Get Real About Down Syndrome: New Research on Family Attitudes
Karolinska University Hospital, Stockholm, Sweden
- 2013 What the Other Children Without Down Syndrome are Thinking: Sibling Issues
Karolinska University Hospital, Stockholm, Sweden
- 2013 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Japan Society for Human Genetics, Sendai, Japan
- 2013 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers / Lecture
St. Luke's International Hospital, Tokyo, Japan
- 2014 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers / Lecture
Shinshu University, Matsumoto, Japan
- 2014 Keeping Children and Adolescents with Down Syndrome Healthy: Building the Down Syndrome Program at Massachusetts General Hospital
Kinki University, Osaka, Japan
- 2015 Predicting Obstructive Sleep Apnea in People with Down Syndrome / Plenary presentation
Trisomy 21 Research Society, Brain and Spine Institute, Hôpital Pitié Salpêtrière, Paris,

France

- 2016 New Prenatal Tests for Down Syndrome: Updates and What This All Means for Professionals and Families / Lecture
Royal Society of Paediatrics and Child Health, Liverpool, United Kingdom
- 2016 Latest and Greatest Research About Down Syndrome
Royal Society of Paediatrics and Child Health, Liverpool, United Kingdom
- 2016 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Healthcare Professionals Need to Know / Lecture
Department of Pediatrics and Adolescent Medicine, Queen Mary Hospital, The University of Hong Kong, China
- 2016 Prenatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers / Lecture
Department of Pediatrics, Ospedale Bambino Gesù, Rome, Italy
- 2017 Prenatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers / Lecture
Aga Khan University Hospital, Karachi, Pakistan
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Healthcare Professionals Need to Know / Lecture
59th Annual Meeting of Japanese Society of Child Neurology, Osaka, Japan
- 2017 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings with Down Syndrome / Lecture
Konan University, Kobe, Japan
- 2018 Prenatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers / Lecture
Onaizah Association for Human Services, Onaizah, Saudi Arabia
- 2018 Latest and Greatest Research About Down Syndrome
Onaizah Association for Human Services, Onaizah, Saudi Arabia
- 2018 Adult Brothers and Sisters Who Have Siblings with Down Syndrome
Onaizah Association for Human Services, Onaizah, Saudi Arabia
- 2019 Prenatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Healthcare Providers / Lecture
Indus Health Network, Karachi, Pakistan
- 2019 Down Syndrome Clinic to You (DSC2U): An Online Personalized Care Plan for Caregivers and Primary Care Providers
International Conference on Communication in Healthcare, San Diego, CA

Report of Clinical Activities and Innovations

Current Licensure and Certification

2004	Passed USMLE Step 1
2006	Neonatal Resuscitation (NALS) Provider
2006	Passed USMLE Step 2
2006	Basic Life Support (BLS) for Healthcare Providers
2006	Pediatrics Advanced Life Support Program (PALS) Provider
2008	Passed USMLE Step 3
2008	Massachusetts Registered Physician
2011	Design and Implementation of Clinical Trials, Harvard Catalyst
2011	American Board of Medical Genetics
2017	Basic Life Support (BLS) for Healthcare Providers

Practice Activities

2012-	Ambulatory Care	Medical Genetics, Massachusetts General Hospital	2-3 sessions per week
2012-	In-patient consultations	Medical Genetics, Massachusetts General Hospital	1 week every 2 months and 1 weekend per month

Clinical Innovations

Creating a Virtual Down Syndrome Clinic (2016 to current)	As the P.I. on this PCORI grant, I am overseeing a multidisciplinary team of clinicians and caregivers from around the country to transform the way that healthcare can be delivered to the overwhelming majority of patients with Down syndrome who do not have access to a specialty clinic like the one that I co-direct at MGH. We will be building a Virtual Clinic, seeking to automate personalized responses to caregivers who answer questions online. We will be testing the efficacy of our model in a randomized, controlled national trial.
Creating a New Screening Tool for Obstructive Sleep Apnea in Patients with Down Syndrome (2010 to current)	As the P.I. on grants from the NIH and HRSA, I am overseeing a team of researchers who have identified a novel assessment that may rule out the presence of significant sleep apnea in patients with Down syndrome. As the future of reimbursements is uncertain in our country, we hope that this simple tool, which can be performed by primary care physicians, can be game changing. This work has been published, and we are currently validating our findings at MGH.
Creating a Mobile App for Nutrition and Exercise Goals for Patients with Down	As the P.I. on an institutional award, I am overseeing a multidisciplinary team of clinicians and software engineers to design a mobile app which will empower patients with Down syndrome, and their caregivers, to

Syndrome (2015 to current)	address obesity and its co-morbidities. We have engaged a team of patients with Down syndrome and caregivers in focus group and design workshops to help create this first-of-its-kind application for a population with intellectual disabilities.
Creation of Electronic intake form at Down Syndrome Program at Massachusetts General Hospital (2014 to current)	This first-ever HIPAA-compliant electronic intake form allows families and caregivers to input clinically relevant information in advance of clinical appointments at the MGH Down Syndrome Program. This resulted in more efficient and effective care and higher satisfaction levels from our patients. The data collected through these intake forms have also been used for quality improvement projects. This intake form has also served as a national model, and we have plans to license the form to other healthcare systems.
Creation of Registry of Patients in Medical Genetics Division at Massachusetts General Hospital (2013 to current)	I created an online, secure registry, using REDCap software, for our Division to track all of the patients seen in our general genetics and metabolism out-patient clinics. This clinical advancement has allowed our Division to perform quality improvement projects and link families whose members have similar diagnoses.
Creation of multidisciplinary Down Syndrome Program at Massachusetts General Hospital (2012 to current)	As co-Director of the MGH Down Syndrome Program, I helped create a now internationally renowned program of clinical excellence. Each year, my team of 15 healthcare professionals see ~500 patients with Down syndrome, of all ages, during carefully coordinates multidisciplinary evaluations. Our MGH Down Syndrome Program offers the most robust services in the world for people with Down syndrome, and physicians and community leaders from around the globe, including Japan, Sweden, Canada, and the United Arab Emirates, have come to visit our Program in an effort to replicate our programming.
Creation of a National Down Syndrome Patient Database (2012 to current)	I oversee a multi-institutional Patient Database, collecting and tracking clinical data on patients with Down syndrome seen in specialty clinics around the country. Under my leadership, we have published research papers in top-tier medical journals, and I have presented on the Patient Database at national and international conferences. Ultimately, by closely tracking longitudinal prospective clinical data on patients with Down syndrome, we will be able to develop more evidence-based guidelines to improve their overall outcomes.
Creation of multidisciplinary Down Syndrome Program at Boston Children's Hospital (2009-2012)	As an Instructor, I expanded the Down Syndrome Program at Boston Children's Hospital to offer, for the first time, multidisciplinary services to patients with Down syndrome between the ages of 3 to 18. Our team of 8 healthcare professional saw about 300 patients with Down syndrome each year. During this time, I also helped oversee an online series of ~25 one-hour Webinars for caregivers and families on a diversity of topics related to their children's needs (e.g., celiac disease, toilet training).

Report of Education of Patients and Service to the Community

Activities

No activities below were sponsored by outside entities.

- 2006 Celebrating Life with Down Syndrome / Lecture
 YMCA of Wykoff, NJ
- 2007 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings
 with Down Syndrome / Seminar
 Massachusetts Down Syndrome Congress, Worcester, MA
- 2007 What Your Other Children are Thinking / Lecture
 Down Syndrome Association of Middle Tennessee, Nashville, TN
- 2008 Brothers and Sisters Who Have Siblings with Down Syndrome: Questions and
 Answers / Lecture
 Affiliates In Action Conference, Scottsdale, AZ (national)
- 2008 Compassionate Delivery of a New Diagnosis to New and Expectant Parents of
 Children with Down Syndrome/ Lecture
 Affiliates In Action Conference, Scottsdale, AZ (national)
- 2008 Prenatal and Postnatal Medical Support: Capturing the Sentiments of Parents Who
 Have Children with Down Syndrome / Keynote
 National Down Syndrome Congress, Boston, MA (national)
- 2008 What Your Other Children Are Thinking/ Keynote
 National Down Syndrome Congress, Boston, MA
- 2008 Celebrating Life with Down Syndrome / Lecture
 Down Syndrome Advocacy Foundation, Long Island, NY
- 2008 What Your Other Children Are Thinking / Lecture
 Down Syndrome Advocacy Foundation, Long Island, NY
- 2008 What Your Other Children are Thinking / Lecture
 Fundación Síndrome de Down de Nuevo León, Monterrey, Mexico (international)
- 2008 Brothers and Sisters: Lifelong Teachers/ Lecture
 Fundación Síndrome de Down de Nuevo León, Monterrey, Mexico (international)
- 2008 What Your Other Children Are Thinking / Lecture
 Down Syndrome Association of Central Florida, Orlando, FL
- 2008 Giving the Diagnosis of Down Syndrome: The 'How To' Guide / Lecture
 Down Syndrome Association of Central Florida, Orlando, FL

- 2008 Celebrating Life with Down Syndrome / Lecture
Down Syndrome Network of West Virginia, Charleston, WV
- 2008 What Your Other Children Without Down Syndrome Are Thinking: Sibling
Issues / Lecture
Down Syndrome Association of Atlanta, Atlanta, GA
- 2008 Celebrating Life with Down Syndrome / Lecture
Down Syndrome Association of Atlanta, Atlanta, GA
- 2008 Compassionate Delivery of a New Diagnosis to New and Expectant Parents of
Children with Down Syndrome / Lecture
Down Syndrome Association of Atlanta, Atlanta, GA
- 2009 What Your Other Children Without Down Syndrome Are Thinking: Sibling
Issues / Lecture
Down Syndrome Guild of Greater Kansas City, Kansas City, MO
- 2009 What your other children without Down syndrome are thinking: Sibling issues / Lecture
Down Syndrome Association of Peterborough, Ontario, Canada (international)
- 2009 What your other children without Down syndrome are thinking: Sibling issues / Lecture
Down Syndrome Affiliates in Action, Washington, D.C. (national)
- 2009 Prenatal and postnatal medical support: How your group can make a difference/ Lecture
Down Syndrome Affiliates in Action, Washington, D.C. (national)
- 2009 Celebrating Life with Down Syndrome / Lecture
The Down Syndrome Association of Greater St. Louis, St. Louis, MO
- 2009 What your other children without Down syndrome are thinking: Sibling issues / Lecture
National Down Syndrome Congress, Sacramento, CA (national)
- 2009 What your other children without Down syndrome are thinking: Sibling issues / Lecture
Massachusetts Down Syndrome Congress, Worcester, MA
- 2009 What your other children without Down syndrome are thinking: Sibling issues / Lecture
Down Syndrome Association of Memphis & the Mid-South, Memphis, TN
- 2009 Celebrating Life with Down Syndrome / Lecture
Down Syndrome Association of Memphis & the Mid-South, Memphis, TN
- 2009 Down Syndrome / Lecture
Loveland, Inc., Lincoln, MA
- 2009 A Challenging Dynamic: Delivering a Diagnosis of Down Syndrome to New and
Expectant Parents / Lecture
Kennedy Willis Center on Down Syndrome, Cooperstown, NY

- 2009 Celebrating Life with Down Syndrome / Lecture
Down Syndrome Association of Northern Virginia, Fairfax, VA
- 2009 What your other children without Down syndrome are thinking: Sibling issues / Lecture
Down Syndrome Association of Northern Virginia, Fairfax, VA
- 2009 What your other children without Down syndrome are thinking: Sibling issues / Lecture
Down Syndrome Association of Delaware, Wilmington, DE
- 2009 What your other children without Down syndrome are thinking: Sibling issues / Lecture
Down Syndrome Association of Central Ohio, Columbus, OH
- 2009 What your other children without Down syndrome are thinking: Sibling issues / Lecture
Down Syndrome Information Alliance, Sacramento, CA
- 2010 Making a Diagnosis of Down Syndrome: New Advances and Updates for Healthcare Professionals / Lecture
Down Syndrome Network of Montgomery County, Bethesda, MD
- 2010 Down Syndrome Prenatal Testing: Everything Your Group Should Know and Should Be Doing / Lecture
National Down Syndrome Society, New York City, NY (national)
- 2010 Celebrating Life with Down Syndrome / Lecture
Greater Clear Lake Families Exploring Down Syndrome, Houston, TX
- 2010 What your other children without Down syndrome are thinking: Sibling issues / Lecture
Greater Clear Lake Families Exploring Down Syndrome, Houston, TX
- 2010 Celebrating Life with Down Syndrome / Lecture
Canadian Down Syndrome Society, Regina, Saskatchewan, Canada (international)
- 2010 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues/ Lecture
Canadian Down Syndrome Society, Regina, Saskatchewan, Canada (international)
- 2010 Down Syndrome Prenatal Testing: Everything Your Group Should Know and Should Be Doing / Lecture
Down Syndrome Affiliates in Action, Kansas City, MO
- 2010 Sibling Issues: What Your Organization Can Do to Support Brothers and Sisters/ Lecture
Down Syndrome Affiliates in Action, Kansas City, MO
- 2010 What Your Other Children without Down Syndrome are Thinking / Lecture
GRAAL, Krakow, Poland (international)

- 2010 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know/ Lecture
GRAAL, Krakow, Poland (international)
- 2010 Therapies for Children and Adolescents with Down Syndrome/ Lecture
GRAAL, Krakow, Poland (international)
- 2010 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
Down Syndrome Association of Roanoke, Roanoke, VA
- 2010 Challenging Dynamic: Delivering a Diagnosis of Down Syndrome to New and Expectant Parents / Lecture
Down Syndrome Association of Roanoke, Roanoke, VA
- 2010 Celebrating Life with Down Syndrome / Lecture
Down Syndrome Association of Roanoke, Roanoke, VA
- 2010 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
National Down Syndrome Congress, Orlando, FL
- 2010 New Prenatal Tests for Down Syndrome: Local Updates, National Strategies, and What This All Means for Your Family / Lecture
Massachusetts Down Syndrome Congress, Worcester, MA
- 2010 Celebrating Life with Down Syndrome / Lecture
Eastern Pennsylvania Down Syndrome Center, Center Valley, PA
- 2010 Crash Course on Down Syndrome for Brothers & Sisters / Lecture
Eastern Pennsylvania Down Syndrome Center, Center Valley, PA
- 2010 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings with Down Syndrome / Lecture
Down Syndrome Aim High Resource Center, Albany, NY
- 2010 REDCap Registry: A practical solution for the Down Syndrome Program at Children's Hospital Boston / Lecture
National Down Syndrome Society, Washington, D.C. (national)
- 2010 New Prenatal Tests for Down Syndrome: Local Updates, National Strategies, and What This All Means for Your Family / Lecture
Down Syndrome Guild of Southeast Michigan, Dearborn, MI
- 2010 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings with Down Syndrome / Lecture
Down Syndrome Guild of Southeast Michigan, Dearborn, MI

- 2010 Current Research on Prenatal Testing and Parent Responses / Lecture
Down Syndrome Guild of Greater Kansas City, Kansas City, KS
- 2010 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings
with Down Syndrome / Lecture
Down Syndrome Guild of Greater Kansas City, Kansas City, KS
- 2010 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings with
Down Syndrome / Lecture
Down Syndrome Association of West Michigan, Grand Rapids, MI
- 2010 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings
with Down Syndrome / Lecture
Wisconsin Upside Down, Milwaukee, WI
- 2010 Crash Course on Down Syndrome for Brothers & Sisters / Lecture
Down Syndrome Association of Houston, Houston, TX
- 2010 Celebrating Exceptionalities / Keynote
North Coast Community Homes, Cleveland, OH
- 2011 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Parents Need to Know / Lecture
Massachusetts Down Syndrome Congress, Worcester, MA
- 2011 Celebrating Life with Down Syndrome / Keynote
Down Syndrome Association of Central Oklahoma, Oklahoma City, OK
- 2011 What Your Other Children Without Down Syndrome are Thinking: Sibling Issues /
Lecture
Down Syndrome Association of Central Oklahoma, Oklahoma City, OK
- 2011 Keeping Children and Adolescents with Down Syndrome Healthy: All of the Medical
Updates that Parents Need to Know / Lecture
Down Syndrome Association of Central Oklahoma, Oklahoma City, OK
- 2011 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings
with Down Syndrome / Lecture
Fundación Down Tigre, Buenos Aires, Argentina (international)
- 2011 Prenatal Testing for Down Syndrome: National Updates and What Your Affiliate Can
and Should Be Doing Right Now / Lecture
Down Syndrome Affiliates in Action, Dallas, TX (national)
- 2011 Healthy Students with Down Syndrome: All the Medical Updates that Educators Need to
Know / Lecture
Down Syndrome Network of Montgomery County, Gaithersburg, MD

- 2011 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings with Down Syndrome / Lecture
National Down Syndrome Society, New York City, NY (national)
- 2011 Keeping Children and Adolescents with Down Syndrome Healthy: All of the Medical Updates that Parents Need to Know / Lecture
Family Resources Associates, Inc., Colts Neck, NJ
- 2011 Brothers and Sisters with Rett Syndrome / Lecture
International Rett Syndrome Foundation, Boston, MA (national)
- 2011 Crash Course on Down Syndrome for Brothers & Sisters / Lecture
Down Syndrome Information Alliance, Sacramento, CA
- 2011 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings with Down Syndrome / Lecture
Down Syndrome Information Alliance, Sacramento, CA
- 2011 Down Syndrome Patient Registry Pilot Project / Lecture
National Down Syndrome Congress, San Antonio, TX (national)
- 2011 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
National Down Syndrome Congress, San Antonio, TX (national)
- 2011 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings with Down Syndrome/ Lecture
National Down Syndrome Congress, San Antonio, TX (national)
- 2011 Keeping Children and Adolescents with Down Syndrome Healthy: All of the Medical Updates that Parents Need to Know / Lecture
Down Syndrome Alliance of the Midlands, Omaha, NE
- 2011 Keeping Children and Adolescents with Down Syndrome Healthy: All of the Medical Updates that Parents Need to Know / Lecture
Massachusetts Down Syndrome Congress, Exeter, NH
- 2011 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings with Rett Syndrome / Webinar
International Rett Syndrome Association, Boston, MA (national)
- 2011 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues for Parents / Lecture
St. Clair County Community Mental Health Authority, Port Huron, MI
- 2011 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know/ Lecture
St. Clair County Community Mental Health Authority, Port Huron, MI

- 2011 Celebrating Life with Down Syndrome/ Lecture
St. Clair County Community Mental Health Authority, Port Huron, MI
- 2011 New Prenatal Tests for Down Syndrome: National Updates and What This All Means for Your Family/ Lecture
St. Clair County Community Mental Health Authority, Port Huron, MI
- 2011 Celebrating Life with Down Syndrome / Keynote
Connecticut Down Syndrome Congress, Westbrook, CT
- 2011 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know/ Keynote
Connecticut Down Syndrome Congress, Westbrook, CT
- 2011 What Your Other Children Without Down Syndrome are Thinking: Sibling Issues for Parents / Lecture
Connecticut Down Syndrome Congress, Westbrook, CT
- 2012 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Down Syndrome Alliance of the Midlands, Omaha, NE
- 2012 What Your Other Children Without Down Syndrome are Thinking: Sibling Issues / Lecture
Down Syndrome Alliance of the Midlands, Omaha, NE
- 2012 Prenatal Tests for Down Syndrome: All of the Updates that Your Group Needs to Know / Lecture
Down Syndrome Affiliates in Action, Washington, D.C. (national)
- 2012 Let's Get Real About Down Syndrome: How You Can Use New Research for Families/ Lecture
Down Syndrome Affiliates in Action, Washington, D.C. (national)
- 2012 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Massachusetts Down Syndrome Congress, Worcester, MA
- 2012 What your other children without Down syndrome are thinking: Sibling Issues/ Lecture
Massachusetts Down Syndrome Congress, Worcester, MA
- 2012- Massachusetts Down Syndrome Congress / Chair, Medical and Science Advisory Board
Provide guidance to organization on clinical and research trends for people with Down syndrome
- 2012 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture

- South Carolina Down Syndrome Family Conference, Colombia, SC
- 2012 What your other children without Down syndrome are thinking: Sibling issues/ Lecture
South Carolina Down Syndrome Family Conference, Colombia, SC
- 2012 Celebrating Life with Down Syndrome / Lecture
Upper Peninsula Down Syndrome Association, Marquette, MI
- 2012 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Parents Need to Know / Lecture
Upper Peninsula Down Syndrome Association, Marquette, MI
- 2012 New Prenatal Tests for Down Syndrome: National Updates and What This All
Means for Your Family / Lecture
Massachusetts Down Syndrome Congress, Ipswich, MA
- 2012 Down Syndrome Patient Database / Lecture
Down Syndrome Medical Interest Group, Washington, DC (national)
- 2012 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
National Down Syndrome Congress Convention, Washington, DC (national)
- 2012 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
World Down Syndrome Congress Convention, Cape Town, South Africa (international)
- 2012 New Prenatal Tests for Down Syndrome: National Updates and What This All
Means for Your Family / Lecture
World Down Syndrome Congress Convention, Cape Town, South Africa (international)
- 2012 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Parents Need to Know / Lecture
World Down Syndrome Congress Convention, Cape Town, South Africa (international)
- 2012 Celebrating Life with Down Syndrome / Lecture
World Down Syndrome Congress Convention, Cape Town, South Africa (international)
- 2012 Celebrating Life with Down Syndrome / Keynote
National Association for Down Syndrome, Chicago, IL (national)
- 2012 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
National Association for Down Syndrome, Chicago, IL (national)
- 2012 Celebrating Life with Down Syndrome / Keynote
The Early Childhood Direction Center, Edmeston, NY (national)

- 2012 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
The Early Childhood Direction Center, Edmeston, NY (national)
- 2012 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
The Early Childhood Direction Center, Edmeston, NY (national)
- 2012 Celebrating Life with Disabilities / Keynote
The Moore Center, Manchester, NH (national)
- 2012 Celebrating Life with Disabilities / Keynote
Down Syndrome Awareness Group of East Tennessee (national)
- 2013 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
Down Syndrome Association of Greater Charlotte, Charlotte, NC (national)
- 2013 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Down Syndrome Association of Singapore, Singapore (international)
- 2013 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Gigi's Playhouse, Rockford, IL (national)
- 2012 Celebrating Life with Disabilities / Keynote
Gigi's Playhouse, Rockford, IL (national)
- 2012 Celebrating Life with Disabilities / Keynote
Down Syndrome Association of Acadiana, Lafayette, LA (national)
- 2013 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
Down Syndrome Association of Acadiana, Lafayette, LA (national)
- 2013 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Down Syndrome Association of Acadiana, Lafayette, LA (national)
- 2013 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Educators Need to Know / Lecture
Down Syndrome Association of Acadiana, Lafayette, LA (national)
- 2013 Let's Get Real About Down Syndrome: How You Can Use New Research on Families / Lecture
Down Syndrome Association of Acadiana, Lafayette, LA (national)

- 2013 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Massachusetts Down Syndrome Congress, Worcester, MA (regional)
- 2013 The Brave New World of Prenatal Testing / Lecture
The Jack Kent Cooke Foundation (regional)
- 2013 Celebrating Life with Disabilities / Keynote
Down Syndrome Association of San Diego, San Diego, CA (national)
- 2013 Celebrating Life with Disabilities / Keynote
Canadian Down Syndrome Society, Winnipeg, Canada (international)
- 2013 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
Canadian Down Syndrome Society, Winnipeg, Canada (international)
- 2013- National Center for Prenatal and Postnatal Down Syndrome Diagnoses Resources / Professional Advisory Committee
Provide guidance to the national organization on accurate, up-to-date, balanced materials for expectant women
- 2013 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
Wyoming Down Syndrome Association, Casper, Wyoming (national)
- 2013 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Wyoming Down Syndrome Association, Casper, Wyoming (national)
- 2013 Adult Brothers and Sisters Who Have Siblings with Down Syndrome / Lecture
National Down Syndrome Congress Convention, Denver, CO (national)
- 2013 Prenatal and Postnatal Diagnoses of Down Syndrome: New Advances and Updates for Today's Physicians / Lecture
Department of Developmental Services, Boston, MA
- 2013 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
Chesapeake Down Syndrome Parents Group (national)
- 2013 Celebrating Life with Disabilities / Lecture
Trisomy 21 Foundation of Northern New York (national)
- 2013 Celebrating Life with Down syndrome / Lecture
Svenska Downforeningen, Stockholm, Sweden (international)
- 2013 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues /

- Lecture
Maine Down Syndrome Network, Portland, ME (regional)
- 2013 Celebrating Life with Disabilities / Plenary talk
Maine Down Syndrome Network, Portland, ME (regional)
- 2013 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Maine Down Syndrome Network, Portland, ME (regional)
- 2013 Crash Course on Down Syndrome for Brothers & Sisters / Workshop
Maine Down Syndrome Network, Portland, ME (regional)
- 2014 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
Father's Joy, Houston, TX (national)
- 2014 Crash Course on Down Syndrome for Brothers & Sisters / Workshop
Father's Joy, Houston, TX (national)
- 2014 Celebrating Life with Disabilities / Keynote talk
Down Syndrome Association of Northeast Indiana, Ft. Wayne, IN (regional)
- 2014 Adult Brothers and Sisters Who Have Siblings with Down Syndrome / Lecture
Down Syndrome Association of Northeast Indiana, Ft. Wayne, IN (regional)
- 2014 What is the New Prenatal Testing, and How Can Affiliates Respond? / Webinar Lecture
National Center for Prenatal and Postnatal Down Syndrome Resources (national)
- 2014 Celebrating Life with Disabilities / Keynote talk
Down Syndrome Parent Group of Western New York, Buffalo, NY (regional)
- 2014 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
Down Syndrome Parent Group of Western New York, Buffalo, NY (regional)
- 2014 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Down Syndrome Parent Group of Western New York, Buffalo, NY (regional)
- 2014 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Family Resource Associates, Inc., Shrewsbury, NJ
- 2014 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
National Down Syndrome Congress Annual Convention, Indianapolis, IN (national)

- 2014 Adult Brothers and Sisters who Have Siblings with Down Syndrome
National Down Syndrome Congress Annual Convention, Indianapolis, IN (national)
- 2014 What Your Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
Down Syndrome Association of Greater Charlotte, Charlotte, NC (national)
- 2014 Celebrating Life with Disabilities / Keynote talk
Down Syndrome Association of Greater Charlotte, Charlotte, NC (national)
- 2014 Crash Course on Down Syndrome for Brothers & Sisters / Lecture
Down Syndrome Association of Greater Charlotte, Charlotte, NC (national)
- 2014 Down Syndrome / Lecture
Wells Fargo, Charlotte, NC (national)
- 2014 Celebrating Life with Disabilities / Keynote talk
CCARC, New Britain, CT (regional)
- 2014 Celebrating Life with Disabilities / Keynote Remarks
ACDS, Plainview, NY (national)
- 2014 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
Human First, Westbury, NY (national)
- 2014 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Service Providers Need to Know / Lecture
Human First, Westbury, NY (national)
- 2014 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
The 21 Club, New Hartford, NY (national)
- 2014 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Parents Need to Know / Lecture
The 21 Club, New Hartford, NY (national)
- 2014 New Prenatal Tests for Down Syndrome: National Updates and What This All
Means for Your Family / Lecture
The 21 Club, New Hartford, NY (national)
- 2014 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
Down Syndrome Association for Families of Nebraska, Lincoln, NE (national)
- 2014 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Parents Need to Know / Lecture

- Down Syndrome Association for Families of Nebraska, Lincoln, NE (national)
- 2014 New Prenatal Tests for Down Syndrome: National Updates and What This All Means for Your Family / Lecture
Down Syndrome Association for Families of Nebraska, Lincoln, NE (national)
- 2014 Celebrating Life with Disabilities / Lecture
Down Syndrome Association for Families of Nebraska, Lincoln, NE (national)
- 2014 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Down Syndrome Advocacy Foundation of Long Island, Brookville, NY (national)
- 2014 Adult Brothers and Sisters who Have Siblings with Down Syndrome / Lecture
Down Syndrome Advocacy Foundation of Long Island, Brookville, NY (national)
- 2014 Keeping Students with Down Syndrome Healthy: All the Medical Updates that Educators Need to Know / Lecture
Down Syndrome Advocacy Foundation of Long Island, Brookville, NY (national)
- 2015 Celebrating Life with Disabilities / Keynote
GiGi's Playhouse, Syracuse, NY (national)
- 2015 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows / Lecture
Down Syndrome Association of Wisconsin, La Crosse, WI (national)
- 2015 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Down Syndrome Association of Wisconsin, Marshfield, WI (national)
- 2015 The Latest and Greatest Research About Down Syndrome / Lecture
Family Resources Associates, Inc., Shrewsbury, NJ (national)
- 2015 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Massachusetts Down Syndrome Congress, Worcester, MA (regional)
- 2015 The Latest and Greatest Research About Down Syndrome / Lecture
Massachusetts Down Syndrome Congress, Worcester, MA (regional)
- 2015 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows / Lecture
Down Syndrome Association of Central Ohio, Columbus, OH (national)
- 2015 New Prenatal Tests for Down Syndrome: New Advances in Genetic Testing / Lecture
Down Syndrome Association of Central Ohio, Columbus, OH (national)
- 2015 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture

- Down Syndrome Alliance of the Midlands, Omaha, NE (national)
- 2015 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
Down Syndrome Association for Families of Nebraska, Lincoln, NE (national)
- 2015 Crash Course on Down Syndrome for Brothers & Sisters / Workshop
Down Syndrome Association for Families of Nebraska, Lincoln, NE (national)
- 2015 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
World Down Syndrome Congress conference, Chennai, India (international)
- 2015 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
World Down Syndrome Congress conference, Chennai, India (international)
- 2015 Adult Brothers and Sisters who Have Siblings with Down Syndrome / Lecture
World Down Syndrome Congress conference, Chennai, India (international)
- 2015 Celebrating Life with Down Syndrome / Keynote
World Down Syndrome Congress conference, Chennai, India (international)
- 2015 Weight Management Challenges for People with Down Syndrome / Lecture
World Down Syndrome Congress conference, Chennai, India (international)
- 2015 New Prenatal Tests for Down Syndrome: National Updates and What This All Means for Your Family
World Down Syndrome Congress conference, Chennai, India (international)
- 2015 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows
World Down Syndrome Congress conference, Chennai, India (international)
- 2015 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Down Syndrome Association of Wisconsin, Green Bay, WI (national)
- 2015 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Family Connections of South Carolina, Lexington, SC (national)
- 2015 The Latest and Greatest Research About Down Syndrome / Lecture
Family Connections of South Carolina, Lexington, SC (national)
- 2016 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Down Syndrome Awareness Group (DSAG) of Baton Rouge, Baton Rouge, LA (national)

- 2016 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows / Lecture
Down Syndrome Awareness Group (DSAG) of Baton Rouge, Baton Rouge, LA
(national)
- 2016 The Latest and Greatest Research About Down Syndrome / Lecture
Down Syndrome Awareness Group (DSAG) of Baton Rouge, Baton Rouge, LA
(national)
- 2016 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
Down Syndrome Awareness Group (DSAG) of Baton Rouge, Baton Rouge, LA
(national)
- 2016 Crash Course on Down Syndrome for Brothers & Sisters / Lecture
Down Syndrome Awareness Group (DSAG) of Baton Rouge, Baton Rouge, LA
(national)
- 2016 Celebrating Life with Down Syndrome / Keynote
GiGi's Playhouse Weschester, Tarrytown, NY (national)
- 2016 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows / Lecture
Down Syndrome Awareness Group of East Tennessee, Knoxville, TN (national)
- 2016 Keeping Students with Down Syndrome Healthy: All the Medical
Updates that Educators Need to Know / Lecture
Down Syndrome Association of Delaware, Wilmington, DE (national)
- 2016 The Latest and Greatest Research about Down Syndrome / Lecture
Down Syndrome Association of Delaware, Wilmington, DE (national)
- 2016 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Parents Need to Know / Lecture
Canadian Down Syndrome Society, Montreal, Canada (international)
- 2016 The Latest and Greatest Research about Down Syndrome / Lecture
Canadian Down Syndrome Society, Montreal, Canada (international)
- 2016 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
National Down Syndrome Congress Convention, Orlando, FL (national)
- 2016 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
Chesapeake Down Syndrome Parent Group, Baltimore, MD (national)
- 2016 Brothers and Sisters Workshop
Chesapeake Down Syndrome Parent Group, Baltimore, MD (national)

- 2016 Prenatal Diagnoses of Down Syndrome: Delivering Results in our New Age of Genetic Testing
Upside for Down Foundation, De Vervolmaakte Mens, Leiden, Netherlands (international)
- 2016 Common Threads: Celebrating Life with Down Syndrome / Keynote Lecture
Metrocare Services, Dallas, Texas (national)
- 2016 The Latest and Greatest Research about Down Syndrome / Lecture
Metrocare Services, Dallas, Texas (national)
- 2016 My Life as a Clinical Geneticist / Lecture
The Hotchkiss School, Salisbury, Connecticut (regional)
- 2016 Building a Virtual Clinic for Patients with Down Syndrome / Lecture
Ariadne Labs, Boston, MA (regional)
- 2017 Medical Updates: Keeping Children and Adolescents with Down Syndrome Healthy / Lecture
Blessed by Downs, Thibodaux, LA (regional)
- 2017 The Latest and Greatest Research about Down Syndrome / Lecture
Blessed by Downs, Thibodaux, LA (regional)
- 2017 Celebrating Life with Down Syndrome / Keynote
Blessed by Downs, Thibodaux, LA (regional)
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Down Syndrome Association of Central Florida, Orlando, FL (regional)
- 2017 The Latest and Greatest Research about Down Syndrome / Lecture
Down Syndrome Association of Central Florida, Orlando, FL (regional)
- 2017 Common Threads: Celebrating Life with Down Syndrome / Keynote Lecture
Down Syndrome Association of Central Florida, Orlando, FL (regional)
- 2017 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues / Lecture
Down Syndrome Association of Central Florida, Orlando, FL (regional)
- 2017 Adult Brothers and Sisters who Have Siblings with Down Syndrome / Lecture
Down Syndrome Association of Central Florida, Orlando, FL (regional)
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Massachusetts Down Syndrome Congress, Worcester, MA (local)

- 2017 Promising Developments in Alzheimer's Research & Down Syndrome / Lecture
Massachusetts Down Syndrome Congress, Worcester, MA (local)
- 2017 Celebrating Life with Down Syndrome / Keynote
GiGi's Playhouse NYC, New York City, NY (national)
- 2017 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows / Lecture
Down Syndrome Association of Middle Tennessee, Nashville, TN (regional)
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Parents Need to Know / Lecture
Flower City Down Syndrome Network, Rochester, NY (regional)
- 2017 Down Syndrome: Not Just an Extra Chromosome (lecture)
Yokohama Project, Yokohama, Japan (international)
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Parents Need to Know / Lecture
Yokohama Project, Yokohama, Japan (international)
- 2017 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
National Down Syndrome Congress, Sacramento, CA (national)
- 2017 Our New Age of Genetic Testing / Lecture
Anne Carlsen Center, Medora, ND (national)
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Parents Need to Know / Lecture
Designer Genes of North Dakota, Bismarck, ND (national)
- 2017 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Opening Lecture
Down Syndrome Federation of India, New Delhi, India (international)
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Parents Need to Know / Lecture
Down Syndrome Federation of India, New Delhi, India (international)
- 2017 The Latest and Greatest Research about Down Syndrome / Lecture
Upside of Downs, Cleveland, OH (national)
- 2017 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical
Updates that Parents Need to Know / Lecture
Deutsches Down-Syndrom InfoCenter, Nuremberg, Germany (international)
- 2017 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues /

- Lecture
Down Syndrome Association of Wisconsin, Milwaukee, WI (national)
- 2017 Crash Course on Down Syndrome for Brothers & Sisters
Down Syndrome Association of Wisconsin, Milwaukee, WI (national)
- 2018 The Latest and Greatest Research about Down Syndrome / Lecture
F.R.I.E.N.D.S., Johnson City, TN (national)
- 2018 Celebrating Life with Down Syndrome / Keynote
F.R.I.E.N.D.S., Johnson City, TN (national)
- 2018 What the Other Children Without Down Syndrome Are Thinking: Sibling Issues /
Lecture
Massachusetts Down Syndrome Congress annual conference, Worcester, MA (regional)
- 2018 Obstructive Sleep Apnea in People with Down Syndrome / Lecture
Massachusetts Down Syndrome Congress annual conference, Worcester, MA (regional)
- 2018 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings
with Down Syndrome / Lecture
World Down Syndrome Congress, Glasgow, Scotland (international)
- 2018 Adult Brothers and Sisters Who Have Siblings with Down Syndrome
World Down Syndrome Congress, Glasgow, Scotland (international)
- 2018 Down Syndrome & Unexplained Regression
World Down Syndrome Congress, Glasgow, Scotland (international)
- 2018 Prenatal Diagnoses of Down Syndrome: New Advances and Updates for Today's
Healthcare Providers / Lecture
World Down Syndrome Congress, Glasgow, Scotland (international)
- 2018 Down Syndrome Clinic to You (DSC2U): An Online Personal Care Plan for Caregivers
and Primary Care Providers
World Down Syndrome Congress, Glasgow, Scotland (international)
- 2018 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings
with Down Syndrome / Lecture
World Down Syndrome Congress, Glasgow, Scotland (international)
- 2018 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings
with Down Syndrome / Lecture
National Down Syndrome Congress Convention, Dallas, TX (national)
- 2018 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows / Grand
Rounds
Jack's Basket, Minneapolis, MN (national)

- 2018 What Your Other Children Are Thinking: Brothers and Sisters Who Have Siblings with Down Syndrome / Lecture
Down Syndrome Federation of India, Pune, India (international)
- 2018 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Down Syndrome Federation of India, Pune, India (international)
- 2018 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
Canadian Down Syndrome Society Annual Conference, Hamilton, Canada (international)
- 2018 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows / Grand Rounds
Canadian Down Syndrome Society Annual Conference, Hamilton, Canada (international)
- 2018 Obstructive Sleep Apnea in People with Down Syndrome / Lecture
LuMind Down Syndrome Research Foundation, Boston MA (regional)
- 2018 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
21 Connect, Buffalo, NY (national)
- 2019 Down Syndrome Clinic to You (DSC2U): An Online Personalized Care Plan for Caregivers and Primary Care Providers
Massachusetts Down Syndrome Congress, Worcester, MA (regional)
- 2019 Down Syndrome Clinic to You (DSC2U): An Online Personalized Care Plan for Caregivers and Primary Care Providers
National Down Syndrome Congress, Pittsburgh, PA (national)
- 2019 Brain Train: SMART Actions that Might Boost Cognition and Prevent Alzheimer's Disease in Patients with Down Syndrome
National Down Syndrome Congress, Pittsburgh, PA (national)
- 2019 Brain Train: SMART Actions that Might Boost Cognition and Prevent Alzheimer's Disease in Patients with Down Syndrome / Lecture
Down Syndrome Association of Wisconsin, Milwaukee, WI (national)
- 2019 Brain Train: SMART Actions that Might Boost Cognition and Prevent Alzheimer's Disease in Patients with Down Syndrome / Lecture
UP21 Foundation, Covington, LA (national)
- 2019 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture

UP21 Foundation, Covington, LA (national)

- 2019 Brain Train: SMART Actions that Might Boost Cognition and Prevent Alzheimer's Disease in Patients with Down Syndrome / Lecture
Down Syndrome Advocacy Foundation, Brookville, NY (national)
- 2019 Down Syndrome: All People Deserve to be Healthy
Fidelity, Boston MA (regional)
- 2019 Brain Train: SMART Actions that Might Boost Cognition and Prevent Alzheimer's Disease in Patients with Down Syndrome / Lecture
Down Syndrome Association of Minnesota, Minneapolis, MN (national)
- 2019 Down Syndrome Clinic to You (DSC2U): An Online Personalized Care Plan for Caregivers and Primary Care Providers
Down Syndrome Association of Minnesota (national)
- 2019 Down Syndrome Clinic to You (DSC2U): An Online Personalized Care Plan for Caregivers and Primary Care Providers
Special Needs Initiative, United Healthcare, Minneapolis, MN (national)
- 2019 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Clinicians Need to Know / Lecture
Down Syndrome Association of Northeast Indiana, Ft. Wayne, IN (regional)
- 2019 Down Syndrome Clinic to You (DSC2U): An Online Personalized Care Plan for Caregivers and Primary Care Providers
Down Syndrome Association of Northeast Indiana, Ft. Wayne, IN (regional)
- 2019 Keeping Children and Adolescents with Down Syndrome Healthy: All the Medical Updates that Parents Need to Know / Lecture
The 21 Cub of Central New York, Utica, NY (regional)
- 2019 Taking Care of Your Family: Celebrating the Highs and Surviving the Lows / Lecture
The 21 Cub of Central New York, Utica, NY (regional)

Educational Material for Patients and the Lay Community

No educational materials below were sponsored by outside entities.

Books, monographs, articles

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|------|--|-----------|--|
| 2001 | <i>Common Threads:
Celebrating Life with
Down Syndrome</i> | Co-author | Award-winning book for families about success stories about people with Down syndrome (Band of Angels Press) |
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2009	“Driving Forward”	Author	Chapter in <i>Thicker than Water: Essays by Adult Siblings of People with Disabilities</i> (Woodbine House)
2009	<i>Fasten Your Seatbelt: A Crash Course on Down Syndrome for Brothers and Sisters</i>	Co-author	Book for brothers and sisters who have siblings with Down syndrome (Woodbine House)

Presentations in other media

2001-current	<i>New York Times, The Wall Street Journal, Boston Globe, Boston Herald, L.A. Times, and others</i>	Print Appearances	Numerous quoted interviews in national publications each year.
2005	ABCNews, Boston, MA	Television Appearance	“Study: Down Syndrome Diagnosis Talk Should Change”
2005	ABC’s Good Morning America (national)	Television Appearance	“Mothers: Doctors Too Negative When Diagnosing Down Syndrome”
2005	WBUR’s “On Point” (NPR)	Radio Appearance	“Down Syndrome & Doctors”
2006	PBS’s Religion and Ethics Newsweekly (national)	Television Appearance	“Down Syndrome”
2008	CNN (national)	Television Appearance	“Down syndrome decision”
2008	NPR (national)	Radio Appearance	“Prenatal Down Syndrome Screening”
2008	NPR’s Brian Lehrer Show (national)	Radio Appearance	“Down syndrome”
2008	CNN’s House Call with Dr. Sanjay Gupta (national)	Television Appearance	“Couples Faced with the News That Their Child May Have Down Syndrome”
2009	NPR (WPBS)	Radio Appearance	“A San Diego Company Develops a Test for Down's”

2009	The Parents' Journal with Bobbi Conner	Radio Appearance	"Guidance for Kids Who Have a Sibling with Down Syndrome"
2009	NPR (WBUR)	Radio Appearance	"Living with a Sibling with Down Syndrome"
2009	FOXNews, Boston	Television Appearance	"Down syndrome and siblings"
2009	FOXNews, Boston	Television Appearance	"Down Syndrome Research"
2009	CNN	Television Appearance	"Una leccion para todos"
2009	ABCNews NOW	Television Appearance	"Special Needs Kids and Their Siblings"
2010	ABC World News with Diane Sawyer (national)	Television Appearance	"The R Word"
2011	WBUR (NPR), Boston	Radio Appearance	"Down Syndrome Test Raises Questions for Future Moms"
2011	WBUR (NPR), Boston	Radio Appearance	"New, Safer Test for Down Syndrome Sparks Ethical Concern"
2011	WGBH, Boston	Television Appearance	"A New Down Syndrome Test Raises Ethical Issues"
2011	WPCC (NPR), South Carolina	Radio Appearance	"How much do you want to know--and should you know--about your unborn baby?"
2011	FOX News, Boston	Television Appearance	"GQ controversy: Down Syndrome Comment"
2011	MSNBC, New York (national)	Television Appearance	"Down Syndrome's Rewards Touted as New Test Looms"
2011	ABCNews with Diane Sawyer (national)	Television Appearance	"Safer Down Syndrome Test Hits Market Monday"
2012	WNPR (NPR), Connecticut	Radio Appearance	"Prenatal Genetic Testing"

2012	CBS's "This Morning," New York City (national)	Television Appearance	"Early prenatal test raises ethical questions"
2012	"Think Out Loud," Oregon Public Broadcasting	Radio Appearance	"Living with Down Syndrome"
2012	"Chronicle," ABC News, Boston	Television Appearance	"Down Syndrome"
2012	NBC News, Knoxville	Television Appearance	"Down Syndrome"
2013	FOX News, Boston	Television Appearance	"Doctors finding it easier to diagnose Down syndrome"
2013	ABC News, Boston	Television Appearance	"UMass Researchers block Down syndrome chromosome"
2013	"The Takeaway," NPR	Radio Appearance	"New Genetic Therapy Provides Breakthrough for Down Syndrome"
2013	"Here and Now," NPR	Radio Appearance	"New Genetic Therapy Could Erase Down Syndrome"
2013	"Chronicle," ABC News, Boston	Television Appearance	"Potential End to Down Syndrome Leaves Parents Torn"
2013	WGN Radio (Chicago)	Radio Appearance	"Major Breakthroughs in Down Syndrome Treatment"
2013	KKSF Radio (San Francisco)	Radio Appearance	Down Syndrome Research
2013	Today Show, NBC News	Television Appearance	"How Down Syndrome Might Help Unravel the Alzheimer's Puzzle"
2014	NPR	Radio Appearance	"Blood Test Provides More Accurate Prenatal Testing For Down Syndrome"
2014	WCVB, ABCNews Boston	Television Appearance	"Why lower rates of breast cancer in women with Down syndrome?"

2015	WCVB, ABCNews, Boston	Television Appearance	“Man hoping cutting edge genetic technology can identify disability”
2017	“The Pulse,” NPR	Radio Appearance	“People with Down syndrome living longer, but at higher risk for Alzheimer’s”
2017	WCVB, ABCNews, Boston	Television Appearance	“New study examines medical cost of caring for a Down syndrome child”
2017	“Brains On!,” NPR	Radio appearance	“What is Down Syndrome?”
2018	WCVB, ABCNews, Boston	Television Appearance	“Clinical Trial Could Give Answers to Potential Link Between Down Syndrome, Alzheimer’s”
2018	WBZ, CBSNews, Boston	Television Appearance	“Vaccine Being Tested to Prevent Dementia in People with Down Syndrome”
2018	“Dateline,” NBC	Television Appearance	“Meet Jamie Foxx’s Superstar Sister.”
2019	FOX25 News, Boston	Television Appearance	“Report: Labs not following all guidelines for noninvasive prenatal screening”
2019	“The Source,” Texas Public Radio	Radio Appearance	“1 In Every 700 Babies Is Born With Down Syndrome In The U.S.”

Patient educational material

2012-current	Numerous handouts about co-occurring conditions with Down syndrome	Co-author	Patient education handouts on www.massgeneral.org/downsyndrome
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Recognition

2002	National Media Award	National Down Syndrome Congress
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2003	National Book Award	TASH
2003	Diversity Ambassador Award	American Red Cross
2005	Down Syndrome Community Service Award	National Down Syndrome Society
2006	President's Award	National Down Syndrome Society
2008	Christian Pueschel Memorial Research Award	National Down Syndrome Congress
2011	Gunnar Dybwad Leadership Award	MA Department of Developmental Services, Executive Office of Health & Human Services
2012	Allen C. Crocker Award of Excellence	Massachusetts Down Syndrome Congress
2012	Allen Crocker, M.D., Health Services Award	MA Department of Developmental Services, Executive Office of Health & Human Services
2013	Down Syndrome Association Star Award	Down Syndrome Association of San Diego
2014	ACDS Inspirational Service Award	ACDS
2016	Patriots Difference Maker of the Week	New England Patriots Charitable Foundation

Report of Scholarship

Publications

Peer reviewed publications in print or other media

Research Investigations

1. Koppenhaver, D., Erickson, K., **Skotko, B.** (2001). Supporting communication of girls with Rett syndrome and their mothers in storybook reading. *International Journal of Disability, Development, and Education*, 48: 395-410.
2. Koppenhaver, D., Erickson, K., Harris, B., McLellan, J., **Skotko, B.**, & Newton, R. (2001). Storybook-based communication intervention for girls with Rett syndrome and their mothers. *Disability and Rehabilitation*, 23: 149-159.
3. **Skotko, B.**, Kensinger, E., Locascio, J., Einstein, G., Rubin, D., Tupler L., Krendl, A., & Corkin, S. (2004). Puzzling thoughts for H.M.: Can new semantic information be anchored to old semantic memories? *Neuropsychology*, 18: 756-69.

4. **Skotko, B.**, & Canal, R. (2004). Apoyo postnatal para madres de niños con síndrome de Down. *Revista Síndrome de Down*, 21: 54-71.
5. **Skotko, B.**, Koppenhaver, D., & Erickson, K. (2004). Parent reading behaviors and communication outcomes in girls with Rett syndrome. *Exceptional Children*, 70(2): 145-66.
6. **Skotko, B.**, Bedia, R. (2005). Continuing a Pregnancy After Receiving a Prenatal Diagnosis of Down Syndrome in Spain. *Progresos en Diagnostico y Tratamiento Prenatal*, 17: 189-92.
7. **Skotko, B.**, Andrews, A., & Einstein, G. (2005). Language and the Medial Temporal Lobe: Evidence from H.M.'s Spontaneous Discourse. *Journal of Memory and Language*, 53: 397-415.
8. **Skotko, B.** (2005). Communicating the postnatal diagnosis of Down syndrome: An international call for change. *Italian Journal of Pediatrics*, 31: 237-243.
9. **Skotko, B.**, & Canal, R. (2005). Postnatal support for mothers of children with Down syndrome. *Mental Retardation*, 43: 196-212.
10. **Skotko, B.** (2005). Prenatally diagnosed Down syndrome: Mothers who continued their pregnancies evaluate their health care providers. *American Journal of Obstetrics & Gynecology*, 192: 670-77.
11. **Skotko, B.** (2005). Mothers of children with Down syndrome reflect on their postnatal support. *Pediatrics*. 115: 64-77.
12. **Skotko, B.**, & Levine, S. P. (2006). What the other children are thinking: Brothers and sisters of persons with Down syndrome. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*, 142C:180-6.
13. **Skotko, B.**, Rubin, D., & Tupler, L. (2008). H.M.'s personal crossword puzzles: Understanding memory and language. *Memory*, 16: 89-96.
14. Rosen, D., Lombardo, A., **Skotko, B.**, & Davidson, EJ (2011). Parental perceptions of sleep disturbances and sleep-disordered breathing in children with Down syndrome. *Clinical Pediatrics*, 50: 121-125.
15. **Skotko, B.G.**, Levine, S.P., Goldstein, R. (2011). Having a Son or Daughter with Down Syndrome: Perspectives from Mothers and Fathers. *American Journal of Medical Genetics Part A* 155:2335-2347.
16. **Skotko, B.G.**, Levine, S.P., Goldstein, R. (2011). Having a Brother or Sister with Down Syndrome: Perspectives from Siblings. *American Journal of Medical Genetics Part A*: 155:2348-2359.
17. **Skotko, B.G.**, Levine, S.P., Goldstein, R. (2011). Self-perceptions from People with Down Syndrome. *American Journal of Medical Genetics, Part A*: 155:2360-2369.
18. **Skotko, B.G.**, Davidson, E.J., Weintraub, G.S. (2013). Contributions of a specialty clinic for

- children with Down syndrome. *American Journal of Medical Genetics, Part A*: 161(3): 430-437.
19. Grieco, J., Pulsifer, M., Seligsohn, K., **Skotko, B.**, Schwartz, A. (2015). Down Syndrome: Cognitive and Behavioral Functioning Across the Lifespan. *American Journal of Medical Genetics, Part C* 169C:135-149.
 20. **Skotko, B.G.**, Levine, S.P., Macklin, E.A., Goldstein, R.D. (2015). Family Perspectives about Down Syndrome. *American Journal of Medical Genetics Part A*, 170(4): 930-941.
 21. de Graaf, G., Buckley, F., **Skotko, B.G.** (2015). Estimates of the live births, natural losses, and elective terminations with Down syndrome in the United States. *American Journal of Medical Genetics, Part A*: 167A:756-767.
 22. Lavigne, J., Sharr, C., Ozonoff, A., Prock, L.A., Baumer, N., Brasington, C., Cannon, S., Crissman, B., Davidson, E., Florez, J.C., Kishnani, P., Lombardo, A., Lyerly, J., McCannon, J.B., McDonough, M.E., Schwartz, A., Berrier, K.L., Sparks, S., Stock-Guild, K., Toler, T.L., Vellody, K., Voelz, L., **Skotko, B.G.** (2015). National Down syndrome patient database: Insights from the development of a multi-center registry study. *American Journal of Medical Genetics Part A* 167A:2520-2526.
 23. de Graaf, G., Buckley, F., **Skotko, B.G.** (2016). Live Birth, Natural Losses, and Elective Terminations with Down Syndrome in Massachusetts. *Genetics in Medicine*. 18:459-466.
 24. Sharr C, Lavigne J, Elsharkawi IMA, Ozonoff A, Baumer N, Brasington C, Cannon S, Crissman B, Davidson E, Florez JC, Kishnani P, Lombardo A, Lyerly J, McDonough ME, Schwartz A, Berrier KL, Sparks S, Stock-Guild K, Toler TL, Vellody K, Voelz L, **Skotko BG.** (2016). Detecting celiac disease in patients with Down syndrome. *American Journal of Medical Genetics, Part A* 170A:3098-3105.
 25. Allareddy V, Ching N, Macklin E, Voelz L, Weintraub, G, Davidson E, Albers Prock L, Rosen D, Bruun R, **Skotko BG.** (2016). Craniofacial features as assessed by lateral cephalometric measurements in children with Down syndrome. *Progress in Orthodontics* 17:35-47.
 26. Kageleiry A, Samuelson D, Duh MS, Lefebvre P, Campbell J, **Skotko BG.** (2016). Out-of-pocket medical costs and third-party healthcare costs for children with Down syndrome. *American Journal of Medical Genetics Part A* 173(6):627-637.
 27. de Graaf G, Buckley F, **Skotko BG.** (2017). Estimation of the number of people with Down syndrome in the United States. *Genetics in Medicine*. 19(4):439-447.
 28. **Skotko BG,** Macklin EA, Muselli M, Voelz L, McDonough ME, Davidson E, Allareddy V, Jayaratne YSN, Bruun R, Ching N, Weintraub G, Gozal D, Rosen D. (2017). A predictive model for obstructive sleep apnea and Down syndrome. *American Journal of Medical Genetics Part A*. Apr;173(4):889-896.
 29. Elsharkawi I, Gozal D, Macklin EA, Voelz L, Weintraub G, **Skotko BG.** (2017). Urinary biomarkers and obstructive sleep apnea in patients with Down syndrome. *Sleep Medicine*. 34:84-89.

30. Rafii MS, **Skotko BG**, McDonough ME, Pulsifer M, Evans C, Doran E, Muranevicib G, Kessler P, Abushakrah S for the ELND005-DS Study Group (2017). A Randomized, Double-Blind, Placebo-Controlled, Phase II Study of Oral ELND005 (scyllo-Inositol) in Young Adults with Down Syndrome without Dementia. *Journal of Alzheimer's Disease*, 58: 401-411.
31. Lavigne J, Sharr C, Elsharkawi I, Ozonoff A, Baumer N, Brasington C, Cannon S, Crissman B, Davidson E, Florez JC, Kishnani P, Lombardo A, Lyerly J, McDonough ME, Schwartz A, Berrier K, Sparks S, Stock-Guild K, Toler T, Vellody K, Voelz L, **Skotko BG**. (2017). Thyroid dysfunction in patients with Down syndrome: Results from a multi-institutional registry study. *American Journal of Medical Genetics Part A*, 173(6):1539-1545.
32. De Graaf G, Buckley F, Dever J, **Skotko BG** (2017). Estimation of live birth and population prevalence of Down syndrome in nine U.S. states. *American Journal of Medical Genetics Part A*, 173(10):2710-2719.
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34. Hart S, Visootsak J, Tamburri P, Phuong P, Baumer N, Hernandez M-C, **Skotko BG**, Ochoa-Lubinoff C, D'Arduy X, Kishnani PS, Spiridigliozzi GA (2017). Pharmacological interventions to improve cognition and adaptive functioning in Down syndrome: Strides to date. *American Journal of Medical Genetics Part A*, 173(11):3029-3041.
35. Allyse, M., Aypar, U., Bonhomme, N., Darilek, S., Dougherty, M., Farrell, R., Grody, W., Highsmith, W.E., Michie, M., Nunes, M., Otto, L., Pabst, R., Palomaki, G., Runke, C., Sharp, R.R., **Skotko, B.**, Stoll, K. Wick, M. (2017). Offering Prenatal Screening in the Age of Genomic Medicine: A Practice Guide. *Journal of Women's Health*, 26(7):755-761
36. Diercks GR, Wentland C, Keamy D, Kinane TB, **Skotko BG**, de Guzman V, Grealish E, John Dobrowski J, Soose R, Hartnick CJ (2018). Hypoglossal Nerve Stimulation in Adolescents With Down Syndrome and Obstructive Sleep Apnea. *JAMA Otolaryngology-Head & Neck Surgery*, 2018;144(1):37-42.
37. Lazar J, Woglom C, Chung J, Schwartz A, Hsieh Y, Moore R, Crowley D, **Skotko B** (2018). Co-Design process of a smart phone app to help people with Down syndrome manage their nutritional habits. *Journal of Usability Studies* 13(2): 73-93
38. Santoro SL, Bartman T, Cua CL, Lemle S, **Skotko BG** (2018). Use of Electronic Health Record Integration for Down Syndrome Guidelines. *Pediatrics* 142(3). pii: e20174119.
39. Mucha BE, Banka S, Ajeawung NF, Molidperee S, Chen GG, Koenig MK, Adejumo RB, Till M, Harbord M, Perrier R, Lemyre E, Boucher RM, **Skotko BG**, Waxler JL, Thomas MA, Hodge JC, Gez J, Nicholl J, McGregor L, Linden T, Sisodiya SM, Sanlaville D, Cheung SW, Ernst C, Campeau PM (2018). A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. *Genetics in Medicine*. 21(5):1058-1064.

40. De Graaf G, Levine SP, Goldstein R, **Skotko BG** (2019). Parents' perceptions of functional abilities in people with Down syndrome. *American Journal of Medical Genetics Part A*. 179(2):161-176.
41. **Skotko BG**, Allyse, MA, Bajaj K, Best RG, Klugman S, Leach M, Meredith S, Michie M, Stoll K, Gregg, AR (2019). Adherence of cell-free DNA noninvasive prenatal screens to ACMG recommendations. *Genetics in Medicine*, 21(10): 2285-2292.
42. Tarui T, Im K, Madan N, Madankumar R, **Skotko BG**, Schwartz A, Sharr C, Ralston SJ, Kitano R, Akiyama S, Yun HJ, Grant E, Bianchi DW (2019). Quantitative MRI Analyses of Regional Brain Growth in Living Fetuses with Down Syndrome. *Cerebral Cortex*, [ePub ahead of print].
43. Burkardt DD, Zachariou A, Loveday C, Allen CL, Amor DJ, Ardisson A, Banka S, Bourgois A, Coubes C, Cytrynbaum C, Faivre L, Marion G, Horton R, Kotzot D, Lay-Son G, Lees M, Low K, Luk H-M, Mark P, McConkie-Rosell A, McDonald M, Pappas J, Phillippe C, Shears D, **Skotko B**, Stewart F, Stewart H, Temple IK, Mau-Them FT, Verdugo RA, Weksberg R, Zarate YA, Graham JM, Tatton-Brown K (2019). HIST1H1E heterozygous protein-truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. *American Journal of Medical Genetics Part A*, [ePub ahead of print].
44. Hart SJ, Zimmerman K, Linardic CM, Cannon S, Pastore A, Patsiogiannis V, Rossi P, Santoro SL, **Skotko BG**, Torres A, Valentini D, Vellody K, Worley G, Kishnani PS (2019). Detection of iron deficiency in children with Down syndrome. *Genetics in Medicine* [Epub ahead of print]
45. Coluzzi E, Pannone L, Andreoli C, Clementina Radio F, Pizzi S, Carpentieri G, Bruselles A, Catanzaro G, Pedace L, Miele E, Carcarino E, Ge X, Chijiwa C, Lewis MES, Meuwissen M, Kenis S, Van der Aa N, Larson A, Brown K, Wasserstein MP, **Skotko BG**, Begtrup A, Person R, Karayiorgou M, Louw Roos J, Van Gassen KL, Koopmans M, Bijlsma EK, Santen GWE, Barge-Schaapveld DQCM, Ruivenkamp CAL, Hoffer MJV, Lalani SR, Streff H, Craigen WJ, Graham BH, van den Elzen APM, Kamphuis DJ, Öunap K, Reinson K, Pajusalu S, Wojcik MH, Viberti C, Di Gaetano C, Bertini E, Petrucci S, De Luca A, Rota R, Ferretti E, Matullo G, Dallapiccola B, Sgura A, Walkiewicz M, Kooy RF, Tartaglia M (2019). Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. *Am J Hum Genet.*, Sep 5;105(3):493-508.
46. Ilacqua A, Benedict J, Shoben A, **Skotko BG**, Matthews T, Benson B, Allain DC (2020). Alzheimer's disease development in adults with Down syndrome: Caregivers' perspectives. *Am J Med Genet Part A*, 182(1):104-114.
47. Santoro SL, Cannon S, Capone G, Franklin C, Hart SJ, Hobensack V, Kishnani PS, Macklin EA, Manickam K, McCormick A, Nash P, Oreskovic NM, Patsiogiannis V, Steingass K, Torres A, Valentini D, Vellody K, **Skotko BG** (2019). Unexplained regression in Down syndrome: 35 cases from an international Down syndrome database. *Genet Med*. [Epub ahead of print]

Other Peer-reviewed Publications

1. **Skotko, B.**, Kishnani, P., & Capone, G. for the Down Syndrome Diagnosis Study Group (2009). Prenatal diagnosis of Down syndrome: How best to deliver the news. *American Journal of Medical Genetics, Part A*, 149A: 2361-7.
2. **Skotko, B.**, Capone, G., & Kishnani, P. for the Down Syndrome Diagnosis Study Group (2009). Postnatal diagnosis of Down syndrome: Synthesis of the evidence on how best to deliver the news. *Pediatrics*, 124: e751-e758.
3. **Skotko, B.** (2009). With new prenatal testing, will babies with Down syndrome slowly disappear? *Archives of Disease in Childhood*, 94: 823-826.
4. Diercks, G.R., Keamy D., Kinane, T.B., **Skotko, B.**, Schwartz, A., Grealish, E., Dobrowski, J., Soose, R., Hartnick, C. (2016). Hypoglossal Nerve Stimulator Implantation in an Adolescent with Down Syndrome and Sleep Apnea. *Pediatrics* 137(5):e20153663.
5. Jacobs, J., Schwartz, A., McDougle, C., **Skotko, B.G.** (2016). Rapid Clinical Deterioration in an Individual with Down Syndrome. *American Journal of Medical Genetics Part A*. Early view online.

Non-peer reviewed scientific or medical publications/materials in print or other media

Reviews, chapters, monographs and editorials

1. **Skotko, B.**, Tenenbaum, A. (2016). Down syndrome. In Rubin, L., Merrick, J. (eds.) *Health Care for People with Intellectual and Developmental Disabilities Across the Lifespan*. 4th edition. Medford, MA: Springer Publishing.
2. Roberts, M., **Skotko, B.** (2016, 2017, 2018, 2019). Down syndrome. In Domino FJ (Ed.), www.5MinuteConsult.com, Philadelphia: Wolters Kluwer.

Letters to the Editor

1. **Skotko, B.** (2006). A surprising postnatal diagnosis. (Letter to the Editor) *Obstetrics and Gynecology*, 108: 1297.
2. **Skotko, B.** (2006). Words matter: The importance of nondirective language in first-trimester assessments for Down syndrome. (Letter to the Editor). *American Journal of Obstetrics and Gynecology*. 195:625-26.
3. **Skotko, B.** (2006). Comparing Three Screening Strategies for Combining First- and Second-Trimester Down Syndrome Markers. (Letter to the Editor). *Obstetrics & Gynecology*. 107:1170.
4. **Skotko, B.** (2007). First- and Second-Trimester Evaluation of Risk for Down Syndrome. (Letter to the Editor) *Obstetrics and Gynecology*, 110: 1426.

5. **Skotko, B.G.**, Leach, M. (2011). Physicians need to offer up-to-date information about Down syndrome to expectant couples to inform decision-making [E-letter]. *Pediatrics*. October 17, 2011. http://pediatrics.aappublications.org/content/128/2/393.full/reply#pediatrics_el_51755 (Accessed October 17, 2011.)
6. Leach, M., **Skotko, B.G.** (2012). Resources available for informed prenatal decisions. (Letter to the Editor). *Genetics in Medicine*: 14:348-349.
7. **Skotko, B.G.**, Samuelson, D., Kageleiry, A., Lefebvre, P., Hellstern, M., Campbell, J. (2019). Comment on “The price of abandoning diagnostic testing for cell-free DNA screening.” (Letter to the Editor). *Prenatal Diagnosis*: 39(2):130.

Clinical Guidelines and Reports

1. Gregg, A.R., Gross, S.J., Best, R.G., Monaghan, K.G., Bajaj, K., **Skotko, B.G.**, Thompson, B.H., Watson, M.S., are the Noninvasive Prenatal Screening Working Group of the American College of Medical Genetics (2013). ACMG Statement on Noninvasive Prenatal Screening for Fetal Aneuploidy. *Genetics in Medicine* 15(5): 395-398. [Consensus Statement, which is now being implemented nationally by providers. This was developed as part of a Working Group. My contribution was to help review the literature, discuss recommendations with national expert colleagues, and draft/review/approve final manuscript.]
2. Gregg, A.R., **Skotko, B.G.**, Benkendorf, J.L., Monaghan, K.G., Bajaj, K., Best, R.G., Klugman, S., and Watson, M.S.; on behalf of the ACMG Noninvasive Prenatal Screening Work Group. (2016). Noninvasive prenatal screening for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics. *Genetics in Medicine* 18(10):1056-1065. [Consensus Statement, which is now being implemented nationally by providers. This was developed as part of a Working Group and an update to the previous one in 2013. My contribution was to help review the literature, discuss recommendations with national expert colleagues, and draft/review/approve final manuscript.]

Abstracts, Poster Presentations and Exhibits Presented at Professional Meetings

1. Skotko, B.G., Samuelson, D., Kagleiry, A., Duh, M.S., Lefebvre, P. Campbell, J., DerSarkissian, M., Hellstern, M. (2017). Income growth trajectory for parents of children with Down syndrome in the United States. Poster presented at the American College of Medical Genetics and Genomics annual conference in Phoenix, AZ.

Narrative Report

I have dedicated my professional energies toward children with cognitive and developmental disabilities, with a particular focus and expertise on Down syndrome. I spend approximately 80% of my time dedicated to research activities, 15% on clinical excellence, and 5% on administrative responsibilities.

In the area of **Clinical Expertise and Innovation**, I helped launch two nationally recognized Down syndrome clinics, first at Boston Children’s Hospital and now at Massachusetts General Hospital. I am

frequently invited to give lectures and Grand Rounds on topics related to Down syndrome around the world. My scholarship has also included two books about Down syndrome, and more than 20 other first- or senior-author, peer-review manuscripts about clinical care, many of which have resulted in national television and radio appearances. I most recently was first author on a chapter about clinical care for patients Down syndrome in a definitive internationally utilized textbook for medical provides. Under my leadership, our clinic also built the first-ever HIPAA-compliant electronic intake form allows families and caregivers to input clinically relevant information in advance of clinical appointments at the MGH Down Syndrome Program. I also created and oversee a national, longitudinal, multi-center Down syndrome patient registry, which has resulted in peer-review publications, helping to change the way that clinical care is delivered by providers across the country. I have also served on committees for the American College of Medical Genetics and Genomics, helping to write practice guidelines for prenatal testing for Down syndrome and other genetic conditions.

In the area of **Teaching and Education**, I have formally supervised numerous trainees, and I have also educated fellows on an annual basis about genetic and metabolic disorders. In addition to the two nationally available CME courses that I have created on topics related to Down syndrome, I have given over the past five years more than 150 lectures and Grand Rounds in academic settings, including many invited international presentations in Sweden, Japan, Brazil, Singapore, Paris, and UK, among others. My clinical supervision including precepting in my medical genetics clinic and Down Syndrome Program at Massachusetts General Hospital. I also serve as one of the supervising attendings to the fellows in our Harvard Medical School Medical Genetics Training Program during their rotations at Massachusetts General Hospital. Overall, I find my teaching and mentorship responsibilities to be deeply gratifying. What a privilege to be involved with so many accomplished and eager students and residents!

In the significant supporting activity of **Investigation**, I have served as principal investigator in two main areas of research: (1) interdisciplinary, translational research for patients with Down syndrome and (2) informed decision-making with prenatal genetic testing.

(1) With nearly \$5 million in NIH, HRSA, PCORI, and industry funding, I am leading a 12-person research team investigating a novel method to screen for obstructive sleep apnea in patients with Down syndrome. I have also been the Principal Investigator on multiple significant Phase II clinical drug trials, the first in the country for patients with Down syndrome. My team and I are building and testing a virtual Down syndrome clinic, enabling patients with Down syndrome across the country to have access to high level clinical care.

(2) Combining robust datasets, my team and I have also looked at the impact of prenatal testing over the years, estimating the number of livebirths, natural losses, and selective terminations with Down syndrome from 1900–2010. I also led a 29-member research team of Down syndrome experts around the country to write the most definitive research review on how expectant parents can make informed decisions regarding prenatal testing for Down syndrome. My team and I also conducted a robust assessment of family life when a member has Down syndrome. I also helped co-author a policy statement, now widely used, for the American College of Medical Genetics and Genomics on noninvasive prenatal screening.

In the significant supporting activity of **Education of Patients and Service to the Community**, I have co-authored two award-winning books about Down syndrome for families and am frequently asked to provide expert commentary in national print, radio, and television media, including the *New York Times*, *The Wall Street Journal*, ABC News, NPR, and CNN, among others. I have enjoyed the deeply fulfilling experience of speaking to parents, caregivers, and patients at conferences organized by non-profit

disability organizations. My more than 200 talks over the years have provided meaningful opportunities to engage with families as far away as Japan, Netherlands, India, Sweden, and South Africa, among others. Humbled, I have been honored with the highest honors from the Down syndrome community including awards from the National Down Syndrome Society and National Down Syndrome Congress. I have also enjoyed serving as Chairman of the Board of Directors for the Massachusetts Down Syndrome Congress and have also served on the non-profit boards of the American Red Cross, the National Down Syndrome Society, and the Band of Angels Foundation.