

## CURRICULUM VITAE

Susan E Howell, M.S., C.G.C., M.B.A

Current Position: Associate Professor, Section of Genetics and Metabolism  
Department of Pediatrics, University of Colorado School of Medicine  
Board Certified Genetic Counselor, eXtraordinary Kids Clinic, Fragile X Clinic and  
Turner Syndrome Clinic, Children's Hospital Colorado  
Address: 13123 East 16th Ave Box 140, Aurora, CO 80045  
Phone/Fax: Ph: 720-777-8361; Fax: 720-777-7868  
Email: susan.howell@childrenscolorado.org

### EDUCATION

1992-1996 Bachelor of Arts in Molecular, Cellular, and Developmental Biology  
University of Colorado - Boulder, Colorado  
1997-1999 Master of Science in Medical and Molecular Genetics  
Indiana University Purdue University, Indianapolis, Indiana  
2002-2004 Master of Business Administration, University of Denver- Denver, Colorado

### ACADEMIC APPOINTMENTS

2016– 2021 Sr. Clinical Instructor, Department of Pediatrics  
University of Colorado School of Medicine, Aurora, CO  
2021-2023 Assistant Professor, Department of Pediatrics, Section of Developmental-Behavioral  
Pediatrics, University of Colorado School of Medicine  
2023- 2025 Assistant Professor, Department of Pediatrics, Section of Genetics and Metabolism,  
University of Colorado School of Medicine  
2025-current Associate Professor, Department of Pediatrics, Section of Genetics and Metabolism,  
University of Colorado School of Medicine

### HOSPITAL POSITIONS

2008-2023 Board-Certified Genetic Counselor, Clinic and Research Coordinator, eXtraordinary  
Kids Clinic, and Fragile X Clinic, Children's Hospital Colorado, Section of  
Developmental-Behavioral Pediatrics, Department of Pediatrics, University of Colorado  
School of Medicine  
2010- current Director of Clinic Development, Marketing and Community Outreach, eXtraordinary  
Kids Clinic, Children's Hospital Colorado, Section of Developmental-Behavioral  
Pediatrics, Department of Pediatrics, University of Colorado School of Medicine  
2015-2018 Board-Certified Genetic Counselor and Clinic Coordinator, eXtraOrdinary Kids Turner  
Syndrome Clinic, Children's Hospital Colorado, Section of Developmental-Behavioral  
Pediatrics, Department of Pediatrics, University of Colorado School of Medicine  
2018- current Board-Certified Genetic Counselor, eXtraOrdinary Kids Turner Syndrome Clinic,  
Children's Hospital Colorado, Section of Endocrinology, Department of Pediatrics,  
University of Colorado School of Medicine  
2023- current Board-Certified Genetic Counselor, Clinic and Research Coordinator, eXtraordinary  
Kids Clinic, and Fragile X Clinic, Children's Hospital Colorado, Section of Genetics and  
Metabolism, Department of Pediatrics, University of Colorado School of Medicine

### GOVERNMENT AND OTHER PROFESSIONAL POSITIONS

1999 Prenatal Genetic Counselor, Washington University School of Medicine, St. Louis,  
MO  
1999-2002 Regional Genetic Counselor, Department of Health and Human Services, State of  
North Carolina  
2003-2004 Marketing and Business Development Associate, OSI Pharmaceuticals, Inc.,  
Boulder, CO  
2004-2006 Director of Clinical Development, SmartCare Family Medical Centers, Greenwood  
Village, CO  
2006-2007 Healthcare Analyst, Redstone Venture Partners, Inc., Englewood, CO

2007-2008 Genetic Counselor and Marketing Director, Sciona, Inc., Boulder, CO

### **HONORS, SPECIAL RECOGNITIONS AND AWARDS**

2002-2004 Alma C. Swanson Academic Achievement Scholarship, University of Denver-Daniels College of Business, Denver, CO

2022 Carolyn Mills Lovell Genetic Counselors Award presented to genetic counseling mentee for primary mentored capstone project abstract, American College of Medical Genetics 2022 Annual Clinic Genetics Meeting, Nashville, TN

2023 Top Rated Poster Award, American College of Medical Genetics 2023 Annual Clinic Genetics Meeting, Salt Lake City, UT

2024 Western Society of Pediatric Research (WSPR) Mead Johnson Nutrition Junior Faculty Award, most meritorious abstract in section of Genetics per peer review, 2024 WSPR meeting, Carmel, CA (>100 abstracts submitted to section of Genetics)

2024 Top Rated Poster Award, American College of Medical Genetics 2024 Annual Clinic Genetics Meeting, Toronto, Canada

2025 Team Science Award, Colorado Child Health Research Institute, CU Department of Pediatrics, and ACCORDS, Aurora CO

### **MEMBERSHIPS IN PROFESSIONAL ORGANIZATIONS**

1998-current National Society of Genetic Counselors (NSGC)

2007-2010 American Society of Human Genetics (ASHG)

2008-2010 American College of Medical Genetics (ACMG)

2008-2024 Mountain States Genetics Network (MOSTGENE)

2013-current Colorado Genetic Counseling Educational Symposium, Founder and Co-Chair (<https://sites.google.com/view/cogc/home>)

### **MAJOR COMMITTEE AND SERVICE RESPONSIBILITIES**

#### Hospital/Departmental

2017 Chair for the Search Committee for the FORWARD Fragile X Research coordinator

2021-current Co-chair for 6 Search Committees for the eXtraordinary Kids Clinic Professional Research Assistant

2022-current Co-chair, eXtraordinary Kids Program semi-annual Journal Club

2023 Co-chair for Search Committee for the eXtraordinary Kids Clinic and Fragile X Clinic Clinical Advanced Practice Provider

#### University of Colorado School of Medicine

2023-current Invited member of the Mid-Career Faculty Working Group (MCFWG), dedicated to the mission of providing crucial support to colleagues, Department of Pediatrics, School of Medicine, and Children's Hospital Colorado.

2013-current Founder and Co-Chair, Colorado Genetic Counseling Education Symposium (<https://sites.google.com/view/cogc/home>)  
Key responsibilities: Organizing and directing full day, semi-annual professional educational events for genetic counselors in the Rocky Mountain region (Two events per year, ~10 speakers/event, ~95 attendees, coordination of onsite lunch, poster sessions, hybrid format to provide for both virtual attendees and virtual presenters, website announcements and materials, CEU management), including submission/approval for continuing educational credits from the National Society of Genetic Counselors (NSGC) and website announcements and materials

2015- current Genetic Conditions Consensus Group- Turner Syndrome Working Group

2023- current Founder and Co-Chair for the Colorado Genetic Counseling Grant Awards Committee  
Key responsibilities: Recruitment, organizing and co-directing monthly committee meetings (10 members, 60 min per meeting) for the development of a Colorado funded Genetic Counseling grant funding mechanism funded by fees and sponsorships accumulated through the Genetic Counseling Educational Symposium beginning in 2024 for the purpose of increasing the impact of work being done by genetic counselors, both students and practicing counselors. Awards given since inception:

2024- \$500 GC student capstone research award, Olivia Isaacks  
2025- \$300 GC student capstone research award, Gabriela Ramirez-Renta  
2025- \$500 PEAK GC Professional Award, Janet Bell, CGC

#### Local Community Committees and Advisory Boards

2010- current Advisory Board Member, Fragile X Colorado Community Support Network  
2015- current Advisory Board Member, Rocky Mountain Chapter, AXYS regional support group

#### National Organizations and Committees

2008- current National Fragile X Foundation Clinic and Research Consortium (FXCRC), Clinical Committee, Key responsibilities: Authorship, coordination and review of consensus clinical care documents guiding clinical practice for 26 national Fragile X clinics and dissemination to local providers

2008-2011 Klinefelter Syndrome & Associates – (National) Conference Committee, Key responsibilities: Agenda development, speaker recruitment, hosting and logistic coordination for event at CHCO for national attendance (~180 attendees)

2014- current Association for X & Y Syndromes (AXYS) - Scientific Advisory Committee  
Key responsibilities: Authorship, coordination and review of manuscripts and research proposals requesting recruitment through the AXYS membership

2014- current Association for X & Y Syndromes (AXYS) - (National) Conference Committee, Key responsibilities: Agenda development, speaker recruitment, logistic coordination for event for national attendance (~180 attendees)

2015- 2024 Association for X & Y Syndromes (AXYS) – Board of Directors  
Key responsibilities: Participate in monthly board of director meetings with deliverables of fundraising initiatives, budget oversight, spending approvals, as well as updates on efforts from personal participation in advocacy, research and support initiatives.

2019- current Association for X & Y Variations (AXYS), Clinic and Research Consortium (ACRC)- Advisory Committee. Key responsibilities: Authorship, coordination and review of research proposals and consensus clinical care documents guiding development and practice for 8 national sex chromosome specialty clinics

2020- current Turner Syndrome Global Alliance (TSGA) INSIGHTS (*Inspiring New Science in Guiding Healthcare in Turner Syndrome*) Research Registry- Steering Committee  
Key responsibilities: Participate in monthly steering committee meetings discussing the development, policies and procedures for the INSIGHTS registry launch and ongoing recruitment, with manuscript contributions from the registry data and review of submitted research proposals and manuscripts per the registry policies.

2020 Research Support for National Fragile X Foundation, Virtual Patient-Focused Drug Development meeting with Federal Drug Administration

2021- current GALAXY (*Generating Advancements through Longitudinal Analysis in X and Y Syndromes*) Research Registry- Steering Committee member  
Key responsibilities: Participate in monthly steering committee meetings discussing the development, policies and procedures for the GALAXY registry launch and ongoing national recruitment, with manuscript contributions from the registry data and review of submitted research proposals and manuscripts per the registry policies. Present to local and national audiences about why the registry is important in advancing the science of sex chromosome aneuploidies/variations.

2023- current National Fragile X Foundation- Board of Directors member  
<https://fragilex.org/blog/welcome-class-of-2023-board/>  
Key responsibilities: Participate in quarterly Board of Director meetings (4/year, alternating virtual and in-person) with deliverables of fundraising initiatives as well as review of budgets, financial oversight, guiding initiatives of advocacy, research and development.

2023 FXCRC Annual Meeting Committee.  
Key responsibilities: participate in the organization and support for hosting the 2023 FXCRC Annual meeting (50 attendees from all FXCRC clinics across the US),

- including facility reservations, coordination of lunch, tech support on site, and day of logistical needs.
- 2025 NASCARR Communications and Dissemination Committee, Co-Chair with Dr. Chijioke Ikomi

#### International Organizations and Committees

- 2025-2026 Klinefelter Syndrome International Clinical Practice Guidelines, Co-Lead for the Clinical Care Practices sub-committee alongside Dr. Michael Zitzmann.

### **PROFESSIONAL CERTIFICATIONS AND LICENSURE**

#### **Board Certified Genetic Counselor**

- 2002 American Board of Genetic Counseling (ABGC) Comprehensive Certification (by exam)
- 2012,2017,2022 Recertification, American Board of Genetic Counseling (ABGC)
- 2017 Pharmacogenomics Certificate, Skaggs School of Pharmacy, University of Colorado School of Pharmacy

### **PROFESSIONAL DEVELOPMENT**

- 2017 *Pharmacogenomics Certificate Program*, Skaggs School of Pharmacy, University of Colorado School of Pharmacy.  
Summary: An 8-week practice-based continuing education program focused on the application of pharmacogenetic information to clinical practice and appropriate therapeutic interventions, including interpretation of pharmacogenomic test results, literature evaluation and use of evidence-based guidelines, to facilitate the implementation of pharmacogenomics in different health care settings
- 2020 – 2021 *Teaming 101, Teaming for Early Career Research Scientists*, Colorado Clinical and Translational Sciences Institute (CCTSI), University of Colorado Anschutz  
Summary: curriculum incorporates online coursework followed by 6 individual workshops designed to build capacity for participating in and leading effective interdisciplinary and translational scientific teams, addressing a teamwork model of conducting clinical and translational research
- 2021 *Sitefinity Training*, University of Colorado, Anschutz  
Summary: 4-hour online training provided by the university OIT Web Services + Web DevOps teams for the utilization of the Sitefinity web design software to develop university websites specific to the promotion and support of clinical and research programs, dissemination of publications and recruitment and retention of participants for research initiatives
- 2022 *Updates in Health Disparities in Medical Genetics*, American College of Medical Genetics (ACMG), 3 hour online continuing education course addressing various topics of implicit bias in genetic counseling.
- 2023 *Communicating Your Science to the Public* workshop series, Colorado Clinical and Translational Sciences Institute (CCTSI), University of Colorado Anschutz. 5 hour virtual course addressing communication topics for interfacing live with a public audience.
- 2024 *Mentorship Academy*. Hosted by University of Colorado Department of Medicine and Michigan Medicine. In person, all day event that includes Ted Talk-style sessions, a keynote speaker, and breakout rooms to facilitate discussion and provide actionable advice and guidance for faculty and staff interested in developing their mentorship skills.
- 2024 *Social Media Strategies for Effective Fundraising*. The Fund Raising School IU Lilly Family School of Philanthropy. 9-hour online training course addressing various topics, skills, and tool for successful fundraising utilizing social media platforms.

### **REVIEW AND REFEREE WORK**

#### Grant Review Committees

- 2019 All of US Genetic Counseling Resource grant reviewer (NIH). Key responsibilities: Prepare reviews for three primary submissions as well as participate onsite for a single day the review and discussion of proposals to the National Institutes of Health, Funding Opportunity: [OT-PM-19-001](#). This funding opportunity was designed to establish the All of US Genetic Counseling Resource (an awarded national network of genetic

counselors) to support the National Institutes of Health's *All of Us* Research Program in returning personalized, actionable health-related DNA results to more than 155,000 participants nationwide.

Ad Hoc Reviewer for Professional Peer Reviewed Journals and Websites

2019	Genetics Home Reference (U.S. Library of Medicine)
2021	PeerJ
2021	Prenatal Diagnosis
2023	Birth Defects Research
2024	Clinical and Experimental Obstetrics and Gynecology
2024	European Child & Adolescent Psychiatry
2024	Reproductive BioMedicine Online
2025	Case Reports in Pediatrics
2025	American Journal of Medical Genetics, part A

**INVITED LECTURES AND PRESENTATIONS**

\* Indicates that presentation was recorded and is available as educational material for families or online.

National/International

2008, Aug	"How to talk with your kids about X&Y chromosome variations," KS&A Mile Hi X&Y Chromosome Variations Conference, Denver, CO
2009, July	"How To Talk With Your Child About His or Her Diagnosis," I Love L.A. Families Conference with KS&A, AAKSIS and the XXYY Project. Los Angeles, CA.
2010, July	"Trisomy X Roundtable- Girls Only," Call of the Wild Families Conference with KS&A and AAKSIS. Mason, OH
2010, July	"How Did This Happen? Genetics 101," Call of the Wild Families Conference with KS&A, AAKSIS. Mason, OH
2010, July	"How to Talk With Your Child About His or Her Diagnosis," Call of the Wild Families Conference with KS&A, AAKSIS. Mason, OH
2011, July	"SibShop for Siblings of Children with 48,XXYY," XXYY Project Symposium. Boston, MA
2012, March	"Trisomy X: Expanding the Phenotype", Abstract Oral Presentation, American College of Medical Genetics Annual Clinical Genetics Meeting, Charlotte, NC
2012, July	"SibShop for Siblings of Children with 48,XXYY," XXYY Project Symposium. Estes, CO
2013, March	"From Screening to Treatment: A Paradigm Shift in Understanding Fragile X and FMR1 Disorders" Concurrent Session, American College of Medical Genetics Annual Clinical Genetics Meeting, Phoenix, AZ
2013, July	"Genetics 101" * KS&A Families Conference. Aurora, CO <a href="https://youtu.be/JBUK8EYYPvo">https://youtu.be/JBUK8EYYPvo</a>
2013, July*	"Trisomy X: An Overview" KS&A Families Conference. Aurora, CO <a href="https://youtu.be/IAKp8-sg0ql">https://youtu.be/IAKp8-sg0ql</a>
2014, July	"SibShop for Siblings of Children with 48,XXYY," XXYY Project Symposium. Estes, CO
2014, Sept	"Sex Chromosome Aneuploidies: A Multidisciplinary Perspective on Counseling and Current Treatment Recommendations," 2024 National Society of Genetic Counselors (NSGC) Annual Meeting, New Orleans, LA
2015, July	"How to Talk With Your Child About The Diagnosis," 2015 AXYS Families Conference, Baltimore, MD
2016, July	"From FX101 to Agenda Planning," 15 <sup>th</sup> International Fragile X Conference, San Antonio, TX
2017, June	"Trisomy X" AXYS Families Conference Keynote Address, Aurora, CO. Trisomy X Panel, with S Howell MS, R Wilson PhD, K Wigby MD, 2017 AXYS Families Conference Keynote Address, Aurora, CO, Jun 2017
2017, June*	"Disclosing the Diagnosis," 2017 AXYS Families Conference. Aurora, CO <a href="https://youtu.be/ADwzWD0KmT0">https://youtu.be/ADwzWD0KmT0</a>
2017, June	"From Diagnosis to Agenda Planning," AXYS Families Conference. Aurora, CO.

2017, July "The Genetics of Turner Syndrome," Turner Syndrome of the United States (TSSUS) Families Conference, Greenwood Village, CO

2018, May\* "Disclosing the Diagnosis," AXYS Webinar Series <https://youtu.be/KVbD0jG1BjE>

2019, June "From Diagnosis to Agenda Planning for First Time Attendees," 2019 AXYS Family Conference, Atlanta, GA

2019, June "Infant Retreat," AXYS Families Conference. Atlanta, GA. Empowering Parents of Infants and Toddlers with X&Y Chromosome Variations: Retreat, with Shanlee Davis, MD, Rebecca Wilson PsyD, and Nicole Tartaglia, MD, 2019 AXYS Family Conference, Atlanta, GA

2019, June "Interview With Self-Advocates," 2019 AXYS Family Conference, Atlanta, GA,

2021, Feb\* "Risk for Early Ovarian Failure in Girls with Trisomy X Syndrome" AXYS Virtual Webinar series, Co-Presentation with N Tartaglia and S Davis, 2021 AXYS Webinar Series, <https://youtu.be/YyC-tbjrZl8>

2021, April "Impacting eXtraordinary: Klinefelter Syndrome at the Intersection of Research, Clinical Care and Community," Keynote presentation for the American Society of Andrology, Virtual

2021, June "GALAXY Registry: Generating Advancements through Longitudinal Analysis in X and Y Syndromes," 2021 AXYS Families Conference, Virtual

2021, June "X and Y Chromosome Disorders: A Growing Need for Understanding These Common Conditions." Invited speaker for Temple University, College of Education, Early Childhood and Interventions course, Virtual

2022, Feb "Prenatal Diagnosis for Turner Syndrome: An Update" Presenter for the State of the Art symposium for the Pediatric Endocrine Society Annual Meeting, Virtual

2022, April "Multidisciplinary Turner Syndrome Care: Updates for the Pediatrician and Pediatric Subspecialist", Pediatric Academic Societies Annual Meeting, Turner Syndrome Panel Symposium, Denver, CO, April 2022

2022, April "An Extra X or Y Chromosome: Implications, Clinical Care and Hot Topics for Sex Chromosome Trisomies", Pediatric Academic Societies Annual Meeting, Panel Discussion, Denver, CO, April 2022

2022, Jul\* Females with Fragile X Syndrome Panel Discussion, with N Tartaglia, R Shaffer, J Barber, B Haas-Gilver, 18<sup>th</sup> International Fragile X Conference, San Diego, CA Jul 2022

2022, Sept\* "Non-invasive Prenatal Testing (NIPT) Results for Participants of the eXtraordinary Babies Study: Screening, Counseling, Diagnosis, and Discordance" 3<sup>rd</sup> International Workshop on Klinefelter Syndrome, Trisomy X and XYY, Leiden, Netherlands, Sept 2022

2022, Oct "From Pathogenesis to Targeted Treatments: A Paradigm Shift in Understanding Fragile X and Family Experiences Navigating this Inherited Condition" Canadian Association of Genetic Counsellors Annual Education Conference, Virtual Oct 2022, >500 Genetic counselor attendees nationwide for an 80 minute presentation alongside Francois Balduc, MD, and two representatives of families on the Board of Directors for the Canadian Fragile X Advocacy Organization

2023, March "The eXtraordinary Babies Study: a Natural History of Prenatally Identified Infants with Sex Chromosome Trisomies (SCTs): Analysis of Birth Defects and Medical Problems in the First Year of Life" Abstract Oral Presentation, American College of Medical Genetics Annual Clinical Genetics Meeting, Salt Lake City, UT. Approximately 50 attendees

2023, June\* "Introductions to X&Y Chromosome Variations" Series of presentations on XXY, XXX, XYY, XXYY, XXXY and mosaicism. 2023 AXYS Families Conference, Virtual.  
<https://www.youtube.com/watch?v=f3XxOZ26eeg>,  
[https://www.youtube.com/watch?v=xYqhyR8MR\\_w](https://www.youtube.com/watch?v=xYqhyR8MR_w),  
<https://www.youtube.com/watch?v=ItyG7J42dP0>,  
<https://www.youtube.com/watch?v=GK0ESgtsN10>,  
<https://www.youtube.com/watch?v=Vx3nB3YwUBs>,  
<https://www.youtube.com/watch?v=WlnypVgiPqA>

- 2023, June "Updates from the eXtraordinary Babies Study: 290 Infants with a Prenatal Diagnosis of Sex Chromosome Trisomy (XXY, XXX, XYY) and XXYY." 2023 AXYS Families Conference, Virtual.
- 2023, Oct "Genetic Counseling for FMR1 Associated Disorders" In person presentation about pedigree analysis and cascade testing and counseling in families affected with Fragile X and associated disorders (FXTAS, FXPOI, FXAND) when indicated to prompt group discussions about expansion of clinical services to meet various needs among families. FXCRC Professionals Meeting, approx. 50 in attendance. Aurora, CO.
- 2023, Dec\* National Fragile X Foundation (NFXF) MasterClass™ - "Fragile X Genetics" a recorded webinar for genetic counselors about the genetics and implications in FMR1 associated conditions, co-presented with Matthew Walsh, LCGC at Emory University. Approved for category 1 CEUs for continuing education of genetic counselors through the National Society of Genetic Counselors (NSGC), on demand until 2025, [www.nsgc.org](http://www.nsgc.org).
- 2024, May "Genetic Counseling in XXYY." 2024 XXYY UK Families Conference, approximately 45 in attendance. Castleton, United Kingdom.
- 2024, July "Enabling Choices: Navigating Genetic Counseling and Reproductive Options for Individuals with an FMR1 mutation." 19<sup>th</sup> International Fragile X Conference, Orlando, FL Jul 2024
- 2024, Sept "Not Just a Carrier: Medical and Emotional Impact of Being a Carrier for an X-Linked Disorder." Educational Breakout Session Presentation, 2024 National Society of Genetic Counselors (NSGC) Annual Conference, New Orleans, LA, September 2024.
- 2024, Dec "Fragile X Syndrome Expert Preceptorship" Onsite educational training experience to Shionogi colleagues teaching how care is delivered, to allow Shinogi to better understand and meet the educational and research needs for Fragile X treatments and better understand patient experiences to inform strategy and tactics throughout the Shionogi organization.
- 2025, July "Understanding X and Y Variations: Genetic Diagnosis, Terminology & Next Steps Planning" 2025 AXYS Families Conference, Atlanta, GA, ~45 attendees
- 2025, July "Disclosing the Diagnosis- Panel Discussion" 2025 AXYS Families Conference, Atlanta, GA, ~35 attendees
- 2025 Oct "Genetic Counseling for Sex Chromosome Trisomies: Insights from Parental Experiences." 4<sup>th</sup> International Workshop on Klinefelter Syndrome, Trisomy X and XYY, Padua, Italy Oct 2025 ~100 attendees
- 2025 Nov "A Twist on a Classic: Management and Counseling Nuances in Turner Syndrome and Incidental Mosaicism." 2025 National Society of Genetic Counselors (NSGC) Annual Meeting, Seattle, WA. ~200 attendees
- 2026 Feb\* "Turner Syndrome Prenatal Counseling" Pediatric Endocrine Society, Turner Syndrome Special Interest Group (PES SIG) February 2026, Virtual. 41 attendees.

#### Regional

- 2017, April "Delineating the Phenotype in Triple X Syndrome," Seminars in Genetics Webinar, Kaiser Permanente Northern California Regional Genetics Department\* >150 attendees
- 2019, May "Infants with Sex Chromosome Trisomies Identified by NIPT: Preliminary Profile of Neurodevelopmental Skills in Participants with 47,XXY in The eXtraordinary Babies Study", 3rd Annual Spring Pennsylvania Association of Genetic Counselors Meeting, Harrisburg, PA
- 2019, June "eXtraordinary Kids: Perspectives on Counseling Sex Chromosome Aneuploidies," 11th Philadelphia Prenatal Conference, Philadelphia, PA, >500 attendees
- 2019, July "Clinical and Research Update on Klinefelter Syndrome and Sex Chromosome Aneuploidies," Webinar for Kaiser Permanente California CME, Northern California Regional Genetics Department
- 2021, April "Fragile X Premutation and Associated Disorders" Webinar with Dr. Maureen Leehey for Natera Rocky Mountain Region, Virtual

- 2023, May "Sex Chromosome Aneuploidies: Overview of Clinical Features and Care" Virtual presentation to the California State Department of Health and Education to the Prenatal Screening SCA Workgroup to establish the noninvasive prenatal screening mandate in SCAs, 35 attendees
- 2023, Nov "The eXtraordinary Babies Study: a Natural History of Prenatally Identified Infants with Sex Chromosome Trisomies (SCTs): Analysis of Birth Defects and Medical Problems in the First Year of Life" In person 60min presentation at the Maryland/DC genetic counseling conference (MDCGC), approximately 100 attendees
- 2024, Jan "eXtraordinary Babies Study: Genetic Counseling Implications of Discordant Prenatal Noninvasive Screening Results in Sex Chromosome Aneuploidies" 2024 Western Society for Pediatric Research, Carmel, CA, approximately 50 attendees \*Mead Johnson Nutrition Junior Faculty Award, most meritorious abstract in Genetics (>100 abstracts submitted to Genetics)
- 2024, Dec "Sex Chromosome Aneuploidies: Overview of Clinical Features and Care" Virtual presentation to the California State Department of Health and Education to the Prenatal Screening SCA Workgroup to inform postnatal care for infants identified by noninvasive prenatal screening mandate in SCAs, ~50 attendees

Local

- 2013, April "Research Updates on Sex Chromosome Aneuploidies and Fragile X Syndrome," Clinical Genetics and Metabolism Grand Rounds, University of CO, Aurora, CO
- 2013, Nov "Fragile X Syndrome: An Update on Counseling and Treatment Options for Patients with Fragile X Syndrome," 1st Semi-Annual Colorado Genetic Counselors Symposium, Aurora, CO
- 2013, Nov "Genetics of Fragile X syndrome," 2<sup>nd</sup> Bi-Annual Colorado Fragile X Conference, Aurora, CO
- 2017, April "Applying Supervision Models to Your Practice Panel Discussion," Semi-Annual Colorado Genetic Counselors Education Symposium, Aurora, CO
- 2016, Nov "Fragile X Genetics and Inheritance," Annual Colorado Fragile X Fall Conference, Aurora, CO
- 2017, Oct "Increasing Prenatal Diagnosis of Sex Chromosome Aneuploidies," Kaiser CO Maternal Fetal Medicine, Denver, CO
- 2017, Nov "Prenatal Diagnosis of Sex Chromosome Aneuploidies," Colorado Center for Reproductive Medicine, Lone Tree, CO
- 2018, March "Fragile X, Denver Clinic and Research Update", Genetic Grand Rounds, University of Colorado, Aurora, CO
- 2018, Sept "The Genetics of Turner Syndrome," Turner Syndrome Global Alliance (TSGA) Colorado Families Conference, Littleton, CO
- 2018, Oct "Latest Treatments in SMA and Clinical Advances in Sex Chromosome Aneuploidies (SCA)" Colorado Dinner Lecture Series, Centennial, CO
- 2018, Oct "Sex Chromosome Aneuploidy: Genetic Counseling and Follow-up Care and Research for Infants with a Prenatal Diagnosis, with Dr. Nicole Tartaglia, MD, MS, Natera CME Lecture: Northern Colorado OB/GYN and Maternal Fetal Medicine, 24 attendees, Firestone, CO
- 2019, June "Introduction to Fragile X Syndrome and Sex Chromosome Abnormalities"- eXtraordinary Kids Clinic Summer Intern Didactic, University of Colorado, Aurora, CO.
- 2019, Oct "Genetic Counseling for Sex Chromosome Aneuploidies: Updates, Nuances, and Considerations" Semi-Annual Colorado Genetic Counselors Education Symposium, Children's Hospital Colorado, Aurora, CO
- 2019, Dec "Clinical and Research Update on Klinefelter Syndrome and Sex Chromosome Aneuploidies," Maternal Fetal Medicine Education Day, University of Colorado, Aurora, CO
- 2020, Sept "Turner Syndrome Clinical Care Updates", Pediatric Grand Rounds, Department of Pediatrics School of Medicine, University of Colorado, Aurora, CO
- 2020, Oct "Sex Chromosome Aneuploidies and Cancer," Semi-Annual Colorado Genetic Counselors Education Symposium, Aurora, CO

2020, Nov	“Introduction to Sex Chromosome Abnormalities and the eXtraordinary Kids Clinic”- eXtraordinary Kids Clinic Student Intern Didactic, University of Colorado, Aurora, CO
2021, June	“Introduction to Fragile X Syndrome and Sex Chromosome Abnormalities”- eXtraordinary Kids Clinic Summer Intern Didactic, University of Colorado, Aurora, CO
2021, Nov	“Introduction to the eXtraordinary Kids Clinic, Turner Syndrome Clinic and Fragile X Clinic” presentation to the University of California, Los Angeles, Genetic Counseling Student Interest Group (GCSIG), virtual
2022, June	“Introduction to eXtraordinary Kids and Sex Chromosome Abnormalities”- eXtraordinary Kids Clinic Summer Intern Didactic, University of Colorado, Aurora, CO
2023, April	“Transition to Adulthood in Turner Syndrome” presentation for families at the Turner Syndrome Educational Day, Children’s Hospital Colorado (approximately 45 attendees)
2023, May	“Prenatal Identification of Sex Chromosome Aneuploidies: Perspectives of Diagnosis, Clinical Care and Research Findings,” Perinatology Grand Rounds for Northern Colorado, Department of OB/GYN, University of Colorado, virtual, approximately 12 attendees
2023, June	“Introduction to eXtraordinary Kids and Sex Chromosome Abnormalities”- eXtraordinary Kids Clinic Summer Intern Didactic, University of Colorado, Aurora, CO
2024, May	“A Look at Advocacy: The National Fragile X Foundations Advocacy Day on The Hill” Semi-Annual Colorado Genetic Counselors Education Symposium, Aurora, CO. Approximately 150 in attendance, hybrid.
2024, June	“Understanding Sex Chromosome Abnormalities, XXY, XYY, XXX, 45X and more”- eXtraordinary Kids Clinic Summer Intern Didactic, University of Colorado, Aurora, CO. 7 attendees
2025, June	“Understanding Sex Chromosome Abnormalities, XXY, XYY, XXX, 45X and more”- eXtraordinary Kids Clinic Summer Intern Didactic, University of Colorado, Aurora, CO. 6 attendees

## TEACHING EXPERIENCE

### Resident/Fellows Teaching

2008-present	Outpatient specialty clinic supervision and bedside teaching of Developmental-Behavioral Pediatric, Genetic, Genetic Counseling, and Maternal-Fetal Medicine fellows, pediatric residents, medical students and UC Anschutz graduate student clinical trainees (2-3 trainees/month, 60 clinical hours/month)
2011, Feb	“Fragile X Syndrome and Sex Chromosome Abnormalities – What Every Pediatrician Should Know,” Children’s Hospital Developmental-Behavioral Pediatrics Resident Rotation Didactic, 4 attendees
2015, Sept	“Fragile X Syndrome and Sex Chromosome Abnormalities – What Every Pediatrician Should Know,” Children’s Hospital Developmental-Behavioral Pediatrics Resident Rotation Didactic, 5 attendees
2016, March	“Sex Chromosome Aneuploidies and Fragile X Syndrome - Medical, Neurodevelopmental, and Psychological Features,” Children’s Hospital Developmental-Behavioral Pediatrics Resident Rotation Didactic, 3 attendees
2017, May	“Sex Chromosome Aneuploidies and Fragile X Syndrome - Medical, Neurodevelopmental, and Psychological Features,” Children’s Hospital Colorado Developmental-Behavioral Pediatrics Resident Rotation Didactic, 3 attendees
2017, Nov	CLSC 6653 Lecturer, “Turner Syndrome,” Key Concepts in Neurodevelopmental Disabilities I, LEND Fellowship lecture, University of Colorado, Aurora, CO, 20 attendees
2022, Feb	“Fragile X Syndrome” Children’s Hospital Colorado Developmental-Behavioral Pediatrics Fellowship Didactic, 3 attendees

### Medical/Graduate Student Teaching

2011, Jan "Sex Chromosome Abnormalities – Presentations in the School Setting," Lecture to University of Denver School Psychology Students, Low Incidence Disabilities Course, Denver, CO, 12 attendees

2011, Nov GENC 6170 Lecturer, "Research Projects in the Fragile X and eXtraordinary Kids Clinics," University of Colorado Genetic Counseling Graduate Students, Aurora, CO

2012, Nov GENC 6170 Lecturer, "Research Projects in the Fragile X and eXtraordinary Kids Clinics," University of Colorado Genetic Counseling Graduate Students, Aurora, CO

2013, Nov GENC 6170 Lecturer, "Research Projects in the Fragile X and eXtraordinary Kids Clinics," University of Colorado Genetic Counseling Graduate Students, Aurora, CO

2014, Nov GENC 6170 Lecturer, "Research in the Field of Genetic Counseling, Projects in the Fragile X and eXtraordinary Kids Clinics", University of Colorado Genetic Counseling Graduate Students, Aurora, CO, 8 attendees

2015, Nov GENC 6170 Lecturer, "Research in the Field of Genetic Counseling, Projects in the Fragile X and eXtraordinary Kids Clinics," University of Colorado Genetic Counseling Graduate Students, Aurora, CO, 8 attendees

2016, Jan "Fragile X," CU Student Chapter of the American Academy of Developmental Medicine and Dentistry, Aurora, CO, 35 attendees

2016, Nov GENC 6170 Lecturer, "Research in the Field of Genetic Counseling," University of Colorado Genetic Counseling Graduate Students, Aurora, CO, 8 attendees

2017, Nov GENC 6170 Lecturer, "Research in the Field of Genetic Counseling", University of Colorado Genetic Counseling Graduate Students, Aurora, CO, 8 attendees

2019, Nov GENC 6170 Lecturer, "Research in the Field of Genetic Counseling" University of Colorado Genetic Counseling Graduate Students, Aurora, CO, 8 attendees

2020, Nov GENC 6170 Lecturer, "Research in the Field of Genetic Counseling" University of Colorado Genetic Counseling Graduate Students, Aurora, CO, 8 attendees

2021, June "X&Y Chromosome Disorders: A Growing Need for Understanding These Common Conditions" Temple University, School Psychology Graduate Students, Philadelphia, PA, virtual, 20 attendees

2021, Nov GENC 6170 Lecturer "Research in the Field of Genetic Counseling" University of Colorado Genetic Counseling Graduate Students, Aurora, CO, 8 attendees

2022, Feb "Sex Chromosome Aneuploidies: Early Interventions and Preschool Supports" Lecture to University of Denver School Psychology Graduate Students, Denver, CO. 6 attendees, virtual

2022, Dec GENC 6170 Lecturer "Research in the Field of Genetic Counseling", University of Colorado Genetic Counseling Graduate Students, Aurora, CO. 8 attendees, in person with Melissa Gibbons, MS, CGC, Kami Schneider, MS, CGC and Curtis Coughlin, MS, CGC, PhD

2023, Dec GENC 6170 Panel discussion "Research in the Field of Genetic Counseling", University of Colorado Genetic Counseling Graduate Students, Aurora, CO. 8 attendees, in person with Melissa Gibbons, MS, CGC, Emily Todd, MS, CGC, Marrissa Lafeniére, MS, CGC and Curtis Coughlin, MS, CGC, PhD

2024, Oct GENC 6170, Presentation with Talia Thompson, PhD, "Qualitative and Mixed Methods in Genetic Counseling Research" University of Colorado Genetic Counseling Graduate Students, Aurora, CO. ~ 8 attendees

2024, Dec GENC 6170 Panel discussion "Research in the Field of Genetic Counseling", University of Colorado Genetic Counseling Graduate Students, Aurora, CO. 8 attendees, in person with Melissa Gibbons, MS, CGC, Kami Schneider, PhD, MS, CGC, Marrissa Lafeniére, MS, CGC and Curtis Coughlin, MS, CGC, PhD

2025, May "Understanding Fragile X Syndrome: Genetics, Clinical Insights, and Interventions", Key Concepts in Neurodevelopmental Disabilities, LEND Fellowship recorded Webinar, University of Colorado, Aurora, CO, 35 attendees

Trainees and Mentees:

High school:

- 2018-2019 Madison Foonberg, high school sponsored research internship, Fragile X multidisciplinary clinic
- 2019 – 2020 Sonoma Carlos, CHCO Child Health Research Internship, George Washington HS
- 2022 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Saige Ju, Denver School of the Arts
- 2025 Natalie Rooks, High School research intern, eXtraordinary Kids Clinic

Undergraduate/Pre-Med Students:

- 2016 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Meeka Nagamoto, Summer research internship, University of California, Los Angeles. Project produced the video documentary "The XXYY Sibling Project: A Voice to be Shared," highlighting a sibling's perspective of having a brother with XXYY, giving siblings a voice that can be heard and shared, <https://www.youtube.com/watch?v=GvU186EEw4s>
- 2016 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Jenna Nagamoto, University of Arizona. Project produced the video documentary "The XXYY Sibling Project: A Voice to be Shared," highlighting a sibling's perspective of having a brother with XXYY, giving siblings a voice that can be heard and shared, <https://www.youtube.com/watch?v=GvU186EEw4s>
- 2018 Co-Mentor for eXtraordinary Kids Clinic Summer, Will Karakash, Summer research internship, University of North Carolina, Chapel Hill
- 2019 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Isani Singh, CHCO Child Health Research Internship, Harvard University, abstract accepted for presentation at the National Organization for Rare Disease meeting. Peer-reviewed published manuscript, PMID: 35785515
- 2019 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Sophia Deklotz, CHCO Child Health Research Internship, University of Denver. Project abstract accepted for oral presentation at Western Society for Pediatric Research, peer-reviewed published manuscript, PMID: 32542985
- 2020 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Brisa Avilla, for eXtraordinary Babies study, University of Colorado, School of Medicine
- 2021 Co-mentor for Child Health Research Summer Internship program, Jennifer Rubin, University of North Carolina
- 2021 Co-mentor for Undergraduate Research Project in eXtraordinary Kids Clinic, Natalia Klamut, University of Colorado, Boulder
- 2022 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Kayla Nocon, for eXtraordinary Babies study, Regis University
- 2022 Co-Mentor for eXtraordinary Kids Clinic Intern, Stefany Contreras, XXYY dedicated project, Regis University
- 2022 Primary mentor for eXtraordinary Kids Clinic Summer Intern, Chloe Alviz,, UCLA student
- 2022 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Sarah Huang, for eXtraordinary Babies study, University of California, Los Angeles. Accepted into Genetic Counseling Graduate Program at Stanford University, class of 2025
- 2022 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Helina Seyoum, University of Colorado, Boulder
- 2022 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Tiana Grosskreuz, University of Rochester, New York
- 2023 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Geetali Lal, for eXtraordinary Babies study, University of Colorado, Boulder
- 2023 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Fathia Farah, for eXtraordinary Babies study, Cornell University
- 2023 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Emma Todd, for eXtraordinary Babies study, Colorado School of Mines
- 2023-current Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Mia Middleton, for eXtraordinary Babies study, University of Washington. Hired after the Summer to continue on as a student employee

- 2023 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Barbara Hilton, for eXtraordinary Babies study, University of Colorado, Boulder
- 2023 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Ariana Sanchez, for eXtraordinary Babies study, St. Thomas University, Minnesota
- 2023 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Aleks Radunovic, for eXtraordinary Babies study, 1<sup>st</sup> yr med student at Tilman J Feritta Family College of Medicine
- 2023 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Mahelet De Anda, for eXtraordinary Babies study, University of Colorado, Denver
- 2023-current Primary mentor for eXtraordinary Kids Clinic Summer Intern, Kayla Molison, University of Colorado, Boulder graduate.\* Hired after the Summer to continue on as a student employee and genetic counseling assistant
- 2024 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Emma Todd, for eXtraordinary Babies study, Colorado School of Mines
- 2024 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Emma Schaffer, for eXtraordinary Babies study, University of Colorado, Boulder 2024 graduate
- 2024 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Alyssa Keller, for eXtraordinary Babies study, University of Colorado, Boulder
- 2024 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Maria Despradel, for eXtraordinary Babies study, Colorado State University
- 2024 Primary Mentor for eXtraordinary Kids Clinic Summer Intern, Jeffery Greenfield, for eXtraordinary Babies study, Arizona
- 2024 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Greyson Seale, for eXtraordinary Babies study, University of Colorado
- 2024 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Giovanna Ruffolo, for eXtraordinary Babies study, University of Colorado, Boulder
- 2024 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Anastasia Zhivotov, for eXtraordinary Babies study, University of Colorado, Denver
- 2025 Primary Mentor for eXtraordinary Kids Clinic Summer Intern, Amity Howard, for eXtraordinary Babies study
- 2025 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Alyssa Keller, for eXtraordinary Babies study,
- 2025 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Aminata Diatta, for eXtraordinary Babies study,
- 2025 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Sam Daley, for eXtraordinary Babies study,
- 2025 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Sanvi Tummalapenta, for eXtraordinary Babies study,
- 2025 Co-Mentor for eXtraordinary Kids Clinic Summer Intern, Valerie Sandino-Murphy, for eXtraordinary Babies study,

Graduate Students: Capstone Project Mentor for Genetic Counseling Graduate Students

- 2011 Kali Messer, University of Colorado Genetic Counseling Program, "Genetic Counseling for Trisomy X Syndrome" Poster presentation at the 2013 American College of Medical Genetics annual meeting
- 2012 Anna Dennis, University of Colorado Genetic Counseling Program, Aurora, CO, "Parent to Child Disclosures of a Sex Chromosome Aneuploidy Diagnosis: Experiences of Parents and Individuals with Diagnoses" Poster presentation at the 2013 National Society of Genetic Counselors annual meeting and peer-reviewed published manuscript, PMID 25179748
- 2013 Arlene Colvin, University of Colorado Genetic Counseling Program, Aurora, CO, "Children's Books for Sex Chromosome Aneuploidy" published on Amazon. As of June 2024, 1,741 copies of the books have been ordered.

- 2015 Kali Schreiner, University of Colorado Genetic Counseling Program, Aurora, CO, "Development of Visual Aids for use in Genetic Counseling for Fragile X Associated Disorders." Poster presentation at the 2016 International Fragile X Conference
- 2015 Amy Reicks, University of Colorado Genetic Counseling Program, Aurora, CO, "Characterizing Quality of Life in Children and Adults with Sex Chromosome Aneuploidy using Patient-Centered Outcome Measures: A pilot study of the AXYS AXYON Patient Registry" Poster presentation at the 2016 International conference on Klinefelter syndrome
- 2015 Heather Wiles, Emory University, Genetic Counseling Program, Atlanta GA "Stress, Anxiety, and Adaptation to Genetic Information: Parental Experiences Receiving a Prenatal Diagnosis of Klinefelter Syndrome."
- 2016 Hillary Weinphal, University of Colorado Genetic Counseling Program, Aurora, CO, "Psychological functioning and behavioral profiles in boys with XYY syndrome" Poster presentation at the 2017 American College of Medical Genetics annual meeting.
- 2017 Briana Zieba, University of Colorado Genetic Counseling Program, Aurora, CO, "Pilot study of health, fitness, and quality of life in adolescent girls with Turner syndrome" poster and oral presentation at a national Turner syndrome conference and regional WSPR conference; peer-reviewed published manuscript, PMID 31894218
- 2018 Rhianna Urban, University of Colorado Genetic Counseling Program, Aurora, CO, "Metabolomic Signature in Adolescents with XXY with and without Testosterone Replacement." Poster presentation at the International Klinefelter Conference and Peer-reviewed published manuscript, PMID 36884262
- 2018 Catherine Buchanan, University of Colorado Genetic Counseling Program, Aurora, CO, "Eosinophilic Esophagitis in Sex Chromosome Aneuploidies" Poster presentation at 2019 American College of Medical Genetics annual conference and peer-reviewed published manuscript, PMID: 34738344
- 2018 Madison LaFleur, University of Colorado Genetic Counseling Program, Aurora, CO, "From Sibling to Guardian in Fragile X syndrome"
- 2019 Lindsey Guzewicz, University of Colorado Genetic Counseling Program, Aurora, CO, "The Spectrum of Features in Males and Females with Mosaic 45,X/46,XY," Poster presentation at the 2020 American College of Medical Genetics annual meeting, Peer reviewed manuscript published, PMID: 33616298
- 2020 Jessica Tisher, University of Colorado Genetic Counseling Program, Aurora, CO, "The Lived Parental Experience Having a Daughter with Trisomy X", 2021 ACMG abstract accepted for poster presentation, peer-review published manuscript, PMID 37691399
- 2021 Lylan Ho, University of Colorado Genetic Counseling Program, Aurora, CO, "Disparities Among Families with a Prenatal Diagnosis of a Sex Chromosome Aneuploidy who are Participating in the eXtraordinary Babies Study"
- 2021 Carly Peterson, University of Colorado Genetic Counseling Program, Aurora, CO, "Parent Wellbeing While Raising A Child with a Prenatal Diagnosis of a Sex Chromosome Aneuploidy." Abstract: Winner of the Carolyn Mills Lovell Genetic Counselors Award, American College of Medical Genetics 2022 Annual Meeting
- 2022 Christina Miller, University of Colorado Genetic Counseling Program, Aurora, CO, "Information needs in adolescents with differences of sex development (DSDs)". Accepted Educational breakout session platform presentation at the National Society of Genetic Counselors (NSGC) annual conference, Fall 2023, Chicago.
- 2023 Megan Bettencourt, University of Colorado Genetic Counseling Program, Aurora, CO, "Autism Spectrum Disorder Diagnoses for Participants of the INSIGHTS Turner Syndrome National Registry". Poster presentation at the American College of Medical Genetics (ACMG) annual meeting, 2024, Toronto, Canada
- 2023 Aaron Holbert, University of Colorado Genetic Counseling Program, Aurora, CO, "Landscape of Needs: Transitioning to Adulthood for Patients with XXY/Klinefelter Syndrome"
- 2024 Laura George, University of Colorado Genetic Counseling Program, Aurora, CO, "Decoding Language Development in Sex Chromosome Trisomy: Early Predictors and Clinical Implications"

- 2025 Madeline Halseth, University of Colorado Genetic Counseling Program, Aurora, CO, "Parent Perceptions of Social Support During Prenatal Diagnosis of Sex Chromosome Trisomy: Implications for Genetic Counseling"
- 2025 Brittany McVicar, University of Colorado Genetic Counseling Program, Aurora, CO, "Genetic Counseling in Prenatally Identified 47,XYY"

Graduate Students: Capstone Mentor for LEND Fellowship

- 2022-2023 Jillian Kirk, University of Colorado JFK LEND Trainee Fellowship, Aurora, CO "Evaluating the Impact of Early Intervention Speech Therapy on Expressive and Receptive Communication Development in Prenatally Identified Children with Klinefelter Syndrome (XXY) participating in the eXtraordinary Babies Study."
- 2024-2025 Olivia Isaacks, University of Colorado JFK LEND Trainee Fellowship, Aurora, CO, "Investigating the relationship between Hearing loss in girls with Turner syndrome and Developmental Concerns."

Graduate Students: Capstone Mentor for Masters in Public Health

- 2022-2023 Mariah Brown, University of Colorado MPH in Health Systems, Management & Policy + Global Health. "Abortion Bans: Policy Analysis on Genetic Counseling and Prenatal Genetic Testing in the US."

Post-Doctoral Fellows:

- 2019 Talia Thompson, PhD, Post-Doc Psychology Research Fellow, Co-mentor for a mixed methods project in Turner syndrome
- 2022 Agnethe Berglund, MD, PhD, University of Aarhus, Denmark, Co-mentor for 6-month post-doctoral clinical/research fellowship in SCAs
- 2024 Karli Swenson, PhD, MPH(c), Post-Doc Clinical-Translational Research Fellow, Co-mentor for 12-month post-doctoral clinical/research fellowship in SCAs

**GRANT SUPPORT**

Current Support:

Dates: 9/15/2025 - 8/31/2030

**Network for Advancing Sex Chromosome Aneuploidy Research Readiness (NASCARR)**

Funding Source: NIH NCATS 1U54HD121580-01

Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 25%

4/20/2023- 3/31/2028

***The eXtraordinary Babies-Childhood Study: Natural History of Health and Neurodevelopment in Infants and Young Children with Sex Chromosome Trisomy***

Funding Source: NIH NICHD R01 HD091251

Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 25%

7/2022-3/2023

Funding Source: NIH NICHD R01 HD091251-05S1, NIH NICHD R01 HD091251-06A1

Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 30%

9/2017-6/2022

Funding Source: NIH NICHD R01 HD091251

Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 30%

07/01/2022 – 06/30/2025

**Interrogating Fatty Acid Metabolism Impairment and Clinical Correlates in Males with Klinefelter Syndrome**

Funding Source: NIDDK 1R01DK133292  
 Role: Co-Investigator Effort: 5%

9/1/2021- 8/31/2026  
**Fragile X Clinic and Research Consortium Research Registry, FORWARD**  
 Funding Source: CDC NCBDDD – Subcontract for Denver clinic site  
 Role: Co-Investigator Denver PI: Nicole Tartaglia, MD, MS Effort: 5%

7/1/2018-present  
**Access to Care for Communities through Education, Service and Supports (Project Access)**  
 Funding Source: Upper Payment Limit/Medicaid, HCPF/UCSOM, #53005  
 Role: Co-Investigator PI: Sandy Friedman Effort: 20%

Prior Support:  
 5/1/2021- 4/30/2022  
**Noninvasive Prenatal Testing Laboratory Development Grant**  
 Funding Source: Perkin-Elmer  
 Role: Principal Investigator Effort: 5%

03/01/2021 – 12/31/2022  
**AXYS Registry Seed Grant**  
 Project Title: GALAXY Registry Development  
 Funding Source: AXYS  
 Role: Co-Investigator PI: Shanlee Davis, MD, PhD Effort: 2%

8/15/2019 – 8/14/2022  
**Turner Syndrome Global Alliance Kaleidoscope Award**  
 Project Title: Longitudinal Clinical Data Registry for Girls with Turner Syndrome  
 Funding Source: Turner Syndrome Global Alliance  
 Role: Co-Investigator PI: Shanlee Davis, MD, PhD Effort: 2%

7/1/2020-6/30/2021  
**Adult SCA Clinic Development Grant**  
 Funding Source: AXYS grant from WITH Foundation  
 Role: Principal Investigator Effort: 35%

6/2017-4/2021  
**LEARN NN107: Effects of AFQ056 on Language Learning in Young Children with Fragile X Syndrome (FXS)**  
 Funding Source: NIH NINDS NeuroNext,  
 Role: Co-Investigator PI: Elizabeth Berry-Kravis, MD PhD Effort: 5%

1/2017-12/2020  
**TRIXY Study: Neurodevelopmental Risks in Young Children with an Extra X or Y Chromosome**  
 Funding Source: NWO Vidi, University of Leiden, The Netherlands  
 Role: Co-Investigator PI: Sophie van Rijn, PhD Effort: 5%

9/1/2015- 8/30/2020  
**Colorado: Testing Longitudinal Outcome Measures and Improving Minority Participation in the Fragile X FORWARD Database**  
 Funding Source: Centers for Disease Control (CDC) U01 DD001190  
 Role: Co-Investigator Denver PI: Nicole Tartaglia, MD, MS Effort: 5%

2009-2020  
**Fragile X Clinic and Research Consortium Research Registry, FORWARD**  
 Funding Source: CDC NCBDDD – Subcontract for Denver clinic site.

Role: Co-Investigator Denver PI: Nicole Tartaglia, MD, MS Effort: 5%

2017 – 2020

**National Fragile X Foundation Fragile X Biobank**

Funding Source: National Fragile X Foundation

Role: Co-Investigator Denver PI: Nicole Tartaglia, MD, MS Effort: 5%

07/2014-06/2019

**Cognitive Outcome Measure Battery for Children with Intellectual Disability**

Funding Source: NIH NICHD R01 HD0761893

Role: Co-Investigator PI: David Hessel, PhD (UC-Davis) Effort: 5%

2009-Present

**Psychology, Genes, and Hormones in X&Y Chromosome Variations**

Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 5%

2009-2011

Funding Source 1: Children's Hospital Colorado Research Institute Research Scholar Award

Funding Source 2: CCTSI Pediatric CTSC support

2011-Present

Funding Source: eXtraordinary Kids Gift Funds

Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 5%

2018-2019

**National Fragile X Foundation Clinic Support Grant**

Funding Source: National Fragile X Foundation

Role: Principal Investigator Effort: 2%

2017-2018

**National Fragile X Foundation Clinic Support Grant**

Funding Source: National Fragile X Foundation

Role: Principal Investigator Effort: 2%

10/2017-9/2018

**BabyGenes Laboratory Development Grant**

Funding Source: BabyGenes, Inc.

Role: Principal Investigator Effort: 2%

8/2011-7/2017

**Effects of Testosterone and Genetic Factors on Psychological and Motor Skills in XXY**

8/2011-7/2016: Funding 1: NINDS NIH K23NS070337

Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 5%

8/2011-7/2014: Funding 2: CCTSI Pediatric CTSC Microgrant support

Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 5%

8/2016-7/2017: Funding 3: CHCO Research Institute Bridge Funding

Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 5%

2014-2015

**A Randomized, Double-Blind, Placebo-Controlled, Parallel-Group, Fixed-Dose Study of NNZ-2566 in Fragile X Syndrome**

Funding Sponsor: Neuren Pharmaceuticals

Role: Co-Investigator Denver PI: Nicole Tartaglia, MD, MS Effort: 5%

2014-2015

**A 6-week, Randomized, Multicenter, Double-Blind, Parallel, Flexed- and Fixed-dose Study of MC01C1 (Metadoxine Extended-release) 700mg and 1400mg Compared with Placebo in Adults and Adolescents with Fragile X Syndrome**

Funding Sponsor: Alcobra Pharmaceuticals  
Role: Co-Investigator Denver PI: Nicole Tartaglia, MD, MS Effort: 5%

2013-2015

**FRAGXIS: A randomized, double-blind, 12-week, parallel group, placebo-controlled study of the efficacy and safety of RO4917523 in patients with Fragile X Syndrome**

Funding Sponsor: Hoffmann-LaRoche  
Role: Co-Investigator Denver PI: Nicole Tartaglia, MD, MS Effort: 5%

2013-2014

**FOXTAIL: A study of RO4917523 in pediatric patients with Fragile X syndrome**

Funding Sponsor: Hoffman-LaRoche  
Role: Co-Investigator Denver PI: Nicole Tartaglia, MD, MS Effort: 5%

7/2012-6/2013

**Colorado Fragile X Clinic Grant**

Sponsor: National Fragile X Foundation, Colorado LINKS group  
Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 5%

2012-2013

**An Open-Label Extension Study to Evaluate the Safety, Tolerability, and Pharmacokinetics of STX209 (Arbaclofen) in Subjects with Fragile X Syndrome**

Sponsor: Seaside Therapeutics Multicenter Trial  
Role: Co-Investigator Denver PI: Nicole Tartaglia, MD, MS Effort: 5%

2011-2013

**A Randomized, Double-Blind, Placebo-Controlled Fixed-Dose Study of the Efficiency, Safety, and Tolerability of STX209 (Arbaclofen) Administered for the Treatment of Children with Fragile X Syndrome**

Funding Sponsor: Seaside Therapeutics Multicenter Trial  
Role: Co-Investigator Denver PI: Nicole Tartaglia, MD, MS Effort: 5%

2011-2013

**A Randomized, Double-Blind, Placebo-Controlled Study of the Efficiency, Safety, and Tolerability of STX209 (Arbaclofen) Administered for the Treatment of Social Withdrawal in Adolescents and Adults with Fragile X Syndrome**

Sponsor: Seaside Therapeutics Multicenter Trial  
Role: Co-Investigator Denver PI: Nicole Tartaglia, MD, MS Effort: 5%

2009-2014

**Visual Spatial Cognitive Deficits in 22q11.2 Deletion Syndrome**

P.I. Tony Simon PhD, University of California Davis  
Funding Source: NIMH NIH R0142974HD  
Role: Co-Investigator P.I. Tony Simon PhD (UC- Davis) Effort: 5%

2009-2011

**Neuromotor Outcome Measures for Clinical Trials in Fragile X Syndrome**

Funding Source: FRAXA Research Foundation  
Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 5%

2008-2010

**Psychological and Motor Effects of Testosterone Therapy in XXY and XYY – Pilot study**

Funding Source: Society for Developmental-Behavioral Pediatrics  
Role: Co-Investigator PI: Nicole Tartaglia, MD, MS Effort: 5%

2008-2012

### **eXtraordinary Kids Clinic Program**

Funding Sponsor: Weckbaugh Family Foundation Program Grant  
Role: Co-Investigator

Effort: 5%

2007-2010

### **eXtraordinary Kids Clinic Program Development Award**

Funding Sponsor: Hagemann Charitable Trust  
Role: Co-Investigator PI: Nicole Tartaglia, MD, MS

Effort: 5%

2007-2011

### **Fragile X Syndrome Clinic Program Development Gift**

Funding Sponsor: Judith Cole Estate  
Role: Co-Investigator PI: Nicole Tartaglia, MD, MS

Effort: 5%

## **BIBLIOGRAPHY**

### Papers published in peer-reviewed journals

1. N Tartaglia, **S Howell**, R Wilson, A Sutherland, L Wilson, "Trisomy X Syndrome: A Review," *Orphanet Journal of Rare Diseases*, 2010 May 5:8. Distinguished as a "Highly Accessed" Article with over 371,000 views of open access version since publication. (PMID 20459843; PMCID PMC2883963) <https://ojrd.biomedcentral.com/counter/pdf/10.1186/1750-1172-5-8.pdf>
2. N Tartaglia, L Cordiero, **S Howell**, R Wilson, J Janusz, "The Spectrum of the Behavioral Phenotype in 47,XXY Klinefelter syndrome," *Pediatric Endocrine Reviews*, 2010 Dec;8 Suppl 1:151-9. (PMID 21217607) [https://cuanschutz.primo.exlibrisgroup.com/discovery/openurl?institution=01UCOHS\\_INST&vid=01UCOHS\\_INST:01UCOHS&id=pmid:21217607&sid=Entrez:PubMed](https://cuanschutz.primo.exlibrisgroup.com/discovery/openurl?institution=01UCOHS_INST&vid=01UCOHS_INST:01UCOHS&id=pmid:21217607&sid=Entrez:PubMed)
3. N Tartaglia, **S Howell**, N Ayari, C D'Epagnier, P Zeitler, "48,XXYY, 48,XXXY and 49,XXXXY syndromes: Not just variants of Klinefelter syndrome," *Acta Paediatrica*, 2011 Jun;100(6):851-60. (PMID 21342258; PMCID PMC3314712) <https://onlinelibrary-wiley-com.proxy.hsl.ucdenver.edu/doi/epdf/10.1111/j.1651-2227.2011.02235.x>
4. K Hager, K Jennings, S Hosono, **S Howell**, J Gruen, S Rivkees, N Tartaglia, H Rinder, "Molecular Diagnostic Testing for Klinefelter Syndrome and Other Male Sex Chromosome Aneuploidies," *International Journal of Pediatric Endocrinology*, 2012 April 23 (1): 8. (PMID 22524164; PMCID 3411476) <https://ijpeonline.biomedcentral.com/counter/pdf/10.1186/1687-9856-2012-8.pdf>
5. J Visootsak, N Ayari, **S Howell**, J Lazarus, N Tartaglia, "Timing of Diagnosis of 47,XXY and 48,XXYY: A Survey of Parent Experiences," *American Journal of Medical Genetics Part A*, 2013 March 161A:268-272. (PMID 23322622; PMCID PMC3558746) [https://cuanschutz.primo.exlibrisgroup.com/permalink/01UCOHS\\_INST/lo7itt/cdi\\_pubmedcentral\\_primary\\_oai\\_pubmedcentral\\_nih\\_gov\\_3558746](https://cuanschutz.primo.exlibrisgroup.com/permalink/01UCOHS_INST/lo7itt/cdi_pubmedcentral_primary_oai_pubmedcentral_nih_gov_3558746)
6. A Dennis, **S Howell**, L Cordeiro, N Tartaglia, "How Should I Tell My Child? Disclosing the Diagnosis of Sex Chromosome Aneuploidies" *Journal of Genetic Counseling*, 2014 Sep 3, epub ahead of print (PMID:25179748) <https://onlinelibrary.wiley.com/doi/epdf/10.1007/s10897-014-9741-4>
7. N Tartaglia, **S Howell**, R Wilson, J Janusz, R Boada, J Frazier, M Pfeiffer, S Martin, K Regan, S McSwegin, P Zeitler, "The eXtraordinary Kids Clinic: A Model of Interdisciplinary Care for Children and Adolescents with Sex Chromosome Aneuploidy", *Journal of Multidisciplinary Health Care*, 2015 June 23 (PMID: 26229481) [https://cuanschutz.primo.exlibrisgroup.com/discovery/openurl?institution=01UCOHS\\_INST&vid=01UCOHS\\_INST:01UCOHS&id=pmid:26229481&sid=Entrez:PubMed](https://cuanschutz.primo.exlibrisgroup.com/discovery/openurl?institution=01UCOHS_INST&vid=01UCOHS_INST:01UCOHS&id=pmid:26229481&sid=Entrez:PubMed)
8. S Davis, **S Howell**, R Wilson, T Tanda, J Ross, P Zeitler, N Tartaglia, "Advances in the Interdisciplinary Care of Children with Klinefelter Syndrome," *Adv Pediatr*. 2016 Aug 63(1):15-46. (PMID: 27426894) [https://cuanschutz.primo.exlibrisgroup.com/permalink/01UCOHS\\_INST/lo7itt/cdi\\_pubmedcentral\\_primary\\_oai\\_pubmedcentral\\_nih\\_gov\\_5340500](https://cuanschutz.primo.exlibrisgroup.com/permalink/01UCOHS_INST/lo7itt/cdi_pubmedcentral_primary_oai_pubmedcentral_nih_gov_5340500)
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2. Children's Books: "Gregory and His Extra X," "Elizabeth and Her Extra X," "Kyle and his Extra X and Y," and "Jack and his Extra Y." Series of 4 illustrated children's books authored by genetic counseling student A. Colvin for parents and children with X&Y chromosome disorders. Copyright 2014, University of Colorado. As of June 2024, 1,741 copies of the books have been ordered. [https://www.amazon.com/s?i=stripbooks&rh=p\\_27%3AArlie+Colvin&s=relevancerank&text=Arlie+Colvin&ref=dp\\_byline\\_sr\\_book\\_1](https://www.amazon.com/s?i=stripbooks&rh=p_27%3AArlie+Colvin&s=relevancerank&text=Arlie+Colvin&ref=dp_byline_sr_book_1)
3. A Dennis, **S Howell**, N Tartaglia, "Talking with your Child about..." series of handouts for families discussing strategies to disclose the diagnosis of X&Y variations to their children. Handouts created for: XXY/Klinefelter syndrome, XYY, Trisomy X, XXYY syndrome. Informed from A Dennis graduate project. Distributed through the eXtraordinary Kids Clinic, AXYS, and The XXYY Project. 2014
4. "The XXYY Sibling Project Video" - The XXYY Project funded our group to make a video about the effect of XXYY syndrome on siblings. Two undergraduate students Mika Nagamoto (UCLA) and Jenna Nagamoto (Univ of Arizona) were mentored for developing content, interview topics, and editing. Interview also featured in video. Uploaded to YouTube Sep 2017. <https://www.youtube.com/watch?v=GvU186EEw4s&feature=youtu.be>
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14. Original Book: "What You Need to Know When You are Expecting or Have a New Baby with XXY," book authored by **Susan Howell**, Nicole Tartaglia, Shanlee Davis, Chloe Alviz, edited by Kayla Molison, for parents who have or are expecting a new baby with XXY. Copyright Nov 2024, University of Colorado. As of Dec 2024, 19 copies of the book have been ordered. [https://www.amazon.com/What-Need-Know-When-Expecting/dp/B0DPFVYDTG#detailBullets\\_feature\\_div](https://www.amazon.com/What-Need-Know-When-Expecting/dp/B0DPFVYDTG#detailBullets_feature_div)

*Scientific original abstracts published or presented at scientific meetings (all competitive)\*denotes my mentee as first author*

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7. K Messer\*, **S Howell**, N Tartaglia, *Genetic Counseling for Trisomy X Syndrome: Consideration of Parental Perspectives and Experiences*, Poster Presentation, 2013 American College of Medical Genetics Annual Clinical Genetics Meeting, Phoenix, Arizona, March 2013
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13. **S Howell**, S Close, S Terry, A Reicks, C Riby, N Tartaglia, J Moore, *AXYON: An online patient registry including Patient Reported Outcome measures for individuals with Sex Chromosome Aneuploidies*, Poster Presentation, 2<sup>nd</sup> International Conference on Klinefelter Syndrome, Munster, Germany, March 2016
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16. K Regan, S McSwegin, **S Howell**, J Nagamoto, N Tartaglia, *The Important Role of Nursing and Social Work in a Fragile X Clinic*, Poster Presentation, 15<sup>th</sup> International Fragile X Conference, San Antonio, TX, July 2016
17. K Wigby, T Tanda, **S Howell**, L Cordeiro, N Tartaglia, *Exploring Preliminary Relationships of Psychological Symptoms to Adaptive Functioning in Triple X Syndrome*. Oral Platform Presentation, Western Society for Pediatric Research Annual Meeting, Carmel, CA, January 2017
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22. J Barstein, R Nolan, **S Howell**, T Hall, P Lavesser, A Blakeley-Smith, *Characterization of the Broader Developmental Profile in an Individual with 6q16.1q16.2 Microdeletion*, Department of Psychiatry, Poster Presentation, University of Colorado Denver, Denver, Colorado March 2018
23. **S Howell**, M LaFleur, N.Tartaglia, *My Baby Has What? Genetic Counseling Pilot Study to Understand the Experience of Parents Prenatally Diagnosed with Sex Chromosome Aneuploidies*. Poster Presentation, 2018 American College of Medical Genetics Annual Clinical Genetics Meeting, Charlotte, North Carolina, April 2018
24. N.Tartaglia, **S Howell**, L Cordeiro, *The eXtraordinary Babies Study: Preliminary Profile of Neurodevelopmental Skills at the 6-month Visit in Infants with Sex Chromosome Trisomy Identified by*

- NIPT. Poster Presentation, 2018 American College of Medical Genetics Annual Clinical Genetics Meeting, Charlotte, North Carolina, April 2018 \*Featured as part of ACMG NBSTRN Poster Tour\*, co-presented as a poster presentation, Society for Developmental Behavioral Pediatrics Annual Meeting, Anaheim, CA, Sep 2018.
25. Zieba B\*, **Howell S**, Davis S. *Mixed Methods Study of Quality of Life and Physical Activity in Adolescent Girls with Turner Syndrome*, Turner Syndrome Research Network, July 2018, Poster Presentation in Arlington, VA, co-presented as an oral presentation at the Western Society for Pediatric Research, Jan 2019, Carmel, CA.
  26. L Cordeiro, L Huddleston, **S Howell**, N Tartaglia, *Promoting minority family participation in a Fragile X clinic and the FORWARD registry and database*, Oral Presentation, 16th International Fragile X Conference, Cincinnati, OH, Jul 2018
  27. Karakash W, **Howell S**, Davis S. Exercise Capacity and Cardiometabolic Health in Adolescent Girls with Turner Syndrome. Western Society for Pediatric Research, Jan 2019, Oral presentation in Carmel, CA.
  28. Davis S, **Howell S**, Buyers E, Dovey S, Tartaglia N. Evidence for Diminished Ovarian Reserve in Youth with Trisomy X. Endocrine Society Annual Meeting, Mar 2019, Poster presentation in New Orleans, LA.
  29. **S Howell**, C Buchanan, H Miyazawa, G Furuta, N Nguyen, S Davis, N Tartaglia, *Eosinophilic Esophagitis in Patients with Sex Chromosome Aneuploidies*, Poster presentation, 2019 American College of Medical Genetics Annual Clinical Genetics Meeting, Seattle, Washington, April 2019
  30. JA Haven, D Xu, CM Schwanke, T Schwalbe, A Zearfoss, L Pickler, **S Howell**, AF Elias, *A Unique Case of Mosaic Turner Syndrome and Partial Trisomy 17p*, Poster presentation, 2019 American College of Medical Genetics Annual Clinical Genetics Meeting, Seattle, Washington, April 2019
  31. N Tartaglia, **S. Howell**, C. Harrison,, L Cordeiro, J Janusz, J Ross, R Wilson, *The eXtraordinary Babies Study: Preliminary Profile of Neurodevelopmental Skills at the 6 and 12 month visit in infants with SCT Identified by NIPT*, Poster presentation, 2019 AXYS Family Conference, Atlanta, GA, Jun 2019
  32. G Cover, C Meerschaert, R Miller, **S Howell**, N Tartaglia, S Close, A Talboy, *Adult Specialty Clinics for Sex Chromosome Aneuploidy: A WITH Foundation Grant to AXYS to support development of two pilot models at AXYS Clinic & Research Consortium clinic sites*, Poster presentation, 2019 AXYS Family Conference, Atlanta, GA, Jun 2019
  33. T Thompson, **S Howell**, N Tartaglia, *Early therapies, school supports, and educational outcomes for students with X & Y variations: Preliminary results from a national survey*, Poster presentation, 2019 AXYS Family Conference, Atlanta, GA, Jun 2019
  34. Thompson T, Davis S, **Howell S**, Tartaglia N. *Early childhood intervention services for children with sex chromosome aneuploidies*. Western Society for Pediatric Research, Jan 2020. Oral presentation in Carmel, CA. Published in: Journal of Investigative Medicine, January 2020, 68(1), p. 255, <https://afmr.org/Western/multimedia/files/2020/Program.pdf>
  35. J Janusz, C Harrison, **S Howell**, S Davis, T Tanda, L Cordeiro , J Ross, T Thompson, M Brown, R Wilson, N Tartaglia, *Profile of Developmental Skills in Infants with Klinefelter Syndrome (XXY) Identified by NIPT*, Poster presentation, International Neuropsychological Society Annual Meeting, Feb 2020.
  36. L Guzewicz\*, **S Howell**, N Nokoff, J Barker, S Davis, *The Spectrum of Features in Males and Females with Mosaic 45,X/46,XY*, Poster presentation, 2020 American College of Medical Genetics Annual Clinical Genetics Meeting, San Antonio, TX, March 2020
  37. N Tartaglia, S Davis, **S Howell**, K Kowal, J Ross, *The eXtraordinary Babies Study: Medical Features in 137 Prenatally Diagnosed Infants With Sex Chromosome Trisomy*, Poster presentation, 2020 American College of Medical Genetics Annual Conference. Virtual, March 2020
  38. N Tartaglia, S Williams, L Crawford, K Regan, **S Howell**, *Medical Problems in Fragile X Syndrome: Results from the FORWARD 4.0 database*, CDC Fragile X FORWARD All-Clinics Meeting oral presentation, Jul 2020 *Updated data presented as oral virtual presentation: 17<sup>th</sup> International Fragile X Conference, NFXF Research Round-Up, Jul 2020*
  39. L Guzewicz\*, **S Howell**, N Nokoff, J Barker, S Davis, *The Spectrum of Features in Males and Females with Mosaic 45,X/46,XY*, Poster presentation, 2020 National Society of Genetic Counselors 39<sup>th</sup> Annual Meeting, Virtual, Nov 2020

40. Thompson, T., Davis, S., **Howell, S.**, & Tartaglia, N. *Supporting Young Children with X & Y Chromosome Variations Through Early Intervention*. Oral session presented at the Rocky Mountain Early Childhood Conference, Virtual. March 2021
41. Thompson, T., Janusz, J., Boada, R., Wilson, R., Davis, S., **Howell, S.**, & Tartaglia, N. *School Supports and Educational Outcomes for Students with Sex Chromosome Aneuploidies*. Poster session presented at the 54<sup>th</sup> Annual Gatlinburg Conference, Virtual, April 2021, co-presented as an oral presentation, Western Society for Pediatric Research Conference, Carmel CA, Jan 2022 and Poster presentation, 3rd International Workshop on Klinefelter Syndrome, Trisomy X and XYY, Leiden, The Netherlands, September 2022
42. Tisher, J.\* , **Howell, S.**, Tartaglia, N., & Thompson, T. (2021, April). *Parenting young daughters with a prenatal diagnosis of Trisomy X: A phenomenological analysis of mothers' experiences in the early years*. Poster session presented at the 2021 annual American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting, Virtual. Published in: *Molecular Genetics and Metabolism*, April 2021, 132, p.S204-S205, co-presented as a poster presentation, 3rd International Workshop on Klinefelter Syndrome, Trisomy X and XYY, Leiden, The Netherlands, September 2022
43. Louderman, M, Thompson, T, Wilson, R, Davis, S, **Howell, S**, Tartaglia, N. *The Easy Baby: Exploring Temperment Profiles in Young Children with Prenatally Identified Sex Chromosome Trisomies*. Western Society for Pediatric Research, Jan 2022, Oral presentation in Carmel, CA.
44. Davis S, Bamba V, Brickman W, Carl A, Dowlut-McElroy T, Good M, **Howell S**, Kanakatti Shankar R, Law J, Prakash S, Ranallo K. Stakeholder Engagement Efforts to Improve Diversity in Rare Disease Research: Inspiring New Science in Guiding Healthcare in Turner Syndrome (InsighTS) Registry, Western Medical Research Conference 2022, oral presentation, Carmel CA.
45. Peterson, C\*, Thompson, T, Tartaglia, N, Davis, S, **Howell, S**. *Parenting Stress Raising Children with Sex Chromosome Aneuploidies: First Year of Life Results from the eXtraordinary Babies Study*. Oral Platform Presentation, 2022 American College of Medical Genetics Annual Clinical Genetics Meeting, Nashville, TN, March 2022, \*Recipient of the Carolyn Mills Lovell Genetic Counselors Award\*
46. Carl A\*, Law J, Kanakatti Shankar R, Brickman W, Bamba V, Prakash S, Klamut N, Ranallo K, Dowlut-McElroy T, Good M, **Howell S**, Davis S. *Description of Turner Syndrome Diagnosis in Nationally Representative Sample: Inspiring New Science in Guiding Healthcare in Turner Syndrome (InsighTS) Registry*. Oral presentation, Pediatric Endocrine Society Annual Meeting, April 2022
47. Klamut N\*, Bamba V, Law J, Kanakatti Shankar R, Brickman W, Prakash S, Dowlut-McElroy T, **Howell S**, Carl A, Davis SM. Comparison of Diagnostic, Fetal, Maternal and Neonatal Information in Trisomy X Syndrome with Turner Syndrome and Turner Syndrome Mosaicism: Inspiring New Science in Guiding Healthcare in Turner Syndrome (InsighTS) Registry, Pediatric Endocrine Society, 2022, poster presentation, virtual.
48. **S Howell**, T Grosskreuz, S Villalta, H Kraaijenbrink, N Tartaglia, *Moms with the Full Mutation*, Poster, 18<sup>th</sup> NFXF International Fragile X Conference, San Diego, CA, July 2022
49. **S Howell**, K Regan, E Floyd, *Primary Care in the World of Fragile X*, Poster presentation, 18<sup>th</sup> NFXF International Fragile X Conference, San Diego, CA, July 2022
50. **S Howell**, C Buchanan, S Davis, H Miyazawa, G Furuta, N Nguyen, N Tartaglia, *Eosinophilic Esophagitis in Patients with Sex Chromosome Aneuploidies*, Poster Presentation, 3<sup>rd</sup> International Workshop on Klinefelter Syndrome, Trisomy X and XYY, Leiden, Netherlands, Sept 2022.
51. Harrison C, Boada R, **S Howell**, Tanda T, Cordeiro L, Pyle L, Zeitler P, Tartaglia N, *Psychological Effects of Testosterone in Tanner 2-3 Males with XXY: Results of a randomized, placebo-controlled trial*. Oral Presentation, 3<sup>rd</sup> International Workshop on Klinefelter Syndrome, Trisomy X and XYY, Leiden, Netherlands, Sept 2022.
52. **Howell, S.**, Davis, S. M., Thompson, T., Brown, M., Tanda, T., Kowal, K., Alston, A., Ross, J., & Tartaglia, N. R. Noninvasive prenatal screening (NIPS) results for participants of the eXtraordinary babies study: Screening, counseling, diagnosis, and discordance. Oral Presentation, 3<sup>rd</sup> International Workshop on Klinefelter Syndrome, Trisomy X and XYY, Leiden, Netherlands, Sept 2022.
53. M Louderman, Thompson T, Wilson R, Davis S, **Howell S**, Tartaglia N, *The Easy Baby? Exploring temperament profiles in young children with prenatally identified sex chromosome trisomies*, Oral presentation, Western Society for Pediatric Research Conference, Carmel CA, Jan 2022  
*Updated data presented as poster presentation:*  
3rd International Workshop on Klinefelter Syndrome, Trisomy X and XYY, Leiden, The Netherlands,

September 2022 & 24<sup>th</sup> Occasional Temperament Conference, Oct 2023, oral presentation, Quebec Canada

54. S Davis, R Urban, A D'Alessandro, J Haines, C Chan, M Kelsey, **S Howell**, N Tartaglia, P Zeitler, P Baker II, *Unique Plasma Metabolite Signature for Adolescents with Klinefelter Syndrome Reveals Altered Fatty Acid Metabolism*, Poster presentation, 3rd International Workshop on Klinefelter Syndrome, Trisomy X and XYY, Leiden, The Netherlands, September 2022
55. T Thompson\*, S Davis, S Takamatsu, **S Howell**, N Tartaglia, *An International Survey of Academic and Character Strengths in Youth with Sex Chromosome Aneuploidies*, Poster presentation, 3rd International Workshop on Klinefelter Syndrome, Trisomy X and XYY, Leiden, The Netherlands, September 2022
56. Kirk J\*, Davis S, **Howell S**, Brown M, Tartaglia N. The eXtraordinary Babies Study: Analysis of Physical Features of Prenatally Identified Infants with Sex Chromosome Trisomies. Western Society for Pediatric Research, Jan 2023, oral presentation, Carmel, CA.
57. C Miller\*, C Buchanan, **S Howell**, M Vogiatzi, V Miller, A Carl, S Strine, S Davis, *"Don't Paint Us All with the Same Brush" Information Needs in Adolescents with Differences in Sex Development*, Poster presentation, 2023 American College of Medical Genetics Annual Clinical Genetics Meeting, Salt Lake City, UT
58. N Tartaglia, T Thompson, J Janusz, R Wilson, J Ross, **S Howell**, S Davis, L Pyle, *Creating Developmental Growth Curves and Milestone Charts for Infants with Sex Chromosome Trisomy (XXY/Klinefelter, Trisomy X, XYY) Using Data from the eXtraordinary Babies Study*, Poster presentation, 2023 American College of Medical Genetics Annual Clinical Genetics Meeting, Salt Lake City, UT
59. S Davis M Brown, **S Howell**, J Janusz, N Lahlou, L Pyle, R Reynolds, T Thompson, R Wilson, P Zeitler, N Tartaglia, *Testosterone Effects on Short Term Physical, Hormonal, and Neurodevelopmental Outcomes in Infants with 47,XXY/Klinefelter syndrome: The TESTO Study*, Poster presentation, 2023 American College of Medical Genetics Annual Clinical Genetics Meeting, Salt Lake City, UT
60. **S Howell**, S Davis, J Kirk, M Brown, N Tartaglia, *The eXtraordinary Babies Study: a Natural History of Prenatally Identified Infants with Sex Chromosome Trisomies (SCTs): Analysis of Birth Defects and Medical Problems in the First Year of Life*, Poster and Rapid-Fire Oral Presentation, American College of Medical Genetics Annual Clinical Genetics Meeting, Salt Lake City, UT (\*\*Awarded Top Rated Abstract, 20 awardees of 675+ accepted abstracts), co-presented as a poster at the 2023 Annual Spring Pediatric Research Poster Session, CCTSI, CUAnschutz, Aurora, CO
61. Davis SM, Lahlou N, Brown M, Thompson T, Janusz J, **Howell S**, Pyle L, Tartaglia N. *Mini-Puberty Hormone Concentrations do not Predict Physical or Neurodevelopmental Outcomes in Infants with 47,XXY / Klinefelter Syndrome at 2 or 5 months of age*. Pediatric Endocrine Society Annual Meeting, May 2023, poster presentation, San Diego, CA
62. Klamut N\*, Bamba V, Law J, Kanakatti Shankar R, Brickman W, Prakash S, Dowlut-McElroy T, **Howell S**, Carl A, Davis SM. Diagnostic, Perinatal, Medical, and Gynecological Features of Turner Syndrome with Trisomy X Syndrome: Inspiring New Science in Guiding Healthcare in Turner Syndrome (InsighTS) Registry, National Conference on Undergraduate Research, 2023, Eau Claire, WI.
63. **Howell S**, Davis SM, Ross J, Tartaglia N. *eXtraordinary Babies Study: Genetic Counseling Implications of Discordant Prenatal Noninvasive Screening Results in Sex Chromosome Aneuploidies*, 2024 Western Society for Pediatric Research, Oral Presentation, Jan 2024, Carmel, CA. \*Award-winning abstract- Mead Johnson Nutrition Junior Faculty Award, most meritorious abstract in Genetics (>100 abstracts submitted to Genetics) per peer review
64. Wilson, R; Davis, S; Peterson, C; Thompson, T; **Howell, S**, Tartaglia, N. *The eXtraordinary Babies Study: Parenting Stress in Families with a Prenatal Diagnosis of Sex Chromosome Trisomies, 36-month-old Follow-up*, 2024 Western Society for Pediatric Research, Oral Presentation, Jan 2024, Carmel, CA
65. Molison K\*, **Howell S**, Tartaglia N, Davis S. Initial Data from the GALAXY Registry: Generating Advancements in Longitudinal Analysis in X & Y Chromosome Variations, Jan 2024, Oral presentation at the Western Medical Research Conference, Carmel CA.

66. Middleton M\*, Prakash Siddharth, Khanna A, **Howell S**, Carl A, Davis S. Cardiac and Aortic Surgery Outcomes in Patients with Turner Syndrome. Jan 2024, Oral presentation at the Western Medical Research Conference, Carmel CA.
67. **Howell S**, Molison K, Davis S, Carl A, Tartaglia N. Generating Advancements in Longitudinal Analysis in X&Y Chromosome Variations: Initial Data from the Multicenter GALAXY Clinical Data Registry and Biorepository, March 2024, Poster Presentation at the American College of Medical Genetics Meeting, Toronto, Canada, \*Top Rated Poster Award Recipient
68. Klamut, N\*, Carl A, Bothwell S, Bamba V, Law J, Brickman W, Klein K, Kanakatti Shankar R, Pinnaro C, Prakash S, **Howell S**, Tartaglia N, Davis S, Prevalence, Diagnostic Features, and Medical Outcomes of Females with Mosaic Turner Syndrome (45,X/47,XXX): Results from the InsightS Registry, March 2024, Poster Presentation at the American College of Medical Genetics Meeting, Toronto, Canada
69. George, L\*, **Howell S**, Bothwell, S, Tartaglia, NR, Language Development in Children with Sex Chromosome Trisomies: Analysis using the Macarthur-Bates CDI-III, Jan 2025, Oral presentation at the Western Medical Research Conference, Carmel CA.
70. Keller A, Carl A, Bothwell S, Swenson K, **Howell S**, Tartaglia NR, Davis SM. Prenatal, Birth, and Neonatal Outcomes In Sex Chromosome Aneuploidies: Data from the INSIGHTS and GALAXY Registries. Jan 2025, Oral presentation at the Western Medical Research Conference, Carmel CA.
71. Louderman M, Thompson T, Wilson R, Bothwell S, **Howell S**, Swenson K, Tartaglia NR. Using Temperment to Inform Anticipatory Guidance for Young Children with Sex Chromosome Trisomies. Jan 2025, Oral presentation at the Western Medical Research Conference, Carmel CA.
72. **Howell S**, Molison K, Thompson T, Tartaglia NR. The eXtraordinary Babies Study: Evolving Parental Perspectives and Priorities While Raising a Child Prenatally Diagnosed with Sex Chromosome Trisomy. March 2025 Poster Presentation at the annual American College of Medical Genetics Meeting, Los Angeles, CA
73. Nocon K, Swenson K, **Howell S**, Davis, SM, Bothwell S, Ross J, Tartaglia NR. The eXtraordinary Baby Study: Natural History of Infants Identified with XXYY in the Prenatal Period. March 2025 Poster Presentation at the annual American College of Medical Genetics Meeting, Los Angeles, CA
74. Molison K\*, **Howell S**, Thomspson T, Bothwell, S, Davis, SM, Tartaglia NR. The eXtraordinary Babies Study: Parental Emotional Experiences and Coping Strategies During Prenatal Sex Chromosome Aneuploidy Diagnosis. March 2025 Poster Presentation at the annual American College of Medical Genetics Meeting, Los Angeles, CA
75. Tartaglia, NR, Davis SM, **Howell S**, Janusz, J, Thompson, T, Wilson R, Ross J. Correlations of Parental Cognitive Skills with 36-month Developmental Outcomes in Children with Prenatal Diagnoses of XXY, XYY, and Trisomy X. March 2025 Poster Presentation at the annual American College of Medical Genetics Meeting, Los Angeles, CA
76. Davis SM, **Howell S**, Janusz J, Thomspson T, Wilson R, Ross J, Tartaglia NR. Infant Testosterone Treatment Does Not Improve Neurodevelopment at 12, 24, or 36 Months for Boys with 47,XXY. March 2025 Platform Presentation at the annual American College of Medical Genetics Meeting, Los Angeles, CA
77. Grzybacz L, **Howell S**, Davis SM, Ross J, Tartaglia NR, The eXtraordinary Babies Study: Breastfeeding Trends and Challenges in the First Year of Life, Poster Presentation 2025 AXYS Families Conference, Atlanta, GA
78. Molison K, Halseth M, McVicar B, **Howell S**, Bothwell S, Davis SM, Tartaglia NR, Thompson T Parental Experiences During Prenatal Sex Chromosome Trisomy Diagnosis: Results from a Mixed Methods Analysis. Jan 2026, Oral presentation at the Western Medical Research Conference, Carmel CA.
79. Bothwell, S, **Howell S**, Davis SM, Tartaglia NR. An Interactive Dashboard to Monitor Developmental Progression in Children with Sex Chromosome Trisomies. Jan 2026, Oral presentation at the Western Medical Research Conference, Carmel CA.
80. McVicar B, Molison K, Halseth M, **Howell S**, Davis SM, Tartaglia NR, Thompson T. Understanding Parental Experiences from Prenatal cfDNA Results to XYY Diagnosis: Implications for Genetic Counseling from the eXtraordinary Babies Study. March 2026, (pending) Poster Presentation at the American College of Medical Genetics Meeting, Baltimore, MD

81. Halseth M, Molison K, McVicar B, **Howell S**, Davis SM, Tartaglia NR, Thompson T. Parent Perceptions of Social Supports Following Prenatal Identification of Sex Chromosome Trisomy: Genetic Counseling Implications from the eXtraordinary Babies Study. March 2026, (pending) Poster Presentation at the American College of Medical Genetics Meeting, Baltimore, MD