CURRICULUM VITAE

Melissa Gibbons, MS, CGC

Personal history

Current position:	Assistant Professor, Pediatric Genetics and Metabolism, University of Colorado School of Medicine and Children's Hospital Colorado
Professional address:	13123 East 16th Avenue, B395 Aurora, CO 80045
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Education

Institution	Degree	Location	Dates
Simmons University	BS	Boston, MA	1997-2001
University of North Carolina- Greensboro	MS	Greensboro, NC	2003-2005

Academic appointments

Senior Instructor, University of Colorado SOM, Department of Pediatrics	2014-2016
Assistant Professor, University of Colorado SOM, Department of Pediatrics	2016-present

Hospital, government or other professional positions

Molecular Genetics Coordinator, Berman Gund Laboratory, MEEI Boston, MA	2001-2003
Reasearch Associate, Denver Health, Denver, CO	2005-2006
Genetic Counselor, MFM Center at Memorial Hospital, Colorado Springs, CO	2006-2007
Genetic Counselor, University of Colorado, SOM at Childrens's Hospital Colorado	2007-present
Consultant, Sarepta, Duchenne Muscular Dystrophy Advisory Board	2017
Consultant, Avexis, Spinal Muscular Atrophy Advisory Board	2017
Speaker, Biogen Medical Forum Series – Genetics of Spinal Muscular Atrophy	2019-2020
Consultant, Novartis, Genetic Counselor Advisory Board	2020-2021
Consultant, Taysha Gene Therapy Advisory Board	2021

Honors, special recognitions and awards

Paul Moe Excellence in Child Neurology	2018
Nursing Excellence Award for Empiricial Outcomes (interprofessional team award to the department of Neurology Complex Drug Team)	2018

Memberships in professional organizations

National Society of Genetic Counselors	2003- 2020
Membership Committee	2003-2008
Mentor Program Committee	2005-2008
Co-Chair	2005-2007
Neurogenetics SIG	2016-2020
Practice Guidelines Committee	2016-2019

Major committee and service responsibilities

Departmental:

NIH Congenital Muscular Dystrophy Undiagnosed Clinic On-Site Clinic Coordinator	2012
Host for the SMA Research Update Session and the Information Session on Stem Cells in Muscular Dystrophies	2013
Congenital Muscular Dystrophy TLC Conference On-Site Co-Organizer	2013
Genetic Counselor Guidelines Committee	2019-2020
Neuromuscular Genetic Counselor Fellowship Selection Committee	2019-present

Hospital:

Laboratory Therapeutics and Diagnostic Committee	2012-2017
Gene Transfer Therapy Group	2020-present

Regional:

Co-Founder, Co-Chair of the Semi-Annual Colorado Genetic Counselors Symposium	2013-present

National:

Essay Judge for the Annual DNA Day Essay Contest Hosted by ASHG	2009-2016
Reviewer for the NCHPEG Genetics for Child Neurologist Educational Documents	2012
Duchenne Muscular Dystrophy Care Considerations Committee	2015-2016
Member of the Advisory Committee for the DMD Carrier Project	2015-2016
Member, Medical Advisory Council, Cure SMA	2014-present

Licensure and board certification

Diplomate, American Board of Genetic Counselors	2009-present

Inventions, intellectual property and patents held or pending

None

Review and referee work

1. Ad hoc Manuscript Reviewer, Journal of Genetic Counseling 2014

Invited extramural lectures, presentations and visiting professorships

Local

1. Fragile X and the impact of the diagnosis on the family, Swedish Medical Center, Englewood, Colorado (2009)
2. Clinical Trial Preparedness for the Family, Parent Project Muscular Dystrophy– F.A.C.E.S Group, Aurora, Colorado (2012)
3. Local Research Update and Clinical Trial Preparedness for the Family, MDA Parent Group, Aurora, Colorado (2012)
4. Clinical Trial Preparedness for the Family, CURECMD- TLC, Aurora, Colorado (2013)
5. Spinal Muscular Atrophy Beyond 1, 2, 3, Genetics Grand Rounds, University of Colorado (2013)
6. Clinical Trial Readiness for the Family and Ask the Expert Q&A Session, Denver Educational Roundtable for Duchenne Muscular Dystrophy hosted by Jett Foundation, Denver, Colorado (2017)
7. Update of Carrier Testing in Spinal Muscular Atrophy and New Treatments, Kaiser Permanent MFM Department, Denver, Colorado (2017)
8. Spinal Muscular Atrophy: An Update on Therapeutics, Genetics Grand Rounds, University of Colorado, Aurora, Colorado (2017)

9. Latest Treatments in SMA. Colorado Dinner Lecture Series, Loveland, Colorado (2018)
10. Latest Treatments in SMA. Colorado Dinner Lecture Series, Centennial, Colorado (2018)
11. Clinical Trial Readiness for the Family. Denver Educational Roundtable for Duchenne Muscular Dystrophy sponsored by the Jett Foundation, Denver, Colorado (2018)
12. Emerging Treatments in Spinal Muscular Atrophy. Grand Rounds, Evans Community Hospital (2018)
13. Genetics of Neuromuscular Disease. University of Colorado Neuromuscular Symposium, (2018)
14. Emerging Treatments in Spinal Muscular. Pediatric Grand Rounds, Denver Health, Denver, Colorado (2018)
15. The Changing Landscape of Neonatal Neuromuscular Disease: New Diagnostic Paradigms and Treatments, Neonatal Noon Conference, Children's Hospital Colorado, Aurora, Colorado (2019)
16. Genetics and Gene Therapy 101, MDA Engage Event, Aurora, Colorado (2019)
17. Neurocognitive function in carriers of dystrophinopathies, Haberfield Lecture (2020)
18. Spinal Muscular Atrophy and Therapeutic Approaches to Genetic Disorders Including Handling Considerations, AveXis Roundtable Program, Denver, Colorado (2020)

Regional

1. Genetic Counseling: What in the World is That, Connections Conference, Grand Junction, Colorado (2012)
2. Congenital Muscular Dystrophy and Congenital Myopathy, Semi-Annual Colorado Genetic Counselor Symposium , Aurora, Colorado (2013)
3. Utilizing the NextGxDx.com website, Semi-Annual Colorado Genetic Counselor Symposium, Aurora, Colorado (2015)
4. Services Overview and Research Update, MDA Firefighter Boot Camp, Blackhawk, CO (2015)
5. Feedback in Supervision. Semiannual Colorado Genetic Counselor Symposium, Aurora, CO (2017)
6. Neuromuscular Disease Research Update, MDA Firefighter Boot Camp, Blackhawk, Colorado (2017)
7. Therapeutic Approaches to Spinal Muscular Atrophy. Colorado Genetic Counselor Symposium, Aurora, CO (2017)
8. Pediatric Neuromuscular Disease Update. MDA Firefighter Boot Camp, Blackhawk, Colorado (2018)
9. Navigating Genetic Testing. Cheyenne Regional Grand Rounds, Cheyenne, WY (2018)
10. Spinal Muscular Atrophy and Newborn Screening, MSRGN Summit, San Antonio (2018)
11. Direct to Consumer Testing. Semiannual Colorado Genetic Counselors Symposium (2019)
12. Treating the Cause, New York Genetic Task Force Meeting (2020)
13. Secondary Findings in Pediatric Settings. Semiannual Colorado Genetic Counselors Symposium (2020)
14. Update on Colorado Newborn Screening, Semiannual Colorado Genetic Counselors Symposium (2021)
15. Gene Therapy: What is it and how does it impact physical therapy?, APTACO Annual Conference (2021)

National

1. Clinic Coordination, MDA Clinical Conference, Chicago, Illinois (2014)
2. Implications of Genetic Testing on Families with Ataxia, National Ataxia Foundation Annual Meeting, Denver, Colorado (2015)
3. From Diagnosis to Treatment – A Collaborative Approach to SMA Management, American Association of Neuromuscular & Electrodiagnostic Medicine (AANEM) Annual Meeting, Phoenix, Arizona (2017)

4. Emerging Treatments in Spinal Muscular Atrophy. Christopher Ward Neuroscience Nursing Conference, Aurora, Colorado (2018)
5. Genetics and Reproductive Options. Cure SMA Annual Conference, Dallas, Texas (2018) Lasting education on CureSMA website: available at: https://curesma.wpengine.com/wp-content/uploads/2019/07/genetics-and-reproductive-2018-conf.pdf
6. Navigating Genetic Testing in 5q Related Spinal Muscular Atrophy. Cure SMA Annual Clinical Care Meeting, Dallas, Texas (2018)
7. Spinal Muscular Atrophy & Therapeutic Approaches to Genetic Disorders. Peer to Peer Program (Avexis), Atlanta, GA (2018)
8. Gene therapy has arrived: updates, FDA approval, and roles for genetic counselors. NSGC 37th Annual Conference Plenary Platform Presentation, Atlanta, Georgia (2018)
9. Gene Therapy for Spinal Muscular Atrophy. ACMG Annual Clinical Genetics Meeting, Scientific Concurrent Session, Seattle, Washington (2019)
10. Are You Ready for Gene Therapy? Examples from Spinal Muscular Atrophy. ACMG Annual Clinical Genetics Meeting, Satellite Symposium, Seattle, Washington (2019)
11. Genetics 101, Association of Virtual Childhood Neurology Nurses Annual Conference (2020)
12. The Clinicians and Families Experience with the First Year of Newborn Screening in Colorado and Wyoming, CureSMA Clinical Care Virtual Meeting (2021)

Webinars:

1. Emerging Treatments in Spinal Muscular Atrophy, NSGC Prenatal/Pediatric SIG (2018)
2. Spinal Muscular Atrophy and Newborn Screening, Newborn Screening Virtual Education Series (2019) https://www.youtube.com/watch?v=pFXNzpUN3c8&feature=youtu.be
3. Spinal Muscular Atrophy (SMA) Treatment and Outcomes Webinar, APHL New Disorders Workgroup (2019) https://www.newsteps.org/resources/webinars-events/spinal-muscular-atrophy-treatment-and-outcomes
4. The Era of Disease-Modifying Therapy for Duchenne Muscular Dystrophy: Outlining Care Pathways for the Evolving Needs of the Patient, NeuroScience Live (2020) https://www.neuroserieslive.com/presentation-cme/?q=2020_DMD_CME
5. Collaborative Care in SMA: Evolving Roles for Neurologists, Ob-Gyns, PCPs, and Counselors, NeuroScience Live (2021) https://cme.epocrates.com/a/YJVQIS
6. "Treatment and Post-treatment Decisions in SMA, NeuroScience Live (2021) https://www.neuroserieslive.com/presentation-cme/?q=2021_SMA_CME
7. Introduction to the Diagnostic Odyssey, MDA Webinar (2021) https://www.mda.org/care/mda-engage/community-webinars/introduction-to-the-diagnostic-odyssey-of-neuromuscular-disease-2021
8. Facilitating Care in Spinal Muscular Atrophy: Getting the Most out of Newborn Screening, MedScape (2021) https://www.medscape.org/viewarticle/969009

Teaching record

Elementary, Middle School, High School Presentations

1. Genetics & Genetic Counseling, Mackintosh Academy Middle School Student, Aurora, Colorado (2017)

Undergraduate/Graduate/Medical Students/Professional Staff

19. Genetics and Research, First Friday Neuromuscular Lecture Series, Children's Hospital Colorado, Aurora, Colorado (2009)
20. Mitochondrial Myopathies, First Friday Neuromuscular Lecture Series, Children's Hospital Colorado, Aurora, Colorado (2011)

21. Pompe Disease, First Friday Neuromuscular Lecture Series, Children's Hospital Colorado, Aurora, Colorado (2011)
22. Genetics and Ethics, PhD Student Group at University of Colorado Anschutz Campus, Aurora, Colorado (2011)
23. Clinical Utility of the GeneDx epilepsy panels, Genetics in Neurology Brown Bag Lunch, Children's Hospital Colorado, Aurora, CO (2012)
24. Ethics and Genetic Testing, Neurology Resident Lecture Series, Children's Hospital Colorado, Aurora, Colorado (2012)
25. Past, Present and Future of Genetic Testing in Neuromuscular Disease, First Friday Neuromuscular Lecture Series, Children's Hospital Colorado, Aurora, Colorado (2012)
26. Test Ordering at CHCO, Neurology Resident's Lecture Series, Children's Hospital Colorado, Aurora, Colorado (2012)
27. Genetics and Neurology, Department of Neurology, Children's Hospital Colorado, Aurora, Colorado (2012)
28. Test Ordering at CHCO, Neurology Resident's Lecture Series, Children's Hospital Colorado, Aurora, Colorado (2013)
29. Exceptional Children, Red Rocks Community College, Lakewood, Colorado (2013)
30. Genetic Counseling as a Career, Careers in Science Club, University of Colorado, Aurora, Colorado (2014)
31. Congenital Muscular Dystrophy and Congenital Myopathy, First Friday Neuromuscular Lecture Series, Children's Hospital Colorado, (2014)
32. Schwartz-Jampel Syndrome, First Friday Neuromuscular Lecture Series, Children's Hospital Colorado, Aurora, CO (2015)
33. Genetic Testing and Utilization Management, Neurology Residents Lecture Series, , Children's Hospital Colorado, Aurora, CO (2015)
34. Congenital Muscular Dystrophy and Myopathies, Neurology Resident's Lecture Series, Children's Hospital Colorado, Aurora, CO (2015)
35. Types of Genetic Testing and Indications, Academic Half Day Lecture for Pediatric Residents, Aurora, Colorado, (2015)
36. Introduction to Genetic Testing, Academic Half Day Lecture for Pediatric Residents, Aurora, Colorado, (2016)
37. Genetics 101, Neurology Faculty Education Series, Children's Hospital Colorado, Aurora, Colorado (2016)
38. Genetic Testing in Neurology, Children's Hospital Colorado Neurology Advanced Practice Provider Lecture Series, Aurora, Colorado (2016)
39. Introduction to Genetic Testing, Academic Half Day Lecture for Pediatric Residents, Aurora, Colorado (2017)
40. Insurance Process at CHCO, Children's Hospital Colorado Pulmonary Faculty Meeting, Aurora, Colorado (2017)
41. Atypical Cases and the Impact on genetic testing recommendations, First Friday Neuromuscular Lecture Series, Children's Hospital Colorado, Aurora, CO (2017)
42. Spinal Muscular Atrophy Update on Disease and Treatment, CIMFM Faculty Lecture, Children's Hospital Colorado, Aurora, Colorado (2017)
43. Spinal Muscular Atrophy Update on Disease and Treatment, Children's Hospital Colorado Neurology Nursing Education Series Lecture, Aurora, Colorado (2017)
44. Emerging Treatments for Spinal Muscular Atrophy, Neonatology and MFM Fellows Physiology Course Lecture, University of Colorado, Aurora, Colorado (2018)
45. Emerging Treatments for Spinal Muscular Atrophy, University of Colorado Denver, Statistical Genetics Lab Meeting, Denver, Colorado (2018)
46. Introduction to Genetic Testing, Academic Half Day Lecture for Pediatric Residents, Aurora, Colorado, (2019)
47. Neuromuscular Disease for the Pediatric Pulmonologist, Weekly Pulmonary Conference, Aurora, Colorado (2021)

Didactic Teaching, graduate and medical school students, residents, and fellows:

1. Congenital Malformation of the Newborn GENC6150, Lecturer, University of Colorado Anschutz Medical Campus (2009-present)
2. Introduction to Research GENC6170, Lecturer, University of Colorado Anschutz Medical Campus (2011-present)
3. Interprofessional Education and Development (IPED), Facilitator for 8 session, University of Colorado Anschutz Medical Campus (2017)

Clinical Teaching:

General Genetics Weekly Clinics, (2007-2009)
General Genetics Greeley Regional Clinics (2007–2008)
Neuromuscular Clinics (2007-present)
Neurology Clinic (2010-present)
Neurogenetics 2011 - Present
Neuromuscular Carrier Clinic (2015-present)
Neuromuscular New Patient Clinic (2018–present)

Mentorship:

MS Candidate: Kelsey Zegar (degree awarded 2012) 2011-2012
Program: UCD-AMC, Genetic Counseling Program
Role: Mentor, Capstone Committee

MS Candidate: Kali Schreiner (degree awarded 2015) 2015-2016
Program: UCD-AMC, Genetic Counseling Program
Role: Mentor
Abstract: 2016 MDA Clinical Care Conference

MS Candidate: Jessica Goldsberry (degree awarded 2018) 2017-2018
Program: UCD-AMC, Genetic Counseling Program
Role: Mentor, Capstone Committee
Abstract: 2018, National Society of Genetic Counselors

MS Candidate: Mackenzie Blaile (degree awarded 2019) 2018-2019
Program: UCD-AMC, Genetic Counseling Program
Role: Mentor, Capstone Committee Chair
Abstract: 2019, Cure SMA Clinical Care Meeting (Podium Presentation)

MS Candidate: Marrisa Lafreniere (degree awarded 2020) 2019-2020
Program: UCD-AMC, Genetic Counseling Program
Role: Mentor, Capstone Committee
Abstract: 2020, Cure SMA Research Meeting (Poster Presentation)

Neuromuscular Genetic Counseling Fellowship: Kaitlin Smith 2020-2021
Neuromuscular Genetic Counseling Fellowship: Marrisa Lafreniere 2021-present

Grant support

Active: None
Inactive:

1. Increasing Site Capacity for Spinal Muscular Atrophy Patients in the United States, CureSMA. Role: Site Co-PI, 3/2018-3/2019

Bibliography

Peer-reviewed publications:

1. Sharon D, Sandberg MA, Rabe VW, **Stillberger M**, Dryja TP, Berson EL. RP2 and RPGR mutations and clinical correlations in patients with X-linked retinitis pigmentosa. Am J Hum Genet. 2003 Nov;73(5):1131-46. doi: 10.1086/379379. Epub 2003 Oct 16. PubMed PMID: 14564670; PubMed Central PMCID: PMC1180492. https://library-cuanschutz.hosted.exlibrisgroup.com/primo-explore/openurl?sid=Entrez:PubMed&id=pmid:14564670&vid=01UCOHS&institution=01UCOHS&url_ctx_val=&url_ctx_fmt=null&isSerivcesPage=true
2. Wada Y, Sandberg MA, McGee TL, **Stillberger MA**, Berson EL, Dryja TP. Screen of the IMPDH1 gene among patients with dominant retinitis pigmentosa and clinical features associated with the most common mutation, Asp226Asn. Invest Ophthalmol Vis Sci.2005 May;46(5):1735-41. doi: 10.1167/iovs.04-1197. PubMed PMID: 15851576. https://library-cuanschutz.hosted.exlibrisgroup.com/primo-explore/openurl?sid=Entrez:PubMed&id=pmid:15851576&vid=01UCOHS&institution=01UCOHS&url_ctx_val=&url_ctx_fmt=null&isSerivcesPage=true
3. Donkervoort S, Papadaki M, de Winter JM, Neu MB, Kirschner J, Bolduc V, Yang ML, **Gibbons MA**, Hu Y, Dastgir J, Leach ME, Rutkowski A, Foley AR, Krüger M, Wartchow EP, McNamara E, Ong R, Nowak KJ, Laing NG, Clarke NF, Ottenheijm C, Marston SB, Bönnemann CG. TPM3 deletions cause a hypercontractile congenital muscle stiffness phenotype. Ann Neurol. 2015 Dec;78(6):982-994. doi: 10.1002/ana.24535. Epub 2015 Nov 13. PubMed PMID: 26418456; PubMed Central PMCID: PMC5154623. https://library-cuanschutz.hosted.exlibrisgroup.com/primo-explore/openurl?sid=Entrez:PubMed&id=pmid:26418456&vid=01UCOHS&institution=01UCOHS&url_ctx_val=&url_ctx_fmt=null&isSerivcesPage=true
4. Zambonin JL, Bellomo A, Ben-Pazi H, Everman DB, Frazer LM, Geraghty MT, Harper AD, Jones JR, Kamien B, Kernohan K, Koenig MK, Lines M, Palmer EE, Richardson R, Segel R, Tarnopolsky M, Vanstone JR, **Gibbons M**, Collins A, Fogel BL, Dudding-Byth T, Boycott KM. Spinocerebellar ataxia type 29 due to mutations in ITPR1: a case series and review of this emerging congenital ataxia. Orphanet J Rare Dis. 2017 Jun 28;12(1):121. doi: 10.1186/s13023-017-0672-7. Review. PubMed PMID: 28659154; PubMed Central PMCID: PMC5490223. https://library-cuanschutz.hosted.exlibrisgroup.com/primo-explore/openurl?sid=Entrez:PubMed&id=pmid:28659154&vid=01UCOHS&institution=01UCOHS&url_ctx_val=&url_ctx_fmt=null&isSerivcesPage=true
5. Colvin MK, Poysky J, Kinnett K, Damiani M, **Gibbons M**, Hoskin J, Moreland S, Trout CJ, Weidner N. Psychosocial Management of the Patient With Duchenne Muscular Dystrophy. Pediatrics. 2018 Oct;142(Suppl 2): S99-S109. doi: 10.1542/peds.2018-0333L. PubMed PMID: 30275254 https://library-cuanschutz.hosted.exlibrisgroup.com/primo-explore/openurl?sid=Entrez:PubMed&id=pmid:30275254&vid=01UCOHS&institution=01UCOHS&url_ctx_val=&url_ctx_fmt=null&isSerivcesPage=true
6. Lee AJ, Jones KA, Butterfield RJ, Cox MO, Konersman CG, Grosmann C, Abdenur JE, Boyer M, Beson B, Wang C, Dowling JJ, **Gibbons M**A, Ballard A, Janas JS, Leshner RT, Donkervoort S, Bönnemann CG, Malicki DM, Weiss RB, Moore SA, Mathews KD.Clinical, genetic, and pathologic characterization of *FKRP* Mexican founder mutation c.1387A>G. Neurol Genet. 2019 Apr;5(2):

e315. doi: 10.1212/NXG.0000000000000315. eCollection 2019 Apr. PubMed PMID: 31041397; PubMed Central PMCID: PMC6454397. https://library-cuanschutz.hosted.exlibrisgroup.com/primo-explore/openurl?sid=Entrez:PubMed&id=pmid:31041397&vid=01UCOHS&institution=01UCOHS&url_ctx_val=&url_ctx_fmt=null&isSerivcesPage=true

7. The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia. 2019 May;60(5):797-806. doi: 10.1111/epi.14698. Epub 2019 Apr 5. PubMed PMID: 30951195; PubMed Central PMCID: PMC6519344. https://library-cuanschutz.hosted.exlibrisgroup.com/primo-explore/openurl?sid=Entrez:PubMed&id=pmid:30951195&vid=01UCOHS&institution=01UCOHS&url_ctx_val=&url_ctx_fmt=null&isSerivcesPage=true
8. Angione K, **Gibbons M,** Demarest S. An Objective Method for Evaluating Next-Generation Sequencing Panels. J Child Neurol. 2018 Dec 20:883073818815036. doi: 10.1177/0883073818815036. PubMed PMID: 30569809. https://library-cuanschutz.hosted.exlibrisgroup.com/primo-explore/openurl?sid=Entrez:PubMed&id=pmid:30569809&vid=01UCOHS&institution=01UCOHS&url_ctx_val=&url_ctx_fmt=null&isSerivcesPage=true
9. **Gibbons MA,** Stratton A, Parsons J. Spinal Muscular Atrophy (SMA) in the Therapeutic Era. Current Genetic Medicine Reports. 2019 July 16; doi: 10.1007/s40142-019-00172-9. https://link.springer.com/article/10.1007/s40142-019-00172-9
10. Kour S, Rajan DS, Fortuna TR, Anderson EN, Ward C, Lee Y, Lee S, Shin YB, Chae JH, Choi M, Siquier K, Cantagrel V, Amiel J, Stolerman ES, Barnett SS, Cousin MA, Castro D, McDonald K, Kirmse B, Nemeth AH, Rajasundaram D, Innes AM, Lynch D, Frosk P, Collins A, **Gibbons M** et al . Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nat Commun. 2021 May 7;12(1):2558. doi: 10.1038/s41467-021-22627-w. PubMed PMID: 33963192. https://library-cuanschutz.hosted.exlibrisgroup.com/primo-explore/openurl?sid=Entrez:PubMed&id=pmid:33963192&vid=01UCOHS&institution=01UCOHS&url_ctx_val=&url_ctx_fmt=null&isSerivcesPage=true

Books and Monographs:

1. None

Book Chapters and Reviews:

1. None

Clinical Scholarship:

1. None

Lay Articles:

1. Gibbons, M Generations (Official Publication of the National Ataxia Foundation) Implications of Genetic Testing and Ataxia on Families. Summer 2015

Other:

1. Children's Hospital Colorado public website, sections on Spinal Muscular Atrophy https://www.childrenscolorado.org/conditions-and-advice/conditions-and-symptoms/conditions/spinal-muscular-atrophy-sma/

Scientific Abstracts:

1. AR Gaudio AR, MA Stillberger MA, Disease Course of Patients with Pericentral Retinitis Pigmentosa. Presented at the Association of Research in Vision and Ophthalmology Annual Meeting. May 2002, Tampa Bay (Poster Presentation)
2. D Sharon, MA Sandberg, V.W Rabe, M. Stillberger, T.P Dryja, E.L Berson

Mutation Survey of the *RP2* and *RPGR* Genes (Including ORF15) and Evidence for Genotype-Phenotype Correlations. Presented at the Association of Research in Vision and Ophthalmology Annual Meeting. May 2003, Fort Lauderdale (Poster Presentation)

3. C. Rivolta, T.L. McGee, M.A. Stillberger, E.L. Berson and T.P. Dryja. RP11 Transcripts With Premature Stop Codons May Escape Nonsense-Mediated Decay; Possible Role in Explaining Reduced Penetrance. Presented at the Association of Research in Vision and Ophthalmology Annual Meeting, May 2003, Fort Lauderdale (Poster Presentation)
4. Wade Y, Sandberg, MA, McGee, TL, Stillberger, MA, Berson EL, Dryja Tp. Screen of the IMPDH1 gene among patients with dominant retinitis pigmentosa and clinical features associated with the most common mutation**.** Presented at the Association of Research in Vision and Ophthalmology Annual Meeting, May 2003. Fort Lauderdale (Poster Presentation)
5. Stillberger, M. What is the Student Experience with the National Society of Genetic Counselors? Presented at the National Society of Genetic Counselor Annual Meeting, November 2005, Los Angeles (Poster Presentation)
6. G. Scharer, D. Manchester, G. Bellus, L.Pickler, M. Saenz, M. Raymond, K. McKelvie, M. Stillberger et. al., Copy Number Variations (CNV's) is well defined region of Xp22.31 are associated with syndromic phenotypes and intellectual disability. Further delineation of a new microdeletion/duplication syndrome. Presented at the American Society of Human Genetics. November 2008, Philadelphia, PA (Poster Presentation)
7. K. Swisshelm, D. Bertrand, J. LeRoux, B. Lunt, M. Crespin, M. Springer, M. Gibbons, et al. Genomic 10.4 Mb duplication of 7q36.1q36.3 segregating in a family with a maternal finding of somatic-gonadal mosaicism for the duplication. Presented at American College of Medical Genetics. March 2012, Charlotte, NC (Poster Presentation)
8. Gibbons M, Lohr NM, Yang M. LMNA Congenital Muscular Dystrophy: A Case Report of Two Unrelated Patients with a R249W Mutation and Review of the Literature. Presented at American College of Medical Genetics. March 2012, Charlotte, NC (Poster Presentation)
9. Gibbons, MA, Schreiner, KM, Ballard AL Barriers to Care for Carriers of Duchenne and Becker Muscular Dystrophy, One Clinic's Experience with a Neuromuscular Carrier Clinic, Presentation at the MDA Clinical Conference. March 2016, Washington DC. (Poster Presentation)
10. Gibbons, M., Fox, J., Dixon, S. Diagnosis of 5q-related SMA in an asymptomatic woman undergoing routine SMN1 carrier screening during pregnancy Presented at Clinical Care Conference at Cure SMA Annual Meeting. June 2018, Dallas, TX (Poster Presentation)
11. Goldsberry, J Gibbons, M., Ballard, A. Factors that are predictors of recommendation adherence among carriers of dystrophinopathies. Presented at NSGC 37th Annual Meeting. November 2018, Atlanta, GA (Poster Presentation)
12. Blaile M, Parsons, J, Gibbons, M. How new, effective treatment for SMA alters reproductive decision-making, Presented at Cure SMA Researcher Meeting. June 2019, Anaheim, CA (Platform Presentation)
13. Laurel Reed MD, Dylan Brock MD, Ryan Kammeyer MD, Melissa Gibbons MS, Katie Angione MS, Timothy Bernard MD, Scott Demarest MD. What's All the VUS About?: A Retrospective Look at Genetic Panels for Pediatric-Onset Epilepsies, Presented at AES Virtual Meeting, December 2020, Virtual Meeting (Poster Presentation)
14. Kaitlin Smith, MS, CGC, Debra O'Reilly, PAC, Melissa Gibbons, Stacey Dixon, MD, PhD. Unique Genetic Experiences from a Large Adult SMA Clinic, Presented at CureSMA Clinical Care Virtual Meeting, June 2021(Poster Presentation)
15. Marrisa Lafreniere, Mackenzie Blaile, Melissa Gibbons, Alison Ballard, Anne Stratton, The Changing Natural History of Spinal Muscular Atrophy: Perspectives From Young Adults and Their Parent Caregivers, Presented at CureSMA Clinical Care Virtual Meeting, June 2021(Poster Presentation)
16. Taylor Schwab, RN, Kaitlin Haug, RN, Christine Caneva, BS, Jennifer Coffman, DNP, CPNP-AP, Stefanie Leonard, MSN, RN, Melissa Gibbons, MS, CGC, Scott Demarest, MD, Julie Parsons, MD,

Meeting the care coordination needs of complex therapies for rare neuromuscular/neurogenetic disorders: the development of a complex drug program, Presented at MDA Clinical Care Meeting, March 2021 (Poster and Platform Presentation)