



Inaugural O'Donnell Brain Institute Symposium: Autism Spectrum Disorders | March 25-26, 2021

REFERENCES

Session 1 addresses the current need in advancing clinical phenotyping, genotyping, and biomarker development in ASD as critical tools for assessing patients. This session will also address racial disparities that exist in healthcare delivery and clinical research in ASD. See References #1 and #2:

1. Hyman SL, Levy SE, Myers SM, Council On Children With Disabilities SOD, Behavioral P. Identification, Evaluation, and Management of Children With Autism Spectrum Disorder. *Pediatrics*. 2020;145(1).
2. McPartland JC, Bernier RA, Jeste SS, et al. The Autism Biomarkers Consortium for Clinical Trials (ABC-CT): Scientific Context, Study Design, and Progress Toward Biomarker Qualification. *Front Integr Neurosci*. 2020;14:16.

Session 2 addresses the need to accurately analyze neural circuit dysfunction in neurodevelopmental disorders, including ASD. It will update attendees on which circuits are involved in ASD and how identified circuit dysfunction may impact behavior. See Reference #3:

3. Müller RA, Fishman I. Brain Connectivity and Neuroimaging of Social Networks in Autism. *Trends Cogn Sci*. 2018 Dec;22(12):1103-1116. doi: 10.1016/j.tics.2018.09.008. Epub 2018 Oct 31. PMID: 30391214; PMCID: PMC7080636.

Session 3 addresses the need to identify the forms of ASD that are classified as “syndromic” autism that are typically caused by loss of function mutations in single genes. Examples of these are: Fragile X Syndrome, Rett Syndrome, Phelan-McDermid Syndrome and Tuberous Sclerosis. Diagnosis and treatment of the different forms and genetic causes of autism requires clinicians to be educated in their clinical presentation, causes and the latest treatments. See References #4 and #5:

4. Bagni C, Zukin RS. A Synaptic Perspective of Fragile X Syndrome and Autism Spectrum Disorders. *Neuron*. 2019 Mar 20;101(6):1070-1088. doi: 10.1016/j.neuron.2019.02.041. PMID: 30897358.
5. Rotaru DC, Mientjes EJ, Elgersma Y. Angelman Syndrome: From Mouse Models to Therapy. *Neuroscience*. 2020 Oct 1;445:172-189. doi: 10.1016/j.neuroscience.2020.02.017. Epub 2020 Feb 21. PMID: 32088294.

Session 4 addresses the current practice gap of under-utilizing novel treatments for ASD such as gene replacement therapy for monogenic forms of ASD, cerebellar circuit modulation, and small molecule and neuropsychopharmacology approaches for ASD. See Reference #6.

6. Erickson CA, Davenport MH, Schaefer TL, Wink LK, Pedapati EV, Sweeney JA, Fitzpatrick SE, Brown WT, Budimirovic D, Hagerman RJ, Hessler D, Kaufmann WE, Berry-Kravis E. Fragile X targeted pharmacotherapy: lessons learned and future directions. *J Neurodev Disord*. 2017 Jun 12;9:7. doi: 10.1186/s11689-017-9186-9. PMID: 28616096; PMCID: PMC5467059.

ADDITIONAL REFERENCES

7. Satterstrom FK, Kosmicki JA, Wang J, et al. Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. *Cell*. 2020;180(3):568-584 e523.

8. Zablotsky B, Black LI, Maenner MJ, et al. Prevalence and Trends of Developmental Disabilities among Children in the United States: 2009-2017. *Pediatrics*. 2019;144(4).

9. Rubenstein E, Croen L, Lee LC, et al. Community-based service use in preschool children with autism spectrum disorder and associations with insurance status. *Res Autism Spectr Disord*. 2019;66.

10. Lindly OJ, Zuckerman KE, Kuhlthau KA. Healthcare access and services use among US children with autism spectrum disorder. *Autism*. 2019;23(6):1419-1430.

11. Sherman SL, Kidd SA, Riley C, et al. FORWARD: A Registry and Longitudinal Clinical Database to Study Fragile X Syndrome. *Pediatrics*. 2017;139(Suppl 3):S183-S193.

12. Cutri-French C, Armstrong D, Saby J, et al. Comparison of Core Features in Four Developmental Encephalopathies in the Rett Natural History Study. *Ann Neurol*. 2020;88(2):396-406.

13. De Rubeis S, Siper PM, Durkin A, et al. Delineation of the genetic and clinical spectrum of Phelan-McDermid syndrome caused by SHANK3 point mutations. *Mol Autism*. 2018;9:31.

14. Dickinson A, Varcin KJ, Sahin M, Nelson CA, 3rd, Jeste SS. Early patterns of functional brain development associated with autism spectrum disorder in tuberous sclerosis complex. *Autism Res*. 2019;12(12):1758-1773.

15. Smith HS, Swint JM, Lalani SR, et al. Clinical Application of Genome and Exome Sequencing as a Diagnostic Tool for Pediatric Patients: a Scoping Review of the Literature. *Genet Med*. 2019;21(1):3-16.

16. Gadalla KK, Bailey ME, Spike RC, et al. Improved survival and reduced phenotypic severity following AAV9/MECP2 gene transfer to neonatal and juvenile male Mecp2 knockout mice. *Mol Ther.* 2013;21(1):18-30.
17. Gross C, Hoffmann A, Bassell GJ, Berry-Kravis EM. Therapeutic Strategies in Fragile X Syndrome: From Bench to Bedside and Back. *Neurotherapeutics.* 2015;12(3):584-608.
18. Osorio AAC, Brunoni AR. Transcranial direct current stimulation in children with autism spectrum disorder: a systematic scoping review. *Dev Med Child Neurol.* 2019;61(3):298-304.
19. Autism Statistics and Facts <https://www.autismspeaks.org/autism-statistics-asd>
20. Eneriz-Wiemer M, Sanders LM, Barr DA, Mendoza FS. Parental limited English proficiency and health outcomes for children with special health care needs: a systematic review. *Acad Pediatr.* 2014 Mar-Apr;14(2):128-36. doi: 10.1016/j.acap.2013.10.003. PMID: 24602575. <https://pubmed.ncbi.nlm.nih.gov/24602575/>
21. Hansen M, Quintero D. Scrutinizing equal pay for equal work among teachers. Brookings Institute. Thursday September 7th. <https://www.brookings.edu/research/scrutinizing-equal-pay-for-equal-work-among-teachers/>
22. Racial and Ethnic Differences in Children Identified with Autism Spectrum Disorder (ASD). Autism and Developmental Disabilities Monitoring (ADDM) Network, from <https://www.cdc.gov/ncbddd/autism/addm.html>
23. Prevalence of Autism Spectrum Disorder Among Minorities, CDC, ADDEM, at <https://www.cdc.gov/ncbddd/autism/addm-community-report/differences-in-children.html>
24. Miron O, Delgado RE, Delgado CF, Simpson EA, Yu KH, Gutierrez A, Zeng G, Gerstenberger JN, Kohane IS. Prolonged Auditory Brainstem Response in Universal Hearing Screening of Newborns with Autism Spectrum Disorder. *Autism Res.* 2021 Jan;14(1):46-52. doi: 10.1002/aur.2422. Epub 2020 Nov 2. PMID: 33140578; PMCID: PMC7894135. <https://pubmed.ncbi.nlm.nih.gov/33140578/>
25. Miron O, Beam AL, Kohane IS. Auditory brainstem response in infants and children with autism spectrum disorder: A meta-analysis of wave V. *Autism Res.* 2018 Feb;11(2):355-363. doi: 10.1002/aur.1886. Epub 2017 Oct 31. PMID: 29087045; PMCID: PMC5836986. <https://pubmed.ncbi.nlm.nih.gov/29087045/>
26. PEDs tests and tools: <https://www.pedstest.com/AboutOurTools/>
27. Ages and Stages Questionnaire (ASQ-3): <https://agesandstages.com/products-pricing/asq3/>
28. Modified Checklist for Autism in Toddlers <https://cms.m-chat.org/LineagenMChat/media/Lineagen-M-Chat-Media/mchatDOTorg.pdf>