2020 Summary of Advances Nominations

Question 1: Screening and Diagnosis

NIMH	Carbone PS, Campbell K, Wilkes J, Stoddard GJ, Huynh K, Young PC, Gabrielsen TP. Primary Care Autism Screening and Later Autism Diagnosis. Pediatrics. 2020 Aug;146(2):e20192314. doi: 10.1542/peds.2019-2314. Epub 2020 Jul 6. PMID: 32632024; PMCID: PMC7397730.
	The three main goals of this research study sought to illustrate the proportion of children screened by the Modified Checklist for Autism in Toddlers (M-CHAT), identify characteristics associated with screen completion, and examine associations between autism spectrum disorder (ASD) screening and later ASD diagnosis. The data were drawn from toddlers (n=36,233) who were seen during their 18- and 24-month well-child visits between 2013-16. Within the sample, the findings showed that approximately 73% of children were screened and that 1.4% were eventually diagnosed with ASD. And among certain subgroups, the results showed that Hispanic children were less likely to be screened and that family physicians were also less likely to screen for ASD. The researchers also noted that while the American Academy of Family Physicians does not currently recommend universal ASD screening, they account for 16-21% of the provision of pediatric care, and thus represent an important opportunity to increase ASD screening in the US.
OARC	Carpenter KLH, Hahemi J, Campbell K, Lippmann SJ, Baker JP, Egger HL, Espinosa S, Vermeer S, Sapiro G, Dawson G. Digital Behavioral Phenotyping Detects Atypical Pattern of Facial Expression in Toddlers with Autism. Autism Res. 2021 Mar;14(3):488-499. doi: 10.1002/aur.2391. Epub 2020 Sep 14. PMID: 32924332; PMCID: PMC7920907.
	Behavioral observation as a tool to screen for ASD, while more objective, is expensive, time-consuming, and requires significant expertise. In this study, the authors test a tablet-based behavioral assessment to detect differences in facial expression of children with and without ASD while watching brief movies. Children without ASD more often displayed raised eyebrows and an open mouth, indicative of engagement/interest, while children with ASD more frequently displayed a neural expression. The results from this study suggest that computational coding of facial movements and expression via a tablet-based assessment can detect differences in affective expression, one of the early, core features of ASD, and potentially be scaled for more objective ASD screening.
OARC	Constantino JN, Abbacchi AM, Saulnier C, Klaiman C, Mandell DS, Zhang Y, Hawks Z, Bates J, Klin A, Shattuck P, Molholm S, Fitzgerald R, Roux A, Lowe JK, Geschwind DH. Timing of the Diagnosis of Autism in African American Children. Pediatrics. 2020 Sep;146(3):e20193629. doi: 10.1542/peds.2019-3629. PMID: 32839243; PMCID: PMC7461218.
	African American (AA) children affected by ASD experience delays in diagnosis and obstacles to service access, as well as a disproportionate burden of intellectual disability (ID). This study analyzed data from the largest-available repository of diagnostic and phenotypic information on AA children with ASD to examine the wide variation in outcome within the cohort. Researchers found that the average age of ASD diagnosis was around 65 months, which was on average 42 months after parents' first concerns about their children's development. These findings serve as a significant opportunity to expedite diagnosis and identify effective approaches to resolve the disparities in severity-of-outcome for AA children with autism.
OARC	Eisenhower A, Martinez Pedraza F, Sheldrick RC, Frenette E, Hoch N, Brunt S, Carter AS. Multi-stage Screening in Early Intervention: A Critical Strategy for Improving ASD Identification and Addressing Disparities. J Autism Dev Disord. 2021 Mar;51(3):868-883. doi: 10.1007/s10803-020-04429-z. PMID: 32144605; PMCID: PMC7483311.

	Health disparities in ASD detection affect children's access to subsequent interventions. Here, researchers examined potential disparities in implementation of a multi-stage ASD screening and diagnostic evaluation protocol with 4943 children ages 14–36 months. Logistic regressions identified predictors of screening participation and outcomes at each stage; demographic differences (race, language, public insurance) were observed only at first-stage screening and reflected higher participation for children of color and higher positive screens for publicly insured children. Results suggest the multi-stage screening protocol shows promise in addressing disparities in early diagnosis.
OARC	Harris JF, Coffield CN, Janvier YM, Mandell D, Cidav Z. Validation of the Developmental Check-In Tool for Low-Literacy Autism Screening. Pediatrics. 2021 Jan;147(1):e20193659. doi: 10.1542/peds.2019-3659. Epub 2020 Dec 10. PMID: 33303635.
	Significant disparities exist in identification of ASD in children from low-income families and are racial/ethnic minorities where English is not the primary language. The Developmental heck-In (DCI) is a visual ASD screening tool that has been shown to be effective at discriminating between ASD versus non-ASD in a young, underserved sample at high-risk for ASD. This study tests the DCI among a general sample of young underserved children recruited through Heah Start and Early Head Star programs. The results demonstrated that 24 of 26 DCI items discriminated ASD from non-ASD, indicating that the DCI is a promising ASD screening tool for young, underserved children.
OARC	Hiruma L, Pretzel RE, Tapia AL, Bodfish JW, Bradley C, Wiggins L, Hsu M, Lee LC, Levy SE, Daniels J. A Distinct Three-Factor Structure of Restricted and Repetitive Behaviors in an Epidemiologically Sound Sample of Preschool-Age Children with Autism Spectrum Disorder. J Autism Dev Disord. 2021 Jan 2. doi: 10.1007/s10803-020-04776-x. Epub ahead of print. PMID: 33387232.
	This report from the Study to Explore Early Development (SEED) found that the presentation of restricted and repetitive behaviors may be distinct for children with ASD between approximately 3 to 6 years of age. Specifically, repetitive speech, repetitive sensorimotor behavior, and insistence on sameness were more clinically relevant for the ASD group versus those in with non-autism developmental concerns. A better understanding these symptoms could improve clinical diagnosis and understanding of ASD symptomatology in this age range.
OARC	Kaat AJ, Shui AM, Ghods SS, Farmer CA, Esler AN, Thurm A, Georgiades S, Kanne SM, Lord C, Kim YS, Bishop SL. Sex differences in scores on standardized measures of autism symptoms: a multisite integrative data analysis. J Child Psychol Psychiatry. 2021 Jan;62(1):97-106. doi: 10.1111/jcpp.13242. Epub 2020 Apr 20. PMID: 32314393.
	This study combines several available databases to create the largest sample of girls with ASD diagnoses and evaluates if sex affects ASD severity estimates using ASD diagnostic tools such as Autism Diagnostic Observation Schedule (ADOS) severity scores, Autism Diagnostic Interview-Revised (ADI-R) raw scores, and raw scores from two indices on the Social Responsiveness Scale (SRS). The authors found that boys received more severe RRB scores than girls, while girls received more severe scores on both SRS indices than boys. While account for sex significantly improved model fit for half of the outcomes, the least square mean differences were generally negligible. Minimal differences due to sex were found beyond other known influences on ASD severity indicators. These results suggest that among children who receive a clinical diagnosis, systematic sex-specific scoring procedures may not be necessary.
OARC	Wallis KE, Guthrie W, Bennett AE, Gerdes M, Levy SE, Mandell DS, Miller JS. Adherence to screening and referral guidelines for autism spectrum disorder in toddlers in pediatric

	primary care. PLoS One. 2020 May 7;15(5):e0232335. doi: 10.1371/journal.pone.0232335. PMID: 32379778; PMCID: PMC7205236.
	This study estimated factors associated with physicians completing the follow-up interview for the Modified Checklist for Autism in Toddlers with Follow-up (M-CHAT-F), and referring children to diagnostic services, audiology, and Early Intervention (EI) after a positive screen. The study of nearly 4,500 children found that toddlers who screen positive for autism do not receive the recommended referrals for follow-up tests and therapies. Understanding pediatrician decision-making about ASD screening is critical to improving care and reducing disparities.
Question 2: Biology	
OARC	Casingal CR, Kikkawa T, Inada H, Sasaki Y, Osumi N. Identification of FMRP target mRNAs in the developmental brain: FMRP might coordinate Ras/MAPK, Wnt/β-catenin, and mTOR signaling during corticogenesis. Mol Brain. 2020 Dec 16;13(1):167. doi: 10.1186/s13041-020-00706-1. PMID: 33323119; PMCID: PMC7739466.
	Fragile X mental retardation protein (FMRP) is an RNA-binding protein responsible for FXS, but its function during brain development remains largely unknown. This study provides insight into the fetal development of the brain and the potential causes of Fragile X syndrome, of which symptoms include ASD. The results provide further insight into the critical roles of FMRP in the developing brain, where dysfunction of FMRP may influence the regulation of its mRNA targets affecting signaling pathways and epigenetic modifications.
OARC	Fazel Darbandi S, Robinson Schwartz SE, Pai EL, Everitt A, Turner ML, Cheyette BNR, Willsey AJ, State MW, Sohal VS, Rubenstein JLR. Enhancing WNT Signaling Restores Cortical Neuronal Spine Maturation and Synaptogenesis in Tbr1 Mutants. Cell Rep. 2020 Apr 14;31(2):107495. doi: 10.1016/j.celrep.2020.03.059. PMID: 32294447; PMCID: PMC7473600.
	The transcription factor Tbr1 has been identified as a risk factor for ASD, and researchers have previously shown that its function is critical for the development of typical deep-layer cortical neurons. Here, two WNT-signaling agonists rescued the dendritic spines and the synaptic and axonal defects, suggesting that these therapeutic approaches could have relevance in some forms of ASD.
OARC	Fields VL, Soke GN, Reynolds A, Tian LH, Wiggins L, Maenner M, DiGuiseppi C, Kral TVE, Hightshoe K, Ladd-Acosta C, Schieve LA. Association between pica and gastrointestinal symptoms in preschoolers with and without autism spectrum disorder, Study to Explore Early Development. Disabil Health J. 2020 Dec 13:101052. doi: 10.1016/j.dhjo.2020.101052. Epub ahead of print. PMID: 33358227.
	Children with autism spectrum disorder and other developmental disabilities (DDs) are disproportionately affected by pica, the persistent ingestion of nonfood items. This study shows an association between gastrointestinal symptoms and pica in children with and without ASD and DDs. However, pica does not fully explain the increased risk for gastrointestinal symptoms among children with ASD and DDs. These findings inform the specialized healthcare needs of children with ASD and other developmental disabilities.
OARC	Gonatopoulos-Pournatzis T, Niibori R, Salter EW, Weatheritt RJ, Tsang B, Farhangmehr S, Liang X, Braunschweig U, Roth J, Zhang S, Henderson T, Sharma E, Quesnel-Vallières M, Permanyer J, Maier S, Georgiou J, Irimia M, Sonenberg N, Forman-Kay JD, Gingras AC, Collingridge GL, Woodin MA, Cordes SP, Blencowe BJ. Autism-Misregulated eIF4G Microexons Control Synaptic Translation and Higher Order Cognitive Functions. Mol Cell. 2020 Mar 19;77(6):1176-1192.e16. doi: 10.1016/j.molcel.2020.01.006. Epub 2020 Jan 29. PMID: 31999954.

	Alternative splicing of microexons (short exons of 3–27 nucleotides) has previously been linked to autism spectrum disorder, but few studies focused on the functional impact of individual microexons. A new study of eIF4G1 and eIF4G3 microexons in genes encoding translation initiation factors fills this gap, linking microexon splicing to translation, synaptic plasticity and ASD-relevant behaviors such as social behavior. This suggests an autism-disrupted mechanism by which alternative splicing specializes neuronal translation to control higher order cognitive functioning.
OARC	Jao Keehn RJ, Pueschel EB, Gao Y, Jahedi A, Alemu K, Carper R, Fishman I, Müller RA. Underconnectivity Between Visual and Salience Networks and Links With Sensory Abnormalities in Autism Spectrum Disorders. J Am Acad Child Adolesc Psychiatry. 2021 Feb;60(2):274-285. doi: 10.1016/j.jaac.2020.02.007. Epub 2020 Feb 29. PMID: 32126259; PMCID: PMC7483217.
	Areas of the brain involved in processing vision are more weakly connected to those that process sensory information in autistic children than in typically developing children, according to this study. Researchers used fMRI to examine resting-state functional connectivity (FC) patterns of salience and visual networks in children and adolescents with ASD compared with typically developing controls, and to relate them to behavioral measures. In some autistic children, this underconnectivity tracks with fewer impairments in social behaviors. The team also found variations among the autistic children, indicating that no one connectivity pattern applies across the spectrum.
NINDS	Lutz AK, Pfaender S, Incearap B, Ioannidis V, Ottonelli I, Föhr KJ, Cammerer J, Zoller M, Higelin J, Giona F, Stetter M, Stoecker N, Alami NO, Schön M, Orth M, Liebau S, Barbi G, Grabrucker AM, Delorme R, Fauler M, Mayer B, Jesse S, Roselli F, Ludolph AC, Bourgeron T, Verpelli C, Demestre M, Boeckers TM. Autism-associated SHANK3 mutations impair maturation of neuromuscular junctions and striated muscles. Sci Transl Med. 2020 Jun 10;12(547):eaaz3267. doi: 10.1126/scitranslmed.aaz3267. PMID: 32522805.
	Heterozygous mutations of the gene encoding the postsynaptic protein SHANK3 are associated with syndromic forms of ASD, including Phelan-McDermid syndrome. Neonatal skeletal muscle hypotonia is an early clinical symptom in SHANK3-associated ASD. Here, researchers used patient-derived human induced pluripotent stem cells (hiPSCs), Shank3Δ11(-/-) mice, and muscle biopsies from patients with Phelan-McDermid syndrome to analyze the role of SHANK3 on muscle motor unit development. They also show that motor defects in Shank3Δ11(-/-) mice could be rescued with Tirasemtiv, an experimental drug (in development for amyotrophic lateral sclerosis) that sensitizes muscle fibers to calcium.
OARC	Marrus N, Grant JD, Harris-Olenak B, Albright J, Bolster D, Haber JR, Jacob T, Zhang Y, Heath AC, Agrawal A, Constantino JN, Elison JT, Glowinski AL. Genetic architecture of reciprocal social behavior in toddlers: Implications for heterogeneity in the early origins of autism spectrum disorder. Dev Psychopathol. 2020 Oct;32(4):1190-1205. doi: 10.1017/S0954579420000723. PMID: 33161906.
	Impairment in reciprocal social behavior (RSB) is an important clinical part of ASD. However, the behavioral and genetic architecture of RSB in early childhood has not been fully characterized. Researchers studied toddlers' RSBs in a group of toddlers from a community-based volunteer research registry and an ethnically diverse, longitudinal twin sample from two state birth registries. The authors found that variation in RSB was continuously distributed, strongly associated with ASD risk, and temporally stable at 18 months. Genetic analysis at 24 months indicated substantial heritability for all RSB factors. These results demonstrate that substantially overlapping RSB domains may serve as markers of specific pathways to autism.

OARC	Reed MD, Yim YS, Wimmer RD, Kim H, Ryu C, Welch GM, Andina M, King HO, Waisman A, Halassa MM, Huh JR, Choi GB. IL-17a promotes sociability in mouse models of neurodevelopmental disorders. Nature. 2020 Jan;577(7789):249-253. doi: 10.1038/s41586-019-1843-6. Epub 2019 Dec 18. PMID: 31853066.
	Fevers have sometimes been reported to temporarily improve autistic children's behavior. Here, an immune molecule produced during a fever called interleukin 17A (IL-17A) improved sociability in three mouse models of autism. This study supports a neuroimmune mechanism in which the production of IL-17a during inflammation can ameliorate the expression of social behavior deficits by directly affecting neuronal activity in the central nervous system.
OARC	Resendez SL, Namboodiri VMK, Otis JM, Eckman LEH, Rodriguez-Romaguera J, Ung RL, Basiri ML, Kosyk O, Rossi MA, Dichter GS, Stuber GD. Social Stimuli Induce Activation of Oxytocin Neurons Within the Paraventricular Nucleus of the Hypothalamus to Promote Social Behavior in Male Mice. J Neurosci. 2020 Mar 11;40(11):2282-2295. doi: 10.1523/JNEUROSCI.1515-18.2020. Epub 2020 Feb 5. PMID: 32024781; PMCID: PMC7083279.
	Oxytocin is known to play a role in social behaviors in many species, but the neural circuits that mediate the effects of oxytocin are not well understood. In this study, researchers performed in vivo calcium imaging of oxytocin-positive (OT) neurons in mice. They found that OT neurons in the paraventricular nucleus of the hypothalamus (PVH) responds preferentially to social stimuli and is necessary for social behavior. Deficits in this population of neurons may underlie social deficits in *Shank3b* knockout (KO) mice.
OARC	Reynolds AM, Soke GN, Sabourin KR, Croen LA, Daniels JL, Fallin MD, Kral TVE, Lee LC, Newschaffer CJ, Pinto-Martin JA, Schieve LA, Sims A, Wiggins L, Levy SE. Gastrointestinal Symptoms in 2- to 5-Year-Old Children in the Study to Explore Early Development. J Autism Dev Disord. 2021 Jan 4. doi: 10.1007/s10803-020-04786-9. Epub ahead of print. PMID: 33394243.
	Gastrointestinal symptoms (GIS) are commonly reported in children with ASD. Here, the prevalence of GIS in preschool-aged children with autism, other developmental delays (DOD), and other children in the general population (POP) was evaluated. Children in the ASD group were over 3 times more likely to report GIS than the POP group, and almost 2 times more likely to report GIS than the DOD group. These findings have implications for the clinical management of ASD.
OARC	Simonoff E, Kent R, Stringer D, Lord C, Briskman J, Lukito S, Pickles A, Charman T, Baird G. Trajectories in Symptoms of Autism and Cognitive Ability in Autism From Childhood to Adult Life: Findings From a Longitudinal Epidemiological Cohort. J Am Acad Child Adolesc Psychiatry. 2020 Dec;59(12):1342-1352. doi: 10.1016/j.jaac.2019.11.020. Epub 2019 Dec 19. PMID: 31863881.
	IQ is generally expected to remain stable over time, but this study suggests that the measure increases significantly for people with ASD during adolescence and early adulthood. In this long-term study of 126 people on the spectrum, IQ scores increased by an average of 7.48 points between the ages of 12 and 23. In this same period, symptoms of autism in the participants remained unchanged. These findings suggest continued cognitive increments for many people with autism in adolescence, but a lack of improvement in autism symptoms.
NINDS	Tai C, Chang CW, Yu GQ, Lopez I, Yu X, Wang X, Guo W, Mucke L. Tau Reduction Prevents Key Features of Autism in Mouse Models. Neuron. 2020 May 6;106(3):421-437.e11. doi: 10.1016/j.neuron.2020.01.038. Epub 2020 Mar 2. PMID: 32126198; PMCID: PMC7210056.
	This paper presents evidence that reducing the protein tau prevents behavioral signs of autism in two mouse models with distinct genetic causes (Scn1aRX/+ and Cntnap2−/− mice), but with other shared features associated with some ASDs, including epilepsy, abnormally

	enlarged brains, and overactivation of the phosphatidylinositol 3-kinase (PI3K)/Akt (protein kinase B)/ mammalian target of rapamycin (mTOR) signaling pathway. These abnormalities were prevented or diminished by partial or complete genetic removal of tau, perhaps through disinhibition of PTEN, a negative PI3K regulator controlled by tau. The results suggest a role for tau in some types of ASD and point to tau reduction as a potential therapeutic strategy.
OARC	Trakoshis S, Martínez-Cañada P, Rocchi F, Canella C, You W, Chakrabarti B, Ruigrok AN, Bullmore ET, Suckling J, Markicevic M, Zerbi V; MRC AIMS Consortium, Baron-Cohen S, Gozzi A, Lai MC, Panzeri S, Lombardo MV. Intrinsic excitation-inhibition imbalance affects medial prefrontal cortex differently in autistic men versus women. Elife. 2020 Aug 4;9:e55684. doi: 10.7554/eLife.55684. PMID: 32746967; PMCID: PMC7402681.
	Autistic men show a greater imbalance between excitatory and inhibitory signaling in the medial prefrontal cortex than autistic women. This brain area is involved in cognition and social behavior and may be why autistic women seem to camouflage their autism more commonly than men. This finding builds on the signaling imbalance theory of autism, which proposes that autism is the result of too much excitation or inhibition in the brain.
	Question 3: Risk Factors
OARC	Ali AA, Cui X, Pertile RAN, Li X, Medley G, Alexander SA, Whitehouse AJO, McGrath JJ, Eyles DW. Developmental vitamin D deficiency increases foetal exposure to testosterone. Mol Autism. 2020 Dec 10;11(1):96. doi: 10.1186/s13229-020-00399-2. PMID: 33298169; PMCID: PMC7727109.
	The prenatal sex steroid hypothesis links excessive prenatal sex steroid exposure to the behavioral differences observed in ASD. This paper suggests that a deficiency in maternal vitamin D in rats caused an increase in testosterone in the developing brains of males. This could explain why ASD is three times more common in boys.
OARC	Breuss MW, Antaki D, George RD, Kleiber M, James KN, Ball LL, Hong O, Mitra I, Yang X, Wirth SA, Gu J, Garcia CAB, Gujral M, Brandler WM, Musaev D, Nguyen A, McEvoy-Venneri J, Knox R, Sticca E, Botello MCC, Uribe Fenner J, Pérez MC, Arranz M, Moffitt AB, Wang Z, Hervás A, Devinsky O, Gymrek M, Sebat J, Gleeson JG. Autism risk in offspring can be assessed through quantification of male sperm mosaicism. Nat Med. 2020 Jan;26(1):143-150. doi: 10.1038/s41591-019-0711-0. Epub 2019 Dec 23. PMID: 31873310; PMCID: PMC7032648.
	De novo mutations on the paternal chromosome make the largest known contribution to autism risk, and correlate with paternal age at the time of conception. However, the potential impact of assessing parental gonadal mosaicism has not been considered. Here, sperm mosaicism was measured using deep-whole-genome sequencing, for variants both present in an offspring and evident only in father's sperm. The study found that mosaicism quantification can stratify autism spectrum disorders recurrence risk due to de novo mutations into a vast majority with near 0% recurrence and a small fraction with a substantially higher and quantifiable risk. This suggests that genetic counseling would benefit from the addition of sperm mosaicism assessment.
OARC	Chiang AH, Chang J, Wang J, Vitkup D. Exons as units of phenotypic impact for truncating mutations in autism. Mol Psychiatry. 2020 Oct 27. doi: 10.1038/s41380-020-00876-3. Epub ahead of print. PMID: 33110259.
	While multiple gene-disrupting mutations in the same gene can often lead to variable ASD phenotypes, this study found that people with autism who carry truncating mutations in the same exon have similar cognitive abilities and behaviors. Analogous patterns were observed for two independent proband cohorts and several other important ASD-associated phenotypes. The researchers found that exons biased toward prenatal phenotypes

	preferentially contribute towards lower IQ phenotypes, while exons biased towards postnatal phenotypes contribute towards higher IQ phenotypes. These results suggest that exons, rather than genes, often represent a unit of effective phenotypic impact for truncating mutations in autism
OARC	Cummings BB, Karczewski KJ, Kosmicki JA, Seaby EG, Watts NA, Singer-Berk M, Mudge JM, Karjalainen J, Satterstrom FK, O'Donnell-Luria AH, Poterba T, Seed C, Solomonson M, Alföldi J; Genome Aggregation Database Production Team; Genome Aggregation Database Consortium, Daly MJ, MacArthur DG. Transcript expression-aware annotation improves rare variant interpretation. Nature. 2020 May;581(7809):452-458. doi: 10.1038/s41586-020-2329-2. Epub 2020 May 27. Erratum in: Nature. 2021 Feb;590(7846):E54. PMID: 32461655; PMCID: PMC7334198.
	DNA sequencing has resulted in extensive catalogues of human genetic variation, but the interpretation of rare genetic variants in patients remains problematic. This study uses a new tool that helps researchers determine how mutations in an exon affect the number of protein isoforms a gene can express. This expression filter was used to analyze de novo variants in patients with ASD and developmental disorders to show that putative loss-of-function (pLoF) variants in weakly expressed regions have similar effect sizes to those of synonymous variants, whereas pLoF variants in highly expressed exons are most strongly enriched among cases. This will be valuable for the genetic diagnosis of rare diseases, the analysis of rare variant burden in complex disorders, and the curation and prioritization of variants in recall-by-genotype studies.
NINDS	Jensen M, Smolen C, Girirajan S. Gene discoveries in autism are biased towards comorbidity with intellectual disability. J Med Genet. 2020 Sep;57(9):647-652. doi: 10.1136/jmedgenet-2019-106476. Epub 2020 Mar 9. PMID: 32152248; PMCID: PMC7483239.
	Intellectual disability (ID) is often comorbid with autism, which presents challenges for diagnosis and for identifying genetic factors specifically associated with autism. In this study, researchers analyzed pathogenic de novo genetic variants in individuals with autism who had either ID or normal cognitive function. The results suggest that pathogenic de novo variants disrupting autism-associated genes contribute towards autism and ID comorbidity, but that other genetic factors (e.g., missense variants, common variants, variants in regulatory and non-coding regions, or the combinatorial effects of inherited variants) are likely to be causal for high-functioning autism. The study also highlights the importance of stratifying phenotypic heterogeneity in sequencing studies.
OARC	Mordaunt CE, Jianu JM, Laufer BI, Zhu Y, Hwang H, Dunaway KW, Bakulski KM, Feinberg JI, Volk HE, Lyall K, Croen LA, Newschaffer CJ, Ozonoff S, Hertz-Picciotto I, Fallin MD, Schmidt RJ, LaSalle JM. Cord blood DNA methylome in newborns later diagnosed with autism spectrum disorder reflects early dysregulation of neurodevelopmental and X-linked genes. Genome Med. 2020 Oct 14;12(1):88. doi: 10.1186/s13073-020-00785-8. PMID: 33054850; PMCID: PMC7559201.
	This study investigates how differences in the neonatal epigenome, which reflect past interactions between genetic and environmental factors during early development, may discriminate ASD diagnosis. The authors sequenced the methylome of umbilical cord blood samples from the MARBLES and EARLI high-familial risk prospective cohorts to identify an epigenomic signature of ASD at birth. At a region level, 7 differentially methylated regions (DMRs) were identified in males, and 31 DMRs were identified in females across two independent groups of subjects. Autosomal ASD DMRs were significantly enriched for promoter and bivalent chromatin states, and sex differences were observed for X-linked ASD DRMs. Genes associated with DMRs were significantly enriched for brain and embryonic

	expression and binding sites of methyl-sensitive transcription factors relevant to fetal brain development.
NINDS	Niesler B, Rappold GA. Emerging evidence for gene mutations driving both brain and gut dysfunction in autism spectrum disorder. Mol Psychiatry. 2020 May 27. doi: 10.1038/s41380-020-0778-5. Epub ahead of print. PMID: 32461615.
	Gastrointestinal (GI) dysfunction is common in individuals with neurodevelopmental and neuropsychiatric diseases including autism. This commentary posits that genetic factors associated with neuronal dysfunction in the brain and central nervous systems (CNS) also affect GI development and function, contributing directly to GI symptoms in autism spectrum disorders. Further research may lead to treatments that improve both GI and behavioral symptoms.
NINDS	Satterstrom FK, Kosmicki JA, Wang J, Breen MS, De Rubeis S, An JY, Peng M, Collins R, Grove J, Klei L, Stevens C, Reichert J, Mulhern MS, Artomov M, Gerges S, Sheppard B, Xu X, Bhaduri A, Norman U, Brand H, Schwartz G, Nguyen R, Guerrero EE, Dias C; Autism Sequencing Consortium; iPSYCH-Broad Consortium, Betancur C, Cook EH, Gallagher L, Gill M, Sutcliffe JS, Thurm A, Zwick ME, Børglum AD, State MW, Cicek AE, Talkowski ME, Cutler DJ, Devlin B, Sanders SJ, Roeder K, Daly MJ, Buxbaum JD. Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell. 2020 Feb 6;180(3):568-584.e23. doi: 10.1016/j.cell.2019.12.036. Epub 2020 Jan 23. PMID: 31981491; PMCID: PMC7250485.
	In this landmark study, the international Autism Sequencing Consortium (ASC) analyzed the DNA of over 35,000 people from around the world. They identified variants in 102 genes associated with increased risk of developing ASD, up from 65 identified previously. Of the 102 genes, 60 had not been previously linked to ASD and 53 were associated more specifically with ASD as opposed to intellectual disability or developmental delay.
NINDS	Trost B, Engchuan W, Nguyen CM, Thiruvahindrapuram B, Dolzhenko E, Backstrom I, Mirceta M, Mojarad BA, Yin Y, Dov A, Chandrakumar I, Prasolava T, Shum N, Hamdan O, Pellecchia G, Howe JL, Whitney J, Klee EW, Baheti S, Amaral DG, Anagnostou E, Elsabbagh M, Fernandez BA, Hoang N, Lewis MES, Liu X, Sjaarda C, Smith IM, Szatmari P, Zwaigenbaum L, Glazer D, Hartley D, Stewart AK, Eberle MA, Sato N, Pearson CE, Scherer SW, Yuen RKC. Genome-wide detection of tandem DNA repeats that are expanded in autism. Nature. 2020 Oct;586(7827):80-86. doi: 10.1038/s41586-020-2579-z. Epub 2020 Jul 27. PMID: 32717741.
	Tandem DNA repeats are associated with more than 40 monogenic disorders, but their role in diseases with complex genetics is largely unknown. This study investigated tandem repeats in 17,231 genomes of families with individuals with ASD and in population control individuals. At 2,588 loci, they found gene-associated expansions of tandem repeats that were rare among controls and more prevalent among individuals with ASD than their siblings without ASD. Overall, the results estimate that tandem repeat expansions make a collective contribution to the risk of ASD of 2.6%.
Question 4: Treatments and Interventions	
OARC	Driscoll K, Schonberg M, Stark MF, Carter AS, Hirshfeld-Becker D. Family-Centered Cognitive Behavioral Therapy for Anxiety in Very Young Children with Autism Spectrum Disorder. J Autism Dev Disord. 2020 Nov;50(11):3905-3920. doi: 10.1007/s10803-020-04446-y. PMID: 32146598.
	There is little research on the effectiveness of cognitive-behavioral therapy (CBT) protocols on very young children with ASD. This study piloted a family-centered CBT protocol in 16 children aged 3-7 years old with ASD and anxiety disorders. At post-treatment, 81% of children were rated "very much" or "much improved" on the CGI-Anxiety, decreased

	clinician- and parent-rated anxiety, and improved family function and coping, and gains were maintained at the 4-months follow-up.
OARC	González-Domenech PJ, Díaz Atienza F, García Pablos C, Fernández Soto ML, Martínez-Ortega JM, Gutiérrez-Rojas L. Influence of a Combined Gluten-Free and Casein-Free Diet on Behavior Disorders in Children and Adolescents Diagnosed with Autism Spectrum Disorder: A 12-Month Follow-Up Clinical Trial. J Autism Dev Disord. 2020 Mar;50(3):935-948. doi: 10.1007/s10803-019-04333-1. PMID: 31813108.
	Gluten-free and casein-free (GFCF) diets are frequently used as alternative interventions for ASD. This study recruited 37 patients for a crossover trial. Each patient consumed a normal diet for 6 months and a GFCF diet for 6 months; the order (beginning with the normal diet or GFCF diet) was randomly assigned. Questionnaires regarding behavior and autism and dietary adherence were completed and urinary beta-casomorphin concentrations were determined at the beginning of the study, after normal diet, and after GFCF diet. No significant behavioral changes and no association with urinary beta-casomorphin concentrations were found after GFCF diet.
OARC	Lindgren S, Wacker D, Schieltz K, Suess A, Pelzel K, Kopelman T, Lee J, Romani P, O'Brien M. A Randomized Controlled Trial of Functional Communication Training via Telehealth for Young Children with Autism Spectrum Disorder. J Autism Dev Disord. 2020 Dec;50(12):4449-4462. doi: 10.1007/s10803-020-04451-1. PMID: 32300910; PMCID: PMC7572463.
	This randomized controlled trial compares the effects of parent-conducted functional communication training (FCT) on 38 young children with ASD aged 21-84 months. Parents received FCT training from behavioral consultants via telehealth. FCT treatment resulted in a mean reduction in problem behavior of 98% compared to limited behavioral improvement in children receiving "treatment as usual" during a 12-week period. Improvements were also seen in in social communication and task completion.
NINDS	McCamphill PK, Stoppel LJ, Senter RK, Lewis MC, Heynen AJ, Stoppel DC, Sridhar V, Collins KA, Shi X, Pan JQ, Madison J, Cottrell JR, Huber KM, Scolnick EM, Holson EB, Wagner FF, Bear MF. Selective inhibition of glycogen synthase kinase 3α corrects pathophysiology in a mouse model of fragile X syndrome. Sci Transl Med. 2020 May 20;12(544):eaam8572. doi: 10.1126/scitranslmed.aam8572. PMID: 32434848.
	Fragile X syndrome (FXS) is among the most common single gene causes of autism. Studies in a mouse model of FXS implicate aberrant protein synthesis downstream of signaling through the mGluR5 glutamate receptor as a key disease mechanism. However, clinical trials of mGluR5 inhibitors have not been successful, prompting research on alternative treatment strategies. This study shows that selective inhibition of the enzyme glycogen synthase kinase 3 alpha (GSK3α) corrected protein synthesis in a FXS mouse model and improved brain hyperexcitability, learning and memory deficits, and other measures. The results point to GSK3α inhibitors as a potential therapeutic approach for FXS.
OARC	Pellecchia M, Marcus SC, Spaulding C, Seidman M, Xie M, Rump K, Reisinger EM, Mandell DS. Randomized Trial of a Computer-Assisted Intervention for Children With Autism in Schools. J Am Acad Child Adolesc Psychiatry. 2020 Mar;59(3):373-380. doi: 10.1016/j.jaac.2019.03.029. Epub 2019 Apr 3. PMID: 30953731; PMCID: PMC6996249.
	Computer-assisted interventions (CAIs) are popular for educating children with autism, but their effectiveness is not well established. This study evaluated the effectiveness of TeachTown, a CAI designed to improve children's language, cognitive, and academic skills. There were no statistically significant differences in outcomes for children who received TeachTown or treatment as usual. Increased time spent using TeachTown was associated with worse receptive language outcomes for children. Despite growing enthusiasm for CAIs in

	autism treatment, these findings indicate that the decision to implement CAIs in schools should be carefully balanced against the effectiveness of these programs.
OARC	Rogers SJ, Yoder P, Estes A, Warren Z, McEachin J, Munson J, Rocha M, Greenson J, Wallace L, Gardner E, Dawson G, Sugar CA, Hellemann G, Whelan F. A Multisite Randomized Controlled Trial Comparing the Effects of Intervention Intensity and Intervention Style on Outcomes for Young Children With Autism. J Am Acad Child Adolesc Psychiatry. 2020 Aug 24:S0890-8567(20)31350-2. doi: 10.1016/j.jaac.2020.06.013. Epub ahead of print. PMID: 32853704.
	This study examined the effects of 2 levels of treatment intensity (number of hours) and 2 treatment styles on the progress of young children with autism spectrum disorder (ASD). Study results showed that toddlers performed the same regardless of treatment intensity or teaching approach, and that child characteristics did not predict outcomes either. These findings highlight the importance of the rigorous implementation and study of autism intervention science.
OARC	Solish A, Klemencic N, Ritzema A, Nolan V, Pilkington M, Anagnostou E, Brian J. Effectiveness of a modified group cognitive behavioral therapy program for anxiety in children with ASD delivered in a community context. Mol Autism. 2020 May 13;11(1):34. doi: 10.1186/s13229-020-00341-6. PMID: 32404180; PMCID: PMC7218559.
	Youths on the autism spectrum often experience high rates of comorbid anxiety issues. Cognitive behavioral therapy (CBT) has been demonstrated in controlled research settings to be effective in both individual and group formats for treating anxiety disorders across the lifespan. This study examines the effectiveness of a modified group CBT program (Face Your Fears) delivered in a tertiary care hospital and across six community-based agencies providing services for youths with ASD. Results show significant improvements in anxiety levels from baseline to post-treatment, with medium effect sizes.
OARC	Wood JJ, Kendall PC, Wood KS, Kerns CM, Seltzer M, Small BJ, Lewin AB, Storch EA. Cognitive Behavioral Treatments for Anxiety in Children With Autism Spectrum Disorder: A Randomized Clinical Trial. JAMA Psychiatry. 2020 May 1;77(5):474-483. doi: 10.1001/jamapsychiatry.2019.4160. PMID: 31755906; PMCID: PMC6902190.
	Researchers compared the impacts of 2 variants of cognitive behavioral therapy (CBT) on children with ASD and maladaptive and interfering anxiety. Study results showed that CBT adapted for children with ASD led to a greater reduction in anxiety than standard CBT, and both forms of CBT were more efficacious than treatment as usual.
	Question 5: Services
OARC	Bellesheim KR, Kizzee RL, Curran A, Sohl K. ECHO Autism: Integrating Maintenance of Certification with Extension for Community Healthcare Outcomes Improves Developmental Screening. J Dev Behav Pediatr. 2020 Aug;41(6):420-427. doi: 10.1097/DBP.0000000000000796. PMID: 32735419.
	Recent data indicates that most pediatricians do not consistently meet the American Academy of Pediatrics developmental screening guidelines, contributing to delays in diagnosis and interventions for ASD. This study investigated whether the use of Maintenance of Certification (MOC) Quality Improvement (QI) training improves developmental screening rates in underserved, rural primary care practices. At the end of the 12-months learning module, screening rates increased to 88.6% (versus the initial 53.3%) for general developmental training and 99% (versus the initial 68.3%) for ASD-specific developmental screenings. At the one-year follow-up, the rates for general and ASD-specific screening was 96.7% and 97.1%, respectively.
OARC	Brookman-Frazee L, Chlebowski C, Villodas M, Roesch S, Martinez K. Training Community Therapists to Deliver an Individualized Mental Health Intervention for Autism Spectrum

	Disorder: Changes in Caregiver Outcomes and Mediating Role on Child Outcomes. J Am Acad Child Adolesc Psychiatry. 2021 Mar;60(3):355-366. doi: 10.1016/j.jaac.2020.07.896. Epub 2020 Aug 2. PMID: 32755632.
	This cluster randomized trial conducted in 29 publicly funded mental health programs examined the impact of training therapists to deliver "An Individualized Mental Health Intervention for Autism Spectrum Disorder (ASD)" (AIM HI). 202 caregivers of children 5 to 13 years of age with ASD were enrolled, and caregiver strain and sense of competence were assessed at baseline and 6 month postbaseline and child behaviors were assessed at baseline and 6, 12, and 18 months postbaseline. Significant training effect was observed for caregiver sense of competence and associated with reduced child challenging behaviors at 6 months and mediated child outcomes at 12 and 18 months.
OARC	Donnelly LJ, Cervantes PE, Guo F, Stein CR, Okparaeke E, Kuriakose S, Filton B, Havens J, Horwitz SM. Changes in Attitudes and Knowledge after Trainings in a Clinical Care Pathway for Autism Spectrum Disorder. J Autism Dev Disord. 2020 Nov 17. doi: 10.1007/s10803-020-04775-y. Epub ahead of print. PMID: 33201422.
	Health care providers often report feeling unprepared to work with individuals with ASD because specialized training for medical and social service providers is limited. This study modified and tested a previously validated half-day ASD-Care Pathway training in five different settings. Significant improvements in staff perceptions of challenging behaviors, increased comfort in working with the ASD population, and increased staff knowledge for evidence-based practices were found after this short training session on strategies for preventing and reducing challenging behaviors of patients with ASD.
OARC	Mazurek MO, Parker RA, Chan J, Kuhlthau K, Sohl K; ECHO Autism Collaborative. Effectiveness of the Extension for Community Health Outcomes Model as Applied to Primary Care for Autism: A Partial Stepped-Wedge Randomized Clinical Trial. JAMA Pediatr. 2020 May 1;174(5):e196306. doi: 10.1001/jamapediatrics.2019.6306. Epub 2020 May 4. PMID: 32150229; PMCID: PMC7063545.
	In this large-scale study, the Extension for Community Health Outcomes (ECHO) telementoring model was evaluated for effectiveness in improving clinical practices for the care of children with autism. Significant changes in autism screening or treatment of comorbidities were not demonstrated. However, primary care clinicians demonstrated significant improvements in knowledge and self-efficacy immediately following and 3 months after completing the ECHO program.
OARC	McBain RK, Cantor JH, Kofner A, Stein BD, Yu H. State Insurance Mandates and the Workforce for Children With Autism. Pediatrics. 2020 Oct;146(4):e20200836. doi: 10.1542/peds.2020-0836. Epub 2020 Sep 8. PMID: 32900876; PMCID: PMC7546088.
	State mandates have required insurance companies to provide coverage for autism-related child health care services. However, it has not been determined if insurance mandates have improved the supply of child health care providers. Here, the researchers investigate the effect of state insurance mandates on access to child psychiatrists, pediatricians, and board-certified behavioral analysts (BCBAs). State insurance mandates were associated with a 16% increase in BCBAs from 2003 to 2017, but the association with child psychiatrists was smaller and nonsignificant among pediatricians. These findings suggest that policies are needed to specifically address workforce constraints in services for children with ASD.
OARC	McConkey R, Cassin MT, McNaughton R. Promoting the Social Inclusion of Children with ASD: A Family-Centred Intervention. Brain Sci. 2020 May 25;10(5):318. doi: 10.3390/brainsci10050318. PMID: 32466092; PMCID: PMC7288007.

	Community inclusion and engagement is an important issue for the autism community. This study involves a project in Northern Ireland that provided post-diagnostic support to nearly 100 families with children aged 3 to 11 years. Over a 12-month period, an experienced ASD practitioner visited the family and children every fortnight to implement a family-centered plan that introduced the child to various community activities in line with their learning targets and wishes. Quantitative and qualitative results from the study showed improvements in the children's social and communication skills, personal safety, participation in community activities, and reduced stress within the families and strengthened relationships.
OARC	Smith KA, Gehricke JG, Iadarola S, Wolfe A, Kuhlthau KA. Disparities in Service Use Among Children With Autism: A Systematic Review. Pediatrics. 2020 Apr;145(Suppl 1):S35-S46. doi: 10.1542/peds.2019-1895G. PMID: 32238530.
	Researchers reviewed evidence related to disparities in service use and quality of care provided to children with autism by race, ethnicity, and socioeconomic status. They found that racial and ethnic minority groups and low-income families face significant barriers to getting needed services. Researchers did not find studies in which differences in intervention effectiveness was examined, suggesting a need for a larger body of literature to document this topic.
OARC	Stadnick NA, Lau AS, Dickson KS, Pesanti K, Innes-Gomberg D, Brookman-Frazee L. Service use by youth with autism within a system-driven implementation of evidence-based practices in children's mental health services. Autism. 2020 Nov;24(8):2094-2103. doi: 10.1177/1362361320934230. Epub 2020 Jul 18. PMID: 32686469; PMCID: PMC7541440.
	Youths on the autism spectrum may receive mental health services in public mental health systems. This case-control study compared 2537 youths with ASD to 2537 matched peers receiving care in the Los Angeles County Department of Mental Health. On average, youths with ASD had significantly higher volume of claims and received care for significantly longer duration than matched peers. In addition, there were group differences in the types of mental health services received and the practices that were delivered. For example, behavioral parent training practices were delivered more to youths with ASD, whereas practices to address trauma were delivered more to matched peers. These results demonstrate that it may be valuable to explicitly provide more tailored care recommendations for youths on the autism spectrum.
Question 6: Lifespan Issues	
OARC	Garcia JM, Lawrence S, Brazendale K, Leahy N, Fukuda D. Brief report: The impact of the COVID-19 pandemic on health behaviors in adolescents with Autism Spectrum Disorder. Disabil Health J. 2021 Apr;14(2):101021. doi: 10.1016/j.dhjo.2020.101021. Epub 2020 Nov 5. PMID: 33221246.
	This study investigated how restrictions associated with the COVID-19 pandemic affected health behaviors, including physical activity, screen-time, and sleep, in nine youths diagnosed with ASD. Both youths and parents filled out electronic surveys in March and April of 2020, and survey responses indicated a significant decrease in days of physical activity and increase in hours of screen-time. No changes were observed in sleep duration. These results indicate that interventions may be needed to promote health behaviors during long periods of less-structured times such as quarantine.
OARC	Graham Holmes L, Zampella CJ, Clements C, McCleery JP, Maddox BB, Parish-Morris J, Udhnani MD, Schultz RT, Miller JS. A Lifespan Approach to Patient-Reported Outcomes and Quality of Life for People on the Autism Spectrum. Autism Res. 2020 Jun;13(6):970-987. doi: 10.1002/aur.2275. Epub 2020 Mar 10. PMID: 32154664.

	The autism community has called for greater emphasis on enhancing quality of life (QoL) for people with autism. This study created and tested a lifespan QoL measurement tool using National Institutes of Health Parent-Reported Outcomes Measurement Information System (PROMIS). This battery provides a comprehensive portrait of QoL for children, teens, and adults with autism compared to the general population. Some reported a good quality of life, while many reported that their lives were not going as well as they wanted. Women and girls reported more challenges in some areas of life than men and boys.
OARC	Kirby AV, Diener ML, Adkins DE, Wright C. Transition preparation activities among families of youth on the autism spectrum: Preliminary study using repeated assessments across a school year. PLoS One. 2020 Apr 16;15(4):e0231551. doi: 10.1371/journal.pone.0231551. PMID: 32298327; PMCID: PMC7161970.
	Little is known about how to best prepare youths in the transition to adulthood, particularly what roles parents can play in the process that would be helpful. This study followed 15 families of youths with ASD (ages 14-17 and 93% male) for 10 months across an academic year. Findings from this preliminary study indicate that discussions about the future were the most commonly endorsed activities, and social activities were perceived by parents to be most associated with transition preparation over time.
OARC	Lei J, Calley S, Brosnan M, Ashwin C, Russell A. Evaluation of a Transition to University Programme for Students with Autism Spectrum Disorder. J Autism Dev Disord. 2020 Jul;50(7):2397-2411. doi: 10.1007/s10803-018-3776-6. PMID: 30315485; PMCID: PMC7308263.
	The application process and transition to college can be anxiety-inducing for individuals on the autism spectrum. This study evaluates the effectiveness of a transition to university pilot program (Autism Summer School) for autistic students aged 16-19 years who are seeking to apply to or attend college. The program consisted of a three-day, two-night stay on a university campus, and the curriculum was designed to prepare autistic students for typical university life and promote self-care and wellbeing at university, covering topics regarding "work" (lectures, socializing with staff and tutors, and accessing student support and disclosing diagnosis), "rest" (stress reduction, management of situational anxiety, and sporting facilities), and "play" (clubs and societies, shared meals on campus, social outings, and informal socializing). Results from the study indicate that the program identified and reduced concerns of autistic students who are considering attending university.
OARC	Lord C, McCauley JB, Pepa LA, Huerta M, Pickles A. Work, living, and the pursuit of happiness: Vocational and psychosocial outcomes for young adults with autism. Autism. 2020 Oct;24(7):1691-1703. doi: 10.1177/1362361320919246. Epub 2020 May 20. PMID: 32431163; PMCID: PMC7541415.
	Longitudinal data on the functioning of adults who were diagnosed with ASD as children are sparse. The authors studied a cohort of 123 young adults, mean age of 26, referred for neurodevelopmental disorders in early childhood. The cohort was divided into those with an ASD diagnosis at some point and a current IQ greater than or equal to 70 (Ever ASD_Higher IQ), those with an ASD diagnosis at some point and a current IQ less than 70 (Ever ASD_Lower IQ), and those who ever received an ASD diagnosis (Never ASD). Results from direct testing, questionnaires, and interviews indicate that verbal IQ accounted for group differences in employment and autistic features were related to adaptive skills and friendships. ASD diagnosis was associated with lower well-being and fewer positive emotions, with the Ever ASD_Lower IQ group having the highest levels of irritability, hyperactivity, and medications. Families played major roles in supporting adults with and without ASD at all intellectual levels.

NIMH	McCauley JB, Pickles A, Huerta M, Lord C. Defining Positive Outcomes in More and Less Cognitively Able Autistic Adults. Autism Res. 2020 Sep;13(9):1548-1560. doi: 10.1002/aur.2359. Epub 2020 Aug 27. PMID: 32851813.
	Utilizing longitudinal data from (n=126) adults diagnosed with ASD (mean age=26), this study examined how several domains of outcomes—namely autonomy, social relationships, and purpose, were correlated with specific aspects of functioning in autistic adults. For more cognitively able adults, outcomes included living independently, having paid employment, and at least one true friend. For autistic adults who were less cognitively able, outcomes included daily living skills above an 8-year-old level, having regular activities outside the home, and having social contacts outside the family. Verbal IQ was a significant predictor of outcomes achieved for individuals within both more and less cognitively able groups. The findings also demonstrated that among adults with less cognitive abilities, having ever received a formal ASD diagnosis was associated with lower odds of positive outcomes. Conversely, among ASD adults with more cognitive skills, there was a higher likelihood of positive outcomes such as increased daily living skills, fewer mental health problems, and self-reported happiness. These findings can provide individuals with ASD, families and service providers greater insight into important factors to consider when planning the transition to adulthood.
OARC	Moseley RL, Druce T, Turner-Cobb JM. 'When my autism broke': A qualitative study spotlighting autistic voices on menopause. Autism. 2020 Aug;24(6):1423-1437. doi: 10.1177/1362361319901184. Epub 2020 Jan 31. PMID: 32003226; PMCID: PMC7376624.
	There is little information on how autistic individuals navigate the menopausal transition and whether there are additional challenges faced by autistic versus neurotypical individuals. The authors conducted an online focus group with seven autistic individuals assigned female at birth and aged 49-63 years. Issues raised include lack of professional knowledge, understanding, and communication about menopause for autistic people, absence of support, and a heightening pre-existing and generating new cognitive, social, emotional, and sensory difficulties as a result of menopause. This study demonstrates that additional research is needed on the subject of menopause in autistic individuals.
NIMH	Nicolaidis C, Schnider G, Lee J, Raymaker DM, Kapp SK, Croen LA, Urbanowicz A, Maslak J. Development and psychometric testing of the AASPIRE Adult Autism Healthcare Provider Self-Efficacy Scale. Autism. 2020 Aug 28:1362361320949734. doi: 10.1177/1362361320949734. Epub ahead of print. PMID: 32859135.
	Healthcare systems throughout the US are underprepared to adequately address the needs and provide the high quality of care that adults with autism require to live independent, healthy, and productive lives. In this paper, researchers developed and tested a measure of healthcare providers' confidence (or "self-efficacy") in providing care to autistic adults, with the aim that this instrument would help researchers better understand the training needs of health care providers in order to serve the adult autistic community. Using a community-based participatory research model the investigators developed a brief survey instrument (AASPIRE Adult Autism Healthcare Provider Self-Efficacy Scale) with input from autistic adults, community supporters, and healthcare providers. The instrument was administered to 143 primary care providers from eight primary care clinics in Oregon and California, United States. Overall, results from the showed that healthcare providers acknowledge lack of proper preparedness to serve adults with autism. For example, only 25% a minority of respondents expressed high confidence in communicating with adult autistic patients (25%); performing physical exams or procedures (43%); accurately diagnosing and treating other medical issues (40%) and only 14% in identifying accommodation needs (14%). Future research is needed to

	further validate this scale and to understand how to meet providers' training needs most effectively.
	Question 7: Infrastructure and Surveillance
CDC	Dietz PM, Rose CE, McArthur D, Maenner M. National and State Estimates of Adults with Autism Spectrum Disorder. J Autism Dev Disord. 2020 Dec;50(12):4258-4266. doi: 10.1007/s10803-020-04494-4. PMID: 32390121.
	There is no surveillance system for population-based estimates of the adult autistic population in the US. Using simulation and statistical modeling, the authors estimated the national and state prevalence of autistic adults 18-84 years. The authors estimate that 2.21% of US adults aged 18 or older, or 5,437,988 adults, have ASD. This estimate can help government officials estimate needs for services.
CDC	Maenner MJ, Shaw KA, Baio J; EdS1, Washington A, Patrick M, DiRienzo M, Christensen DL, Wiggins LD, Pettygrove S, Andrews JG, Lopez M, Hudson A, Baroud T, Schwenk Y, White T, Rosenberg CR, Lee LC, Harrington RA, Huston M, Hewitt A; PhD-7, Esler A, Hall-Lande J, Poynter JN, Hallas-Muchow L, Constantino JN, Fitzgerald RT, Zahorodny W, Shenouda J, Daniels JL, Warren Z, Vehorn A, Salinas A, Durkin MS, Dietz PM. Prevalence of Autism Spectrum Disorder Among Children Aged 8 Years - Autism and Developmental Disabilities Monitoring Network, 11 Sites, United States, 2016. MMWR Surveill Summ. 2020 Mar 27;69(4):1-12. doi: 10.15585/mmwr.ss6904a1. Erratum in: MMWR Morb Mortal Wkly Rep. 2020 Apr 24;69(16):503. PMID: 32214087; PMCID: PMC7119644.
	The Autism and Developmental Disabilities Monitoring (ADDM) network monitors prevalence of ASD among children aged 8 years. In 2016, ASD prevalence was 1 in 54 children aged 8 years and 4.3 times as prevalent among boys than among girls. Prevalence was lower in Hispanic children compared to other ethnic groups. Disparities continue to be evident in early evaluation and diagnosis for black children. This data highlights the continuing need for services for individuals on the spectrum and their families.
OARC	McCormick CEB, Kavanaugh BC, Sipsock D, Righi G, Oberman LM, Moreno De Luca D, Gamsiz Uzun ED, Best CR, Jerskey BA, Quinn JG, Jewel SB, Wu PC, McLean RL, Levine TP, Tokadjian H, Perkins KA, Clarke EB, Dunn B, Gerber AH, Tenenbaum EJ, Anders TF; Rhode Island Consortium for Autism Research and Treatment (RI-CART), Sheinkopf SJ, Morrow EM. Autism Heterogeneity in a Densely Sampled U.S. Population: Results From the First 1,000 Participants in the RI-CART Study. Autism Res. 2020 Mar;13(3):474-488. doi: 10.1002/aur.2261. Epub 2020 Jan 20. PMID: 31957984; PMCID: PMC7060113.
	The Rhode Island Consortium for Autism Research and Treatment (RI-CART) has established a major statewide research registry and summarizes findings of its first 1,000 participants. This is the first analysis of a large, population-based U.S. cohort with ASD. The analysis found a high rate of cooccurring medical and psychiatric conditions in affected individuals. Additionally, researchers found that females received a first diagnosis of ASD at a later age than males, potentially due to more advanced language abilities in females with ASD. This study suggests that new strategies for earlier diagnosis of ASD in females may be warranted.
OARC	McPartland JC, Bernier RA, Jeste SS, Dawson G, Nelson CA, Chawarska K, Earl R, Faja S, Johnson SP, Sikich L, Brandt CA, Dziura JD, Rozenblit L, Hellemann G, Levin AR, Murias M, Naples AJ, Platt ML, Sabatos-DeVito M, Shic F, Senturk D, Sugar CA, Webb SJ; Autism Biomarkers Consortium for Clinical Trials. The Autism Biomarkers Consortium for Clinical Trials (ABC-CT): Scientific Context, Study Design, and Progress Toward Biomarker Qualification. Front Integr Neurosci. 2020 Apr 9;14:16. doi: 10.3389/fnint.2020.00016. PMID: 32346363; PMCID: PMC7173348.

	Limited biomarkers exist for ASD. This paper describes the Autism Biomarkers Consortium for Clinical Trials (ABC-CT), a multi-site study designed to investigate electrophysiological (EEG) and eye-tracking (ET) indices as candidate biomarkers for ASD. This study includes large, deeply phenotyped cohorts of children with ASD and typical development, a longitudinal design, a focus on well-evidenced candidate biomarkers harmonized with an independent sample, high levels of clinical, regulatory, technical, and statistical rigor, adoption of a governance structure incorporating diverse expertise in the ASD biomarker discovery and qualification process, prioritization of open science, including creation of a repository containing biomarker, clinical, and genetic data, and the use of economical and scalable technologies that are applicable in developmental populations and those with special needs.
CDC	Shaw KA, Maenner MJ, Baio J; EdS1, Washington A, Christensen DL, Wiggins LD, Pettygrove S, Andrews JG, White T, Rosenberg CR, Constantino JN, Fitzgerald RT, Zahorodny W, Shenouda J, Daniels JL, Salinas A, Durkin MS, Dietz PM. Early Identification of Autism Spectrum Disorder Among Children Aged 4 Years - Early Autism and Developmental Disabilities Monitoring Network, Six Sites, United States, 2016. MMWR Surveill Summ. 2020 Mar 27;69(3):1-11. doi: 10.15585/mmwr.ss6903a1. PMID: 32214075; PMCID: PMC7119643.
	The Early Autism and Developmental Disabilities Monitoring (Early ADDM) Network is a subset of the larger ADDM Network and estimates ASD prevalence and monitors early identification of ASD among children aged 4 years. Analysis of 2016 Early ADDM data indicate ASD prevalence is around 1 in 64 children aged 4 years, with higher prevalence among boys than girls, and disparity in ASD prevalence between white and black children have decreased. While more children with ASD are receiving earlier diagnosis, the data in this report can continue to be used to improve early identification so services may be accessed as soon as necessary.