

# 2016-2017 IACC Strategic Plan for Autism Spectrum Disorder

**Chapter Title: How can I recognize the signs of ASD, and why is early detection so important?**

**Aspirational Goal: Provide the earliest possible diagnosis for people on the autism spectrum, so they can be linked to appropriate interventions, services, and supports in as timely a manner as possible to maximize positive outcomes.**

## **Introduction**

Observational studies of infants at risk for ASD reveal that although the emergence of ASD features is variable, subtle signs can be detected within the first few years of life. Experienced clinicians, trained to use validated diagnostic tools, are able to diagnose ASD by 18-24 months of age. Still, the median age of diagnosis in the U.S. is 50 months, with disparities related to SES, geographic region, race/ethnicity, and rural versus urban location. Because we know that early behavioral intervention, delivered during the toddler and preschool years, is effective in increasing social orienting, language, and IQ, this lag in diagnosis means that intervention is delayed by months or years for many children. This chapter reviews the state of our knowledge about screening and diagnostic tools as well as the current state of service delivery and challenges families face when trying to access those services.

## **Topic 1: Implementation of ASD Diagnostic and Screening Tools**

### **Service Needs**

The human brain undergoes unprecedented changes during the first three years of life such as a doubling of synaptic density in prefrontal cortex<sup>1</sup>, underscoring the service needs during this critical period of neuroplasticity. It is not surprising that behavioral interventions initiated with ASD toddlers within this time period result in a range of positive changes including increases in social orienting, language ability, and overall IQ<sup>2-4</sup>. Yet, this success is tempered by the fact that the mean age of diagnosis in the US still hovers around 4 years<sup>5</sup>, significantly beyond this transformative period. Screening using validated autism-specific parent-report tools only occurs systematically within about 50% of primary care settings<sup>6</sup>, yet studies consistently report that their use can result in ASD detection as young as 12-18 months<sup>7</sup>. Reliance on using a standardized screening tool has even been shown to be more effective than pediatrician clinical judgment alone<sup>8</sup>. It is for this reason that despite the US Preventative Services Task Force (USPSTF) lack of endorsement of universal ASD screening<sup>9</sup> using standardized tools is still embraced as the gold standard for ASD detection by the American Academy of Pediatrics<sup>10</sup> and is a critical service need to improve early access to care.

### **What is Known**

Given that many parents take their child for well-baby visits within a primary care setting, recent research on screening has utilized this context. To accommodate the dynamic and busy environment of a primary care setting, parent-report screening tools are, by design, very brief and can often be completed in just 5 minutes. The relatively shallow nature of clinical information that can be gleaned within this context may be a contributor to the high false positive rates often associated with parent-report screening tools. Importantly, a new revision to the M-CHAT-R, the most commonly used screening tool, shows that administration of follow up questions of failed items can result in ASD specificity of approximately 50%, and if all developmental delays are also considered a true positive, that estimate increases to >95%<sup>11</sup>. Despite the important achievement of reducing false positives (if all delays are considered a true positive), administering follow-up questions in the M-CHAT R/F procedure can take anywhere from 5 to 30 minutes and as such does not overcome one of the greatest barriers to ASD screening: lack of time<sup>12</sup>. Leveraging

technology, Campbell and colleagues (2017)<sup>13</sup> showed that a full administration of the M-CHAT-R/F on a tablet not only resulted in greater and more accurate documentation of the screening results within electronic medical record systems, but also eliminated the time barrier because parents answered the follow up questions directly on the tablet, thus bypassing the need to engage medical personnel<sup>13</sup>.

Large-scale studies examining the M-CHAT in primary care have implications for understanding how many children may be missed based on the expected prevalence estimate of 1 in 68 children (14.6 per 1,000) at 8 years of age and 1 in 76 children (13.4 per 1,000) at 4 years of age<sup>5, 50</sup>. In studies using the M-CHAT with the follow up interview, rates were reported to be 105 in 16,115 children at a mean age of 20.9 months (6.5 per 1,000)<sup>11</sup> and 40 in 5,007 children at a mean age of 30 months (8.0 per 1,000)<sup>51</sup>. Using the M-CHAT without the follow up interview at a mean age at approximately 18 months results in detection of 60 in 52,026 children (1.2 per 1,000)<sup>14</sup>. Detection rates highlight the fact that the M-CHAT is missing far more children with ASD than it is detecting at younger ages, particularly at 18 months, which may be related to many factors including: the accuracy of the screening tool, ability of parents to notice and report early signs of autism, readiness of parents to act on a positive autism screen, and the heterogeneity in symptom presentation at this young age, suggesting that screening efforts may need to go beyond simple parent-report tools. One such approach is a 2-stage screening model that combines a general developmental screening tool based on parent report, the Infant Toddler Checklist (ITC)<sup>15</sup>, with subsequent observational ratings to screen for ASD. This observational screen was evaluated when completed by trained undergraduate students with no clinical experience, in contrast to other brief observational ratings by expert raters<sup>50</sup>. Using this approach, detection rates have been reported as 82 in 5,419 children at a mean age of 20.8 months (15.1 per 1,000)<sup>16</sup> which is very close to the expected prevalence rates for ASD<sup>5</sup>. Shedding light on which children may be missed, children with a true positive on the M-CHAT R/F display a lower developmental level than children ascertained with the ITC and follow-up observational rating<sup>16, 17</sup>, and lower developmental level than those evaluated in a prospective sample of younger siblings at familial risk for ASD<sup>18</sup>. Continued improvement in screening approaches may be achieved by better understanding the psychometric features of parent-report screening tools in relation to observational measures<sup>17, 19</sup>, and by examining the utility of different screening thresholds in relation to diagnostic accuracy and cost effectiveness<sup>20</sup>.

Additional new innovations in parent report screening approaches include the incorporation of photographs into the questionnaire to illustrate items in a culturally unbiased manner<sup>21</sup> as well as the examination of combining multiple screening tools to improve sensitivity and specificity<sup>22</sup>. Parents can also find free apps on the internet, such as “ASDetect” that augments descriptions of ASD characteristics with video examples and provides a video-led assessment of child behaviors. This potentially useful approach currently lacks research studies examining usability and accuracy of the App itself although there is empirical support demonstrating that the markers highlighted within the App (e.g., pointing and showing) are predictive of an ASD diagnosis<sup>23, 24</sup>.

A growing appreciation of ASD as a condition marked by unique behavioral, neural and genetic signatures that may precede overt clinical symptoms has resulted in a surge of prodromal and biomarker-seeking research which broadens the scope of future screening efforts. Of particular interest are potential biomarkers likely to facilitate gene-brain-behavior studies, diagnosis, or those that may act as prognostic markers. Observational studies continue to reveal that signs of ASD are subtle, but may emerge within the first year of life, particularly in the areas of social communication, attention, and motor development<sup>18, 25, 26</sup>. Studies deploying eye-tracking technology to measure social visual engagement have demonstrated preliminary utility and accuracy in detecting markers of ASD in the first year of life<sup>27, 28</sup>, with some assays shown to be under stringent genetic control<sup>29</sup>, thus offering a potential bridge for gene-behavior studies. Electrophysiologic research continues to reveal group markers of brain atypicality in infancy, including signs of reduced engagement with, and atypical hemispheric specialization for, social stimuli, and generalized brain hyper-connectivity<sup>30-32</sup>. And for the first time, structural and functional magnetic resonance imaging studies of infants are beginning to predict later ASD diagnosis and core characteristics such as language outcome<sup>33-35</sup> and RNA expression profiles can accurately classify toddlers as ASD at levels exceeding 80%<sup>36</sup>. While these findings suggest a future of exciting new tools for screening and diagnosis, they must be

validated in other high-risk groups and in the general population, and they must be adjusted for broader use in order to be beneficial to the wider community.

Recent advances in early screening and diagnosis make it feasible for experienced clinicians trained on validated tools to diagnose ASD by 18-24 months of age<sup>16, 37</sup>. Autism symptoms in toddlers, as measured by the ADOS-T, were found to be separable and best deconstructed into the new 2-factor DSM-5 structure<sup>38</sup>. This study found that deficits in social interaction and communication are best conceptualized along 1 dimension, whereas restricted, repetitive behaviors and unusual language features are also best conceptualized on a distinct second dimension. These findings support the reorganization of symptoms in the DSM-5 for toddlers and suggest that the structure of autism symptoms is similar to older children. The stability of an ASD diagnosis at 18-24 months is high for samples ascertained from community-based screening 1-2 years later<sup>16 39</sup> and from familial-risk samples<sup>15</sup>. A small proportion of toddlers (4%-10%) were not identified with ASD at 18-24 months who were diagnosed later in community ascertained samples<sup>16, 39</sup> compared to almost half of siblings at familial risk (63% at 18 months; 41% at 24 months)<sup>18</sup>. Distinct combinations of eye contact, gestures, play, and repetitive behaviors at 18 months were predictive of ASD outcome at 36 months, suggesting different developmental pathways to ASD in familial-risk siblings<sup>37</sup>. This underscores the importance of developmental surveillance and follow-up for children with early social communication delays. The heterogeneity in developmental unfolding of ASD features over the first two years of life makes diagnosis of ASD by 18 to 24 months challenging in community-based settings and a critical need for future research.

### **Barriers, Gaps, and Opportunities**

Despite the success of screening using validated tools to identify ASD at very young ages<sup>7</sup>, as well as procedural advances made in the past few years<sup>11, 13</sup>, barriers still exist that prevent widespread uptake within primary care settings including: a lack of education and understanding of ASD<sup>40, 41</sup>, lack of familiarity with screeners<sup>40, 42</sup>, uncertainty where to send a toddler with a test-positive screen<sup>12</sup>, lack of effective and timely means of connecting families of individuals with ASD to available resources<sup>41, 43</sup>, and as mentioned above, extra time and resources required to utilize standardized screening tools<sup>12, 40</sup>.

Although engagement in early treatment has been associated with a range of positive changes including increases in social orienting, language ability, and overall IQ<sup>2-4</sup>, no study has directly examined if children with ASD detected by early screening have better outcomes than those detected by other means, (e.g., parent or provider concern) an issue highlighted by the recent US Preventative Services Task Force (USPSTF) report<sup>9</sup>. However, as noted by Dawson (2016)<sup>44</sup>, such a study would require large representative samples from across the country to be randomly assigned to either a screening or non-screening condition, and then followed to determine long term outcomes and societal costs. Given that early treatment for children under age 3 years has been shown to result in positive gains<sup>45</sup>, and has even been associated with an increased potential to lose an ASD diagnosis altogether<sup>46</sup>, such a study is not without controversy. The USPSTF report raised additional research gaps to consider including a lack of studies that examine: (1) factors that might modify the performance characteristics of ASD screening tests such as age at screening or family characteristics; (2) intermediate and long term health outcomes of children identified as ASD through screening; (3) the impact of screening within low socioeconomic status and minority populations that often have more limited access to care than their wealthier counterparts and; (4) outcomes from large samples of screen negative children which would be critical to ascertain more rigorous estimates of screening sensitivity and negative predictive value.

While a considerable timescale would be required to conduct new RCT studies to specifically address USPSTF concerns, there are opportunities and study designs that could be leveraged using existing resources in the short term. First, data could be examined from within sample cohorts that include clinical longitudinal data from toddlers detected via screening *as well as* toddlers detected via other means (e.g., parent or clinician concern). Using an age-matched observational study design, the clinical characteristics and proximal and distal outcomes could be examined and compared between groups, while controlling for as many variables as possible (e.g., gender, SES). Second, exclusively within cohorts of screen detected toddlers, researchers

could examine outcomes of children detected at identical early ages via screening (e.g., at 18 months) but that contained a subgroup of toddlers who started treatment well beyond the screen-detected age (e.g., due to parental denial or state agency delays in funding treatment). Within such a study, both groups of toddlers (i.e., screen-detected/early treatment start versus screen-detected/late treatment start) should come from identical geographic regions, identical clinical settings (most often pediatric offices) and failed the same screening tool, but the time until treatment engagement would be different between groups. In this way, the impact of very early treatment engagement as afforded by screening could be more directly examined.

In terms of new, future studies, in instances where a traditional RCT design (intervention vs no intervention) may not be feasible and the health impact is high, a stepped wedge cluster randomized trial design could be used wherein the rollout of particular screening or intervention feature is introduced across multiple timepoints<sup>48</sup>, and pre and post intervention periods compared. Another innovative methodology is a sequential, multiple assignment, randomized trial (SMART), which can be used to build an adaptive intervention to test the effectiveness of a series of screening conditions or interventions to support families acting sooner by randomizing participants multiple times and develop the best decision rules for a screening protocol. It might also be feasible to utilize administrative data. A number of states collect state-level data on youth who receive ASD screening and subsequent developmental outcomes. This may afford an opportunity to compare children with and without early screening in terms of variations in developmental outcomes (using the Battelle Developmental Inventory which is required for Federal reporting.)

While universal screening and early detection through other mechanisms such as parent or clinician concern is an essential step in the health care process for ASD and deserves more research attention, it merely creates the opportunity for early treatment and services. Screening in and of itself does not determine if and when parents actually follow through with subsequent diagnostic evaluation and treatment engagement, nor does it determine the quality and benefits of such treatment. A key gap in the field, then, is a paucity of studies examining the many important factors that follow *after* screening has occurred<sup>47</sup>. There is indeed a growing appreciation of the importance of implementation science methods to examine contextual factors (e.g., mode of screening delivery) that may impact successful screening uptake. Some studies are currently underway through a NIMH Prevention, Early Detection, Engagement and Services (PEDS) Network. Another important factor is comprehensive tracking of treatment participation, which is essential to determine the long-term outcomes of children detected early by screening. Interventions for ASD toddlers vary in several important dimensions including the underlying theoretical framework (e.g., strict ABA model or one that includes developmental perspective) mode of delivery (e.g., parent mediated, therapist mediated or both), intervention location (e.g., home or preschool) and the presence or absence of additional services (e.g., occupational therapy). To date, most studies do not report treatment engagement, and if it is reported, it is often at a very coarse level (e.g., number of hours). Now that insurance companies in several states are mandated to pay for autism services, this trend may change. Finally, the ultimate goal of research efforts in early detection and intervention is improvement in clinical practice in community settings at large. More effort will be required to develop reliable and reproducible methodologies that could yield, on a yearly basis, local, regional, state and national “scorecard” results, focused on parameters such as rates of universal screening and access to diagnostic and early intervention services. Only such community-wide parameters could reassure stakeholders in these goals that progress is truly being made.

### **Service and Policy Implications**

Given the overlapping clinical phenotype of ASD with other delays during very early development, screening studies highlight the possibility that the goal of early screening and resulting policy statements could be re-conceptualized as one that emphasizes sensitivity (i.e. detecting as many ASD children as truly exist), while the expectation of high specificity for ASD could come at a subsequent in-depth evaluation.

Screening, while important, is just one step on the path to identification and eventual treatment. In order for screening to be effective, ample evaluation centers must be available with appropriate ASD diagnostic

expertise. Indeed, uncertainty regarding where to send a toddler for an evaluation is a barrier to screening noted by over 75% of pediatricians<sup>12</sup>. Therefore, one major policy change would be recommendations for the establishment of evaluation centers throughout the country based on population and expected rates of ASD. Likewise, the process of screening and evaluation only has meaning if high-quality treatment providers are available and affordable once test-positive cases are identified. Investigation of more cost-effective modes of treatment delivery, such as those that are either partially or fully deployed by parents, are being examined<sup>2</sup>.

An increase in the number of toddlers screened and identified as possible ASD (Nygren et al. 2012) also calls for the need to standardize policy regarding eligibility for “Part C” services, the federal program that funds intervention for children showing delays, including autism, between the ages of birth through two years. Generally, toddlers must first qualify for basic Part C services by exhibiting a particular state-mandated level of delay (usually a 25% delay in two or more areas) which often results in just a few hours of speech or occupational therapy. Although autism is an automatic eligibility category, a child must be identified as either ASD or showing signs of ASD in a separate evaluation visit in order to be eligible to receive ASD-specific treatment. Currently, there are no guidelines or regulations mandating that all toddlers receiving Part C services should be examined for possible ASD. Even once a child is suspected as possible ASD and referred for an in-depth evaluation, there are no policies regarding specific diagnostic and other evaluation tools that should be used to determine if a child is eligible for ASD specific services. It is thus not surprising that many toddlers already identified as showing a delay through Part C, have not been properly evaluated for ASD. Even more concerning, the vast majority of toddlers (at least 75%<sup>49</sup>) who will go on to qualify for special education at school-age, are still not identified in time to receive early intervention. Improved policy that provides clear guidelines regarding ASD detection and subsequent treatment eligibility through Part C are needed.

## **Topic 2: Disparities in ASD Screening and Diagnosis**

### **What is known?**

#### **Disparities in ASD Screening**

Evidence demonstrates that screening is a successful strategy to detect ASD in toddlers (e.g., Guevara et al., 2013; Robins et al., 2014), yet there are a number of barriers that limit screening during well-child check-ups. This has immediate implications for access to services for children from diverse backgrounds. Overall, ASD screening rates during primary care visits range from 1-60% (Arunyanart et al., 2012; dosReis et al., 2006; Gillis et al., 2009); some of the variability in use of standardized screening is based on children’s sociodemographic characteristics. For example, screening may occur less frequently among Spanish-speaking families compared to English-speaking families (Zuckerman et al., 2013). Families with low levels of maternal education exhibit higher screen positive rates on the Modified Checklist for Autism in Toddlers (M-CHAT(-R); Robins et al., 1999, 2009), but are more likely to be lost to follow-up, suggesting that these families are at risk for being underserved (Khowaja et al., 2015). Rural vs. urban locale and race and ethnicity also impact reliability and validity (Scarpa et al., 2013) as well as screen positive rates (Khowaja et al., 2015; Windham et al., 2014). Consistent use of screening tools also may depend on reimbursement – children from low-income families may be more likely to be screened during check-ups since it is often reimbursed by Medicaid (Bethell et al., 2011), but may not be covered by private insurance.

Children from minority backgrounds are diagnosed more than a year later than their White peers (see Daniels & Mandell, 2014). However, it has been demonstrated that when physicians follow a standardized screening protocol, including immediate referral for screen positive cases, disparities in age of diagnosis is reduced to approximately one month (Herlihy et al., 2014). Therefore, access to screening for all children, regardless of sociodemographic characteristics, language spoken at home, and geographic locale, is crucial to reduce existing disparities that carry forward from screening to early diagnosis to early intervention, cascading to impact life-long outcomes.

### Validity of Screening Instruments in Diverse Groups

A number of studies have examined ASD screening tools in different languages across the world; an exhaustive review of international studies is beyond the scope of this document, but see Garcia-Primo et al. (2014) for a review of screening in Europe, Soto et al. (2014) for discussion about cultural adaptation of screening tools, and [www.mchatscreen.com](http://www.mchatscreen.com) for studies using the M-CHAT(-R), which has been translated into more than 50 languages. In the US, psychometric properties of validated screening tools have been examined in diverse samples, and factors including low educational attainment, language/literacy, rural vs. urban, race, and ethnicity impact reliability and validity (Scarpa et al., 2013) as well as screen positive rates (Khowaja et al., 2015; Windham et al., 2014). The variability of results from these papers indicate that there is a need for additional research to adapt tools that will be valid (i.e., demonstrate adequate sensitivity and specificity) in diverse populations.

### Disparities in Access to Diagnostic Services and Age of Diagnosis

Differences both in prevalence rates and age of diagnosis by sociodemographic characteristics likely relate to disparities in access to expert services. According to the most recent Autism and Developmental Disorders Monitoring (ADDM) Network study (Centers for Disease Control and Prevention [CDC], 2016), White children were 20% more likely to have indicators of ASD in their school and health records than Black, 40% more likely than Asian and Pacific Islander, and 50% more likely than Latino children. A variety of factors, including economic challenges (e.g., invalid phone numbers; Khowaja, Hazzard, & Robins, 2015), geographic distance between families and service providers (Kiani et al., 2013), reduced professional resources and capacity (Janvier et al., 2015), and characteristics impacted by cultural knowledge, such as stigma (Bates et al., 2014) often contribute to diminished service availability and utilization in rural disadvantaged communities.

The literature is equivocal about racial disparities in age of diagnosis – some studies find that White children are diagnosed younger than non-White children, but others do not (e.g., see Daniels & Mandell, 2014). Alternately, Latinos compared to Whites have consistently been found to be diagnosed later (Fountain et al., 2011; Magaña, Lopez, Aguinaga, & Morton, 2013; Shattuck et al., 2009; Valicenti-McDermott et al., 2012). Children from more affluent families are more likely to be diagnosed with ASD than those from low-income households (Daniels & Mandell, 2014; Thomas et al., 2012) and children of college educated mothers are more likely to receive an ASD diagnosis compared to mothers with low education (Dickerson et al., 2016). Geographic location may also show differences; parent-reported ASD lifetime prevalence rates from the 2011/12 National Survey of Children’s Health were 2.3% for urban versus 1.8% for rural areas of residence (NSCH, 2011/12). Some studies noted as much as an 8-month lag in age of diagnosis for rural compared to urban samples (Mandell, Novak, & Zubritsky, 2005; Rhoades, Scarpa, & Salley, 2007; Rosenberg et al., 2011), but others found no differences (Twyman et al., 2009). An analysis of ADDM data and residential tracts found that ASD diagnosis decreased in tracts that had a higher proportion of Latino residents (Dickerson et al., 2016) which is similar to results found in a study that examined ASD diagnosis in Texas schools (Palmer et al., 2005).

### Validity of Diagnostic Instruments across Cultures

There is general agreement that the best approach to ASD diagnosis includes both parent interview and an observational assessment of the child (Huerta & Lord, 2012), such as the Autism Diagnostic Interview-Revised (ADI-R) and the Autism Diagnostic Observation Schedule (ADOS(-2)). The ADI-R has been translated into 17 languages (Western Psychological Services, 2017) and a small number of studies have examined the validity of the ADI-R in different countries such as Greece (Papanikolaou et al., 2009), Japan (Tsuchiya et al., 2013), Finland (Lampi et al., 2010), and Brazil (Becker et al., 2012) with varying results. With respect to validation studies with diverse populations in the US, researchers found that the sensitivity and specificity

of the ADI-R with a US-based Spanish speaking population of parents of children with ASD were lower (Vanegas, Magaña, Morales, & McNamara, 2016) than values previously reported for mostly White, middle-class respondents (Lord, Rutter, & Le Couteur, 1994). The communication domains were found to be especially problematic for parents whose primary language was Spanish when reporting on children who spoke mainly English (Vanegas et al., 2016). Little is known about the validity of the ADI-R among low-income families in the US. The ADOS-2 has been translated into 19 different languages (Western Psychological Services, 2017), however cross-cultural validation studies of the ADOS(-2) have not been identified.

### **Barriers, Gaps, and Opportunities**

Evidence of ASD screening and diagnosis, including those data that speak to disparities in access to these services, often relies on provider surveys. Studies examining medical or state records for specific mention of ASD screening and diagnosis would be helpful in documenting disparities and also in tracking improvements based on policy changes or improved access to care. One of the specific gaps identified in the recent report from the USPSTF (Siu et al., 2016) calls for health outcomes research for children detected through screening, with particular emphasis on including participants from minority and low-income families. Therefore, it will be critical to evaluate the quality of screening instruments and programs in diverse samples of children with long-term outcomes. Implementation studies examining the translation from research settings to community settings with diverse populations, including examining fidelity of adhering to screening protocols, also is a critical gap in the existing literature. For example, many research and clinical settings that report ASD screening using the M-CHAT, report that the second stage of Follow-Up questions are not employed for children who initially score in the moderate risk range (e.g., Charman et al., 2015; Yama et al., 2012), or that providers use clinical judgment in interpreting results and making selective referrals, rather than relying on the validated scoring algorithms (e.g., Pierce et al., 2011; Windham et al., 2014).

In addition, family level variables such as insufficient financial resources, lack of insurance coverage, language barriers, geographic isolation, and limited knowledge and experience with our complex healthcare systems, may be barriers to the timely diagnostic evaluation of an at-risk child (Zuckerman et al., 2014). Overall, there is limited research that documents these systemic and individual level barriers to get from early ASD or developmental screening to appropriate diagnosis to early intervention (Kavanagh et al., 2012), and gaps remain in our understanding of how to promote effective implementation of evidence-based screening and assessment in resource-poor, underserved settings and populations.

Finally, and perhaps most importantly, there is a need for prospective studies that demonstrate that equal access to high quality screening, with immediate referral for positive screen cases to diagnostic evaluation and early intervention services, will reduce disparities in prevalence, as well as any disparities in long-term outcomes for children with ASD.

### **Service and Policy Implications**

A primary barrier to ASD screening and early diagnosis is the limited availability of diagnostic clinics with providers trained in ASD diagnosis leading to long waiting lists and poor reimbursement for comprehensive diagnosis (Shattuck & Grosse, 2007). In low resource and rural areas, the availability of clinics is even more limited than in more populated regions. Many children are not diagnosed until entry into the school system, yet schools often lack trained professionals who can make informed diagnoses.

One clear policy implication includes dedicating resources to early screening in underserved communities, including rural, low SES, and minority communities, with a corresponding increase to fund adequate evidence-based diagnostic evaluations in order to avoid lengthening waitlists (Shattuck & Grosse, 2007). Other policy implications include increasing psychoeducation to raise awareness and reduce stigma, building external professional networks, promoting continuing education programs for healthcare professionals, such as the Autism Case Training Curriculum developed by CDC (<https://www.cdc.gov/ncbddd/actearly/autism/curriculum/class.html>), using alternative service delivery

models (e.g., telehealth, web-based, community health workers) or settings (e.g., schools, child care centers, mobile clinics) for screening/diagnosis, and providing wraparound services that address additional stresses (e.g., chronic illness, unemployment, lack of insurance) faced by individuals in these communities. Finally, it is clear that children are not often well-tracked from time of ASD screening to receipt of services (Daniels et al., 2014). To fully inform policy, it is imperative to have a system in place that can assure children and families adequate, timely, and appropriate services as they move through the identification, referral, and treatment process.

### **Topic 3: Workforce**

#### **Service Needs**

The increased prevalence of ASD over the past two decades has led to a need for a larger workforce trained in the identification and diagnosis of these disorders. Professionals on the diagnostic front line include psychologists, psychiatrists, developmental pediatricians, neurologists and speech and language pathologists. Early detection of ASD requires training those professionals who come in regular contact with young children, including primary care providers and child care providers, in order that they incorporate effective screening and referrals in their daily practice patterns. In response to this need, CDC developed a web-based education program, the [Autism Case Training](https://www.cdc.gov/ncbddd/actearly/autism/curriculum/class.html), to inform healthcare providers on fundamental components of identifying, diagnosing, and managing ASD through real life scenarios (<https://www.cdc.gov/ncbddd/actearly/autism/curriculum/class.html>). Promoting, refining, and delivering similar education programs is a critical factor in building a workforce that can effectively serve individuals with ASD and their families.

Finally, there is a need to raise public awareness and encourage parents to observe and track their child's development in order recognize early signs of ASD, and encourage them to discuss their concerns with their child's doctor, teachers, and other involved adults. The "Learn the Signs. Act Early" campaign developed by CDC, and the "16 Gestures by 16 Months" series developed by the First Words Project are examples of awareness strategies that can be utilized to raise awareness and facilitate parent-provider collaborations.

#### **What is known?**

Families: Families experience lengthy delays between initial referrals and diagnosis of ASD (CDC 2016 ADDM paper, Guinchat et al. 2012, Chawarska et al. 2007). The average delay was 13 months in a study of surveillance records (Wiggins, Baio, Rice, 2006). Parents may not recognize signs of developmental delay, or may have concerns about their child's development but do not know how or when to act on those concerns. There is a critical research gap on understanding how parent concerns can impact parent engagement in acting on referral for diagnosis and early intervention.

Primary care providers and expert clinical evaluations: Evidence demonstrates that healthcare professionals are less likely to detect ASD using developmental surveillance without the use of screening tools. Even experienced professionals may miss or misjudge symptoms during a brief observation (Gabrielson et al., 2015). However, primary care providers face barriers to implementing screening that include the time necessary to identify ASD, the cost of conducting screening and the reimbursement for this work, and having appropriately trained personnel in their offices or referral networks. These factors have been reported by primary care providers as the major obstacles preventing them from implementing more consistent screening.

## **Barriers, Gaps, and Opportunities**

There continue to be gaps in our understanding of how healthcare professionals can best reach families from underserved communities, especially in populations that have had market growth in the US such as the Somali and Hispanic/ Latino populations. There is an opportunity to improve identification of ASD through materials prepared in proper languages, but more importantly, to better understand how to make connections with diverse populations in culturally competent formats and develop a workforce with greater cultural diversity. For example, outreach activities held in places of worship and other community gatherings where families feel safe and have trust in their care team may improve parent-provider partnerships and lead to increased identification of ASD.

Lack of compliance with ASD screening recommendations in primary care clinics can be partially addressed through continued development of accurate screening instruments that are easily and efficiently implemented in busy primary care settings. [“Birth to Five: Watch Me Thrive!”](#) is a coordinated federal effort to raise awareness about the importance of universal early behavioral and developmental screening. “Birth to Five: Watch Me Thrive!” offers a “Compendium of Screening Measures for Young Children”, which is a collection of research-based screening tools for children under the age of 5. Practitioners in early care and education, primary health care, child welfare, and mental health can use this reference to learn about the cost, administration time, quality level, training required, and age range covered for each screening tool.

Another strategy for increasing compliance with ASD screening recommendations in primary care clinics is with improved reimbursement for the administration of these instruments. Opportunities exist to leverage other forces to encourage change in these practice habits, such as maintenance of certification requirements. Using innovative technology and professional development to support collaboration between the medical home, the IDEA Part C early intervention system, and other members of the family’s care team may offer a mechanism to improve developmental surveillance and monitoring of services (Adams & Tapia, 2013).

Early care and education practitioners may lack the technical training to review and compare complex psychometric information on the quality of developmental screening tools. Training for this workforce is needed to improve their ability to screen effectively, recognize ASD symptoms, communicate clearly with parents, and refer appropriately for evaluation and intervention services.

## **Services and Policy Implications**

The ‘workforce’ necessary for assisting all children to have healthy, meaningful lives is a workforce encompassing families, persons with autism, paraprofessionals, and health providers. Some important service initiatives are ongoing, but there is a need for additional efforts. The CDC’s Learn the Signs. Act Early initiative supports early screening. Multiple federal agencies were engaged in Birth to Five: Watch Me Thrive to raise awareness about the importance of early behavioral and developmental screening. The American Academy of Pediatrics supports universal screening for ASD and provides training to pediatric providers through several formats (publications, webinars, and face-to-face conferences) and the University Centers of Excellence in Developmental Disabilities also provide training to over a dozen health care disciplines. In spite of the recommended guidelines for utilizing these resources, we are not reaching most of the families and children in need of early intervention. Therefore, service-relevant policies should be considered to make professional development and training more available and to dedicate more resources in order to expand the workforce to address unmet service needs of early screening, diagnosis, and access to care. Policies should facilitate the collaboration of community-based programs and social supports with professional services.

## **Topic 4: Service System**

### **Service Needs**

It is critically important that children with ASD are identified early so they can be referred to intervention programs that address their individual needs. Consequently, all children benefit from routine screening at multiple intervals, a medical home, coordinated care, and affordable health insurance that covers ASD screening and a range of interventions and treatments. Eligibility criteria and the lead agency for early intervention, Insurance coverage, and the coordination of healthcare vary by state: some states or regions have more comprehensive and coordinated systems of healthcare than others; the lead agency for early intervention is health in some states and child welfare or education in other states. Even in better-resourced areas, families are often faced with many complex steps from screening to diagnosis to treatment. Given that the vast majority of toddlers (at least 75%<sup>49</sup>) who will go on to qualify for special education at school-age, are still not identified in time to receive early intervention (0-3), there is a continued pressing service need to improve access to early intervention for this age group, through IDEA Part C. This means that most infants and toddlers with developmental delays, including those who will receive a diagnosis of ASD, miss the opportunity to receive early intervention services. This service need is even more pressing for children from minority backgrounds.

#### **What is known?**

Nearly half of children with ASD have private insurance; the other half have insurance provided by Medicaid or the state-based Children's Health Insurance Program (CHIP), or dual private and public coverage (National Survey of Children with Special Health Care Needs). However, about half of families of children with ASD report that their insurance coverage is inadequate to meet their myriad of complex needs and costs. As noted earlier, reimbursement for ASD screening may improve screening results and more readily become a standard procedure in practices. A systemic problem is that some insurance plans do not cover state-of-the-art treatments, such as Applied Behavior Analysis, or may place limits on essential behavioral, medical, or other healthcare. Additionally, as early as possible, family social service supports, which are inexpensive and contribute greatly to families meeting the needs of the child, are not covered. These limitations often leave families struggling in many ways, including to cover the cost of care on their own, which results in significant financial burden and other problems. In fact, nearly half of families of children with ASD say their child's health condition has caused major problems for the family and in some cases bankruptcy and other family disruptions, such as divorce or job loss.

There is a dearth of information available on the complexity of accessing ASD-related services. The [Autism Collaboration, Accountability, Research, Education and Support \(CARES\) Act of 2014](#) is aligned to this purpose. Additionally, the IDEA Annual Report to Congress<sup>49</sup> indicates a continued broader challenge of the under-identification of infants and toddlers with developmental delays including ASD who should be eligible for early intervention through IDEA Part C.

#### **Gaps, barriers and opportunities**

There is a pressing need to improve access to early screening and to increase the accuracy of screening tools because these are the gateway to early intervention services. Coordination of a care team that includes healthcare and childcare providers is critical to address gaps in screening, and begin to break down barriers for families to act on screening results and to support family engagement in intervention services.

There is a continued need for ASD insurance reform. Families of children with ASD who have a medical home report fewer unmet needs and more shared decision making with healthcare providers

(Golnik et al., 2012). The Affordable Care Act of 2010, Section 2703, created an optional Medicaid State Plan benefit for states to establish Health Homes to coordinate care for people with Medicaid who have chronic conditions. Chronic conditions listed in the statute include mental health, substance abuse, asthma, diabetes, heart disease and being overweight. Addition of other chronic conditions, including ASD, is subject to state application, then review and approval. State ASD insurance mandates increase ASD diagnosis and treatment rates by 13%, after controlling for other variables (Mandell et al). This effect increases the longer the insurance mandates are in place. However, the number of children receiving ASD services is still less than would be expected given current prevalence estimates, though this does not control for public versus private service utilization.

There is also a need for systematic analyses of the complexities of accessing the service systems. Given the lack of research in this area, examples of the multifaceted challenges families face to access services may best illustrate these complexities:

*One example is a family that does not suspect anything unusual about their child's development and visits their pediatrician at 24 months of age. This family has private health insurance. A screening is conducted and the child scores positive for possible ASD. The pediatrician is wary of making the diagnosis and in this case, refers the child to a developmental-behavioral pediatrician who is the specialist at the local university hospital. The family pays out of pocket to see the specialist after waiting several months for an appointment. The family must pay the bill and manage their own direct claim processing, often resulting in minimal payment of the bill.*

*The developmental-behavioral pediatrician confirms an ASD diagnosis and the office nurse gives the parents a phone number for the state-mandated city-run Early Intervention Program; again, the family is left alone to coordinate. When the mother calls the phone number, she is assigned an early intervention coordinator who arranges for an in-home assessment to be conducted within the mandated time period (usually less than a month). By now, the child is almost 30 months old; early intervention services only last until he is 36 months old or shortly thereafter. The family and the Early Intervention team then need to refer the child to the local school system, which may be a different "sector" or agency, that will re-assess him for preschool services beginning at 3 years of age, and determine where and how he will receive preschool services.*

*An alternative example is a child whose parents are concerned about their 30-month-old daughter's lack of speech, and do not have a primary care provider or medical home. The family asks about this milestone during a visit to an emergency department when the child has a high fever. The family has Medicaid through the ACA Medicaid expansion. The doctors and staff at the emergency room are not familiar with Early Intervention, but suggest the family see a pediatrician regarding their daughter's speech delay. Several months later, they see a pediatrician, who is concerned about ASD, calls Early Intervention herself and attempts to schedule an assessment. The family is bilingual, so a bilingual evaluator with ASD experience must be located, and this takes several months. The family is unable to arrange transportation and would miss further days of work, therefore the diagnostic assessment does not occur. By this time, the child is nearly 3 years old. The Early Intervention team suggests that the family be referred to the Committee for Preschool Special Education (CPSE) through her local school district. The family is uncomfortable with the idea of special education for such a young child and does not want to participate in anything that labels their child as "different" in the school system, so declines services.*

*A final example is a parent of an older child with autism, who is concerned very early about her next son's delayed development. The mother is well-informed about appropriate services. Her*

*pediatrician is not concerned and tries to reassure her, but the mother calls the Early Intervention hotline directly. A home-based assessment is arranged for the 18-month-old boy. The home-based evaluators from Early Intervention do not attempt to make a diagnosis but recommend minimal treatment - an hour a week of home-based general instruction and an hour a week of physical therapy - for what is classified as a general developmental delay. Early Intervention sends the bills for these services to the family's private insurer. When the family decides, several months later, that they would like more in-depth information about their child's development, they seek out an evaluation at a local hospital using their insurance. The family is told that they have already met the maximum coverage allowed with the services billed by Early Intervention. Their pediatrician must negotiate with forms and a telephone appeal to the insurance company to get approval for a skilled evaluation of the child. When this occurs, the child receives a formal diagnostic evaluation for ASD by a psychologist and child psychiatrist. The team makes a diagnosis, and this is reported back to Early Intervention, at which point more hours and different therapies are negotiated. The child is now 24 months old. In this case, because of the knowledge of the parents and the responsiveness of Early Intervention (but not the medical system), the child began initial treatment early on, but more intense services were not offered until 6 months after when the family pursued further assessment.*

### **Service and Policy Implications**

Expansion of Section 2703 of the Affordable Care Act to include ASD and other developmental disabilities may increase the number of families who have a medical provider and a medical home and improve access to and coordination of care. Changes to, or repeal of, the ACA could jeopardize insurance coverage for ASD and/or types of benefits available for an ASD diagnosis. Additional barriers to adequate insurance coverage could result if the requirement to cover pre-existing conditions is revoked.

Coordination of service sectors is urgently needed. Specific service and policy implications of the illustrated examples can be summarized in any of the following ways:

1. Families must navigate different sectors of service in terms of information, provision, and funding (e.g., medical providers, local government, education) all within a very short period of time (from noted concern to early intervention age eligibility cut-offs).
2. Families must deal with different sources of funding for services, frequently with different rules for who, what and how many services can be provided, with no clear sources of information about what these sources are and how they interact.
3. These sectors are not coordinated and often do not communicate with each other, particularly across health and social service agencies. In most instances, there is no funding to support coordination or an assigned liaison.
4. There are considerable differences in the type and amount of services supported by insurance plans. There are also considerable geographic differences in type and amount of services available; inequities and disparities exist across counties, cities and states.
5. There are very clear rules for when and how families are given certain kinds of information (e.g., Early Intervention mandates how quickly an assessment must occur and meetings must be arranged), but there is also substantial variation in other aspects of communication. Time allowed (or reimbursed) for communication between families and experts is often inadequate.
6. Rules which are intended to make services more patient centered (e.g., access to bilingual assessments when a child is exposed to multiple languages) may sometimes create barriers to appropriate services (e.g., access to autism experts) when alternatives could be considered.
7. Systems do not take into account families' concerns about stigma, the reluctance of professionals to make a diagnosis or share concerns about red flags of ASD in very young children, missed or false

positive diagnoses, and the need for earlier evaluations and re-evaluations of very early assessments as symptoms are unfolding.

### **Summary/Progress towards Aspirational Goal**

Significant advances have been made toward early identification of individuals with ASD, so they can be linked to appropriate interventions, services, and supports in as timely a manner as possible. However, gaps still remain. There is a need to validate tools in diverse settings and populations. There is a need to evaluate the effectiveness of universal screening for improving outcomes in ASD. There is a great need to understand the disparities in access and/or utilization of screening and diagnostic tools, and entry into intervention services. The challenges and barriers include gaps in the evidence base for the benefits of early detection in diverse populations and settings; an insufficient workforce with expertise in ASD diagnosis and intervention; lack of medical home for families of children with ASD; the need for continued insurance reform; disparate and uncoordinated service sectors; and the lack of an infrastructure to track children and families in order to evaluate the efficacy of service systems. There have been important strides in the area of early detection of ASD features, and in demonstrating the impact of early intervention. Yet, there are significant challenges and barriers to implementing screening, diagnostic, and treatment services broadly and reducing disparities in access and utilization. The way forward is reflected in the three Objectives proposed for Question 1.

### **Objectives:**

#### **Objective 1: Strengthen the evidence base for the benefits of early detection of ASD.**

- Implement innovative designs to evaluate the benefit of universal screening for ASD, including research that addresses the specific research gaps noted by the USPSTF report.
- To improve early detection there must be greater attention paid to special autism populations such as girls and intellectually delayed individuals.

#### **Objective 2: Reduce disparities in early detection and access to services**

- Improve family engagement and help build an awareness of healthy developmental milestones and warning signs of concern.
- Demonstrate the validity of different screening and diagnostic tools for culturally-diverse communities.
- Increase services in high poverty and under-served regions; improve inclusion of these populations in research.
- Policy: Addressing differences in state policy requirements for Medicaid and the requirement of a diagnosis to receive services.
- Workforce training and the development of a more diverse workforce.

#### **Objective 3: Improve/validate existing, or develop new tools, methods, and service delivery models for detecting ASD in order to facilitate timely linkage of individuals with ASD to early, targeted interventions and supports.**

- Continue research on the potential translation of biomarker findings into feasible and valid screening or diagnostic tools.
- Increase the number of families with medical home
- Better understand lack of compliance with screening recommendations; address barriers to universal screening
- Consider the impact of insurance reform and national policy on coverage for screening, diagnosis and intervention for children with ASD and their families.

- Evaluate innovative service delivery methods (e.g., use of technology) to improve detection methods and increase access.
- Improve the coordination of service delivery and communication among service delivery sectors; improve continuity of treatment and responsiveness to families' needs and preferences.

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### Topic 1: Implementation of diagnostic and screening tools

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## Topic 2: Disparities

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