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Early diagnosis and adulthood transition in autism spectrum disorders (ASD): How primary care helps to close the gaps

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Abstract

Autism spectrum disorder (ASD), a complex neurodevelopmental condition, is a rapidly expanding public health crisis worldwide. The current care system for ASD is fragmented and inadequate. The medical community recognizes two major gaps as deserving of immediate attention: early screening and the transition to adulthood. In this essay, we emphasize the critical status of these two missing components and their negative impacts on ASD care. We also explore the systemic causes of these unmet needs, discuss how a primary care—based approach is best suited to close the gaps, and briefly introduce some innovative primary care—based interventions currently under way. Finally, we identify obstacles and propose steps to overcome these barriers in order to achieve timely diagnosis and continuity of care for patients with ASD.

Introduction: The ASD "Epidemic": Identify major gaps in management

Autism Spectrum Disorder (ASD) is a fast-growing neuro-developmental disorder characterized by deficits in social communication, as well as restricted or repetitive behaviours and interest [1,2]. Its global prevalence has reached around 1 in 160 [3]. In the United States (U.S.), prevalence is 1 in 68 and continues to rise [4]. Despite significant global efforts to improve healthcare for patients with ASD, this population is still largely underserved [5-7]. In particular, early screening and adult transition services are two major unmet needs [8-10]. The most vulnerable and underserved sub-populations in the current landscape of ASD care include highrisk infants (e.g., siblings of patients with ASD or those with certain prenatal exposures), undiagnosed patients with ASD, and adolescents or young adults with ASD [11]. Recently, leading ASD tertiary-care centers in the U.S., including Boston Children's Hospital, have begun to address these challenges [12].

Early diagnosis and adulthood transition: The unmet needs

Early screening and timely diagnosis of populations at high risk for ASD is currently inadequate. Studies have shown that early detection of ASD, especially before age 2, followed by early intervention, can lead to better outcomes [13,14]. Clinicians can reliably diagnose ASD in children as young as 24 months, yet a recent systematic review demonstrated that the mean age at diagnosis ranges from 38 to 120 months [15]. A key factor in this delay in diagnosis is the lack of endorsement by the US Preventive Services Task Force (USPSTF), which concluded that there is insufficient evidence to support universal ASD screening. This, together with other factors, led to the poor implementation rate of universal, tier-1 screening instruments such as M-CHAT (Modified Checklist for Autism in Toddlers) starting at 18 months [15-18]. In fact, only approximately 50% of all US paediatric

practices adopted routine universal screening for ASD [19]. The stance of USPSTF is controversial; many experts in the field and professional organizations such as the American Academy of Paediatrics believe that strong evidence already exist to support universal screening [20].

Several groups are currently validating tier-2 screening for children at risk [21]. In fact, some paediatric practices already adopted tier-2 screening instruments and recently reported reductions in diagnostic delays via partnership with specialists [22]. However, it is currently largely absent from standard paediatric practices. Furthermore, a growing body of evidence suggests that high-risk infants and toddlers warrant earlier screening or specialist referrals before 18 months [23]. Through observation, parents and clinicians have noted early signs of ASD as well as objective measurements at or before 12 months of age [21,24]. While studies confirmed that paediatricians fail to refer a high proportion of at-risk children for timely evaluation [8,21,22,25,26], some early screening tools have already shown promising sensitivities and specificities for early detection [21]. A shortage of ASD specialists, as well as poor coordination between primary care providers (PCPs) and specialty care, can further delay confirmatory diagnosis and treatment, contributing to suboptimal outcomes [27].

In addition to early detection, paediatric-to-adult transition is another important unmet need [10]. Approximately 50,000 Americans with ASD turn 18 years old each year, requiring structured transition

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services with unique considerations from both the medical and social perspective [28].

Unfortunately, the current U.S. healthcare system has few structured medical transition protocols in place [29,30]. Fewer than 20% of young people with ASD receive healthcare transition services, and fewer than 50% have the opportunity to discuss the transition to an adult provider with their paediatricians [31]. Paediatricians lack transition-related training [32], while Internists report insufficient knowledge in ASD care [33]. Consequently, most patients with ASD continue in paediatric care beyond the age of twenty-two [34]. Unstructured transition may lead to a loss to care [35] which has been associated with increases in emergency room attendance, suicides, homicides, and psychotic breakdowns in this age group [10,35,36]. This period also coincides with an increase in obesity, epilepsy, depression, and anxiety [35,37,38]. The increased morbidity and loss of care has formed a consensus among the medical community that adolescents with ASD are vulnerable and underserved, requiring our immediate attention [10].

"Resuscitating" PCPs' roles in ASD care

Traditionally, psychiatry, developmental paediatrics, and neurology form the cornerstone of ASD management in the U.S. [39] and most other countries [40-42], while PCPs play a very limited role in ASD care [3,4,33,43]. It's notable that PCPs make up nearly 50% of the total physician population in the U.S. [44,45] and in most other developed countries [46-49]. PCPs currently perform well visits, making them well suited for early screening and providing resources for transition to adulthood [50,51]. In contrast, specialists in ASD care are becoming increasingly scarce. Furthermore, their expertise is best utilized for confirmative diagnosis and specific treatments rather than screening and health maintenance. With the fast-growing population of ASD patients and at-risk infants, PCP's roles in ASD routine care and co-management warrant prompt "resuscitation", especially in the areas of early detection and adult transition.

In recent years, the U.S. has seen an expansion of ASD centers affiliated with tertiary-care hospitals. However, none of these have inhouse primary-care services, and there is limited collaboration between ASD centers and primary-care practices in the community [53]. Notably, several primary care–oriented efforts to improve care for patients with ASD have been successfully piloted [8,9], and neuropsychiatry-based ASD centers are working to build stronger alliances with primary-care practices in the community [52]. A primary-care based approach, or "activated autism practice," in the words of Kairys et al. [9] could serve as an example.

How PCPs help to close the gaps in early diagnosis

To improve early diagnosis of ASD, paediatric practices need to adopt evidence-based protocols necessary to provide timely screening during well-child visits. The training goals should be two- fold: to implement universal screening with validated tools [51], and to train paediatricians to recognize high-risk patients, so that these toddlers (and even infants) can be identified and referred for timely evaluation. A study by Kairys et al. [9] illustrates the success of such an educational intervention among PCPs.

A recent systematic review of tier-2 screening research concluded insufficient evidence due to small number of studies related to each published screening tool [21]. However, attempts to validate existing tier-2 screening tools are currently under way [21,54,55]. Similarly, research efforts are growing for the development of effective tools

for early detection [21,56]. These new approaches may allow for ASD diagnosis and intervention as early as 12-15 months of age [21,24,56]. New technologies, such as eye-tracking devices, could be integrated into primary-care practice for early screening of children at high risk of developing ASD [57]. We believe that in the near future, there will be more convincing evidence to recommend early screening for high risk infants/toddlers. Leaders of paediatric practices need to stay up-to-date with the fast-evolving landscape of ASD screening. However, as current early screening tools are undergoing validation, we believe that it is important for paediatricians to proactively refer at-risk infants/toddlers to early intervention services and/or specialists for timely evaluation. Figure 1 presents our proposal for a PCP-led ASD early detection program with consideration of tier-2 screening.

To further expedite diagnosis and management of comorbidities, PCPs can partner with a pre- selected network of developmental paediatricians, psychiatrists, and early intervention clinicians for fast-track diagnosis and treatment. ASD screening of the paediatric population at large will nonetheless be time consuming. PCP offices with trained staff and clear protocols are essential to ensure the success and reliability of early screening.

PCP-led transition team smooths transition into adult care

PCPs, with their large physician base and focus on health maintenance, are ideally positioned to take the lead in coordinating the creation and implementation of transition services. Primary-care practices, with guidance from specialists, can improve transition care via a three-pronged approach. First, they should set up a dedicated transition team bringing together frontline paediatricians and internists assisted by case managers, behavioural therapists, mental health providers, and social workers. Second, the team should design working protocols that clearly delineate workflow and duties for each team member, with both outpatient and inpatient navigation algorithms. Third, all team members and related staff should undergo transition-related training. Internists are often less familiar with ASD care than paediatricians [44] and may require additional ASD-specific training to successfully implement transition protocols.

The creation and implementation of transition protocols should be guided by the following goals. First, they serve to address teen or young adults with ASD's medical and mental health concerns as well as provide sex education in a stepwise, evidence-based, and ASD-friendly manner. The secondary goals are to coordinate with schools, employment training programs, and other community resources to prepare patients for greater functional independence in society. With a dedicated team, systematic protocols, and physician training, the PCP-led transition services we propose (Figure 2) may help to minimize missed days from school and work, emergency room visits, hospitalizations, suicides, and crime rates within the population of adolescents with ASD. U.S. primary-care practices affiliated with major tertiary-care ASD treatment centers, such as that of Boston Children's Hospital, have already begun to address the urgent issues of ASD care transition, although details are still unclear [12].

Challenges and future perspectives

ASD is a complex neurodevelopmental disorder with high prevalence [1,3]. Two major gaps in the current landscape of ASD management are early diagnosis and paediatric-to-adult transition. Based on what we have learned from the experiences of patients, families, and providers, we believe that a primary care-based approach

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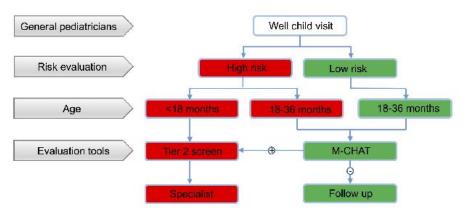


Figure 1. PCP-led early-screening protocol. Early screening teams led by primary care physicians can perform evidence-based universal screening and tier-2 screening to facilitate early diagnosis and intervention for patients with ASD

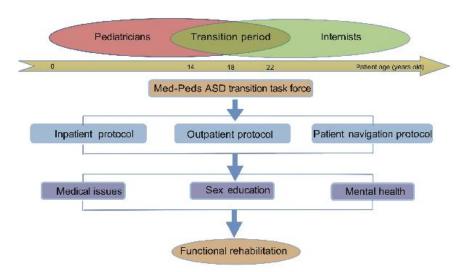


Figure 2. PCP-led adult transition program. During the transition period of patients with ASD (14 to 22 years of age), paediatricians and internists in primary care practices can come together to develop and implement transition protocols (including inpatient, outpatient, and patient navigation components). These protocols should aim to address patients' medical and mental health concerns and improve sex education which tends to be overlooked for adolescents and young adults with ASD

may be the key to fulfilling future needs. However, we must first overcome significant obstacles. Successful early screening requires upto-date paediatrician training, team building, protocol implementation, as well as parental awareness. The same applies to the transition to adult care, with the additional need for ASD-specific training of internists.

Recently, the Center for Disease Control implemented an ASD awareness campaign, reflecting increased governmental support for the issues identified above [58]. The American Association of Paediatrics support the notion of early screening. However, improved ASD care will require changes in insurance reimbursement, policy support such as recommendation from the Task force, and dedicated funding. Ongoing research efforts serve to delineate how PCPs can engage in further co-management roles alongside specialists to fill the current gaps in the management of ASD [59].

Primary care systems are already in place or expanding in most western countries with a high prevalence of ASD [46,47,49]. We call for the incorporation of primary care within existing ASD centers, and better collaboration between ASD centers and PCPs in the community. By promoting early diagnosis and improving transition to adulthood, PCPs worldwide can help individuals with ASD reach their maximal functional status, which in turn will benefit the society as a whole.

Compliance with ethical standards

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