

Review

Autism Screening in Family Medicine Practice: Early Detection, Barriers and Benefits

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Received: 13 May 2022, Accepted: 14 May 2022, Published: 17 May 2022

Abstract

Autism spectrum disorder is a neurodevelopmental disorder marked by social communication difficulties, restricted interests, and repetitive activities. Both genetic and environmental variables impacting the growing brain have an influence on it. Around the world, 1 in 100 children is identified with autism spectrum disorder. Estimates of prevalence has increased over time and varied significantly inside and across sociodemographic groups. Family physicians are the primary healthcare experts to whom children with autism are directed when they have signs and symptoms related to autism or for any other reason, including protective healthcare services. Early diagnosis of autism result in effective and timely management of the patients, hence family medicine practice plays a vital role in autism diagnosis as family physicians are the first contact with the patient. The purpose of this research is to review the available information about the autism screening in family medicine practice regarding early diagnosis, barriers and benefits. The significance of acquiring an early diagnosis of autism and following early therapeutic intervention is well-established in the literature, as late diagnosis is linked to greater parental stress and delayed early intervention, both of which are necessary for long-term beneficial outcomes. Early detection is critical since studies have shown that early diagnosis and intervention programs improve functional results and quality of life. As family physicians are the community's first point of contact, it's vital for them to be able to recognize autism early on in an effort to enhance early intervention and a better outcome.

Keywords: *autism, family physician, early, diagnosis, primary care*

Introduction

Autism spectrum disorder (ASD) is a neurodevelopmental disorder marked by social communication difficulties, restricted interests, and repetitive activities. Both genetic and environmental variables impacting the growing brain have an influence on it. The clinical evaluation process begins with a developmental screening of the general patient group to identify at-risk children, proceeded by a referral to a specialist for a clear diagnosis and a full neuropsychological test (1). Persistent deficiencies in social communication or engagement and confined, repetitive patterns of behaviour, and aberrant sensory responses are major signs of autism. The magnitude of such symptoms varies greatly from patient to patient, resulting in a wide range of clinical manifestations. ASD commonly appears throughout childhood, with symptoms appearing as early as 18 months of age. However, due to the wide range of clinical symptoms, ASD remains difficult to identify. Diagnosis involves both parental and caregiver awareness to recognize indicators and a multidisciplinary medical and paramedical team assessment to confirm signs (2).

Around the world, 1 in 100 children is identified with autism. Estimates of prevalence has increased over time and varied significantly inside and across sociodemographic groups (3). When societal settings begin to confront their constraints in social communication, children begin to show symptoms between the ages of 18 and 24 months. Concerns differ depending on the age at which parents become aware of deviance. Young children are brought in with issues such as delayed or regressive development and speech, as well as improper play or conduct for their age. Academic challenges, social awkwardness, or behaviours significant enough to disturb family life are common in older children. In-depth inquiry in the latter case often finds minor early symptoms that were disregarded because there appeared to be normal growth and even appeared extra sharp due to their extraordinary independence, quick understanding of mechanics, and keen observation (4).

Because of the complexity, severity, and similarity of ASD symptoms with those of other psychiatric conditions, it is critical to appropriately identify ASD using suitable instruments and scales in order to enhance the diagnosis and management of ASD patients. Parents and caregiver interviews, patient interviews, direct

observation of patients, and complete clinical assessments that involve a thorough investigation of family history for ASD or other neurodevelopmental problems are all used as assessment methods to confirm the diagnosis (5). A complete, comprehensive, and structured approach is required to diagnose ASD. The diagnostic assessment not only identifies the disorder but also helps to know the child's corresponding abilities and weak points, recognizes which maladaptive behaviour patterns and co-occurring problems are prevalent, clearly states the effect of the condition of the child on the family, and baselines the patient's developmental skills. Because a solid diagnosis of ASD can be made as soon as 18-24 months of age, primary care physicians should make every effort to detect and intervene in children with ASD as early as symptoms appear (6).

Family physicians are doctors who practice in close proximity to family members' homes or in locations where they can be quickly accessible and seen first, and who provide preventative healthcare as well as first-line diagnosis and treatment (7). As a result, family physicians are the primary healthcare experts to whom children with ASD are directed when they have signs and symptoms related to autism or for any other reason, including protective healthcare services. According to studies, family physicians and paediatrician perform an integral role in the identification of ASD (8, 9). Early therapy has been shown to be beneficial, thus an early detection of autism is crucial. Physicians who conduct developmental surveillance on a regular basis and utilize suitable screening methods boost the likelihood of a timely diagnosis. Screening is giving an autism-specific test to all children, not only those who exhibit autistic behaviors, at their 18- and 24-month clinic visits. Clinicians must refer children for diagnostic evaluations as soon as surveillance or screening test findings raise concerns (10).

The number of studies highlighting the value of early diagnosis and intervention has grown in recent years as the frequency of ASD has increased. Unfortunately, many children with the disease do not obtain a diagnosis until they reach the age of four. Late diagnosis can be due to a variety of factors, including healthcare providers' lack of comprehension of complex early warning indicators and awareness of effective early detection procedure (11). Early diagnosis of autism result in effective and timely management of the patients, hence family medicine practice plays a vital role in

autism diagnosis as family physicians are the first contact with the patient. The purpose of this research is to review the available information about the autism screening in family medicine practice regarding early diagnosis, barriers and benefits.

Methodology

This study is based on a comprehensive literature search conducted on April 11, 2022, in the Medline and Cochrane databases, utilizing the medical topic headings (MeSH) and a combination of all available related terms, according to the database. To prevent missing any possible research, a manual search for publications was conducted through Google Scholar, using the reference lists of the previously listed papers as a starting point. We looked for valuable information in papers that discussed the information about autism screening in family medicine practice regarding early diagnosis, barriers and benefits. There were no restrictions on date, language, participant age, or type of publication.

Discussion

Autism is a spectrum condition, with major differences in patients' social, communication, and cognitive capacities. Multiple aspects of adaptive functioning are significantly harmed as a result of symptoms. Individuals with ASD require varied levels of psychological support to achieve relative independence, and in some situations, they may require ongoing care. Children with autism have varied degrees of communication difficulties, depending on their age and intellectual skills. Speech delays, repetitive speech, echolalia, pronoun reversal, poor understanding, and a complete lack of spoken language are all examples of these deficiencies (12).

Epidemiology

In children aged 4 and 8, the prevalence of ASD was 1.70% and 1.85%, in the United States, while the frequency in Europe ranged from 0.38% to 1.55% (2). In Oman, there were 1.4 per 10,000 cases of ASD, 29 per 10,000 cases in the UAE, and 4.3 per 10,000 cases in Bahrain (13). A study conducted in Riyadh, Saudi Arabia reported a prevalence of 2.51%, 1 ratio 40, 25 per 1000 cases, with a male to female ratio of 3:1 (14). Findings of a cross-sectional study conducted in Saudi Arabia in 2020 revealed that ASD was found in 2.618 children per 1,000 in Jeddah, 3.68 children per 1,000 in Makkah, and 2.81 children per 1,000 in both Jeddah and Makkah (15).

Early diagnosis and Benefits

ASD considered to be one of the most rapidly growing psychiatric disorders in youngsters. According to recent statistics, the prevalence has increased to 1 in 50 to 68 children. According to current evidence, therapies to improve the performance of autistic children are more efficient when started early in life, and the long-term prognosis is better. As a result, healthcare workers must be trained and informed about ASD. Family physicians who have their initial medical encounter with children can play a significant role in diagnosis thus improving the management (16).

The significance of acquiring an early diagnosis of ASD and following early therapeutic intervention is well-established in the literature, as late diagnosis is linked to greater parental stress and delayed early intervention, both of which are necessary for long-term beneficial outcomes. This is especially crucial because research has shown that interventions started before the age of four are linked to considerable improvements in cognition, language, and adaptive behaviour. Early intervention in ASD has also been associated with gains in daily living abilities and social behaviour, according to various studies. Overall, the data suggest that early identification and intervention are critical for children with ASD's long-term prospects and quality of life (17).

Through developmental monitoring and screening, children with ASD can receive ASD-specific behavioural therapies that enhance long-term outcomes. Low rates of ASD screening, inferior performance characteristics of ASD screening instruments in real-world practice, and impediments to screen-positive children being examined for ASD in a timely manner may all contribute to delayed detection (18). Findings of a survey conducted in 2014 among family physicians and paediatricians depicted that 17% of respondents checked for ASD on a regular basis according to defined standards, while 41% screened for ASD but did not follow the guidelines. Paediatricians were much more likely than family physicians to screen for ASD. The tendency to consistently test and be confident in identifying warning indicators of ASD was substantially linked with respondents' pre-professional knowledge in ASD. Physicians who do ASD-specific screenings and recognize early indicators can set families and children on the route to receiving much-needed services (19).

Early detection is critical since studies have shown that early diagnosis and intervention programs improve functional results and quality of life. Unfortunately, early

detection of ASD can be difficult. Despite caregivers voicing concerns about possible ASD between 15–22 months, it is frequently diagnosed beyond the age of 3 years. ASD can manifest itself in a variety of ways, depending on the child. Family physicians are well-positioned to screen for ASD because they are on the front lines of community healthcare. When children visit clinics for acute concerns, family physicians can search for signs of ASD opportunistically, and comprehensively screen for ASD during well-child developmental evaluations (20).

Family physicians or primary care physicians are often the first to notice developmental red flags associated with autism as they are the first point of contact. In order to provide recommendations on the next steps and therapy, Physicians must have a complete understanding of autism symptomatology characteristics. Physicians should not just be knowledgeable about autism; they should also be well-versed in the evidence-based therapies that are available in order to assist caregivers also which would make it easier for people with autism to get access to appropriate, empirically supported treatments that can help them achieve better outcomes in areas like communication, emotional control, and everyday tasks (21).

The widespread implementation of active surveillance systems, which typically involve standardized and particular screening methods for neurodevelopmental disorders, would enable prompt and early assessments, with significant benefits for children's and families' well-being. Furthermore, if a child has been identified as having an ASD risk through screening, a prompt referral to a specialized service expedites the diagnostic procedure, minimizes the time between initial concerns and diagnosis, and enhances family involvement with the evaluation process. Even though the scientific community agrees that early intervention based on early detection is critical, putting this strategy in place and building connections among many stakeholders in this field remains difficult (22).

Barriers

Family physicians' experiences, views, and attitudes around autism have still not been thoroughly investigated, and information about how physicians' beliefs influence their attitudes toward autism screening is also quite scarce in the literature. A qualitative study conducted among family physicians revealed that the routine screening was considered as being hampered by a lack of proper training in child development and

screening procedures, as well as a deficit of community-based resources to handle children with autism which were the major barriers according to the physicians (23). Another literature review study conducted in 2021 reported that there are five major obstacles that hinder children with ASD from receiving early diagnosis and care. Lack of information, social stigma, healthcare providers dismissing parents' initial worries, hurdles to ASD screening and access to ASD services. The author further reported that another obstacle preventing low-income families from receiving ASD services were financial constraints while some parents said that the lack of private or government insurance to pay the price of autism therapy has put most children's chances of receiving treatment out of reach (24).

Physician knowledge and skills are crucial in providing high-quality- quality care to autistic people. Many people with ASD, their families, and even clinicians have complained that doctors lack the specific knowledge needed to screen, diagnose, and refer people with ASD. Autistic people and their caregivers believe that physicians need to learn more about how to help people with severe ASD symptoms, enable more efficient communication, and manage sensory issues. There is need of improving physician expertise and better adapting care surroundings to sensory sensitivity (25). There have been reports of barriers to implementing developmental screening in primary care setting. Time restrictions and insufficient reimbursement are two systemic concerns. In addition, providers' desire to implement routine screening is influenced by appropriate teaching on the value of developmental screening. Furthermore, maintaining adoption in a primary care context necessitates the participation of the entire clinic staff, initial and continuous training (26).

Despite efforts to advocate autism, initiatives to formalize autism training, and particular awareness-raising activities, nearly 40% of physicians report that they have no formal autism training and are unsure how to manage autistic patients. Where physician have personal experience with autism, whether through a family relation or friend on the spectrum or because they are autistic themselves, there is a higher level of autism awareness. The most pressing requirement is communication skills training for physicians. Physicians and hospital professionals admit to having communication issues with autistic patients. Only 25% of primary care physicians said they were confident in talking with autistic adult patients or recognizing and implementing appropriate accommodations (27).

Another study conducted in 2019 reported that lack of knowledge and expertise for diagnosing and managing children with autism, as well as insufficient visit time and compensation, were the most significant barriers among physicians (28).

Several studies have found that early diagnosis of ASD, followed by early intervention, can help to decrease the disorder's negative implications. Implementing an ASD screening program could increase the possibility of an early diagnosis of the illness, as well as the provision of services to the children who are affected. Limited diagnostic resources, a failure of follow-up in screening programs, and societal hurdles such as social stigma are all obstacles to early identification (29). There is limited literature available regarding the diagnosis and management of autism by family physicians. In future more research studies are needed to highlight the role of family physicians in early diagnosis and management of autism.

Conclusion

As family physicians are the community's first point of contact, it's vital for them to be able to recognize autism early on to enhance early intervention and a better outcome. To persuade family physicians to screen for autism, more advocacy, education and training is needed. More resources are needed to establish autism screening in practices and health care systems, with a special focus on the administration of autism screening tools.

Disclosure

Statement:

The authors declare no conflict of interest.

Funding:

No fund.

Ethical consideration:

Non applicable.

Data availability

Data that support the findings of this study are embedded within the manuscript.

Authors' contribution:

All authors contributed equally to the drafting, writing, sourcing, article screening and final proofreading of the manuscript.

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