CAN AUTISM BE DETECTED EARLY IN LIFE?

Lisa Pangemanan[1]
*Corresponding author email: lisa-p@ukwms.ac.id
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ABSTRACT

The neurological defect known as Autism Spectrum Disorder (ASD) usually manifests in early childhood. It is typified by altered social communication, along with repetitive activities. It should be detected as soon as possible, since with early care, the affected person's long-term results can be greatly improved. With an emphasis on early behavioral indicators that may be noticeable during the first year of life, such as reduced eye contact and delayed social communication, the paper explores the prospect of early autism detection. It explores how well the Modified Checklist for Autism in Toddlers Revised/Follow (M-CHAT-R/F) works and how new technologies like eye-tracking and neuroimaging may provide more accurate early detection techniques and diagnostic tools. Furthermore, the role of both genetic and environmental factors in the development of ASD is explored, emphasizing how genetic screening may help identify at-risk groups early. The significance of early intervention programs, which have been demonstrated to improve developmental trajectories when initiated before the age of three, is finally discussed in this study. Better access to screening and care will be made possible by ongoing research into predictive models and technology developments, which should improve early detection techniques.

Keywords: *Autism; Early detection*

1 *Department of Child Health, Faculty of Medicine, Widya Mandala Catholic University, Surabaya, Indonesia*

INTRODUCTION

Autism Spectrum Disorder is increasing in number from year to year.[1] Data from the Ministry of Health in 2021 reported that autism children in Indonesia significantly increased to around 2.4 million, with an estimated increase of 500 people every year.[2] The ratio of normal to autistic children was 1:500, which was reported in 2000. This ratio has increased to 1:150 in 2006. The increased number of autistic children has increased the number of therapy and special schools needed by those children.[2] Early vigilance followed by early diagnosis and management will optimize outcomes and reduce the need for specialized treatment and schools later in life, therefore early identification of autism is a must.[3] This review explains practical and available tools to identify early signs of autism.

Definition

Autism is a developmental disability that impacts social interaction and is characterized by repetitive behavior that typically emerges at an early age but may grow more noticeable as one age, potentially leading to difficulties in social engagement.[4] While the DSM-5 provides a clear basis for diagnosing autism, there are various tools available for the early identification of ASD risk, including the Modified Checklist for Autism in Toddlers Revised/Follow (M-CHAT-R/F), the Survey of Well-Being Young Children (SWYC): Parent's Observations of Social Interactions (POSI), the Screening Tool for Autism in Toddlers and Young Children (STAT), the Social Communication Questionnaire (SCQ), the First Year Inventory (FYI), among many other screening instruments.[5,7] A primary care physician can utilize the M-CHAT-R, a screening instrument mentioned in the manual for Stimulation, Detection, and Early Intervention of Child Development at the Basic Health Service Level (SDIDTK).[8]

Early Signs of Autism in Infancy

Early indications of autism can already be seen in infants. Early indicators of autism include limited eye contact, a lack of social smiles, delayed talking, repetitive motions, atypical reaction to sensory input, and non-reaction to their name. These early warning signs may prompt the worried parents to seek medical attention. By 12-18 months, or even as early as 9 months, many children show these signs.[3,9] Screening for all children during well-child visits at 9, 18, and 30 months is recommended by The American Academy of Pediatrics.[10] It is also advised to specifically screen for autism during well-child exams at 18 and 24 months.[10]

Growth and development monitoring for children 0-6 years old can use The Maternal and Child Health Book (Buku Kesehatan Ibu dan Anak/KIA). To ascertain whether their child's developmental abilities are adequate for their age, parents completed the checklist based on their thoughts. This surveillance is done in 3-month intervals for children below two years of age, and 6-month intervals for children 2-6 years. Fulfillment failure of the age-based checklists, warrants further medical help.[11]

Tools for Early Detection

The Integrated Child Health Examination, also known as Pemeriksaan Kesehatan Anak Terintegrasi (PKAT) has been included in the health service cluster for toddlers and preschoolers. It can be used to detect early signs of ASD, which are part of a child's health, as well as growth and development monitoring for all children aged six months, according to the Decree of the Minister of Health HK.01.07/Menkes/2015/2023 concerning Technical

Guidelines for the Integration of Primary Health Services.[12] A referral for additional evaluation will be made if the PKAT examination reveals any deviations. Early detection of developmental, growth, and clinical issues will allow for better results through early management.

The Modified Checklist for Autism in Toddlers Revised and Follow (M-CHAT R/F) is a checklist specifically used to screen for autism by classifying the risk of a child with the target age 16-30 months, having autism.[13] First invented in 1999 with 23 questions answered by parents regarding the child's behavior and development by Robins, Fein, and Barton; it was revised to 20 questions and added with a Follow-up process to improve the accuracy in 2014 by the same team. The sensitivity and specificity for M-CHAT vs M-CHAT R/F are 85 vs 91% and 87 vs 95 %.[7,14] A "no" response for all questions except 2, 5, and 12 indicates a risk of autism. A"yes" response to those 3 questions implies a risk for autism. The risk of autism is low if the total score is 2 or less; medium if the total score is 3-7; and high if the total score is 8 and above. Low risk for autism in children 2 years and younger requires repeated screening. Medium risk for autism in children requires advanced screening to the next stage (Follow). High risk for autism does not require further screening and is immediately referred for diagnostic and early intervention if needed.[15] The screening took 5-10 minutes and the Follow-up took another 20-30 minutes. It is an ideal initial screening tool that often requires a follow-up with diagnostic tools like the Autism Diagnostic Observation Schedule in Autism Screening (ADOS).

The ADOS is a clinician-administered tool considered the "gold standard" to diagnose ASD, which consists of structured observation of social interactions; communication; play; imagination; and repetitive behaviors. It has 4 modules that cover a wide age range, tailored for children with limited or no speech; children with some phrase speech; children with fluent speech and younger developmental ages; and adolescents and adults with fluent speech. Each behavior is scored on a scale from 0-3, with a total score compared to diagnostic cutoffs that indicate autism spectrum levels. The sensitivity, specificity, PPV, and NPV for ADOS are 88-90%; 80-87%; 78-84%; and 89-94%. It took about 40-60 minutes per module.[16]

Several promising screening tools, such as eye tracking and neuroimaging studies, are still developing.[17,18] Eye tracking is a promising technology for early autism screening. It leverages small, non-invasive cameras to assess a child's gaze patterns that will provide information regarding social and non-social stimuli responses. Their distinct gaze patterns provide valuable information to identify these early behavior indicators. Eye gaze patterns and fixation time are reduced in children with autism.[19] Things consistently found in neuroimaging studies of children below one year with autism, i.e. increased total cerebral volume; early brain overgrowth that will be reduced in size later on; altered growth of amygdala, caudate nucleus, basal ganglia, and corpus callosum; elevated extra-axial cerebrospinal fluid; and altered white matter connectivity in several brain areas, particularly in the corpus callosum and other areas related to social cognition and communication; reduced functional connectivity; reduced activation in regions responsible for social processing (fusiform, prefrontal cortex, and temporal-parietal junction); aberrant connectivity between brain areas related to social and emotional regulation (amygdala-prefrontal cortex); although those studies were done in relatively modest samples; thus further research must be done to yield robust data.[17,18]

Genetic and Environmental Risk Factors

The incidence rate of autism is 2-8% if one of the siblings has autism. This risk will increase to 12-20% if his sibling suffers from a wider domain disorder. Several twin studies found that a shared gene has a greater impact on autism compared to a shared environment.[20] Several genetic variations are suspected to be involved in the occurrence of ASD, such as single nucleotide polymorphism of genes CNTNAP2, MTHFR, OXTR, SLC25A12, RELN, 5-HTTLPR, SHANK3, and VDR.[20–22] The umbrella review published in 2022 found that CNTNAP2, MTHFR, and VDR may confer autism risk[21], while a systematic review published in 2022 found that MTHFR is the only consistent gene related to the occurrence of ASD[22]. Despite consistent and promising results for early detection of autism through genetic testing, inadequate sample numbers result in further research still needs to be done to obtain good results.

Drug and toxic exposure in utero such as prenatal exposure to valproate, SSRI antidepressant, and organophosphate insecticide (chlorpyrifos); especially in the first trimester; increases the risk of autism. High levels of maternal stress, maternal infection in the first trimester, low immunity due to infections, and preterm birth are related to the increased incidence of autism.[20]

The Effect of Prompt Intervention

Twenty-five percent of children with autism will be able to demonstrate considerable improvement, and early intervention will allow them to join regular classrooms (23).[23] Less than 10% of children with ASD continue to be non-verbal after receiving intervention. If the intervention is administered before the age of five, children with non-verbal ASD are more likely to speak. Compared to children diagnosed later in life, those diagnosed and treated before the age of three exhibit superior cognitive, linguistic, and social functioning. In addition to teaching them Functionally Equivalent Replacement Behavior (FERBs), early intervention will prevent the development of secondary symptoms.[23] Tantrums, anxiety, self-harm, and feeling depressed are secondary symptoms that will make matters worse. In a primary care setting, a general practitioner can perform the screening to identify autism risk quicker, which will speed up the diagnosis and intervention initiation.[6,24]

The outcomes of early intervention for autism are significantly influenced by the types of early intervention. Applied Behavior Analysis (ABA), Early Start Denver Model (ESDM), and Discrete Trial Training (DTT) are examples of early intervention available for autism. Applied Behavior Analysis gives positive outcomes in socialization, communication, and expressive language. Unfortunately, such is not true for receptive language, adaptive behavior, and cognitive ability.[25] Applied Behavior Analysis is a validated intervention when started early, with a structured therapy based on learning theory, using positive reinforcement to encourage positive behavior. The sooner intervention is given, the greater impact is seen. A more targeted intervention is needed. Children with more severe symptoms and lower cognitive ability will show slower improvement. Actively involved parents will yield more positive results. Better results will come from spending more time practicing those everyday living skills.[25]

Future Directions in Early Detection

Traditionally, diagnosis based on behavioral assessment can only be made once the child is 18-24 months old. Definitive diagnosis is usually established at 3 years or older. Nowadays, research is directed to be able to diagnose autism as early as

possible, ideally before the age of 1 year. Various biological markers, such as genetic markers, neuroimaging patterns, and physiological responses; machine learning and AI-powered analysis of video recordings, sensor data, and electronic health records; wearable devices and non-invasive sensors such as EEG headbands and wearable heart rate monitors; along with automated behavioral analysis that analyzes facial expressions, vocal tones, and gestures through video are still ongoing.[26,27] With research that uses real-time data, data confidentiality is an ethical issue. Justice is also a moral issue that arises when not all people cannot afford to access these services.[28–30]

CONCLUSION

At-risk newborns can be identified by early behavioral indicators including delayed babbling and poor eye contact. A diagnostic tool should be used after screening tools to identify children who require more testing. Emerging genetic, environmental, and technological insights continue to refine our understanding of ASD. A lot of research is being done to create culturally appropriate, accessible, and effective screening techniques that enable early detection of ASD and encourage prompt intervention for the best possible developmental outcomes.

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