Improving Autism Spectrum Disorders (ASD) in Children Through Health Informatics

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Autism as a syndrome was first delineated in a study conducted at Johns Hopkins University. In this research, 11 children were examined, revealing a core inability to engage with others, an inability to employ language for communication, and an almost compulsive need for sameness. Additionally, a prominent presence of anxiety in the clinical manifestations of autism was observed, with intense fears of commonplace objects being frequently demonstrated by these children (Kanner, 1943). We now have a well-established understanding that autism is a multifaceted neurological and developmental condition characterized by impairments in social communication and the manifestation of restricted interests and repetitive behaviors. These attributes are shaped by a combination of genetic and environmental elements that affect the maturation of the brain (Hodges et al., 2020). Furthermore, studies have shown that autism spectrum disorders (ASD) are present across diverse socioeconomic backgrounds, cultural contexts, as well as racial and ethnic groups (Taylor Dyches et al., 2001).

Early signs of autism in infancy often manifest as deficits in attention, communication, and attachment, which persist and evolve into distinct cognitive, behavioral, and social profiles as children grow (Kientz et al., 2014a). The developmental features of autism during infancy continue into school-age children where they face distinct challenges navigating evolving social norms, different educational environments, interactions with new individuals of the same age group and adults, and adaptations to unfamiliar routines. These challenges can be particularly demanding given their developmental delays and struggles with change. These changes can impact various aspects of their functioning, as they must adapt to more complex social settings, acquire advanced proficiencies, increase their level of communication, and handle greater amounts of information (Kientz et al., 2014a). This progression highlights the significance of prompt recognition and assessment, allowing for timely intervention and support for children with autism.

In this article, my primary emphasis revolves around the examination of various health informatics interventions tailored to enhance the well-being and life quality of children who have ASD. I shall delve briefly into several critical areas, including autism screening tools, the development and application of models for detecting autism based on electronic health record (EHR) data, the application of telehealth technologies in the context of screening, assessment, and diagnosis of autism, and lastly, explore the impact of wearable behavioral aids in addressing the difficulties associated with autism.

Background

Historically, children with ASD have been recognized within the realm of pediatric primary care (PC) through the process of actively seeking parental concerns, engaging in surveillance driven by healthcare providers, and conducting routine developmental screenings employing standardized assessment tools (Levy et al., 2020). One such assessment tool was the Checklist for Autism in Toddlers (CHAT), which was created in the early 1990s. It comprised nine items for parents to report on and an additional five items for direct observation by the child's primary care provider (PCP) or healthcare visitor (Khowaja & Robins, 2013). Although it initially demonstrated promise as one of the earliest validated screening tests for autism with a high positive predictive value (PPV) at 18 months, CHAT's limitations in sensitivity and the need to remove its observation items related to child's behavior led to the development of a modified version, which can be said to be the earliest form of health informatics intervention for children with autism, the Modified CHAT (M-CHAT) (Sturner et al., 2022).

In the late 2000s, Goodwin (2008) highlighted the development of innovative technologies tailored to individuals with autism, their families, and their providers while emphasizing the potential of internet-based telecommunication technology to facilitate remote clinical healthcare and enhance patient education. Regarding recent developments, numerous tools and screening methodologies are currently available for assessing ASD in children as well as infants. This increase is partly due to the median age of autism diagnosis in the United States, which is approximately 4 years, as by this age children in the process of developing ASD, even those without an official diagnosis, significantly lag behind peers of their similar age group (Ruel et al., 2021).

To identify children at a heightened risk of developing ASD, the American Academy of Pediatrics (AAP) and the Centers for Disease Control and Prevention (CDC) advocate the utilization of ASD-specific screening tools during both the 18- and 24-month checkup visits. This recommendation stems from an established connection between early evidence-based interventions and enhanced long-term results for children with autism (Sturner et al., 2022).

Health Informatics Interventions

Screening Tools

The research underscores the significance of timely detection and intervention in ASD for the advancement of verbal and socio-cognitive abilities, particularly through early screening. Children undergoing intensive behavioral intervention before age 4 for at least two years, experience a notable increase in IQ and improved adaptive functioning (Ruel et al., 2021). In this section, I will provide an examination of two screening tools.

The Modified Checklist for Autism in Toddlers Revised with Follow-up (M-CHAT-R/F)

M-CHAT is a widely used screening tool for autism approved for ASD screening by the American Academy of Pediatrics (AAP) and advocacy groups like ‘Autism Speaks' (Sturner et al., 2016). Further investigation into the M-CHAT revealed that conducting a follow-up interview after a positive M-CHAT screening effectively decreased the rate of unnecessary referrals and substantially enhanced the positive predictive value (PPV) from 0.11 to 0.65, underscoring the importance of this procedure. This modified version is known as the Modified Checklist for Autism in Toddlers Revised with Follow-up (M-CHAT-R/F) (Sturner et al., 2022). The M-CHAT-R/F is a quick assessment with 20 yes-or-no questions, taking less than two minutes to score. It includes items that evaluate behaviors often linked with autism (such as diminished social interest, social play, or pretend play) and developmental behaviors typically preserved in children with autism. Scoring ranges from 0-2, indicating very low ASD risk, to 3-7 for moderate risk, and finally 8-20 for high ASD risk (Pop-Jordanova & Zorcec, 2021).

It was found that PCPs preferred screening kids during their 18-month and 24-month routine visits, using an online version of the M-CHAT filled out by caregivers at home or in the waiting room through a web system called the Child Health and Development Interactive System. This online M-CHAT/F allowed PCPs to clarify positive parent responses during well-child visits, streamlining the process instead of requiring additional visits or phone calls with trained interviewers. The utilization of this electronic system provided significant advantages, as electronic decision support offers a vital framework when following an algorithm is suitable and holds clinical benefits (Sturner et al., 2016).

The Quantitative Checklist for Autism in Toddlers (Q-CHAT)

The Q-CHAT is a reliable measure of autistic characteristics with good psychometric properties across diverse settings and cultures (Tartarisco et al., 2021). It introduced ordinal responses (how much/often) instead of the binary yes-or-no responses of the CHAT and M-CHAT, acknowledging autistic traits lie on a dimension. Comprising 25 items relating to a child's development and reflective of autistic behaviors, the Q-CHAT employs a Likert scale ranging from 0 to 4 for each item. The cumulative Q-CHAT score spans from 0 to 100 (Sturner et al., 2022).

Lately, machine learning (ML) has found applications in behavioral science, particularly to enhance the classification accuracy of autism screening and diagnostic tools, with a primary focus on children. This was confirmed by Tartarisco et al. (2021) in his study where five distinct ML algorithms, including logistic regression (LR), support vector machine (SVM), naïve Bayes (NB), and K-nearest neighbors (KNN), were utilized to assess the entire set of Q-CHAT items. Their outcomes demonstrated that ML classifiers can accurately differentiate between typically developing (TD) children and those with autism with a high degree of accuracy, relying on a limited set of Q-CHAT items. More precisely, machine learning algorithms attained autism detection rates surpassing 90% when utilizing 14 items and over 80% with only 3 items. These outcomes confirm the Q-CHAT's suitability as an early quantitative autism screening tool with cross-cultural applicability and underscore machine learning's potential to enhance precision.

Electronic Health Record (EHR)

Although tools like M-CHAT-R/F play a crucial role in early detection, there is a requirement to innovate and integrate supplementary data sources to improve their precision and dependability. Passive surveillance of EHR data presents a promising option for early detection (Engelhard et al., 2023).

The EHR contains various documented early autism correlates, including premature birth, low birth weight, and perinatal complications. Conditions like hyperbilirubinemia and respiratory infections are also documented through diagnostic codes. Moreover, issues related to crying, feeding, and sleeping, which link to potential autism diagnosis, could be found in clinical notes or frequent health service visits. Although these findings may not have substantial predictive value on their own, collectively, EHR data can effectively identify autism in early childhood (Engelhard et al., 2023). A recent study by Onishchenko et al. (2021) demonstrated that predictive models utilizing claims data based solely on prior diagnosis codes could yield valuable autism-related insights as early as 100 weeks of age. In contrast to screening tools like the M-CHAT, EHR-based autism detection occurs much earlier (at 30 days of age) and is completely passive, requiring no additional data collection beyond routine care. This method identified 45.5% (approximately half) of children with autism at 30 days while upholding a high specificity of 90.0% (Engelhard et al., 2023).

In a different study conducted by Lingren et al. (2016), an automated algorithm for identifying a cohort of ASD patients from EHR was developed, evaluated, and validated. They utilized this algorithm to investigate over 20,000 patients, which was the largest ASD cohort assembled thus far, examining the patterns of medical comorbidities that co-occur with ASD. Analyzing large cohorts through computational algorithms allowed for the identification of clinically distinct subgroups of comorbidities associated with ASD. By applying the algorithm across multiple institutions, they demonstrated the feasibility of generating a substantial cohort of ASD patients. This large cohort allowed them to explore ASD comorbidities, confirming previous findings of distinct comorbidity clusters in ASD. The algorithm's automation enabled the creation of high-quality cohorts, offering the potential for further large-scale EHR studies and research into tailored treatment approaches based on comorbidity clusters and genetic characteristics.

Telehealth

Telemedicine offers a practical substitute for conventional in-person procedures in ASD screening, assessment, and diagnosis. It includes both real-time (synchronous) and store-and-forward (asynchronous) approaches. Synchronous telehealth employs live videoconferencing to instruct caregivers through activities and observe the child's behaviors in real-time while asynchronous telehealth relies on uploading videos of interactions between the child and caregiver to internet portals for storage and later sharing with clinicians (Riva et al., 2023). A study by Nazneen et al. (2015) illustrated the positive participation of parents in recording videos of their children's behavior at home and then sharing them with PCPs.

Application of Telehealth: Diagnosis

The Naturalistic Observation Diagnostic Assessment (NODA) is an established application that can be downloaded for direct use. It employs the store-and-forward approach and comprises 2 main parts. The first, called NODA Capture, allows caregivers to upload short videos from their mobile devices. Parents organize the child’s environment according to provided scenario descriptions and given sample videos. Subsequently, they upload all the recordings along with the child's developmental history to the portal. The first three scenarios primarily display the child's behavior associated with play, and social communication abilities, while the final scenario highlights the parent's concerns about the child's behavioral symptoms. The second component is NODA Connect, which is an online platform allowing doctors to diagnose the child by using home videos and the provided developmental history. It also helps in connecting the observed behavioral characteristics in the videos, like a lack of eye contact, with the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5) criteria. This portal can also be used to share the diagnosis findings with parents and other medical professionals. NODA, which relies on videos uploaded by caregivers for ASD diagnosis by clinicians, achieved a specificity of 0.94 and a sensitivity of 0.85 (Liu & Ma, 2022).

Application of Telehealth: Assessment

Telemedicine-based Autism Spectrum Disorder Evaluation Tool for Toddlers and Young Children (TELE-ASD-PEDS) is not a screening or even a diagnostic tool but is utilized to assess core ASD behaviors in children aged 1-3 via real-time video conferencing with trained examiners. It employs an ML algorithm to identify the 12 most predictive age- and communication-appropriate activities, offering support to clinicians. During the assessment, clinicians rate the child on 7 behavior items (e.g., repetitive play or eye contact) via video conferencing, using a binary yes-or-no response and a Likert scale to gauge symptom severity (3 = ASD features present; 2 = probably atypical; 1 = no ASD features) (Liu & Ma, 2022).

Wearable Behavioral Aids

Recent advancements in health monitoring have led to the development of external wearable systems that can discreetly capture real-time behavioral and physiological data. These devices, known as 'wearable technology' (WT), are worn externally and have become increasingly relevant in the context of ASD. It offers various opportunities to assess the physiological and behavioral aspects of individuals with ASD and supports their engagement in different domains of life. Of particular interest is the autonomic nervous system (ANS), which plays a vital role in regulating emotions and arousal levels, often showing atypical patterns in individuals with autism. Wearable devices can measure these physiological markers indicative of ANS activity which include heart rate, body temperature, and electrodermal activity, providing valuable insights into physiological responses that are not easily observed otherwise (Black et al., 2020). There have been many studies utilizing these. An illustrative case is the study by Nguyen et al. (2021) which demonstrated the effectiveness of a wearable ‘Anxiety Meter’ in enhancing awareness of anxiety symptoms in children diagnosed with ASD. Citing another study, Liu et al. (2017) demonstrated the development of a system that employed ‘Google Glass’ and facial recognition software to design interactive games for children with ASD using real-time interactions.

Discussion

Importance

The aforementioned health informatics interventions underscore the need for ongoing research in their respective domains due to the significant benefits they offer. For instance, telemedicine addresses concerns such as reducing delays between parents expressing concerns and obtaining an official assessment and diagnosis of ASD. While expediting the waiting period for parents, telehealth further improves the standardization of ASD diagnosis and assessment within primary care settings with favorable sensitivity and specificity (Riva et al., 2023). Moreover, real-time follow-up telehealth interviews facilitate conversations between caregivers and PCPs regarding a child’s behaviors that might indicate ASD (Sturner et al., 2016).

It has been elucidated that while EHR data may have limited individual significance, their collective value is substantial when it comes to predicting autism diagnoses (Engelhard et al., 2023). Concerning wearable devices designed for health monitoring, they possess the capability to automatically identify instances of stress and tantrums associated with specific situations (Koumpouros & Kafazis, 2019). Furthermore, wearable sensors facilitate ecological validity (i.e., representativeness or naturalness since most assessments and diagnoses are conducted in unfamiliar environments), repeated assessment (to compensate for the heterogeneity in presentation of symptoms and varied developmental trajectories observed in children with ASD), and mitigation of reactivity where reactivity indicates that the act of measurement can influence what is being measured (Kientz et al., 2014b).

These considerations give a tiny glimpse into the critical importance of the interventions and the necessity for their continuous development.

Limitations

The studies mentioned above are accompanied by a few limitations. Firstly, some of these studies, such as the one conducted by Sturner et al. (2016) focusing on autism screening with online decision support using M-CHAT-R/F, encountered relatively small sample sizes due to the relatively low prevalence of autism diagnoses. Additionally, challenges persisted in convincing families about the necessity of diagnostic evaluations. Secondly, the study conducted by Sturner et al. (2022) to determine the specificity and PPV of M-CHAT-R/F witnessed a notable number of parents declining participation, resulting in an overrepresentation of parents with higher levels of education in the sample. Thirdly, the predictions generated by the EHR-based autism detection model in the study conducted by Engelhard et al. (2023) are rooted in data from the Duke University Health System (DUHS). Consequently, these findings may not be readily transferable to settings with differing demographic characteristics or population health profiles, or to environments employing distinct EHR systems or data structures. Furthermore, the viability of these models is contingent upon the availability of infant EHR data, making it improbable for these findings to be applied in settings where EHRs are not the norm, particularly in cases involving individuals lacking access to healthcare.

Conclusion

In conclusion, health informatics interventions have significantly advanced the early identification and treatment of ASD in children. The historical reliance on parental concerns and developmental screening has evolved into a multifaceted approach, incorporating various innovative tools and technologies. The importance of these health informatics interventions is underscored by their ability to bridge gaps in early detection, improve accuracy, and expedite the ASD diagnostic process, ensuring children receive timely interventions for better long-term outcomes.

Recommendations

Continuing forward, the limitations of these interventions should be addressed through larger-scale studies and ongoing research must be conducted to enhance their precision and reliability. I recommend the usage of a combination of these interventions to further increase the efficiency of individual tools. For example, integrating data from dedicated screening tools like M-CHAT with telemedicine diagnostic methods that include real-time interviews, would be advantageous. Furthermore, if available, supplementing EHR data during the period of infancy to any of the interventions for the given child would enhance the effectiveness of the other intervention being used individually. Additionally, I recommend focusing on advancing telehealth applications for the diagnosis and assessment of ASD, with an emphasis on expanding their reach and enhancing their capabilities. Finally, I suggest further investment in the development and integration of wearable technology for real-time monitoring of physiological and behavioral aspects, particularly for individuals with ASD, for favorable outcomes.

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