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Potential for Digital Behavioral Measurement Tools to Transform the Detection and Diagnosis of Autism Spectrum Disorder

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For more than a century, clinicians have used clinical observation to describe complex psychiatric disorders, such as autism spectrum disorder (ASD). From these observations, screening tools and diagnostic methods, such as the *Diagnostic and Statistical Manual of Mental Disorders* (Fifth Edition), have been developed to predict and define overlapping and heterogeneous conditions that affect neurodevelopment. The current standard of care for screening for ASD involves asking caregivers to complete a questionnaire about their toddlers' behavior during a pediatric well-child visit. If the caregivers indicate a certain number of symptoms, the toddlers are referred for diagnostic assessment by a trained clinician who observes the children's behavior using semistandardized behavioral tasks. The clinician then decides whether symptoms are present and their severity. Because of the difficulty in establishing reliable clinical ratings based on subjective observations of behavior, diagnostic training requires extensive background knowledge about child development and several months of training by a criterion standard clinical expert. This rigorous training has influenced the field of autism research over the past several decades, allowing for replication and comparability among studies that was previously impossible.

Although standardized methods for clinical observation have been essential in accurately detecting and defining psychiatric disorders, several features of the current process pose challenges. These include using subjective rather than objective measurements, the lack of precision in measurement, a need for highly trained professionals causing long waiting periods for families seeking diagnostic assessments, difficulty in exporting these methods to natural environments, such as homes or communities lacking access to trained professionals, and the infeasibility of gathering large data sets to clarify the heterogeneity of ASD and its overlap with other comorbid disorders. Digital behavioral measurements have the potential to address each of these challenges and transform clinicians' ability to detect, characterize, diagnose, and monitor symptoms for ASD and other neurodevelopmental disorders. Currently, the use of digital measurement tools is an active area of research in the autism field and has already achieved some success in other areas of psychiatry.¹

Subjective vs Objective Measurement

Although caregiver reports and clinician ratings offer important information about a child's behavior and will remain critical elements of ASD screening and diagnosis, they have several disadvantages. Commonly used screening methods that rely on caregiver reports have lower performance rates when used with caregivers of lower education and minority status, which is likely influenced by caregivers' knowledge about child development.² Although the American Academy of Pediatrics recommends screening all children with ASD at age 18 months, an underuse of ASD screening in pediatric primary care is due, in part, to the complexity of interpreting caregivers' reports and may contribute to the median age of ASD diagnosis in the United States remaining at 40 months despite advancements in effective treatments.³ The difficulty in making reliable judgements about the presence or absence of symptoms and their severity is underscored by the substantial amount of training required to become proficient in diagnosing ASD, which has contributed to few available diagnosticians and few studies conducted in low-resource communities.⁴ Ultimately, such labor-intensive approaches are insufficient to address the worldwide need for the early identification of ASD and other neurodevelopmental disorders.

Improving the Precision of Behavioral Measurement

A clear advantage of digital behavioral measurement is that it offers better precision and resolution. Computers provide quantitative measurements of behavior, allowing clinicians to describe behavior as a dynamic process that varies on a scale of milliseconds. For example, a cardinal symptom of autism in children is the failure to orient when called by name, a symptom present by age 8 to 10 months. Using computer vision analyses to assess a child's orientation by tracking head movements not only detects any failure to orient, but also detects delayed orientation. When children with autism orient to their name, they do so a full second slower than a neurotypical toddler.⁵ Caregivers and clinicians are not able to discern this delay with the naked eye. Digital measurements provide precise, continuous, and quantitative data that capture the dynamic nature of behavior. Initial studies indicate that a computer is able to automatically code ASD symptoms that are associated with attention, gaze, and facial expression as accurately as expert human coders.⁶ Moreover, rather than judging a symptom, such as a lack of a facial expression, on a 3-point scale (1 being normal, 2 being mildly abnormal, and 3 being moderately abnormal), digital approaches allow for more detail. Digital behavioral measurement automatically calculates the latency, amplitude, duration, and frequency of individual facial motor movements that comprise an emotional expression. This increase in precision allows clinicians to discover ways of describing autism symptoms that were previously undetectable.

Large-scale Data Collection in Natural Settings

Large-scale data collection in natural settings has the potential to begin to address several barriers to the progress in understanding and describing neurodevelopmental disorders. The heterogeneity, complexity, and overlap among neurodevelopmental disorders have made detection and diagnosis difficult. Defining reliable subtypes of ASD that map to etiological factors and predict responses to specific treatments has been a key objective of autism

research. However, the lack of adequately sized data sets derived from truly representative populations is one of several challenges in achieving this goal. The current knowledge of ASD, including how to prescribe individualized treatments, is based on small samples that are conducted in laboratory settings. To our knowledge, population-based studies that characterize the complex ASD phenotype that differs by age, sex, racial/ethnic background, treatment history, and etiology are largely absent.

Large data sets that allow iterative feedback and the continual refinement of screening and diagnostic algorithms based on machine learning will provide better measures for ASD symptoms, their variance over time, and overlap with other conditions, such as attention-deficit/hyperactivity disorder and intellectual disability. The authors have recently demonstrated the feasibility of creating stimuli that elicit ASD symptoms in young children. The stimuli can be downloaded by caregivers and shown to their children at home on a smartphone.⁷ Sensors in commonly available devices, such as cameras in smartphones and smart tablets, are able to capture behaviors, such as attention and facial expressions, and differentiate between children who are at a high risk vs a low risk for ASD.⁷ This integrated approach of active stimuli design, consumer-grade sensing exploitation, and automatic analysis via computer vision and machine learning provides reliable quantitative measurements of children's behaviors in natural environments. Such an approach can monitor whether autism symptoms are increasing in severity, as occurs in the case of autistic regression, or improving in response to treatment. The availability of large quantitative data sets allows for empirically derived, bottom-up descriptions of behavior. Rather than approaching diagnosis with a set of predetermined symptoms and rating scales, clinicians can let the data reveal how individuals with differing backgrounds (ie, etiology, age, sex, ethnicity/race, and treatment) vary in a range of behavioral features, such as attention, motor activity, and affective expression.

Conclusions

While clinicians fulfill an essential role in evaluating and treating individuals with ASD, digital behavioral measurement approaches to early detection, diagnosis, and symptom monitoring hold the potential to accelerate science and increase access to early detection and treatment, thereby improving the lives of individuals with ASD and other neurodevelopmental disorders. To realize this potential, future research should focus on the careful and systematic empirical validation of such measures to ensure their accuracy, reliability, interpretability, and true clinical utility.

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