Performance of a Novel Software-based Autism Spectrum Disorder Diagnostic Device* for Use in a Primary Care Setting

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• Approved by IntegReview Institutional Review Board. Statistical analysis independently verified by Bright Research Partners.

STUDY FLOW

RESULTS

For study completers with a determinate device result, performance was:

<table>
<thead>
<tr>
<th>Performance Metric</th>
<th>% (95% CI)</th>
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<tr>
<td>PPV</td>
<td>80.8% [70.3, 88.6]</td>
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<tr>
<td>NPV</td>
<td>98.3% [90.6, 100]</td>
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<tr>
<td>Sensitivity</td>
<td>98.4% [91.6, 100]</td>
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<tr>
<td>Specificity</td>
<td>79.9% [67.6, 87.7]</td>
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• There was no evidence of device performance inconsistency across subjects’ sex, race/ethnicity, household income, parent education level, or geographic location as determined by examining the overlapping of corresponding 95% CIs.

• Of the study completers with ASD, 52% received a determinate result and all were correctly classified by the device with the exception of a single false negative.

• The prevalence of diagnoses among study completers as determined by specialist agreement was:
  - Neurotypical (9%)
  - ASD (29%)
  - Non-ASD Neurodevelopmental Condition (62%)

DISCUSSION

Compared to the current ASD diagnostic process, the device could allow for timelier initial evaluation,2,3 has the potential to address disparities that exist in time to diagnosis for minoritized children,9,10 and can be applied to a primary care setting unlike existing diagnostic instruments which are likely to over-diagnose if used in populations with lower ASD prevalence.8,10

• Using this device in conjunction with clinical judgment and DSM-5 criteria, PCPs could efficiently and accurately assess children 18-72 months old for ASD in the primary care setting, whereas currently nearly all children with ASD are diagnosed in specialty care.

• This could enable earlier diagnosis and earlier ASD-specific interventions, thereby increasing the chances of optimal outcomes.

• Shifting a portion of ASD diagnosis to PCPs could allow specialists to focus efforts on diagnosis and treatment of children with more complex presentations.

• The device is user friendly, and results are rapidly available upon completion of the inputs. The device’s safety feature of providing an indeterminate output conveys actionable information to a PCP indicating high likelihood of a complex neurodevelopmental disorder.

• PCP actions could include continued developmental surveillance, referral to speech, occupational, or behavioral therapy services, or specialist evaluation.

REFERENCES

1. Autism spectrum disorder (ASD) is one of the most common developmental disorders with a prevalence of 1.7-2.5.1,2 The average age of ASD diagnosis is 4.3 years in the U.S. and has remained largely unchanged since the CDC began tracking prevalence rates in 2000.3,4 The lack of diagnostic tools for ASD in primary care settings contributes to an average delay of 3 years between first parental concern and diagnosis and to long wait lists for specialty evaluation.1,5,6 This delay is often even longer for children who are non-white, female, of lower socioeconomic status, and/or in rural areas.1,5,7 A diagnostic aid that utilizes technology could enable PCPs to efficiently and effectively streamline the ASD evaluation process allowing for earlier ASD diagnosis and ASD-specific interventions.

OBJECTIVE

This study examined the performance of an artificial intelligence-based (AI) software as a medical device designed to aid in the diagnosis of ASD compared to diagnosis by independent agreement among board-certified specialist clinicians.

DESIGN/METHODS

• This prospective pivotal study used a double-blind active comparator design conducted at 14 sites across 6 states.

• The device collects three sets of age-dependent inputs:
  1. Caregivers completed a questionnaire via a mobile application.
  2. Caregivers used the application to upload two distinct, 1-2 minute videos of their child in natural settings interacting, playing, or talking. Video analysts scored uploaded videos for a variety of ASD features including communication, social interaction, sensory interests, and stereotyped behaviors.
  3. A PCP met with the caregiver and child and completed a brief questionnaire during a 10-minute visit.

• The AI algorithm used the inputs to generate a result of positive or negative for ASD.

• To reduce the risk of false classifications, the algorithm was designed to provide an indeterminate output as a safety feature.

PARTICIPANTS

• 425 study completers (36% female) age 18–72 months old with developmental concern; M = 3.33 years (SD = 1.15)

• Study population mirrored US population across race, ethnicity, and socio-economic status.

• Compared to the current ASD diagnostic process, the device could allow for timelier initial evaluation,2,3 has the potential to address disparities that exist in time to diagnosis for minoritized children,9,10 and can be applied to a primary care setting unlike existing diagnostic instruments which are likely to over-diagnose if used in populations with lower ASD prevalence.8,10

• Of the study completers with a determinate output (68%), 91% had complex determinations, including ASD (20%) and non-ASD neurodevelopmental conditions (71%).

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