Web-based tool produces fast, accurate autism diagnosis

April 10 2012, by Katie DuBoff

Researchers at Harvard Medical School have significantly reduced from hours to minutes the time it takes to accurately detect autism in young children.

The process of diagnosing autism is complex, subjective, and often limited to only a segment of the population in need. With the recent rise in incidence to 1 in 88 children, the need for accurate and widely deployable methods for screening and diagnosis is substantial. Dennis Wall, associate professor of pathology and director of computational biology initiative at the Center for Biomedical Informatics at Harvard Medical School, has been working to address this problem and has discovered a highly accurate strategy that could significantly reduce the complexity and time of the diagnostic process.

Wall has been developing algorithms and associated deployment mechanisms to detect autism rapidly and with high accuracy. The algorithms are designed to work within a mobile architecture, combining a small set of questions and a short home video of the subject, to enable rapid online assessments. This procedure could reduce the time for autism diagnosis by nearly 95 percent, from hours to minutes, and could be easily integrated into routine child screening practices to enable a dramatic increase in reach to the population at risk.

"We believe this approach will make it possible for more children to be accurately diagnosed during the early critical period when behavioral therapies are most effective," said Wall.

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Autism is diagnosed through a careful analysis of an individual's behavior. When children are evaluated for autism, they typically take the Autism Diagnostic Interview, Revised, known as the ADI-R, a 93-question questionnaire, and/or the Autism Diagnostic Observation Schedule, known as the ADOS exam, which measures several behaviors in children. Together these evaluations can take up to three hours to complete and must be administered by a trained clinician. Often, there is a delay of more than a year between initial warning signs and diagnosis because of the waiting times to see a clinical professional who can administer the tests and deliver the formal diagnosis, Wall said.

Using machine learning techniques, an artificial intelligence method where machines are trained to make decisions, Wall and his team studied results of the ADI-R from the Autism Genetic Research Exchange for more than 800 individuals diagnosed with autism to find redundancies across the exam. They found that only seven questions were sufficient to diagnose autism with nearly 100 percent accuracy, equivalent to the full 93-question exam. They validated the accuracy of the seven question survey against answer sets from more than 1,600 individuals from the Simons Foundation and more than 300 individuals from the Boston Autism Consortium.

Wall applied similar techniques to the ADOS exam, this time classifying more than 1,050 individuals with near perfect sensitivity and slightly less than 95 percent specificity. The outcome of this work was not only a shortened mechanism for evaluating a child (8 out of 29 steps), but also a roadmap for evaluating short home video clips. Together these results have tremendous potential to move a substantial percentage of the effort into a mobilized electronic health framework with broad reach and applications.

"This approach is the first attempt to retrospectively analyze large data repositories to derive a highly accurate, but significantly abbreviated classification tool," said Wall, who is also associate professor of pathology at Beth Israel Deaconess Medical Center. "This kind of rapid assessment should provide valuable contributions to the diagnostic process moving forward and help lead to faster screening and earlier treatment," he said.

The traditional diagnostic surveys for autism can be prohibitive for families and caregivers because they are lengthy and have to be administered by a licensed clinician, often in an environment that is unfamiliar to the child, which can be a tremendous burden for families in remote areas, said Wall. "With this mobilized approach, the parent or caregiver will be able to take the crucial first steps to diagnosis and treatment from the comfort of their own home, and in just a few minutes."

Currently, Wall has made a survey and video site available to the public for free to continue evaluating the effectiveness of the shortened approaches and is working on ways to mobilize the overall approach to enable wide reach across the entire population in need. His team has also launched a Facebook page to spread the word and to share the survey more broadly. To date, 2,500 people have taken the Autworks survey.

  More information: "Use of machine learning to shorten observation-based screening and diagnosis of autism" by Wall et al. *Nature Translational Psychiatry*, online publication, April 10, 2012

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