

Researchers Seek Sweet Spot for Early and Accurate Autism Identification



Why We Did This Study

Many chronic conditions, such as Alzheimer's disease and autism, are often diagnosed long after the earliest signs may be present. Earlier identification could help people receive support, screening, and treatment sooner, but acting too early when less information is available can also lead to false alarms.

The researchers wanted to develop a method that uses information collected over time in electronic health records to help determine when there is enough evidence to alert a healthcare provider and when it may be better to wait for more information. They also wanted the method to account for a common challenge in health record data, namely that some people have not been followed long enough to know whether they will eventually receive a diagnosis.

WHO PARTICIPATED IN THIS STUDY?

The study used both simulated patient data and real-world health records. For Alzheimer's disease, researchers analyzed records from 8,803 patients in the National Alzheimer's Coordinating Center dataset. For autism, they analyzed health records from 29,357 children in a pediatric healthcare system.

The autism cohort included children born between 2016 and 2023 who had multiple well-child visits. The researchers used these records to test whether their approach could identify people at high risk before a formal diagnosis was made.

WHAT WERE THE RESULTS?

The authors used a type of artificial intelligence called reinforcement learning to learn when to act and when to wait for more information. Their method performed better than standard prediction methods, especially when important clinical information became available at consistent or predictable time points, for example due to routinely scheduled screening.

The method also accounted for people whose follow-up ended before it was known whether they would eventually receive a diagnosis, helping the system avoid becoming overly cautious and delaying alerts. In real-world testing, the system identified people later diagnosed with Alzheimer's disease an average of 21 months before their diagnosis, later diagnosed with autism about 9 months before diagnosis, while limiting false alerts to about 10% among people who did not receive the diagnosis.

WHAT DO THESE RESULTS MEAN?

These findings suggest that artificial intelligence can help healthcare systems determine when to send an early alert by considering both current information and the value of waiting for future information. The study also shows that accounting for incomplete follow-up is important to avoid overly cautious alerts. With further development and validation, this approach could help clinicians identify people who may need further evaluation or intervention much earlier, creating more opportunities for timely support and care.

Read more about this study: <https://duke.is/AI-autism-identification>