Deep Learning for Autism Diagnosis: Insights into Genomic and Phenotypic Features

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Autism is a group of heterogeneous disorders defined by deficits in social interaction and communication. Even though autism has a significant genetic component, it is solely diagnosed through behavioral examinations. The most extensively used exam, Autism Diagnostic Observation Schedule (ADOS), uses 29 different scoring categories for diagnosis. Diagnosis of autism under the current approach is often significantly delayed, inhibiting access to early intervention programs. A computer-aided diagnostic tool was developed in order to address this delay and incorporate genomic information into the diagnostic process. Specifically, the developed tool utilizes deep learning (an algorithm used for artificial intelligence) to find high-level and diagnostically significant abstractions in the data. This novel tool, coined “deep diagnosis”, predicts autism diagnosis by combining genomic and phenotypic information (single nucleotide polymorphisms and ADOS scores, respectively) into a deep learning framework. Pre-existing genomic and phenotypic datasets were acquired from Autism Genetic Resource Exchange (AGRE) and National Database for Autism Research (NDAR). Using this information, the deep diagnosis tool achieved 99.6% accuracy, 83.3% specificity and 100% sensitivity, when predicting autism diagnosis. This research is the first proof of concept that exploits information from two complementary domains to predict autism diagnosis. It is also the first deep learning framework for computer-aided autism diagnosis. This work has the potential to be used as a low-cost application by clinicians during the diagnostic process, effectively serving as a real-time “second opinion”, and can lead to earlier, more reliable autism diagnosis for many families.

Awards Won:
China Association for Science and Technology (CAST): Award of $1,200