Towards an Ontology-Driven Machine Learning Approach to the Diagnosis Classification Problem in Autism Spectrum Disorder

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Abstract

Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder that is characterized by social, communicational, and behavioral impairment. This research aims to develop an ontology-driven machine learning approach to the ASD diagnosis classification problem with respect to feature selection. As a first step, we analyzed ASD in terms of the Mental Disease Ontology (MD) and discussed from the MD perspective on ASD a machine learning analysis of an ASD-related dataset based on the support vector machine.

Keywords:

autism spectrum disorder, machine learning, Open Biomedical Ontologies (OBO) Foundry

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Introduction

Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder that is characterized by impaired social and communication skills as well as by restricted and repetitive patterns of behavior. One central issue with existing clinical ASD-diagnosis methods is that they require a considerable amount of time. This problem stimulates an increasing interest in the application of machine learning methods to the amelioration of ASD screening and diagnostic instruments [1]. Despite its potentiality, the machine learning approach presently used may have its limitations with respect to feature selection. In this research we aim to develop an ontology-driven machine learning approach [2] to the ASD disgnosis classification.

Methods and Results

We interpreted ASD in terms of the Mental Disease Ontology (MD) [3] which is built in compliance with the Open Biomedical Ontologies (OBO) Foundry, where a mental disease is a disease to undergo pathological mental processes. From the MD perspective on ASD, we considered a machine learning analysis of part of Thabtah's [1] ASD-related dataset representing the predictivity of all the six possible pairs among the following four features with respect to accuracy: (i) Born with jaundice; (ii) Family member with Pervasive Development Disorder (PDD); (iii) Country of residence; and (iv) Screening Score (see Table 1). In this data analysis, we utilized the Support Vector Machine (SVM): a (supervised) learning system based on kernel methods that is used especially for (binary) classification or regression analysis. In particular, we employed the SVM with the (Gaussian) Radial Basis Function (RBF) kernel: one of the most widely used nonlinear kernels.

Table 1 – Accuracy of the ASD Diagnosis Classification

Feature 1	Feature 2	Accuracy
(i) Jaundice	(ii) Jaundice	0.577
(i) Jaundice	(iii) Residence	0.712
(i) Jaundice	(iv) Screening Score	1.000
(ii) Family with PDD	(iii) Residence	0.750
(ii) Family with PDD	(iv) Screening Score	1.000
(iii) Residence	(iv) Screening Score	0.942

Conclusions

Our observations show that the MD interpretation of ASD can be expected to boost the credibility of the SVM approach to the ASD diagnostic performance. Future work includes search for more suitable parameters than RBF kernels and accuracy for machine learning algorithms for the ASD classification which build upon our own data collection and the DSM-V [4] criteria.

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