Autism Diagnostics, LLC

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Patent application: M19-022L Diagnostic for Childhood Risk of Autism Spectrum Disorder

Problem

Autism now affects 2% of children. It is difficult to screen for, and difficult to diagnose at early ages, and the average age of diagnosis is approximately 4 years, resulting in a major delay in obtaining early intervention services that could greatly improve outcomes if started early.

Currently, diagnosis is based solely on behavioral symptoms, and can require multiple visits over years to confirm.

Current diagnosis based on behavior does not provide any information as to underlying medical problems and how to treat them.

The Solution

- Blood/urine based diagnostic tests based on measurements of several key metabolites using primarily Liquid Chromatography with tandem Mass Spectrometry (LC-MS-MS)
- The test would be submitted for CLIA approval, and made available nationally and internationally.
- **This would be the first biomedical test for diagnosing autism.**
- In addition to diagnosing autism, it would yield levels of 30-50 metabolites which will be important for developing individualized metabolic therapy.
- Repeated testing would help guide treatment and assess treatment efficacy.

"The development of a metabolomics-based test for ASD would be a very helpful tool to accelerate the screening and diagnosis process as well as improve its accuracy in order to identify those children in need of critical services much earlier when interventions are known to be more effective."
- Dr. Richard Frye, MD, PhD, Director of Autism Program at Phoenix Children's Hospital.

Pilot Study Results

Pilot study published in fall 2019 found that our model with only 3 metabolites yielded 94% sensitivity and 100% specificity (sufficient accuracy for commercialization if validated in larger cohort).

Now validating test in larger cohort.

Several proprietary methods to further improve sensitivity and specificity.

Received first place for best start-up opportunity in university-wide competition at Arizona State University in Nov 2019.

Opportunity/ Market

Market Size:

Currently, approximately 1.5 million people in the US with autism.
Approximately 80,000 new cases each year in US.
Worldwide, the numbers are about 20x larger

Customers: Physicians who order tests for patients with autism, or suspected of having autism

First Customers: We would work with many autism groups (Autism Research Institute, Medical Academy of Pediatric Special Needs, etc.) to advertise the tests directly to families and to their physicians. Each autism specialty physician usually treats 1000-3000 patients, so a few early adopters would greatly boost enrollment, and we have good connections to many.

Competitive Advantage

Currently, there is no competitor with a marketed test.

BioRosa is approximately 2-3 years from market with a test based on a subset of the metabolites we measure. It will provide much less guidance to families re. medical interventions.

NeuropointDX has developed tests based on amino acid measurements for diagnosing several subsets of ASD, totaling about 30% of ASD cases.

Our pilot tests yielded 96% sensitivity/specificity, and we believe we can substantially improve those results using several proprietary approaches.

Business Model

Marketing: After CLIA approval, we will begin offering the test. We will also publish research papers on our results, to help advertise the test, and present at many autism conferences for families and physicians.

Scalability: With current equipment we could process about 1,000 samples/month, or 12,000/year. We can easily increase scalability by purchasing additional equipment and hiring additional staff.

Critical Metrics: Retail price of $800, expected profit of $500/test;

Investment to date: $100k Catalyst funding from ASU; over 15 years of studies of biochemistry of autism involving millions of dollars.

Future Investment: We need approximately $100k to finalize our sample collection and test validation for CLIA approval. More funding would allow more rapid expansion of testing capabilities

Team

James Adams – autism researcher
Haiwei Gu – metabolomics expert
Juergen Hahn – biostatistician
Rosa Krajmalnik-Brown: microbiome expert
Tapan Audhya – CLIA consultant (director of a CLIA-approved laboratory and has developed many CLIA-approved tests)

Unique Qualifications: We have invested many years of effort to reach this stage, and are near completion of our sample collection for final development and validation of our diagnostic test. We are prepared for CLIA submission after validation of our model.

Return on Investment

New Funding: We need approximately $100k to complete sample collection and test validation, and if successful we will be able to begin marketing the test within several months after CLIA approval.

Financial Opportunity: Test would retail for $800, and net profit of about $500/test after all expenses.
Year 1: conservatively estimate 5,000 sales: $4 M gross, $2.5 M profit
Year 2: conservatively estimate 10,000 sales: $8M gross, $5 M profit
Year 3: potential for much larger volume

Human Impact: As the first diagnostic test for autism, it could reduce age of diagnosis by several years, resulting in much earlier detection and intervention, and much reduced symptoms in children with autism.
As a personalized treatment guide, it will reveal each child's primary metabolic abnormalities, which in many cases are treatable.

Summary

Today, autism is viewed as a puzzling psychiatric disorder.

In reality, there is very strong data from our group and others that it involves many metabolic abnormalities (low glutathione, high oxidative stress, low sulfate, etc.), and those abnormalities are often treatable (low sulfate, low carnitine, high oxidative stress).

We have the ability and opportunity to develop the first diagnostic test for autism, which will become a vital tool for physicians for both diagnosing autism and for guiding treatment.

Questions?