Rainbow Genomics Whole Genome Sequencing Test Determines Co-Occurring Conditions (Comorbidity) in Autistic Children

Establish a diagnosis by confirming spontaneous (de novo) and challenging mutations in over 1000 newly-reported genes associated with autism spectrum disorder

Evidence-Based Genomic Tests

SAN FRANCISCO, CALIFORNIA, USA, October 19, 2021 /EINPresswire.com/ -- Hong Kong and U.S. based Rainbow Genomics announced today the introduction of a multi-genomic testing strategy for children affected by autistic spectrum disorder. The test is especially appropriate for children presented with co-occurring conditions, also known as comorbidity, including speech delay, intellectual disability, developmental regression, movement abnormality, and seizures.

The strategy is to enable rapid detection of challenging spontaneous mutations, especially variants in over 1000 newly-reported autism genes, and simultaneously confirm if the child is affected by co-occurring conditions that exhibit similar autism symptoms.

1.) Clarifying Co-Occurring Conditions - Over the last 10 years, large-scale genetic research has shown that autism spectrum disorder is caused by multiple genes and environmental factors. Plus, the disorder is often accompanied with overlapping disease conditions. It is often difficult to distinguish which genes are conferring deleterious impacts associated with the common autism symptoms. For example, mutations in the UBE3A gene are associated with autism, but also with Angelman syndrome, a condition that is distinct from autism. However, children affected by Angelman syndrome show similar autistic symptoms such as movement and speech abnormalities, and physicians could misdiagnose the patients as affected by autism.

2.) Detecting Challenging Mutations Associated with Autism - To confirm autism and co-occurring conditions, accurate determination of pathogenic mutations in a large number of genes harboring difficult-to-detect gene variations is critical. Rainbow's multi-genomic testing approach is unique because it enables simultaneous detections of multiple types of genetic

mutations from the whole genome, resulted in rapid clarification of the genetic cause associated with the underlying disorder:

o Copy Number Variants (CNVs) – Copy Number Variants are submicroscopic structural changes in chromosomes that include duplications, deletions, translocations, and inversions. Based on recent worldwide autism research, including studies in Asia, CNVs are important contributing factors in autism susceptibility. Whole genome sequencing with confirmation by RNA and Sanger sequencing, and high-resolution microarray (1.6Kb resolution) testing, allows for the determination of these small- and multi-gene CNVs.

o Spontaneous Genetic (de novo) Mutations – A recent large-scale study using whole genome sequencing data estimated that de novo mutations "contribute to 52%-67% of autism arising from low-risk families (with one child affected), and 30%-39% of cases of all autism" (Communications Biology. 2021, 4:1026).

- These spontaneous, de novo mutations occurred with the child, and are not inherited from the parents

- Rainbow whole genome sequencing test is designed to detect these complex de novo mutations, including intronic, splice site and mosaic mutations, which are challenging to determine.

- Curated Gene List – Rainbow also curated over 1000 newly-reported autism genes with associated annotations to enable CNV and de novo mutation analysis in these novel autism genes.

"My team and I have been a part of the pioneering work in clinical genetics in Hong Kong in the past few decades," said Dr. Stephen Lam, Director of Clinical Genetics Service and Specialist in Paediatrics, at the Hong Kong Sanatorium & Hospital. "We have experienced great success with this comprehensive genomic testing approach in diagnosing our patients with autistic spectrum disorder."

"Detecting de novo mutations, especially in newly-discovered autism genes, and clarifying co-occurring disorders in apparently autistic children will increase diagnostic success," said Daniel Siu, CEO of Rainbow Genomics. "We are very pleased to be able to deploy whole genome, whole exome, transcriptome and Sanger sequencing, coupled with high-resolution microarray testing, for families who would like to receive a diagnosis based on a confirmed genetic etiology, in two months or less."

About Rainbow Genomics

Rainbow Genomics (www.rainbowgenomics.com) is committed to providing clinically-validated genomic testing to Asian, Caucasian, mixed-race, and local minority populations. The company

delivers high diagnostic success for physicians, enabling timely-treatment for patients that can benefit from immediate medical interventions.

Utilizing a multi-technology-platform approach, including whole genome, whole exome, RNA, long-read, methylation, single cell and Sanger sequencing, high-resolution microarray testing, and high-density DNA array genotyping, and through multiple international collaborations, Rainbow Genomics delivers a diagnostic yield meeting or exceeding the highest standards reported by leading U.S. and European medical institutions.

In Asia, Rainbow Genomics provides validated and accredited clinical testing services. For clients in the U.S. and European countries, the company provides “For Research Use” genetic analysis, discovery and confirmatory sequencing services.

All Rainbow Genomics tests are performed in CLIA-certified and CAP-accredited high-complexity clinical laboratories. Patient privacy is protected by Rainbow’s HIPAA-compliant clinical testing process.

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