Rainbow Genomics Launches Retinal Dystrophy (Eye Disorders) Test Using Dual Whole-Exome and 300-Gene Deep Sequencing

Dual Sequencing Approach Determines Both Novel Variants Specific to Asian Patients, and Challenging Mutations Not Easily Detected by Routine Sequencing Methods

SAN FRANCISCO, CA, UNITED STATES, September 20, 2022 /EINPresswire.com/ -- U.S. and Hong Kong-based Rainbow Genomics launches a dual whole exome sequencing and 300-gene "challenging mutation" sequencing test for retinal dystrophy (eye disorders). This test determines the genetic etiologies (genetic causes) associated with a wide range of retinal disorders, including retinitis pigmentosa, cone-rod dystrophy and Leber congenital amaurosis.

The Rainbow Dual Testing Approach:

A. Whole-Exome Analysis - Multiple retinal disorders and their associated pathogenic genetic variants will be reviewed.

• Over 1100 genes are known to be associated with abnormal retinal morphology. Physicians may choose to use exome sequencing, which analyzes over 20,000 genes, as the first-line test to quickly determine the causal gene mutations associated with eye disorder symptoms

• Up-to-the-minute, newly-reported genes are automatically included for analysis

• Copy number, duplication, insertion, deletion and single-nucleotide variant analysis is included

B. 300-Gene "Challenging Mutation" Sequencing determines copy number variants (CNVs) and the following "hard-to-detect" mutations:

- RPGR gene exon 15 (ORF15)
- CACNA2D4 gene AJ, exon 31-38, exon 35 deletions
- CLN3 gene 1Kb deletion
- MAK gene Alu insertion
- TRPM1 gene AJ, exons 227 duplications and deletions

Major Eye Disorders Tested by Rainbow Dual Testing

- Retinitis pigmentosa (RP)
- Cone-Rod Dystrophy (CRD)
- Leber congenital amaurosis (LCA)
- Congenital non-progressive cone-rod synaptic disorder (CRSD)
- Achromatopsia
- Bietti crystalline corneoretinal dystrophy (BCD)
- Bradyopsia
- Chediak-Higashi syndrome
- Choroideremia
- Congenital nystagmus type 1
- Congenital stationary night blindness (CSNB)
- Early and Late-onset Retinal degeneration
- Ectopia lentis
- Enhanced S-cone syndrome
- Familial exudative vitreoretinopathy (FEVR)
- Fundus albipunctatus (FA)
- Gyrate atrophy of choroid and retina
- Isolated microphthalmia
- Juvenile retinoschisis
- Microcornea, myopic chorioretinal atrophy, and telecanthus (MMCAT)
- Microcephaly with chorioretinopathy
- Microphthalmia, anophthalmia, coloboma (MAC) spectrum
- Myopia with cataract and vitreoretinal degeneration
- Neovascular inflammatory vitreoretinopathy (ADNIV)
- Oculocutaneous albinism
- Optic atrophy
- Persistent hyperplastic primary vitreous (PHPVAR)
- Snowflake vitreoretinal degeneration
- Stargardt disease
- Sveinsson chorioretinal atrophy (SCRA)
- Vitelliform macular dystrophy (VMD1) and autosomal recessive bestrinopathy
- An additional 1000 disorders (not listed) associated with abnormal retinal morphology are also tested using whole exome sequencing

Benefits of dual whole exome sequencing and gene-specific deep sequencing approach:

- Novel variant analysis - Recently discovered gene variants, usually not covered by gene panel testing (100-900 genes), will be included by whole exome sequencing (20,000 genes)

- Ethnic-specific deep analysis - Chinese, East Asian and South Asian specific variants, often not

reported (or with conflicting classifications) in ClinVar and other international databases, will be carefully reviewed by Rainbow's clinical teams

For more information, please visit Rainbow Genomics website or contact us at info@rainbowgenomics.com

About Rainbow Genomics

Rainbow Genomics (www.rainbowgenomics.com) is committed to providing clinically-validated genomic and proteomic 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 proteomics, 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.

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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