Deep Clinical Assessment with Whole Genome Sequencing Enables Diagnosis of Adult Unexplained Abnormalities

Successful Genetic Assessment Reduces Misdiagnoses and Supports Early Interventions to Prevent Adult Premature Mortality

SAN FRANCISCO, CALIFORNIA, UNITED STATES, March 6, 2022 /EINPresswire.com/ -- Age-related chronic diseases account for the majority of adult premature mortality. Cancers and cardiovascular disorders represent over half of the causes of premature death for both males and females 50-75 years of age, with diabetes, respiratory, neurological and liver diseases accounting for the majority of the remaining deaths.

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>
> *Dr. Stephen Lam*

Most of the adult chronic diseases are asymptomatic at onset, and are often reported as unexplained abnormalities by routine health examination. Although environmental factors and lifestyles affect the onset of these diseases, many of these disorders are associated with genetic causes. Adult unexplained abnormalities caused by genetic disorders are difficult to diagnose using standard-of-care procedures because their symptoms often overlap with other common disorders, and some individuals who carry the pathogenic mutations develop the disease while others do not.

Monogenic and Polygenic Assessment for Patients Affected by Unexplained Symptoms

The genetic risk of developing these chronic or unexplained disorders can be monogenic (caused by a mutation in a single gene) or polygenic (conferred by variations from multiple genes). Whole genome sequencing coupled with comprehensive clinical assessment can provide relatively high diagnostic success in determining these monogenic and polygenic disease causes.

The approach may be useful in diagnosing unexplained but persistent symptoms such as pains, headaches, heart palpitations, repeated infections, paralysis, numbness, and tingling, etc. When test results for these symptoms are all normal, considerations of genetic testing may be warranted.

Clinical Deep Phenotyping

To diagnose and treat a wide-range of unexplained illnesses, the first step is “Deep Phenotyping”. The comprehensive clinical assessment of unexplained impairments, organ-specific abnormalities, and family histories along with laboratory testing are performed by physicians who specialize in genomic medicine prior to genetic testing. The systematic collection of these clinical symptoms (phenotypes) substantially affects the likelihood of a successful genomic analysis leading to a confirmation of genetic causes.

Whole Genome Sequencing Test

Using whole genome sequencing, the entire human genome is analyzed, covering over five million monogenic mutations and polygenic variants, associated with 6000 monogenic disorders and multiple common diseases. Coupled with deep phenotyping, the test substantially improves the diagnosis of adult debilitating disorders.

“Genetic disorders contribute to a significant proportion of adult morbidity and mortality. Diagnosing adult genetic diseases is challenging because these disorders exhibit variable clinical features that overlap with common disease comorbidities, and are often missed by differential diagnosis. Many patients have undergone extensive testing, and remain undiagnosed or misdiagnosed, and without appropriate treatment,” said Dr. Stephen Lam, Director of Clinical Genetics Service at the Hong Kong Sanatorium & Hospital. “Whole genome sequencing of adults with suspected hereditary conditions may provide a rapid diagnosis when standard-of-care testing could not return a definitive finding."

“We are pleased to provide whole genome sequencing with simultaneous monogenic, polygenic, and pharmacogenomic analysis to improve clinical outcome.” Said Daniel Siu, CEO of Rainbow Genomics,” By working with “super specialists” to perform deep phenotyping, we are able to deliver genome-scale analysis that leads to the diagnosis of unexplained abnormalities. And many of these genetic findings are medically-actionable.”

About The Rainbow Whole Genome Sequencing Test

The test provides five clinical reports:

1. Personal Health Assessment

- Cancers
- Cardiovascular Disorders
- Endocrine System Abnormality including Diabetes and Fatty Liver
- Sleep Disorders
- Hearing and Eye Disorders
- Skeletal and Bone Disorders

- Immune System Disorders
- Skin Abnormality
- Auto-inflammatory Disorders
- Behavioral Abnormality
- Neurological Disorders including Dementia and Alzheimer Disease

2. Reproductive Health Assessment

- Male Infertility
- Female Infertility

3. Whole Genome Carrier Status

- Carrier Screening Analysis of Disorders Recommended by the American College of Obstetricians and Gynecologists and the American College of Medical Genetics

- Whole Genome Carrier Analysis - Mutations in 2500 genes associated with recessive disorders that can be passed on to the patient's children

4. Common Disease Risk Assessment

- Common complex diseases such as diabetes, heart attack, and stroke are primarily caused by changes in multiple gene variants (polygenic risks). However, these variants are highly ethnic-specific. Polygenic risk assessment for ethnic-specific groups such as Asians is difficult because very few publications using Asian patients and healthy controls with statistical significance are available.

- Rainbow Genomics' expert-curated polygenic variants are highly ethnic-specific, and are supported by large-scale genome wide association and replication studies using tens of thousands of patients and controls collectively from Asia, U.S. and European countries.

5. Pharmacogenomic Assessment of 185 Medications to Improve Clinical Outcome

- For chronic disorders, achieving risk-reduction targets through improving medication adherence is critical. Pharmacogenomic assessment enables high therapeutic efficacy through minimization of drug side effects, resulting in better treatment outcome.

About Rainbow Genomics

Rainbow Genomics 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.

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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