Whole Genome Sequencing Test Enables Diagnosis of Growth Abnormality and Sex Development Disorders in Children

Affordable First-Tier Testing for Patients Affected by Disorders of Sex Development, Growth Hormone Deficiency, Short Stature, and Early Puberty

SAN FRANCISCO, CALIFORNIA, UNITED STATES, September 8, 2022 /EINPresswire.com/ -- U.S. and Hong Kong-based Rainbow Genomics expands the use of whole genome sequencing and whole exome sequencing tests for diagnosis of children affected by growth and sex development disorders.

Monogenic mutations leading to disruption of hormonal regulations or aberrant development of crucial organs and tissues like gonad, adrenal gland, thyroid gland, or hypothalamo-pituitary tissue have been unraveled since the 1980s. These abnormalities affect the normal growth of children. As a result, growth and sex development disorders are common reasons for referrals to pediatric endocrinologists.

However, with an increasing number of causal genes responsible for growth and sex development disorders being discovered and often redefined, practicing physicians' mastery of such a long and evolving list of implicated genes becomes the top challenge for these precision medicine practitioners. And yet, most genetic test providers still rely on testing of a small number of genes, leading to low diagnostic success.

To meet these challenges, Rainbow Genomics provides the Rainbow Pedi 1000 (TM) Whole Genome Sequencing/Whole Exome Sequencing Test of over 20,000 genes, with multi-omics confirmation (Sanger, RNA and long-read sequencing), that may increase the diagnostic yield by detecting novel genes, and determining challenging mutations in these newly-reported genes. Genetic counselors with clinical genetics training also provide extensive physician support to improve the understanding of the implications and prognosis associated with specific pathogenic mutations.

This approach may be suitable for the following disorders:

• Disorders of sex development - These are congenital conditions in which the development of chromosomal, gonadal and anatomical sex is atypical, including micropenis, cryptorchidism (testes fail to descend into the scrotum), hypospadias at birth (opening of the urethra is not

located at the tip of the penis), labia majora fusion and ambiguous genitalia. Obtaining a diagnosis is important for treatment considerations and also for reaching sex of rearing decisions.

• Precocious puberty, also known as early puberty - Secondary sexual development could occur too early in girls and boys. Affected children may have inappropriately strong body odor, acne and/or pubic hair before age 7 to 8 years, and be taller than their peers, but their abnormal growth would ultimately stop too early. As a result, these individuals are often shorter than their peers in adulthood.

• Growth hormone deficiency - A clinical condition caused by a shortage of growth hormone, and may be congenital or genetic in origin. Depending on the severity of the deficiency, the affected newborn baby could have a small penis, low blood glucose or prolonged jaundice during the infancy. Other symptoms may become apparent by early childhood, such as height progressively falling off the growth reference centiles. Affected individuals commonly experience a failure to grow at expected rate and have unusually short stature.

• Disorders of growth hormone and insulin like growth factor (IGF) system - Short children not deficient in growth hormone secretion could be suffering from inadequate IGF-1 either because of an impaired growth hormone response pathway, or even defects to uphold appropriate IGF-1 levels (normally regulated by growth hormone) and its growth response.

• Other growth disorders could be related to hidden bone growth and development defects – These conditions may not clinically be suspected because skeletal defects are subtle, yet some patients could be responsive to growth hormone treatment when diagnosed. New drugs beyond growth hormone and IGF-1 have been developed as targeted growth promoting agents. As such, definitive genetic diagnosis will be crucial in correctly choosing such therapy in the future.

Managing growth and sex development abnormalities can be emotionally difficult for children, and may lead to psychological and behavioral problems. A genetic diagnosis determines possible co-morbidities, guides future endocrine and imaging tests, and avoids unnecessary treatment. The diagnosis also allows appropriate genetic counselling and, in many cases, supports initiation of early therapy.

“With a child affected by growth or sex development disorders, a definitive genetic diagnosis is invaluable because it brings psychological reassurance to the young patients, a relief for their families, and relevant knowledge for genetic counseling of parents contemplating future pregnancies. In many patient cases, prognosis and treatment strategy are also clarified when the genetic etiology is determined.” Said Daniel Siu, CEO of Rainbow Genomics,” At a comparable cost of testing a few genes, whole exome sequencing may be considered as an effective first-tier diagnostic or rule-out test by clinicians, especially when initial endocrinology and phenotypic examination failed to return a diagnosis.”

About The Rainbow Pedi 1000(TM) Whole Genome Sequencing and Whole Exome Sequencing Tests

1. Diagnostic Report - This is a diagnostic analysis based on the child's symptoms and family history, utilizing over 20,000 genes from the whole genome or whole exome. Pathogenic single nucleotide, splice, intronic, mosaic, and copy number variants are reported. Variant of uncertain clinical significance (VUS) will also be reported, and VUS with some evidence of pathogenicity will be specifically highlighted.

2. Secondary Finding Report - If consented by the patient or parents, secondary findings, which are pathogenic mutations in genes associated with over 1000 childhood on-set diseases and treatable metabolic disorders, will also be reported.

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 US 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.

DANIEL SIU
Rainbow Genomics
+852 3481 0977
email us here