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# Claritas Genomics announces launch of Claritas Clinical Exome, novel diagnostic test

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Claritas Genomics, Inc., announced the launch of its *Claritas Clinical Exome,* at the Annual Meeting of the American Society of Human Genetics running 10/6 – 10/10 in Baltimore, MD. This novel diagnostic test is the first to exploit the strengths of multiple DNA sequencing platforms simultaneously to deliver confirmed results for a patient's clinical whole exome within 4 weeks, compared to the industry standard of 12 weeks or more. Patients with rare disease typically seek a diagnosis for 8-12 years, receiving 3-5 separate tests at a high cost.

The sequential testing approach can now be replaced by a single test. The Claritas approach takes advantage of an innovative dual-capture, dual-platform method that immediately confirms more than 90% of all genetic variants in the exome, facilitating rapid analysis, interpretation, and return of clinically-relevant results. The *Claritas Clinical Exome* encompasses approximately 97% of the entire exonic coding region of the genome at >20X coverage (mean coverage of 100x) by the Illumina NextSeqTM and immediate confirmation on the Life Ion ProtonTM Platform.

"Claritas is focused on delivering answers to help children affected by genetic disorders, and do so rapidly, cost effectively, and at the highest quality standards possible," said Patrice Milos, President and CEO of Claritas Genomics. "The *Claritas Clinical Exome* we launched today offers patients and caregivers the opportunity to engage in pediatric precision medicine as routine clinical care."

**Multiple Options for Clinical Care**

The *Claritas Clinical Exome* is designed for complex pediatric presentations in which overlapping phenotypes, the patient's symptoms, make selection of a defined region of interest gene set, or panel test, difficult. If clinical presentations are clear, the coverage and accuracy of the *Claritas Clinical Exome* gives healthcare providers the option to choose focused *Regions of Interest* tests from a set of pre-defined gene lists, thereby replacing lengthy and expensive sequential gene panel testing. The first gene sets available today cover pediatric neurology, which targets conditions including neuromuscular disorders, epilepsy and seizures. Claritas will launch other tests later this year. If the *Region of Interest* test result is inconclusive, healthcare providers can choose to have their patient's whole exome analyzed.

"Many clinical providers want the focus, coverage, accuracy and price of a small gene panel with the options and flexibility to quickly look more comprehensively at the whole exome if the smaller gene set does not provide answers" said Scott Pomeroy, MD, PhD, Neurologist-in-Chief and Chair of the Department of Neurology at Boston Children's Hospital. "It is highly desirable to look first at a small gene set that is closely tied to clinical presentation on an exome platform and then have the option to look beyond those genes if the results are inconclusive."

Given the incomplete knowledge of the variants underlying rare disease, even the most advanced gene panels will yield inconclusive results for many patients. In these more difficult cases, healthcare providers can have access to Claritas' partner WuXi NextCODE's Clinical Sequence Analyzer™, which facilitates new insight into patients' disorders by providing unrivalled power for systematically scanning the entire exome sequence to identify novel and de novo disease-causing mutations. This integrated solution offers a full range of sophisticated yet intuitive interpretation and data visualization tools, making it possible to determine causal variants in a much higher proportion of cases than panels alone, thus providing healthcare providers and a much greater number of patients with critical information and recommendations for guiding treatment and care.

**About WuXi NextCODE**

WuXi NextCODE offers uniquely comprehensive and integrated capabilities for using the genome to better diagnose disease and create better medicine. These include a full range of sequencing services through our CLIA-certified laboratory; the world's leading genome interpretation system; a novel database architecture that makes it possible to query, manage, store and share massive genomic data with unrivalled speed and efficiency; and the know-how and experience to apply genomics to optimize every aspect of drug discovery and development. With offices in Shanghai; Cambridge, Massachusetts; and Reykjavik, Iceland, we enable clinicians and researchers at institutions and companies worldwide to use the full power of the genome to deliver precision medicine and support health.

WuXi NextCODE is a wholly owned subsidiary of WuXi AppTec, the operating subsidiaries of WuXi PharmaTech (NYSE: WX).

**About Claritas Genomics**

Claritas Genomics serves children affected with complex genetic disorders by providing timely and accurate results, resolving families' long search for answers. By combining clinical expertise of the world's best pediatric specialists with innovative platform solutions, Claritas is working to improve patient care and enable new discoveries.

Source:
www.claritasgenomics.com