GeneDx Adds Buccal Swab as Non-Invasive Whole Genome Sequencing Sample Collection Option

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New Option Enables Easier DNA Sample Collection from Biological Parents to Aid in Disease Diagnosis

STAMFORD, Conn., May 04, 2023 (GLOBE NEWSWIRE) -- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic and clinical insights, today announced the availability of its GenomeXpress® and GenomeSeqDx® whole genome sequencing tests with buccal swabs as an alternative sample collection option for biological parents or other immediate family members. Sequencing biological parent genomes alongside patient genomes – known as trio analysis - aids in disease diagnosis and greatly increases diagnostic yield rates.

"We are continuously looking for ways to broaden adoption of genome sequencing and facilitate convenient access to families to aid in disease diagnosis," said Paul Kruszka, M.D., Chief Medical Officer at GeneDx. "Research shows diagnostic rates are highest when we can include genomic data of biological parents to classify variants of unknown significance based on inheritance patterns. Adding buccal swab as an additional sample collection method for our GenomeSeqDx and GenomeXpress whole genome sequencing tests can make it easier for providers to collect parent samples for trio testing."

Buccal swab is a convenient, non-invasive method to collect DNA from cells found inside a person's cheek. In the case of trio testing, the diagnostic yield for positively identifying a disease-causing variant increases from 19% to 30%.[1] In addition to its whole genome sequencing tests, GeneDx also makes buccal swab available as an alternative DNA collection method for its XomeDx® and XomeDxXpress® whole exome sequencing tests for patients and biological parent samples.

"We continue to turn to GeneDx for whole genome sequencing testing to accurately identify pathogenic variants that explain our patients' illnesses," said Tara Lynn Wenger, M.D., Ph.D., Associate Professor in the Department of Pediatrics at the University of Washington and Associate Medical Director for Inpatient Genetics Services at Seattle Children's Hospital. "Collecting DNA from biological parents or other relatives is not always so easy. Buccal swab DNA collection for parents will streamline the process and prevent delays in testing and enable us to do a more thorough whole genome analysis."

About GeneDx GenomeSeqDx and GenomeXpress Whole Genome Sequencing
GenomeSeqDx and GenomeXpress clinical whole genome tests by GeneDx include evaluation and analysis of both the protein-coding and non-coding regions of the human nuclear genome, allowing for the broadest potential detection of characterized/pathogenic variants contributing to the molecular basis of a genetic disorder in an affected individual. Detecting and characterizing variants that may contribute to the molecular basis of a genetic disorder is most effective when at least one or both biological parents are included in the analysis. Several large, evidenced-based studies have demonstrated that genome sequencing identifies a causal variant in more than 40% of cases, with higher yields for cases that specifically include samples from family members for analysis.[2,3,4]

About GeneDx
GeneDx (Nasdaq: WGS) delivers personalized and actionable health insights to inform diagnosis, direct treatment and improve drug discovery. The company is uniquely positioned to accelerate the use of genomic and large-scale clinical information to enable precision medicine as the standard of care. GeneDx is at the forefront of transforming healthcare through its industry-leading exome and genome testing and interpretation, fueled by one of the world's largest, rare disease data sets. For more information, please visit http://www.genedx.com and connect with us on LinkedIn, Twitter, Facebook, and Instagram.

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[1] Data on file.

[2] Manickam K, McClain MR, Demmer LA, et al. Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2021 Nov;23(11):2029-2037. doi: 10.1038/s41436-021-01242-6. Epub 2021 Jul 1.

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[4] Dimmock D, Caylor S, Waldman B, et al. Project Baby Bear: Rapid precision care incorporating rWGS in 5 California children's hospitals demonstrates improved clinical outcomes and reduced costs of care. Am J Hum Genet. 2021 Jul 1;108(7):1231-1238. doi:10.1016/j.ajhg.2021.05.008.

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