Genomic Profiling

Clinical Utility

Next generation sequencing can provide clinicians with a diagnosis for diseases that would otherwise be impossible to diagnose. This test is designed to identify alterations that underlie genetic forms of diabetes and hyperinsulinism.

Obtaining a genetic diagnosis can provide life changing results for patients and their families by directing treatment options or providing risk assessment for family members and future pregnancies. Additionally, patients found to have genetic forms of diabetes or hyperinsulinism may be able to participate in research studies (or clinical trials) aimed at understanding their disease or offering new therapies.

Tests developed in concert with endocrinologists, medical geneticists, and researchers at Washington University School of Medicine in St. Louis.

Next-generation sequencing for efficient, cost-effective and robust germline variant detection.

Concise, expert interpretations by board-certified clinical genomicists, reported back to the ordering physician.

Professional consultation available to physicians in result interpretation, and other technical and clinical considerations.

Testing covered by most insurance; preauthorization performed by GPS laboratory personnel.

Orderable disease focused gene sets include:

- Monogenic Diabetes and Maturity Onset Diabetes of the Young (MODY) – 43 genes
- Permanent Neonatal Diabetes Mellitus (PNDM) - 29 genes
- Endoplasmic Reticulum (ER) Stress – 5 genes
- Hyperinsulinism – 18 genes
Genetic Diabetes & Hyperinsulinism Testing
At Washington University

Genes Tested

Endoplasmic Reticulum (ER) Stress Gene Set (5 genes) - CISD2, EIF2AK3, IER3IP1, INS, WFS1

Hyperinsulinism Gene Set (18 genes) - ABCC8, AKT2, CACNA1D, FOXA2, GCK, GLUD1, HADH, HNF1A, HNF4A, INSR, KCNJ11, KOM6A, KMT2D, PGM1, PMM2, SLC16A1, TRMT10A, UCP2

Monogenic Diabetes and Maturity Onset Diabetes of the Young (MODY) Gene Set (43 genes) - ABCC8, AGPAT2, AIRE, AKT2, APPL1, BLK, CEL, CISD2, CP, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HFE, HNF1A, HNF1B, HNF4A, IER3IP1, INS, INSR, KCNJ11, KLF11, LRBA, MNIK, NEUROD1, NEUROG3, NXX2-2, PAX4, PAX6, PCBD1, PDX1, PLAG1, PPARG, PTF1A, RFX6, SLC2A2, SLC19A2, STAT3, TRMT10A, WFS1, ZFP57

Permanent Neonatal Diabetes Mellitus (PNDM) Gene Set (29 genes) - ABCC8, CP, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1B, IER3IP1, INS, KCNJ11, LRBA, MNIK, NEUROD1, NEUROG3, NXX2-2, PAX6, PCBD1, PDX1, PLAG1, PPARG, PTF1A, RFX6, SLC2A2, SLC19A2, STAT3, TRMT10A, WFS1, ZFP57

Testing Methodology
Tests are performed using targeted hybridization capture coupled with next-generation sequencing (NGS) in our CAP/CLIA labs for comprehensive coverage of all coding exons of ordered genes. Types of variation detected include single nucleotide variants (SNVs) and small insertions and deletions (indels).

Results and Interpretation
DNA sequence data are analyzed by GPS’ clinically validated bioinformatics pipeline to identify and annotate genetic variants associated with diabetes and hyperinsulinism.

Variants are interpreted by a board-certified clinical genomicsist in the context of the patient’s disease. Those that are most likely to account for the observed clinical phenotype based on evidence from the medical literature are highlighted. Results are returned to the ordering physician in a concise report.

Specimen Requirements
Specimen types accepted include 2-5 mL peripheral blood in a lavender-top EDTA tube. Specimen kits are available upon request.

Please contact us or fill out the NGS supply order form available on our website at gps.wustl.edu/forms-and-resources.

Turnaround Time
The turnaround time for testing and interpretation is four to six weeks from the time a specimen arrives.

Ordering
To order a test, submit a completed requisition form (available at gps.wustl.edu/forms-and-resources) by fax or email. GPS performs insurance preauthorization.

Ancillary Testing
Deletion/Duplication analysis via CGH (comparative genomic hybridization) for the HNF1B gene is performed as a send out for the Monogenic Diabetes and MODY Gene Set and the Permanent Neonatal Diabetes Mellitus (PNDM) Gene Set when NGS is negative.

Selected References


Contact us to order a test of for more info
Tel: (314) 747-7337
Toll Free: (866) 450-7697
Fax: (314) 747-7336

Email: gps@wustl.edu
Website: gps.wustl.edu

Address:
Cortex Building, Suite 302
4320 Forest Park Ave.
St. Louis, MO 63108