

WASHINGTON UNIVERSITY PATHOLOGY SERVICES

DIABETES, HYPERINSULINISM, AND ER STRESS DISORDERS

Genetics and Genomic Services offers innovative testing for neonatal and monogenic diabetes, hyperinsulinism and endoplasmic reticulum (ER) stress disorders, such as Wolfram Syndrome. Testing was developed in collaboration with leading endocrinologists, medical geneticists, and researchers at Washington University School of Medicine – a highly collaborative group available for challenging cases and novel variant assessments.

EXPERT CONSULTATION AND DIAGNOSIS

- Next-generation sequencing (NGS) for efficient, cost-effective testing designed to arrive at timely and accurate diagnoses
- Concise, expert interpretations by experienced faculty board-certified in clinical genomics
- Consultation for results interpretation and other technical/clinical considerations

COMPREHENSIVE DIAGNOSTICS

- Exome capture NGS for comprehensive and deep coverage of all coding exons of ordered genes with robust detection of single nucleotide variants (SNVs) and small insertions and deletions (indels)
- Discrete orderable gene panels organized according to clinical presentations consistent with neonatal hyperinsulinism, pediatric diabetes, or phenotypes associated with ER Stress disorders
- Exome-level data for reanalysis on indeterminate cases, or those with negative findings, as indicated
- Reflex send out testing for deletion/duplication analysis via aCGH, as clinically appropriate

SPECIMENS ACCEPTED

- 2 to 5 mL peripheral blood in a lavender-top EDTA tube
- 2 to 4 buccal swabs
- Specimen kits available upon request

FOR MORE INFORMATION OR TO ORDER A TEST, CONTACT:

Phone: 314-747-7337

Email: gps@wustl.edu

Web: pathologyservices.wustl.edu



DIABETES AND ER STRESS TEST MENU

- Atypical Diabetes and ER Stress Disorders Panel (54 genes)
- Endoplasmic Reticulum (ER) Stress Disorders Panel (5 genes)
- Hyperinsulinism Panel (18 genes)
- Permanent Neonatal Diabetes Mellitus (PNDM) Panel (29 genes)
- Monogenic Diabetes and MODY Panel (44 genes)

STREAMLINED ORDERING, REPORTING AND BILLING

- Four weeks average turnaround time
- Reporting can be done via fax or electronically
- Assistance with the insurance prior authorization process and determination of patient out of pocket costs for most insurances



Pathology & Immunology