

WASHINGTON UNIVERSITY PATHOLOGY SERVICES

MOSAICISM DISORDERS TESTING

Genetics and Genomic Services offers innovative testing for definitive diagnoses of a diverse group of phenotypes and syndromes that include somatic overgrowth, vascular anomalies, atypical nevi, and other skin lesions often in combination with other skeletal or soft tissue anomalies. Our laboratory is a global leader in mosaicism disorders diagnostic testing, which requires a combination of highly sensitive variant detection and expert variant assessment in this rapidly evolving area of disease.

EXPERT CONSULTATION AND DIAGNOSIS

- High depth 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
- Ongoing collaboration with Washington University genetics and dermatology physicians, whose input contributes to accurate variant assessment and case consultation

COMPREHENSIVE DIAGNOSTICS

- Targeted hybrid 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 subsets organized by presenting phenotype
- Assistance in determining the diagnostic specimen with the highest potential for detection of typically low-allelic fraction variants

SPECIMENS ACCEPTED

- Affected tissue in fresh or FFPE form
- Peripheral blood for the IEI panel
- 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



MOSAICISM DISORDERS TEST MENU

- Cortical Malformation and Epilepsy Panel (39 genes)
- Inborn Errors of Immunity Panel (10 genes)
- Maffucci Syndrome Panel (2 genes)
- McCune Albright Syndrome (5 genes)
- Nevus Panel (28 genes)
- *PIK3CA*-Related Overgrowth Spectrum (PROS) (1 gene)
- Rasopathies Panel (26 genes)
- Somatic Overgrowth Panel (49 genes)
- Somatic Undergrowth Panel (6 genes)
- Vascular Anomalies Panel (65 genes)
- *Comprehensive 75 gene reflex panel available for *PIK3CA* and Custom orders*

STREAMLINED ORDERING, REPORTING AND BILLING

- Three 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