DISORDERS OF SOMATIC MOSAICISM (DoSM) 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 DoSM 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), small insertions and deletions (indels)
- Discrete orderable subsets organized by presenting phenotype, with options for focused or comprehensive analysis
- Assistance in determining the diagnostic specimen with the highest potential for detection of typically low-allelic fraction variants
- Sanger sequencing on comparator specimen for the determination of germline, multi-tissue mosaic or somatic status of variants identified in primary affected tissue

FOR MORE INFORMATION OR TO ORDER A TEST, CONTACT:

Phone: 314-747-7337
Email: gps@wustl.edu
Web: pathologyservices.wustl.edu

SPECIMENS ACCEPTED

- Primary affected tissue in fresh or FFPE form
- Secondary peripheral blood comparator
- Specimen kits available upon request

DoSM TEST MENU

- Somatic Overgrowth Panel (25 genes)
- PIK3CA-Related Overgrowth Spectrum (PROS) (1 gene)
- McCune Albright Syndrome (1 gene)
- Nevus Panel (12 genes)
- Maffucci Syndrome Panel (2 genes)
- Rasopathies Panel (14 genes)
- Curry-Jones Syndrome (1 gene)

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 insurers