SOMA Gene Set

Genomic profiling for Segmental Overgrowth, McCune Albright and related syndromes

**Next-generation sequencing** for efficient and cost-effective somatic variant analysis; germline variants also detected

**Concise, expert interpretations** by board-certified clinical genomicists

**Expert consultation** available to physicians in result interpretation and in other technical/clinical considerations

**Testing covered by most insurance:** preauthorization performed by GPS

**Clinical utility**

The **SOMA Gene Set** is designed to identify patients with diagnostic genetic mutations that underlie segmental overgrowth, McCune Albright and related syndromes.

The **PI3K/AKT/mTOR pathway** is critical in regulating cellular proliferation, mobility and survival. Variation in genes of this pathway can result in several disorders characterized by a wide range of phenotypes often making it difficult to make a **definitive diagnosis**.

Most commonly, disorders characterized by these phenotypes are a result of **somatic variants** - those occurring in only a proportion of the body's cells - rather than germline variants that are found in all cells of the body. As a result, testing for these disorders is strongly recommended from affected tissue.

Next-generation sequencing provides clinicians with a **powerful tool** to manage patients with these diagnostically challenging disorders.
Indications for testing

Indications for referral include clinical features of the following:

- Bannayan-Riley-Ruvalcaba syndrome (BRRS)
- Cowden syndrome
- Congenital, lipomatous, overgrowth, vascular malformations, epidermal nevi and scoliosis/skeletal/spinal anomalies (CLOVES)
- Hemimegalencephaly
- Klippel-Trenaunay syndrome (KTS)
- McCune Albright syndrome (MAS)
- Macrocephaly–capillary malformation (M-CM)
- Maffucci Syndrome Gene Set: IDH1 and IDH2.
- Megalencephaly-polymicrogyria-polydactyly-hydrocephalus (MPPH)
- Proteus syndrome
- Schimmelpenning-Feuerstein-Mims (Epidermal Nevus Syndrome)
- Rasopathies Gene Set: BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RIT1, SHOC2, SOS1 and SPRED1.
- Somatic Overgrowth Gene Set: AKT1, AKT2, AKT3, BRAF, FGFR1, GNA11, GNAQ, HRAS, IDH1, IDH2, KRAS, MAP2K1, MAP3K3, MTO1, NRAS, PDGFRB, PIK3CA, PIK3R1, PIK3R2, PSEN1, RASA1, SMO, TEK, TSC1 and TSC2.
- Specimen requirements

Preferred specimen types include tissue from the affected area. In addition, 2-5 mL of peripheral blood in a lavender-top EDTA tube is also required to allow comparative study.

Acceptable materials for submission include disease involved tissue in the form of a formalin fixed paraffin embedded (FFPE) tissue block, fresh tissue in transport or tissue culture media or buccal swab. Acid decalcified samples and heparinized blood are not acceptable.

Kits for testing on peripheral blood and buccal cells are available upon request.

Turnaround time

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

Ordering

To order a test, submit a completed requisition form (available on our website) by fax or email. GPS performs insurance preauthorization. In the case of archival specimens, GPS coordinates sample acquisition. If surgical pathology materials have to be requested from outside BJC HealthCare, include the release form. Please contact us for more information.

Selected references


Contact us to order a test or for more info

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