ALBERTA PRECISION LABORATORIES
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Gastrointestinal Stromal Tumor Panel: Information for Ordering Providers

Gastrointestinal stromal tumors (GISTs) are uncommon tumors that arise predominantly in the stomach and small intestine.[1] GISTs account for 0.1%-3.0% of gastrointestinal malignant tumors with a mean age of diagnosis of 60 years.[1,2] GISTs may also be benign. Features suggestive of a hereditary GIST cancer predisposition include[1]:

- Multiple tumors
- Hyperpigmented patches of skin in various areas
- Hyperplasia of other cells in the gastrointestinal tract
- Relatives affected with the above spanning multiple generations

The syndromes represented on this panel are inherited in an autosomal dominant manner. The actual risk to develop gland dysfunction or an endocrine tumour is dependent upon the gene involved and the penetrance.

Individuals who carry a pathogenic variant in a tumour predisposition gene have an increased risk of developing certain tumours compared to the general population. Tumour risks depend on the gene(s) in which the variant(s) is identified. These individuals are eligible for increased screening and/or risk reducing surgeries and therapeutic interventions. In addition, results may influence treatment plans for individuals with cancer.

If a pathogenic variant is identified in one of these genes, the patient and/or their family members may be at increased risk for specific cancers or other conditions. Genetic counselling is recommended for these families.

Indications for testing

Patients with a personal and/or family history suggestive of a predisposition to gastrointestinal stromal tumours are eligible for testing.

Ordering privileges

This panel may be ordered by Clinical Geneticists.

GIST NGS panel

Gene(s)	Associated cancers and/or clinical features[1,3]	Associated Hereditary Syndrome[3]
SDHA, SDHB, SDHC, SDHD	Head and neck paragangliomas, extra-adrenal paragangliomas and/or pheochromocytomas, GISTs and renal clear cell carcinoma (rare)	Hereditary paraganglioma/ pheochromocytoma syndrome
PRKAR1A	Skin pigment abnormalities, myxomas, endocrine tumours, schwannomas, GISTs	Carney complex
KIT	GISTs, acute myeloid leukemia, seminomas, mastocytosis	N/A

Associated Disorders[1,3]

Some of the genes on this panel are associated with other rare disorders including:

***KIT*-related hematopoietic neoplasms** such as core binding factor acute myeloid leukemia

Piebaldism is an autosomal dominant disorder characterized by patches of skin and hair that entirely lack pigment. It is caused by pathogenic variants in *KIT.*

Mitochondrial complex deficiencies are rare autosomal recessive conditions with highly variable phenotypes. Pathogenic variants have been reported in *SDHA, SDHB,* and *SDHD.*

When can I expect results?
Results may take up to 4 months.

How are results reported?
Results are sent to the ordering provider and available in Netcare and Connect Care.

Contact Information
Genetic Counsellors, Genetics & Genomics
Calgary: 403-955-3097

Requisition forms, contact information and other resources can be found at:
http://ahsweb.ca/lab/if-lab-genetics-and-genomics

References

1. National Library of Medicine (US). Genetics Home Reference [Internet]. Bethesda (MD): The Library; 1993-2018 [cited 2017 Dec]. Available from: https://ghr.nlm.nih.gov/

2. Kameyama, H., Kanda, T., Tajima, Y., Shimada, Y., Ichikawa, H., Hanyu, T., ... & Wakai, T. (2018). Management of rectal gastrointestinal stromal tumor. *Translational gastroenterology and hepatology*, *3*.

3. Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018 [cited 2017 Dec]. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1116/