

# Cquence

## GIST Genotyping Panel

### Test type

NGS panel test

### Background

Cquence GIST Genotyping Panel is designed to classify GI stromal tumors in a molecular level, which can predict the response of kinase inhibitor therapy and, in some cases, the optimal treatment dose. The panel has a sensitivity of  $\leq 10\%$  mutant allele.

The GIST panel delivers information on a variety of treatment-informative mutations in twenty three genes commonly involved in GI stromal tumors<sup>1-9</sup>, covering all of the hotspot mutation sites in the genes listed below.

### Test specifications

Test code	Methodology	Specimen requirements	Turnaround time
OKG	Next-generation sequencing	10 FFPE unstained sections (6 $\mu\text{m}$ ) with at least 20% tumor content	10-14 days

### Genes targeted (23 genes)

*AKT1, AKT2, AKT3, ATM, BRAF, CDKN2A, HRAS, KIT, KRAS, MAP2K1, NF1, NRAS, PDGFRA, PIK3CA, PTEN, PTPN11, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD* and *TP53*

### References

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3. Heinrich et al. *Correlation of kinase genotype and clinical outcome in the North American Intergroup Phase III trial of imatinib mesylate for treatment of advanced gastrointestinal stromal tumor: CALGB 150105 Study by Cancer and Leukemia Group B and Southwest Oncology Group.* *J Clin Oncol* 26:5360-7, 2008.
4. Heinrich et al. *Kinase mutations and imatinib mesylate response in patients with metastatic gastrointestinal stromal tumor.* *J Clin Onc* 21:4342-4349, 2003.
5. Debiec-Rychter et al. *Use of c-KIT/PDGFR $\alpha$  mutational analysis to predict the clinical response to imatinib in patients with advanced gastrointestinal stromal tumours entered on phase I and II studies of the EORTC Soft Tissue and Bone Sarcoma Group.* *Eur J Cancer* 40:689-95, 2004.
6. Janeway et al. *Defects in succinate dehydrogenase in gastrointestinal stromal tumors lacking KIT and PDGFRA mutations.* *Proc Natl Acad Sci U S A.* 2011 Jan 4;108(1):314-8.
7. Pantaleo et al. *Analysis of all subunits, SDHA, SDHB, SDHC, SDHD, of the succinate dehydrogenase complex in KIT/PDGFR $\alpha$  wild-type GIST.* *Eur J Hum Genet.* 2013 Apr 24. doi: 10.1038/ejhg.2013.80. [Epub ahead of print]
8. Corless et al. *Gastrointestinal stromal tumors: origin and molecular oncology.* *Nat Rev Cancer.* 2011 Nov 17;11(12):865-78.
9. Nannini et al, Pantaleo MA. *An overview on molecular biology of KIT/PDGFR $\alpha$  wild type (WT) gastrointestinal stromal tumors (GIST).* *J Med Genet.* 2013 Jul 5.