«TableStart:Samples»

Sample: «sample» Name: «patient» DOB: «dob» URN: «urn»

**GASTROINTESTINAL STROMAL TUMOUR CANCER MUTATION ANALYSIS** «isdraft»

**SPECIMEN**

extref

**PATHOLOGY**

Histological typing: ...........................

The sample was reviewed by a pathologist and was considered to have ....% tumour cells within the area selected for analysis. Please note: This is not a formal pathology review and is based solely on an H&E of the tissue provided and not on ancillary clinical or pathology information that may be available elsewhere.

«TableStart:Variants»

**RESULT**

**Gene** «gene»

**Reference** «refseq»

**cDNA Change** «hgvsc»

**Protein Change** «hgvsp»

**Read Depth** «varreaddepth»/«totalreaddepth» «afpct»%

«TableEnd:Variants»

**TEST DESCRIPTION**

Tumour DNA was tested in duplicate for mutations in exons 9, 11, 13, 17 and 18 of the KIT gene, and exons 12, 14 and 18 of the PDGFRA gene using massively parallel sequencing. This test detects single nucleotide variants and indels in the target exons only. At 1000x coverage, the limit of detection of this assay has been determined to be X%. At 500x coverage the limit of detection has been determined to be X%. The sample was sequenced to an average «ampReads» aligned reads per amplicon with «ampPct»% uniformity. Regions with less than 100x coverage have not been analysed. These are listed below.

**INTERPRETATION**

«TableStart:Variants»«gene»: «mut»

«TableEnd:Variants»

**COMMENTS**

Mutations in KIT or PDGFA are observed in ~85 % of GIST tumours and are associated with response to tyrosine kinase inhibitors such as imatinib. KIT exon 11 is the most commonly mutated exon. Tyrosine kinase inhibitor resistance mutations are located in KIT exons 13 and 17 (1).

Note: Testing of tissue treated with chemo and/or radiotherapy reduces the cellularity of the neoplastic element and reduces the sensitivity of the assay. Where possible tissue derived from untreated tumour should be tested.

**REFERENCES**

1. Lasota, J. and M. Miettinen, Histopathology, 2008. 53(3): p. 245-66.

Low coverage amplicons:

«lowAmps»

Assay region of interest coverage:

«rois»

*«TableEnd:Samples»*