«TableStart:Samples»

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

**MELANOMA 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.

**RESULT**

**FAILED SAMPLE**

**TEST DESCRIPTION**

Tumour DNA was tested in duplicate for mutations in exons 11 and 15 of the BRAF gene, exons 11, 13, 17 and 18 of the c-KIT gene, and exons 2 to 4 of the NRAS 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**

This sample failed to meet predetermined measures of quality/quantity for this assay. DNA has been scheduled for re-analysis by an alternative method. These results will follow.

**COMMENTS**

The BRAF V600E mutation occurs in 40 to 60% of malignant melanomas and confers sensitivity to the targeted small molecule inhibitor vemurafinib (1). The NRAS gene is mutated in 20% of malignant melanomas and may be amenable to treatment with downstream MAPK and PI3K inhibitors (2). The KIT gene is mutated in 17% of cutaneous melanomas, 11% of acral melanomas and 21% of mucosal melanomas. Patients with KIT mutations benefit from imatinib therapy (3).

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. Flaherty, K.T., et al. New Engl J Med, 2010. 363(9): p. 809-819.

2. Kelleher, F.C., McArthur, G.A., Cancer J., 2012. 18(2): p. 132-136.

3. Hodi, F.S., et al., Journal of Clinical Oncology, 2013. 31(26): p. 3182-3190.

Low coverage amplicons:

«lowAmps»

*«TableEnd:Samples»*