

Example of genetic report from Scotland

Tissue sample reports

Example 1:

Reason for referral: Patient is undergoing investigations for lung cancer. Molecular analysis requested.

Conclusion: **No clinically actionable variant detected (see interpretation).**

Results and interpretation:

Sequence analysis of DNA extracted from the pathology specimen from this patient did not detect a clinically actionable sequence variant in EGFR exons 18-21, KRAS codons 12, 13, 59, and 61 or BRAF codon 600.

This result indicates that this patient is unlikely to respond to treatment with EGFR-tyrosine kinase inhibitors, KRAS-G12C inhibitors or BRAF/MEK inhibitors.

Please note sequence analysis has detected the KRAS variant c.35G>T p.(Gly12Val) (G12V). No licensed targeted therapy for this variant is currently available for lung cancer patients.

Comments:

The neoplastic cell content was estimated to be 40%. This specimen has not been macrodissected.

Please also refer to the pathology report associated with this pathology specimen (as noted in Ref Lab Number field above).

In order to avoid error and/or misinterpretation, transcription of the content of this report is not advised; Laboratory Genetics do not take any responsibility for the accuracy of any data/text transcribed from this report.

Example 2:

Reason for referral: Patient is being investigated for a possible Myeloproliferative Neoplasm.

Conclusion: **JAK2 variant c.1849G>T p.(Val617Phe) NOT detected.**

Results and interpretation:

Testing of DNA from this patient did not detect the pathogenic JAK2 variant c.1849G>T p.(Val617Phe).

This result does not support a diagnosis of a Myeloproliferative Neoplasm in this patient.

This result should be interpreted in the context of all clinical, morphological, and haematological findings.

Comments:

This assay will only detect the presence or absence of the c.1849G>T p.(Val617Phe) variant and can not detect other molecular causes of Myeloproliferative Neoplasms.

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Technical Information:

Allele specific PCR to detect the JAK2 variant c.1849G>T p.(Val617Phe) was performed. We estimate that the level of detection for the tests performed is >5%. References: Bench et al, British Journal of Haematology, 2012, 160, 25-34.