AWMGS
All Wales Medical Genomics Service

Non-Small Cell Lung Cancer (NSCLC) – *EGFR* Testing on ctDNA

Background

The recent development of circulating tumour DNA (ctDNA) technologies has presented us with new opportunities to improve the stratification of NSCLC patients for treatment. ctDNA is shed into the patient's blood stream from the tumour, the ctDNA is representative of the tumour and therefore presents a viable alternative to an invasive procedure for molecular analysis for EGFR variants.

1. NSCLC patients with either no biopsy sample available, or where the biopsy sample has been exhausted or has failed molecular analysis. EGFR tyrosine kinase inhibitors (TKIs) have now been licensed for treatment with an EGFR variant detected using ctDNA.
2. Second-line NSCLC patients treated with EGFR TKI, in whom a sensitising EGFR variant has previously been detected. For these patients the p.Thr790Met (known as T790M) resistance variant will be analysed to determine treatment. The T790M variant is estimated to be present in 50% of EGFR TKI-treated patients; those patients with the T790M variant have been shown to be sensitive to third-generation EGFR TKIs.

Recommended Clinical Referral Criteria

- Patients with NSCLC suitable for treatment with biological agent that meet one of the two criteria detailed above.

Molecular Analysis

Variant screen: Droplet digital PCR analysis of deletions in EGFR exon 19, EGFR c.2573T>G p.(Leu858Arg), and c.2369C>T p.(Thr790Met). The analysis will detect ~0.5% of variant in a background of wild-type genomic DNA

Prices* & Turnaround Times (TAT)

*Prices available on request

Requirements	TAT (calendar days)
Analysis for deletions in EGFR exon 19, EGFR variants c.2573T>G p.(Leu858Arg) and c.2369C>T p.(Thr790Met)	7

Contact Details

All Wales Genomics Laboratory,
Institute of Medical Genetics,
University Hospital of Wales,
Heath Park,
Cardiff CF14 4XW
Tel: 029 2184 4023 / **Fax:** 029 2184 4043

lab.genetics.cav@wales.nhs.uk
http://www.medicalgenomicswales.co.uk

Sample Requirements

10-20ml blood in either CellSave© or Streck© tube to reach the laboratory within 24 hours; it is recommended that samples are not taken whilst patient is receiving chemotherapy. ***NB: 20ml blood preferred to maximise test sensitivity.***
Please label samples with three identifiers and date of collection.
All samples must be accompanied by request form
Consent for testing & DNA storage is assumed when request for test received.
Websites for tubes:
www.streck.com www.janssen.com

Links

Orphanet	OMIM	Genetic Testing Registry	Support
http://www.orpha.net/	http://www.omim.org/	http://www.ncbi.nlm.nih.gov/gtr/	British Lung Foundation http://www.blf.org.uk/Home Roy Castle Lung Cancer Foundation http://www.roycastle.org