



NewGene Clinical Services

Utilising the latest technologies to deliver molecular diagnostics and genomic services, NewGene is able to offer significant benefits leading to improved clinical delivery:

- **TURNAROUND** - Clinically relevant turnaround times
- **RESPONSE** - Emerging clinical need can be met with rapid development of new tests
- **SAVINGS** - The high throughput capacity of the technology gives rise to savings in both time and cost
- **QUALITY** - An excellent track record in external quality assessment
- **FLEXIBILITY** - NewGene can develop a bespoke service to meet your specific needs.

All results and interpretation are reported by an HCPC registered Clinical Scientist.

NewGene also works closely with the NHS and pharmaceutical companies on the validation of biomarkers and the development of diagnostic tests.

Personalised Medicine

RAS testing in colorectal cancer

Erbix is used as part of the first line therapy for patients with metastatic colorectal cancer with a wild type *RAS* tumour. *RAS* wild type status is defined as showing no mutation in exons 2, 3 and 4 of the *KRAS* and *NRAS* genes. Activating mutations prevent the effect of Erbix and can be used to predict resistance to treatment.

- NewGene assay includes identification of 23 *KRAS*, 23 *NRAS* and 1 *BRAF* mutation
- Testing for patients in all settings is funded by Merck-Serono
- 90% of all samples are reported within a 5 day turnaround time.

EGFR mutation testing in non-small cell lung cancer

Gefitinib and Erlotinib are cancer therapies that inhibit the EGFR protein, disrupting the EGFR signaling pathway. As a result they are only effective in those patients whose cancer is caused by *EGFR* activating mutations. The NewGene *EGFR* assay covers all mutations related to drug sensitivity.

- Assay includes 17 point mutations in *EGFR*
- Deletions involving exon 19 and insertions in exon 20 are also investigated
- 90% of all samples are reported within a 5 day turnaround time.

BRAF (V600E) testing

The drug Vemurafenib has been approved for treatment of patients with malignant melanoma with a V600E (c.1799T>A) mutation in the *BRAF* gene. NewGene offers screening for this *BRAF* mutation.

Gastro-Intestinal Stromal Tumours

Gastro-intestinal stromal tumours (GIST) affect the connective and supportive tissues of the digestive tract or nearby structures within the abdomen. GIST are rare, accounting for approximately 2% of tumours in the gastro-intestinal region and are highly resistant to chemotherapy. In many cases the cause is found to be activating mutations in the *cKIT* and *PDGFRA* genes and clinical evidence demonstrates that the mutation status can predict the patient response to tyrosine kinase inhibitors such as Imatinib.

The test is a two step screen targeting the most frequently mutated parts of the *cKIT* and *PDGFRA* genes:

- Step 1: exon 9 and exon 11 of *cKIT* and exon 18 of *PDGFRA*
- Step 2: exons 8, 13 and 17 of *cKIT* and exons 12 and 14 of *PDGFRA*.

Together these mutations account for 80-95% of all known mutations in GIST.

Predictive testing for adverse drug reactions – TPMT

Polymorphisms in the thiopurine methyltransferase (*TPMT*) gene give rise to variation in the activity of the *TPMT* enzyme. Reduced *TPMT* activity can lead to adverse patient reaction when treated with thiopurine drugs (occurring in 10-28% of patients receiving the treatment).

These drugs are commonly prescribed across a range of disorders and disciplines that include: leukaemia, post-transplantation, dermatology, gastroenterology, rheumatology, and autoimmune diseases such as Crohn's disease and lupus.

The assay detects 4 SNPs within the *TPMT* gene, which are responsible for 80-95% of deficient *TPMT* activity.

Sample type for Personalised Medicine

NewGene is able to extract DNA from a range of clinical sample types:

- Formalin fixed paraffin embedded tissue curls (5 x 10 μ m curl)
- Formalin fixed paraffin embedded tissue blocks
- EBUS samples
- Cytology samples - bronchial alveolar lavage
- Fine needle aspirates (FNA)
- Samples fixed stained and covered on a slide
- Small biopsy samples
- EDTA blood (*TPMT* testing).

Tissue must contain at least 30% of tumour material.

For more information visit our website:

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