



## NewGene - Pioneering the application of new technologies in molecular diagnostics

The demand for testing of genetic variation in patients is growing year on year. This is being driven by advances in medical genetics coupled with the growth in personalised medicine, which involves the determination of a patient's biological characteristics in order to select the most effective therapy regime.

Established through a partnership between the Newcastle Hospitals NHS Foundation Trust and Newcastle University, NewGene is a pioneer in developing, validating and delivering molecular diagnostics using the latest high throughput sequencing and genotyping.

### Integrated service provision

NewGene's core team has a background in molecular genetics within the NHS. By combining this clinical and laboratory expertise with the use of high throughput sequencing platforms, NewGene is able to deliver a high quality, fast turnaround service at an attractive price. NewGene works in collaboration with clinicians and scientists to deliver optimal services to NHS Trusts and overseas healthcare providers.

### Broad portfolio of tests

NewGene has a portfolio of tests for clinically significant inheritable disorders and for personalised medicine diagnostics for somatic mutations in cancers that are delivered using its high throughput DNA sequencing platforms.

### Personalised medicine

- KRAS / BRAF combined test for colorectal cancer
- EGFR test for non-small cell lung cancer
- BRAF V600E in melanoma
- TPMT screening for adverse reactions in acute lymphocytic leukaemia
- cKIT / PGDPR for gastro-intestinal stromal tumors.

### Hereditary diseases

- BRCA1 and BRCA2 full gene sequencing for breast cancer
- RASopathies testing
- aHUS genotyping to detect hereditary haemolytic uraemia

### Haemato-oncology

- JAK-2 and MPL testing in myeloproliferative disease
- BCR-ABL monitoring for patients with chronic myeloid leukaemia
- CML mutation screening
- Clonality testing
- Diagnostic testing
  - V600E in BRAF for Hairy Cell Leukaemia
  - D816V in cKIT for Mastocytosis

## Noonan spectrum test: RASopathies

Introduced in collaboration with St George's Healthcare NHS Trust, London, NewGene offers a comprehensive diagnostic test for all Noonan spectrum disorders, also known as RASopathies.

These conditions are inheritable diseases that cause delayed development in children and are associated with a wide range of medical problems, including heart defects, speech and hearing issues and growth and behavioural problems. The range and severity of health complications can vary significantly and the syndromes are not always identified at an early age. Once suspected, it can take up to 18 months for a definitive diagnosis to be made using currently available techniques.

The NewGene test simultaneously screens all coding regions and splice sites of 11 genes and a single exon of one further gene for mutations causative of all the RASopathies. This comprehensive test replaces the current standard multi-stage regime, reducing time to diagnosis and cost and increasing the likelihood of identifying the molecular basis of the disease.

## Atypical Haemolytic Uraemia Syndrome

In collaboration with the Northern Genetics Service, Newcastle Hospitals NHS Foundation Trust, NewGene has developed a comprehensive diagnostic test for atypical haemolytic uraemia syndrome (aHUS) and associated disorders.

Inherited renal disorders are characterised by progressive renal failure, anaemia and vascular problems. The causative mutations for many disorders have been identified in the genes that encode the various components and regulators of the alternative complement pathway.

NewGene has developed a single test covering the most common mutations. Five key genes are sequenced in parallel, enabling rapid and low cost diagnosis.

Simultaneous analysis of five key genes:

- *CFH* (factor H)
- *CD46* (membrane cofactor protein)
- *CFI* (complement factor I)
- *C3* (complement factor C3)
- *CFB* (complement factor B)

Mutations in the genes listed above are found in 60% of aHUS cases.

## Research services

In addition to its clinical services, NewGene combines its expertise in the field of genetics with its latest sequencing technologies to offer DNA sequencing and genotyping services to research clients, principally academia and pharmaceutical companies.

For more information visit our website:

[www.newgene.org.uk](http://www.newgene.org.uk)

**Telephone: +44 (0) 191 242 1923**

**Email: [info@newgene.org.uk](mailto:info@newgene.org.uk)**

