NewGene
Next Generation Diagnostics

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.

Hereditary Disorders

Applying the parallel sequencing capability of next generation sequencing to the clinical setting enables rapid diagnosis and timely decisions on appropriate treatment pathways.

Hereditary Breast Cancer - *BRCA 1* and *BRCA 2*

Over 40,000 new cases of breast cancer are recorded each year in the UK. Most of these arise from sporadic mutations, however up to 10% are due to inherited mutations in breast cancer susceptibility genes. Carriers of *BRCA 1* and *BRCA 2* mutations are at increased risk of developing breast and other cancers. NewGene has developed and validated a cost effective full gene sequencing assay for *BRCA 1* and *BRCA 2*, comprising:

- Full gene sequencing across the coding regions and intron / exon boundaries of both *BRCA 1* and *BRCA 2*
- Multiplex Ligation Dependent Probe Amplification (MLPA) to detect deletions and insertions.

Noonan spectrum test – comprehensive screening for RASopathies

Disruption to the regulation of the RAS-mitogen activated protein kinase signaling cascade has been found to be the common molecular basis for several genetically heterogeneous multiple anomaly syndromes with overlapping phenotypes. In collaboration with SW Thames Molecular Genetics Diagnostics Laboratory, St George's Healthcare NHS Trust, London, NewGene has developed a

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comprehensive diagnostic test for all Noonan spectrum disorders (also known as RASopathies):

- Noonan syndrome
- Noonan-like syndrome disorder with or without juvenile myelomonocytic leukaemia (NSLL)
- Noonan-like syndrome with loose anagen hair (NSLH)
- Cardio-facio-cutaneous syndrome (CFC)
- Costello syndrome
- LEOPARD syndrome (Multiple lentigines syndrome)
- Legius syndrome (Neurofibromatosis type 1-like syndrome).

The NewGene Noonan spectrum test simultaneously screens all coding regions and splice sites of 11 genes and a single exon in an additional gene for the mutations causative of all the RASopathy disorders. 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.

Genes included in the assay:
PTPN11, BRAF, SOS1, RAF1, KRAS, HRAS, NRAS, SHOC2 (exon 1 only), *CBL, SPRED1, MAP2K1, MAP2K2*.

Renal disorders - 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. In collaboration with the Northern Genetics Service, Newcastle Hospitals NHS Foundation Trust, NewGene has developed a comprehensive diagnostic test for aHUS and associated disorders.

The tests includes 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 these genes are found in 60% of aHUS cases. While the disorder membrano-proliferative glomerulonephritis (MPGN II) is frequently related to homozygous Factor H deficiency.

Confirmation of the mutation status of all positive samples for hereditary disorders will be carried out by bi-directional Sanger sequencing.

For more information visit our website:
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