Genomics Research Centre Diagnostic Testing

Director: Distinguished Professor Lyn Griffiths

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NATA/RCPA accreditation: 14979

Costs quoted include analysis and interpretation for specified disorders along with preparation and sending of report to referring clinician or pathology laboratory.

The GRC is NATA accredited to provide sequencing diagnostic services for the indications as outlined below. Sequencing services for other conditions employ identical methods but are not NATA accredited. Please contact the GRC directly to enquire about sequencing for other conditions.

Next Generation Sequencing – Whole Exome Sequencing

The GRC is NATA accredited to undertake Whole Exome Sequencing for identification of genetic mutation for the following disorders:

- Familial Hemiplegic Migraine type 1 (FHM1)
- Familial Hemiplegic Migraine type 2 (FHM2)
- Familial Hemiplegic Migraine type 3 (FHM3)
- CADASIL, CARASIL and small vessel diseases
- Episodic Ataxia
- Spinocerebellar Ataxia
- Epilepsy
- Neuromuscular Disorders

Cost: $1100 per sample (including GST)

Timeframe: 6-8 weeks from sample receipt

Next Generation Sequencing – Targeted Gene Panel

The Targeted Gene Panel uses NGS technology to simultaneously interrogate fifteen genes that are implicated in neurogenetic disorders. This approach is cost-effective in cases where causative mutation may exist in a number of known genes which can be analysed concurrently reducing the need for consecutive (and costly) testing.

Cost: $580 per sample (including GST)

Timeframe: 6-8 weeks from sample receipt

Gene	Exon Coverage	Gene	Exon Coverage	Gene	Exon Coverage
CACNA1A	partial coverage for exons 17, 44 and 46	SCN2A	complete coverage	*HRTA1*	complete coverage
ATP1A2	partial coverage for exons 13, 23, 3'UTR	KCNA1	complete coverage	*TREX1*	complete coverage
SCN1A	partial coverage for exons 1, 2, 17, 3'UTR	PRRT2	complete coverage	*FOXC1*	complete coverage
NOTCH3	partial coverage for exon 26	ATP1A3	partial coverage for exon 22	*COL4A1*	partial coverage for exons 19 and 25
TRESK	complete coverage	GLA	partial coverage for exon 22	*COL4A2*	complete coverage

Targeted Mutation Testing

NATA accredited Sanger sequencing method is used for targeted mutation testing of single exons in the genes listed, or a known mutation point for any of our accredited disorders. This approach is cost-effective for a suspected specific genetic mutation.

Cost: $220 per sample per exon (including GST)

MTHFR C677T Polymorphism - $79.20 per sample (including GST)

Timeframe: 6-8 weeks from sample receipt

Gene	NATA accredited Indications
CACNA1A	FHM1; EA2; SCA6
ATP1A2	FHM2
SCN1A	FHM3; Epilepsy
NOTCH3	CADASIL
MTHFR	MTHFR C677T Polymorphism
Other Gene	FHM, EA, SCA, Epilepsy, small vessel disease, other accredited disorder

How to Order

1. Discuss testing with the patient
2. Complete a patient consent form for Specialised/DNA Testing and provide a copy to us with your request
3. Offer the patient genetic counselling as required – Genetic Counselling services can be located on the HGSA website https://www.hgsa.org.au/asgc/asgc
4. Complete the Request form – available on the GRC website: https://research.qut.edu.au/grc/diagnostic-testing/diagnostic-testing-pricing/
5. Collect and send sample
    a. 5mls whole EDTA blood or 5mls of blood in lithium Heparin is required sent on ice for testing;
    OR
    DNA extracted from 10mls blood collected in EDTA or lithium heparin.
    Please indicate:
        1. Storage conditions of the extracted DNA - TE or Water
        2. Method of DNA extraction
        3. 260/280 ratios (acceptable range 1.6-2.1)
        4. 260/230 ration (acceptable range 1.7-2.3)
    b. Specimen labelling must include:
        - patient first name and surname
        - patient date of birth
        - date and time of collection
        - patient signature; and
        - collector's initials
    c. Send samples packed on ice by courier the same day or overnight to:

Genomics Research Centre (GRC) Clinic
Centre for Genomics and Personalised Health
Queensland University of Technology
5 School Street (loading bay)
Kelvin Grove, Brisbane, QLD
AUSTRALIA, 4059

Please note: Any specimens arriving at the GRC after seven (7) days from collection date will be rejected

Availability of Clinical Advice

The GRC may give advice on the nature of any mutations identified and their likely effect on protein production. Where information is available in the scientific literature, clinical advice regarding the nature of genetic variations can also be provided.

Protection of Personal Information

The GRC is committed to protecting the personal identification of the patients using the service. Thus, personal information, including the discussion of results will only take place with the patient themselves, clinicians authorised on the initial request, or those subsequently authorised in writing by the patient or authorised clinicians.