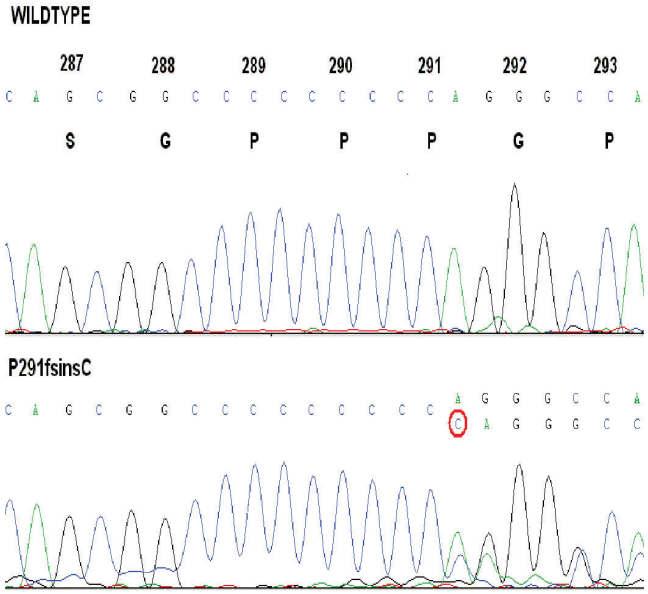
TESTING PROCEDURES

All testing is performed by amplifying the target gene by PCR, followed by capillary sequencing of the DNA product. We examine the coding regions (exons) as well as the adjacent splice sites, in order to facilitate the detection of both common mutations and "private" (rarer) mutations. Mutation detection rates vary from 80 - 95% of affected alleles depending on the gene being studied. Evaluation of intronic regions and distant gene promoters is not cost effective at present.

An example of a sequencing test showing a mutation is given below



PRICING POLICIES

There is currently No Medicare Rebate available on these tests. For pricing information please contact us 07 3840 8500.

ADDRESS TO SEND SPECIMENS

DNA Analysis
Attention: Ivan McGowan
C/o Central Specimen Reception
Pathology Department - Level 6, Mater Adult Hospital
Raymond Terrace
South Brisbane Q 4101.

We require 4 mL EDTA blood or, alternatively, genomic DNA for analysis. Relevant clinical information is necessary for optimal interpretation of results. Parental DNA samples may also be required in some cases. The turnaround time for results is 4 - 8 weeks. Urgent samples can usually be processed in 1 - 2 weeks.

CONTACT PERSONS FOR DISCUSSION

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David.Cowley@mater.org.au
Dr Gary Leong MBBS, FRACP, PhD.
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Mr Ivan McGown BSc (Supervising Scientist).
Ivan.Mcgown@mater.org.au

For enquiries about test availability and results
Phone: 07 3840 8500
Fax: 07 3840 8338

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NATA ACCREDITED FOR TECHNICAL COMPETENCE

RCPA The Royal College of Pathologists of Australasia

NQS INTERNATIONAL CERTIFIED QUALITY MANAGEMENT SYSTEM ISO9001

Mater Pathology

Paediatric Endocrine Genetic Testing Service

Performed exclusively by Mater Pathology

MATER PATHOLOGY

21-Hydroxylase gene analysis for CONGENITAL ADRENAL HYPERPLASIA

We have defined several common 21 hydroxylase gene mutations in the Australian population that occur in 95% of subjects with CAH. Our analysis includes gene copy number evaluation. Samples from the index case are studied in full to identify the causative mutation in that family, and this is then confirmed by analysis of the parents DNA. Subsequent predictive tests of the remaining family members (including the foetus, where applicable) for the specific mutation are then possible with a rapid turnaround time.

Androgen Receptor (AR) gene analysis for ANDROGEN INSENSITIVITY SYNDROME (AIS)

We are able to identify mutations in the AR in 95% of subjects with the complete AIS phenotype, and 15% of those with Partial AIS. The analysis includes sequencing of the tandem repeat portions of the gene.

PIT-1, PROP-1 and Neurophysin 2 (AVP) gene analysis for Congenital Hypopituitarism

We have defined mutations in the PIT-1 gene in Australian children with congenital hypopituitarism, presenting with GH, TSH and PRL deficiency.

More recently we have identified mutations in the Neurophysin 2 (AVP2) gene in subjects with familial Diabetes Insipidus. Identification of the mutation in a family allows early diagnosis and confirmation of the condition.

Sulphonyl-urea receptor (SUR)-1, KIR 6.2, glucokinase, and glutamate dehydrogenase (GDH) gene analysis for PERSISTENT HYPERINSULINISM OF INFANCY (PHHI)

We have shown that mutations in the SUR-1 gene can be identified in 75% of subjects with severe PHHI - these severe cases require pancreatic surgery. In subjects with the typical GDH phenotype, gene analysis is confirmatory in most cases.

MODY 3 (HNF1), MODY 2 (Glucokinase), MODY 1 (HNF 4), and KIR6.2 gene analysis for MATURITY ONSET DIABETES OF THE YOUNG (MODY)

Gene analysis is available in children and their families with a strong family history of type 2 diabetes, including for MODY 3 (HNF1), MODY 2 (Glucokinase) and MODY 1 (HNF 4).

Submission of a relevant genetic family tree is important in MODY cases, and this should include information about the type and age of onset of diabetes. Referring physicians will be asked to complete a patient profile (obtainable from the Mater Pathology website) before genetic testing is carried out: www.mater.org.au/pathology

The detection of MODY3 and some other MODY forms has clinical therapeutic relevance (e.g. use of oral hypoglycaemic agents), and in some cases acts as a guide to screening for other congenital abnormalities (e.g. renal problems).