Yorkhill Division
West of Scotland
Regional Genetics Service

Institute of Medical Genetics
Yorkhill Division
Dalnair Street
Glasgow, G3 8SJ
Tel: 0141 201 0377

NHS
Greater Glasgow and Clyde

SCN1A mutation screening for infantile onset epilepsies

This is a joint clinical epileptology and molecular genetic service providing sequencing of all 26 exons of SCN1A and MLPA (looking for large scale rearrangements) in selected sequencing negative cases. The test is performed within a National Health Service Clinical Pathology Accredited (CPA) Lab and reporting times conform to national standards.

Testing is undertaken when a completed referral form accompanies the DNA/blood sample. The clinician managing the individual's epilepsy may be best placed to complete the form. Completion of the form allows us to prioritise samples, this is necessary for quality control purposes, audit and is essential for the provision of a joint clinical and molecular genetic service. The likelihood of the laboratory identifying a mutation is dependent upon the phenotype of the affected individual. In the first 18 months of the service the overall mutation detection rate has been approximately 30% and in classical SMEI / Dravet Syndrome cases between 80-90%.

Prenatal testing is not offered routinely. Cases can be discussed on an individual basis.

Scottish Referrals – The service is funded directly by the National Services Division and there is no direct cost to the referrer.

UK referrals (outside Scotland) – In view of differing funding arrangements within the NHS in England, Wales and Northern Ireland the charge for the service has to be passed through the referrer. The charge for sequencing the gene is £650.00. There is no additional charge for MLPA. Please supply details of who the invoice should be forwarded to (see below).

Referrals from outside the United Kingdom – The service can accommodate a certain number of samples. The charge is £650.00. Referrers should communicate with us prior to sending samples.

Referring Clinician Name & Institution:

Name:	Institution Address:

(Please circle one of the following)

Child Neurologist Adult Neurologist General Paediatrician
Other Epilepsy Specialist (specify) Clinical Geneticist

Affected Individuals Name and Date of Birth:

Is the test for: Affected individual or carrier status? (Please indicate name and mutation of affected relative)

EPILEPSY PHENOTYPE: *Please delete as appropriate and provide details where necessary:*

Age at first epileptic / febrile seizure	*Details:*
Any factors precipitating first seizure?	*Details:*
Prolonged febrile seizures	**YES / NO** *Age of onset:*
Hemi-clonic or focal febrile seizures	**YES / NO** *Age of onset:*
Hemi-clonic or focal afebrile seizures	**YES / NO** *Age of onset:*
Did different focal seizures affect the opposite side	**YES / NO** *Details:*
Myoclonic seizures	**YES / NO** *Age of onset:*
Generalised tonic-clonic / clonic seizure	**YES / NO** *Age of onset:*

 Please add additional sheets if insufficient space for comments.

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Focal seizures with impairment of awareness	**YES / NO** *Age of onset:*
Atypical absences	**YES / NO** *Age of onset:*
Status epilepticus	**YES / NO** *Details:*
Obtundation status	**YES / NO** *Details:*
Has there been a prolonged seizure free period?	**YES / NO** *If yes, how long for?*
Epileptic encephalopathy	**YES / NO** *Details:*
Any other comments on seizure types:	*Details:*

DEVELOPMENT:

	YES (details)	**NO**
Normal prior to epilepsy onset		
Cognitive decline following epilepsy onset		
Acquired autistic features		
Behaviour problems		

Acquired motor / movement disorder? *(Please circle one of the following)* hypotonia, ataxia, spasticity, dyskinesia.

Is the movement disorder static or paroxysmal? Details:

Current developmental status: *(please circle one of the following)*

Normal **Mild** **Moderate** **Severe** **Profound Learning disability**

Age at which development noted to be abnormal

Continued development regression or plateau at a certain age?

EEG FEATURES: *(Please provide details)*

Please state date of EEG & abnormalities	
Normal interictal EEG	
Generalised spike & wave	
Photosensitivity (any EEG with date)	
Focal / multifocal EEG abnormalities	
Additional comments: Did EEG change over time?	

MRI: *(Please provide details)*

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MEDICATION:

Have any medications increased seizure frequency?

Any medications reduced seizure frequency?

FAMILY HISTORY: (Please provide details/pedigree of family history of febrile seizures or epilepsy in space provided)

SYNDROMIC DIAGNOSIS: *(Please circle one of the following)*

- Severe myoclonic epilepsy in infancy / Dravets Syndrome
- Borderline severe myoclonic epilepsy (SMEB)
- Intractable childhood epilepsy with generalised tonic clonic seizures
- Specific epilepsy syndrome within a family with generalised epilepsy and febrile seizures plus (GEFS+). (Please draw pedigree and state syndromes in space above)
- Other Syndrome- please specify
- Unclassified epilepsy

COMMENTS: (Please provide any other relevant details in space provided below)

INVOICE DETAILS: (Samples from outside Scotland should provide details of billing address below)

* Clinical queries can be directed to Dr Sameer Zuberi, Consultant Paediatric Neurologist (sameer.zuberi@ggc.scot.nhs.uk)
* Queries relating to technical aspects of the mutation analysis can be directed to Rachael Birch (rachael.birch@ggc.scot.nhs.uk).

DNA or whole blood (2-5ml in an EDTA tube) should be forwarded to Rachael Birch, DNA Lab, Duncan Guthrie Institute of Medical Genetics, Royal Hospital for Sick Children, Yorkhill, Glasgow, United Kingdom, G3 8SJ. Tubes must be labelled with name and date of birth.

 Please add additional sheets if insufficient space for comments.