

# TEST REQUEST FORM GENE PANEL



\* Mandatory fields

## PATIENT DETAILS

Forename\* \_\_\_\_\_ Surname\* \_\_\_\_\_  
Hospital ID \_\_\_\_\_ DOB (DD/MM/YYYY)\* \_\_\_\_\_  
Biological Sex\* \_\_\_\_\_

## HEALTH PRACTITIONER DETAILS

Account ID \_\_\_\_\_ Address 1 \_\_\_\_\_  
Full Name\* \_\_\_\_\_ Address 2 \_\_\_\_\_  
Phone \_\_\_\_\_ City/town \_\_\_\_\_  
Email \_\_\_\_\_ Post Code \_\_\_\_\_  
Institution\* \_\_\_\_\_ Country \_\_\_\_\_

## TEST DETAILS

Gene Panel to be analysed\*<sup>1</sup> : \_\_\_\_\_  
\_\_\_\_\_

<sup>1</sup>Please refer to the Gene Panel list on page 3 for available Gene Panels, and refer to our Gene Panel webpage for the contents of each gene panel. Gene panels are based on Whole Exome Sequencing data. Only genes on the panels are analysed after applying a computational filter (*in silico* analysis).

All pathogenic and likely pathogenic variants identified on genes on the panel will be reported. Variants classified as a variant of uncertain significance (VUS) will not standardly be reported. A VUS identified in a clinically relevant gene may be reported as a supplemental finding, where there is a high level of evidence supporting pathogenicity, and where further family history, familial testing or phenotypic evidence may help re-classify the variant. Likely benign and benign variants will not be reported. Single heterozygous variants in genes associated with recessive inheritance may not be reported.

## CLINICAL INFORMATION

Referral Reason\*<sup>2</sup> \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

<sup>2</sup>Detailed clinical information significantly improves the interpretation of identified variants. Please, use HPO terms, when possible: <https://hpo.jax.org/app>

## SAMPLE DETAILS<sup>3</sup>

Sample Type:  Whole Blood (EDTATube)  Genomic DNA, Source: \_\_\_\_\_  
 Saliva (Oragene Tube)  Buccal Swab  
Date Collected (DD/MM/YYYY)\*: \_\_\_\_\_ Time Collected (hh:mm): \_\_\_\_\_

<sup>3</sup>Please refer to our Laboratory User Guide for information on the sample requirements for this test.

## INTERNAL USE ONLY

Sample ID \_\_\_\_\_

# TEST REQUEST FORM GENE PANEL



\* Mandatory fields

## PATIENT'S FAMILY HISTORY

### Mother's Ancestry

- |   |   |   |
|---|---|---|
| <input type="checkbox"/> Admixed American             | <input type="checkbox"/> Ashkenazi Jewish   | <input type="checkbox"/> European (non-Finnish) |
| <input type="checkbox"/> African/African American     | <input type="checkbox"/> East Asian         | <input type="checkbox"/> Middle Eastern         |
| <input type="checkbox"/> Amish                        | <input type="checkbox"/> European (Finnish) | <input type="checkbox"/> South Asian            |
| <input type="checkbox"/> Other, Please specify: _____ |   |   |

### Father's Ancestry

- |   |   |   |
|---|---|---|
| <input type="checkbox"/> Admixed American             | <input type="checkbox"/> Ashkenazi Jewish   | <input type="checkbox"/> European (non-Finnish) |
| <input type="checkbox"/> African/African American     | <input type="checkbox"/> East Asian         | <input type="checkbox"/> Middle Eastern         |
| <input type="checkbox"/> Amish                        | <input type="checkbox"/> European (Finnish) | <input type="checkbox"/> South Asian            |
| <input type="checkbox"/> Other, Please specify: _____ |   |   |

Are the patient's parents consanguineous?

- Yes     No     Unknown

Are there other family members who currently have or have had the same or a similar phenotype as the patient?

- Yes     No

If yes, please list the affected members below:

Relationship to the Patient (e.g.,  
mother, brother, uncle)

Age of  
Onset

Diagnosis/ Symptoms

_____	_____	_____
_____	_____	_____
_____	_____	_____

## CONSENT

\*Please, indicate how long you would like Genseq to store DNA sequencing raw data on your behalf:

- 6 months (default retention time where no option is chosen)     12 months

In addition, in order to (a) fulfil your instructions to the requested perform genetic testing, and (b) for us to provide further cascade genetic tests for you, each as undertaken in the context of an accredited genetic testing service, you understand that other data types, such as patient data received on this Test Request Form, laboratory QC data and report data will be stored for a further period of years taking account of applicable law, regulation and industry guidance. In returning this Test Request Form for processing you are instructing us in writing to undertake such processing on your behalf.

\*I hereby confirm that I have obtained written informed consent from the patient for this test to be performed, including consent for health practitioners registered under my account to access the report.

## BILLING AND REPORT DETAILS

The invoice for this test will be sent to the default billing address associated with your Account ID. If your institution requires a PO number, please insert it here.

PO Number \_\_\_\_\_

Patient Self Pay    Patient phone number: \_\_\_\_\_ Patient email: \_\_\_\_\_

The report for this test will be made available in your account on Genseq's online ordering portal. If you need other health practitioners to have access to the report, please ensure they are registered under your account to ensure appropriate communication.

# TEST REQUEST FORM

## GENE PANEL



\* Mandatory fields

### GENE PANELS

#### Cardiology

- Aortopathy
- Arrhythmogenic Right Ventricular Cardiomyopathy
- Brugada Syndrome - Core
- Brugada Syndrome - Expanded
- Catecholaminergic Polymorphic VT
- Comprehensive Cardiac Arrhythmias
- Comprehensive Cardiomyopathy
- Dilated Cardiomyopathy and Conduction Defects - Core
- Dilated Cardiomyopathy and Conduction Defects - Expanded
- Dyslipidaemia (includes Familial Hypercholesterolaemia)
- Hypertrophic Cardiomyopathy - Core
- Hypertrophic Cardiomyopathy - Expanded
- Left Ventricular Noncompaction Cardiomyopathy
- Long QT syndrome - Core
- Long QT syndrome - Expanded
- Short QT syndrome
- TTR* Single Gene (Sanger sequencing)

#### Hereditary Cancer

- Colorectal Cancer and Polyposis
- Comprehensive Cancer
- Hereditary Breast and Gynaecological Cancer
- Hereditary Breast Cancer
- Hereditary Cancer – *BRCA1/BRCA2*
- Hereditary Cancer High Risk
- Hereditary Cancer – Lynch Syndrome
- Hereditary Endocrine Cancer
- Hereditary Gastrointestinal Cancer
- Hereditary Lung Cancer
- Hereditary Melanoma and Skin Cancer
- Hereditary Pancreatic Cancer
- Hereditary Paraganglioma-Pheochromocytoma
- Hereditary Prostate Cancer
- Hereditary Renal Cancer
- Neurofibromatosis

#### Nephrology

- Alport/Haematuria
- Atypical Haemolytic Uraemic Syndrome
- Bartter Syndrome
- Comprehensive Nephrology
- Cystic Kidney Disease
- Diabetes Insipidus, Nephrogenic
- Extreme Early Onset Hypertension
- Hereditary Systemic Amyloidosis
- Membranoproliferative Glomerulonephritis Including C3
- Glomerulopathy
- Nephrocalcinosis or Nephrolithiasis
- Proteinuric Renal Disease
- Pseudohypoaldosteronism
- Rare Multisystem Ciliopathy
- Renal Malformations (CAKUT)
- Renal Super Panel - Broad
- Renal Tubulopathies
- Tubulointerstitial Kidney Disease
- Unexplained Kidney Failure in Young People
- Unexplained Young Onset End-Stage Renal Disease

#### Neurology

- Acute Rhabdomyolysis
- Adult Onset Dystonia, Chorea or Related Movement Disorder
- Adult Onset Hereditary Spastic Paraplegia (HSP)
- Adult Onset Leukodystrophy
- Adult Onset Neurodegenerative Disorder
- Arthrogyriposis
- Cerebral Malformation
- Cerebral Vascular Malformations
- Childhood Onset Dystonia, Chorea or Related Movement Disorder
- Childhood Onset Hereditary Spastic Paraplegia
- Childhood Onset Leukodystrophy
- Comprehensive Epilepsy
- Comprehensive Neurology
- Congenital Muscular Dystrophy
- Congenital Myaesthetic Syndromes
- Congenital Myopathy
- Dementia
- Early Onset or Syndromic Epilepsy
- Hereditary Ataxia and Cerebellar Anomalies—Childhood onset
- Hereditary Ataxia with Onset in Adulthood
- Hereditary Neuropathy or Pain Disorder
- Holoprosencephaly
- Hydrocephalus
- Intellectual Disability
- Limb Girdle Muscular Dystrophies, Myofibrillar Myopathies and Distal Myopathies
- Macrocephaly
- Malignant Hyperthermia
- Paroxysmal Central Nervous System Disorders
- Rare Neuromuscular Disorders
- Severe Microcephaly
- Skeletal Muscle Channelopathy
- Tuberous Sclerosis

#### Ophthalmology

- Albinism or Congenital Nystagmus
- Bardet Biedl Syndrome
- Bilateral Congenital or Childhood Onset Cataracts
- Blepharophimosis Ptosis and Epicanthus Inversus
- Comprehensive Ophthalmology
- Congenital Fibrosis of the Extraocular Muscles
- Corneal Dystrophies
- Optic Neuropathy
- Pseudoxanthoma Elasticum
- Retinal Disorders
- Sporadic Aniridia
- Stickler Syndrome
- Structural Eye Disease