TEST REQUEST FORM
GENE PANEL

* Mandatory fields

PATIENT DETAILS

Forename* _____ **Surname*** _____

Hospital ID _____ **DOB (DD/MM/YYY)*** _____

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*[1] : _____

_____

[1]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*[2] _____

_____

_____

_____

_____

_____

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

SAMPLE DETAILS[3]

Sample Type: ☐ Whole Blood (EDTATube) ☐ Genomic DNA, Source: _____
☐ Saliva (Oragene Tube) ☐ Buccal Swab

Date Collected (DD/MM/YYY)*: _____ **Time Collected (hh:mm):** _____

[3]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

- ☐ Admixed American
- ☐ African/African American
- ☐ Amish
- ☐ Ashkenazi Jewish
- ☐ East Asian
- ☐ European (Finnish)
- ☐ European (non-Finnish)
- ☐ Middle Eastern
- ☐ South Asian
- ☐ Other, Please specify:_____

Father's Ancestry

- ☐ Admixed American
- ☐ African/African American
- ☐ Amish
- ☐ Ashkenazi Jewish
- ☐ East Asian
- ☐ European (Finnish)
- ☐ European (non-Finnish)
- ☐ Middle Eastern
- ☐ South Asian
- ☐ 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
- ☐ Pseudohypoaldosternism
- ☐ 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
- ☐ Arthrogryposis
- ☐ 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 Myaesthenic 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

INFORMED CONSENT FOR SINGLE GENE TESTING

CONFIRMATION BY HEALTH PRACTITIONER

I the undersigned confirm:

1 I am the health practitioner under whose responsibility genetic testing has been requested in respect of the patient named in the Test Request Form and the above Informed Consent and I owe a professional duty of confidentiality to the patient.
☐ **Yes** ☐ **No**

2 I have provided the patient/ the patient's legal guardian(s) where the patient is a child with all appropriate information concerning genetic testing and processing of genetic data, including indication(s), relevant target disease or condition, purpose and scope, risks, potential outcomes and implications of genetic testing, and alternatives to genetic testing and have provided a copy of the associated Patient Information Leaflet and have discussed the limitations of the requested genetic testing.
☐ **Yes** ☐ **No**

3 I have given the patient / the patient's legal guardian(s) where the patient is a child an opportunity to ask questions and confirm that I have answered all questions asked by the patient/ the patient's legal guardian(s).
☐ **Yes** ☐ **No**

4 I confirm that the patient / the patient's legal guardian(s) where the patient is a child has/have consented to the genetic test results being issued to the health practitioners whose details are provided in the Test Request Form including those registered under the health practitioner's account with Genseq.
☐ **Yes** ☐ **No**

5 I confirm that the patient / the patient's legal guardian(s) where the patient is a child has/have voluntarily given informed consent to genetic testing and processing of genetic data in respect of the patient.
☐ **Yes** ☐ **No**

HEALTH PRACTITIONER

_____ **Professional Registration Number:** _____

Health Practitioner (Full name - BLOCK LETTERS)

X_____ **Profession:** _____

Health Practitioner (Signature)

Date (dd/mm/yyyy)_____

CONFIRMATION AND INFORMED CONSENT OF PATIENT OR LEGAL GUARDIAN(S) [i]

By providing a ☒ yes response to paragraphs 1 to 9 below [ii]and by providing an ☒ yes response to paragraph 10 in respect of data processing and by signing this Consent Form, I/we the undersigned confirm and consent to Genseq performing genetic testing and processing of genetic data in the terms set out below:

1 The patient to whom this informed consent relates is _____(insert name in block capitals).

2 The genetic test to which this informed consent relates to is: [ ] ***DPYD*** [ ] ***TPMT*** [ ] ***HFE*** [ ] ***TTR*** [ ] ***CYP2C19***

3 I /we have received from my health practitioner[iii] /my child /our child's health practitioner all appropriate information concerning genetic testing and processing of genetic data, including indication(s), relevant target disease or condition, purpose and scope, risks, potential outcomes and implications of the above genetic test.

4 I /we have read or have had read to me/us the Patient Information Leaflet relating to the above requested genetic test and understand the information provided to me/us in the information leaflet including the limitations of the requested genetic test.

5 I /we have had an opportunity to ask questions of my/ my child/ our child's health practitioner in respect of the genetic testand processing of genetic data.

6 I/we have received satisfactory answers to all my/our questions from my/my child /our child's health practitioner.

7 I /we have read or have had read to me/us the Test Request Form and confirm that the information provided in the Test Request Form is correct and complete.

8 I/we consent to the genetic testing proposed by my/ my child/our child's health practitioner to be carried out by Genseq on my /my child /our child's biological sample(s) as ordered by my/my child /our child's health practitioner in the Test Request Form[iv].

9 I/we consent to Genseq issuing the report on my/my child/our child's genetic test results to my/my child/our child's health practitioner(s) whose details are provided in the Test Request Form including those registered under the health practitioner's account with Genseq.

10 I/we consent to the disposal of my/my child /our child's residual blood sample (if any) and DNA sample after genetic testing has been performed by Genseq.

I/we agree to the statements and confirm my/our consent to paragraphs 1 –10 above ☐ **Yes** ☐ **No**

EXPLICIT CONSENT TO DATA PROCESSING

11 I/we give my/our explicit consent to the processing by my/my child /our child's health practitioner as controller (and Genseq on their behalf) of my/my child/our child's personal data including health and genetic data for the purpose of the provision of genetic testing services as described here (and in the associated Patient Information Leaflet) to include use of patient clinical and family history, sample receipt, processing, testing, reporting to and correspondence with my/my child/our child's health practitioner(s), retention, storage and disposal of samples, DNA and the processing of related patient payment and billing information. In particular, I/we understand any residual sample[v] of Deoxyribonucleic acid (DNA) will be retained for such period as may be specified by my/my child/our child's health practitioner as controller of my/my child/our child's personal data or as required by law prior to disposal of any such retained DNA. I/we also give consent to the processing of my/our names as guardians in connection with the above. I/we understand that I/we have a right to withdraw my/our consent at any time and that to do so I/we will contact my/ my child / our child's health practitioner.

☐ **Yes** ☐ **No**

PATIENT	**HEALTH PRACTITIONER**
Patient (Full name - BLOCK LETTERS)	**Health Practitioner (Full name - BLOCK LETTERS)**
X_____ **Patient (Signature)**	X_____ **Health Practitioner (Signature)**
Patient DOB (dd/mm/yyyy) _____	**Professional Registration Number:** _____
Date (dd/mm/yyyy) _____	**Date (dd/mm/yyyy)** _____
LEGAL GUARDIAN	
Legal Guardian (1) (Full name - BLOCK LETTERS)	**Legal Guardian (2) (Full name - BLOCK LETTERS)**
X_____ **Legal Guardian (1) (Signature)**	X_____ **Legal Guardian (2) (Signature)**
Date (dd/mm/yyyy) _____	**Date (dd/mm/yyyy)** _____

[i] This genetic testing consent form is for persons who are aged 16 years and over and have capacity to give informed consent to genetic testing, and by signing the consent form give informed consent to genetic testing. This consent form can also be used in the case of children (persons who are under the age of 16 or who are 16 years but not yet 18 years of age and lack capacity to consent) whose legal guardian(s) give informed consent on their behalf to undergo genetic testing. In this consent form where the patient is a child relying on the consent of his or her legal guardian(s), the form refers to the patient as" my child" where one guardian is giving informed consent or "our child " where both of the child's legal guardians give informed consent. The patient's name should be inserted in paragraph 1 and the appropriate deletions made in paragraphs 2 – 10.

[ii] Genseq will not be able to provide Genetic testing services and processing of genetic data where the consent form and or confirmation by Health Practitioner is incomplete or where a negative answer has been given to any of the X boxes. Certain samples are only suitable for testing within a limited period of time and incomplete Test Request Form and / or Confirmation and Consent Form by Patient or Legal Guardian(s) and Health Practitioner will cause delay in testing and may mean that a new sample will be required when submitting the completed Test Request Form and / or Consent Form and Confirmation by Patient or Legal Guardian(s) and Health Practitioner.

[iii] Under section 42 of the Disability Act 2005 (as amended) (**"the 2005 Act"**) the informed consent of an individual undergoing genetic testing must be obtained prior to genetic testing and the processing of genetic data in compliance with the 2005 Act and GDPR. Under Irish law a health practitioner is a registered medical practitioner, dentist, pharmacist, nurse, midwife, optometrist, optician, or a registrant of a profession designated under the Health and Social Care Professionals Act 2005 (as amended) which includes registered psychologists and psychotherapists. See www.coru.ie for the full list of designated professions. In the informed consent and confirmation by Health Practitioner references to health practitioner are to a registrant of one of the designated professions or to a person who is subject to an equivalent duty of confidentiality to the individual whose health or genetic data is to be processed.

[iv] The Test Request Form contains important information relevant to patient consent. The Test Request Form is completed by the patient's health practitioner and specifies the target disease or condition and the test(s) to be performed on the patient's biological sample and provides Genseq with relevant patient information. Genseq relies on the adequacy and accuracy of the information provided by the health practitioner in the Test Request Form. Genseq performs the specific test(s) listed in the Test Request Form and issues a report on the test(s) to the patient's health practitioner(s) whose contact details are set out in the Test Request Form including those registered under the health practitioner's account. By signing the consent form and providing data protection consent, the patient or the patient's legal guardian(s,) as applicable, consent to Genseq performing the test (s) specified in the Test Request Form and to Genseq issuing the report on the test(s) result(s) to the health practitioner(s) whose contact details are provided in the Test Request Form including those registered under the health practitioner's account. Genseq allows access to the test results via its online portal to the patient's health practitioners including those registered under the patient's health practitioner's account with Genseq.