TEST REQUEST FORM
CASCADE TESTING

* Mandatory fields

PATIENT DETAILS

Forename* _____

Surname* _____

Biological Sex* _____

DOB (DD/MM/YYY)* _____

Hospital ID _____

Ancestry[1]

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

[1]As defined by the Genome Aggregation Database (gnomAD).

HEALTH PRACTITIONER DETAILS

Account ID _____
Full Name* _____
Phone _____
Email _____
Institution* _____

Address 1 _____
Address 2 _____
City/town _____
Post Code _____
Country _____

TEST DETAILS

Was the Index (familial) patient tested at Genseq?* ☐ Yes ☐ No / Not known

If Yes:

- Index Patient's Name*: _____
- Genseq ID*: _____
- Index Patient's DOB (DD/MM/YYY)*: _____

If Index was NOT tested at Genseq, please attach a copy of Index (Familial) report* ☐

Additionally, where possible, please send a whole blood sample (EDTA Tube) or Genomic DNA (Source:_____) from the Index with this request form.

Index phenotype*

Age of Onset* _____ Diagnosis/ Symptoms*[1] _____

Relationship between the person being tested and the index patient*

*The person being tested is the index patient's: _____

(e.g. brother, sister, son, daughter, mother, father)

CLINICAL INFORMATION

Reason for testing* ☐Predictive ☐ Carrier ☐ Segregation ☐Other:_____

Clinical details (affected/unaffected)[1]

_____

_____

_____

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

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VARIANT TO BE TESTED

	Gene (e.g. *TTN*)	Transcript (NM_001256850.1)	cDNA change (e.g. c.102917T>A)	Protein Change (e.g. p.(Ile34306Asn))
Variant 1				
Variant 2				
Variant 3				

SAMPLE DETAILS

Sample Type* ☐ Whole Blood (EDTA Tube) ☐ Genomic DNA, **Source:** _____

☐ Saliva (Oragene Tube) ☐ Buccal Swab

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

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

INTERNAL USE ONLY

Sample ID _____

CONSENT TO CASCADE TESTING

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 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 cascade testing for a familial genetic variant.
3 I /we have had an opportunity to ask questions of my/ my child/ our child's health practitioner in respect of the of genetic testing for a familial genetic variant and processing of genetic data.
4 I/we have received satisfactory answers to all my/our questions from my/my child /our child's health practitioner.
5 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.
6 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].
7 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.
8 I/we consent to the disposal of my/my child /our child's residual blood sample (if any) after genetic testing has been performed by Genseq.

I/we agree to the statements and confirm my/our consent to paragraphs 1 –8 above ☐ Yes ☐ No

EXPLICIT CONSENT TO DATA PROCESSING

9 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 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. I understand that giving my explicit consent here is necessary to avail of the genetic testing. ☐ **Yes**

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**
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 Cascade Testing Patient Information Leaflet and have discussed the limitations of genetic testing using Sanger sequencing.
☐ **Yes** ☐ **No**
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**
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**
2 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**

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

CONSENT TO CASCADE TESTING

LEGAL GUARDIAN

Legal Guardian (1) (Full name - BLOCK LETTERS)	**Legal Guardian (2) (Full name - BLOCK LETTERS)**
X_____	X_____
Legal Guardian (1) (Signature)	**Legal Guardian (2) (Signature)**
Date (dd/mm/yyyy) _____	**Date (dd/mm/yyyy)** _____

[i] This Cascade 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 targeted genetic variant 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.

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