THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS
Molecular Genetics Thalassaemia Department
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REQUEST FORM FOR DNA DIAGNOSTIC TESTS – Department Code 25

Patient Identification *(Please tick ☑ and fill in accordingly)*	***Requesting Clinician/Scientist***
Case type: ☐ Inpatient ☐ Outpatient CING No.:	
Name: Surname:	Name: Surname:
I.D. No.: D.O.B.:	Hospital / Clinic: Telephone:
Gender: ☐ Male ☐ Female ☐ Unspecified	E-mail: Fax:
Nationality: Telephone:	Address:
Address:	Code: City: Country:
Code: City: Country:	Reason for Referral:
	Date of Referral: **Signature:**

Patient Status *(Please tick ☑ and fill in accordingly)*

☐ GESY ☐ Government Non-GESY ☐ Hospital Card No: ☐ Private Non-GESY

Requesting Clinician/Scientist Status *(Please tick ☑ and fill in accordingly)*

☐ CING ☐ Government (ΟΚΥπΥ/SHSO) ☐ Private-GESY GESY No: ☐ Private-Non GESY

The referring physician undertakes and confirms understanding of and compliance with mutual obligations as these are determined under the EU GDPR.

Indication for Testing *(Please tick ☑ and fill in accordingly)*

☐ Carrier Testing ☐ Prenatal Diagnosis (PND) ☐ Other (please specify)
☐ Pre-implantation Genetic Test (PGT)

Type of Specimen *(Please tick ☑ and fill in accordingly)*

☐ Whole Blood ☐ Amniotic Fluid ☐ CVS ☐ Blastocyst Biopsy ☐ Blastomere Biopsy
☐ Other (please specify) Date specimen collected:

Sample Receipt *(For laboratory internal use)*

DNA No:	Family No.:	
PND No:	PGT No:	NIPD No:
Sample Receipt Date.:		Amount:
Received by:		**Signature:**

Clinical Features *(Please tick ☑ and fill in accordingly)*

Clinical Feature	Count	Clinical Feature	Count	Clinical Feature	Count	Other Findings
☐ Hb		☐ WBC		☐ Reticulocytes		
☐ MCH		☐ PLT		☐ Ferritin		
☐ MCV		☐ HbA2		☐ TSAT		
☐ MCHC		☐ HbX*		*Notes:		

Test Required (Code No.) *(Please tick ☑ accordingly)*

Molecular Genetic Analysis		Next-Generation Sequencing (NGS) Analysis	
☐ Prenatal diagnosis for Thalassaemia – 1st CVS	1	☐ Targeted NGS panel for the molecular diagnosis of rare anaemias (RA-NGS)	26
☐ Prenatal diagnosis for Thalassaemia – 2nd CVS	2	☐ Targeted in silico gene panel using CES for the molecular diagnosis of non-malignant haematological diseases including iron metabolism and haem synthesis disorders (Haem-NGS)	27.1
☐ Diagnostic samples for Thalassaemia	3		
☐ Alpha and beta locus MLPA analysis	4	☐ Targeted in silico gene panel using WES for the molecular diagnosis of non-malignant haematological diseases including iron metabolism and haem synthesis disorders (Haem-NGS)	27.2
☐ Molecular Diagnosis for Thalassaemia	10		
☐ Sequencing of globin gene	13	☐ Targeted in silico gene panel using CES for the molecular diagnosis of hepatological diseases (Hep-NGS)	28.1
☐ DNA extraction from blood	14	☐ Targeted in silico gene panel using WES for the molecular diagnosis of hepatological diseases (Hep-NGS)	28.2
☐ DNA extraction from tissue	15	☐ Targeted in silico gene panel using CES for the molecular diagnosis of bone diseases (Ost-NGS)	29.1
☐ Pre-implantation genetic test (PGT) (private only)	16	☐ Targeted in silico gene panel using WES for the molecular diagnosis of bone diseases (Ost-NGS)	29.2
☐ Pre-implantation genetic test (PGT) (government)	16.3	☐Targeted in silico gene panel using CES for oxidative stress (OXIS-NGS)	30.1
☐ NIPD for X-linked disorders	17	☐ Targeted in silico gene panel using WES for oxidative stress (OXIS-NGS)	30.2
☐ NIPD for fetal RhD status (private only)	18	☐ Whole exome sequencing (WES) (Single)	31
		☐ Whole exome sequencing (WES) (Trio)	32
		☐ Clinical exome sequencing (CES) (Single)	33
		☐ Clinical exome sequencing (CES) (Trio)	34
		☐ Open up of NGS data from WES after in silico panel	35
		☐ Open up of NGS data from CES after in silico panel	36
		☐ Confirmation of SNVs with Sanger Sequencing (SNV-SANGER)	38
		☐ Trio in silico panel from WES (applied for all above disease-specific panels)	39
		☐ Trio in silico panel from CES (applied for all above disease-specific panels)	40

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Panels/Advanced Evaluation

Please check the box, if diagnostic NGS test will be requested and fill in the following details.

☐ Current NGS request is for diagnostic purposes, only.

Please state if any other diagnostic tests have been performed for this patient.

☐ **Yes** ☐ **No**

If **Yes**, state which tests:

If **No**, state why patient is referred for NGS:

Genetic Testing and Research Consent

I, , hereby authorise to perform genetic testing for diagnostic purposes related to the disease referred to in this request form. I understand that the sample provided may be stored and used anonymised for research directly related to the diagnosed condition, with the aim of advancing scientific knowledge and improving patient care. My identity will be strictly protected, and any data used for research will be de-identified to maintain confidentiality. I have been informed of the potential benefits and risks associated with genetic testing and research and willingly provide my consent.

Patient's Full Name:

Date: **Patient's Signature:**