# TEST REQUISITION FORM

**Customer Code:** .....

REFERENCE LABORATORY GENETICS

Required fields are marked with an asterisk (*)

## ANALYSIS REQUESTED

***Test Code:** ..... ***Test Name:** .....

*Analysis Requested:

- ☐ Single gene testing
- ☐ Array (aCGH)
- ☐ Del/Dup Analysis
- ☐ NGS Based CNVs
- ☐ NIPT
- ☐ Gene panel testing
- ☐ Clinical Exome (CES)
- ☐ Whole Exome (WES)
- ☐ Whole Genome (WGS)
- ☐ Other: .....
- ☐ Carrier testing
  - Gene name: ..... Mutation: .....
  - Relative tested at REFERENCE LABORATORY: ☐ Yes ☐ No
  - If yes, previous order ID: .....
  - Relationship to patient: .....

*Analysis type: ☐ Only idex patient ☐ Trio ☐ Mother ☐ Father ☐ Sibling ☐ Other: .....

*Sample Collection Date: dd/mm/yyyy

*Sample type: ☐ Whole blood ☐ Saliva ☐ DNA ☐ RefLabCard®, number: ..... ☐ Amniotic fluid ☐ Chorionic villi ☐ FFPE

*Prenatal: ☐ No ☐ Yes (please *indicate gestational age: .....)

*Reporting of secondary/incidental findings: ☐ Yes ☐ No

## PATIENT INFORMATION

*Last Name: ..... *First Name: ..... *Date of Birth: dd/mm/yyyy

*Sex: ☐ Female ☐ Male ☐ Unknown

Previously tested at Reference Laboratory: ☐ No ☐ Yes, please specify order ID: .....

*Country: ..... *City: ..... *ZIP code: .....

## FAMILY MEMBER 1 INFORMATION (FILL ONLY IF TRIO ANALYSIS IS ORDERED)

*Last Name: ..... *First Name: ..... *Date of Birth: dd/mm/yyyy

*Sex: ☐ Female ☐ Male ☐ Unknown

*Relationship to patient ☐ Mother ☐ Father ☐ Sibling ☐ Other: .....

*Is Family Member 1 affected with the same phenotype as patient? ☐ No ☐ Yes ☐ Partially ☐ Uncertain

## FAMILY MEMBER 2 INFORMATION (FILL ONLY IF TRIO ANALYSIS IS ORDERED)

*Last Name: ..... *First Name: ..... *Date of Birth: dd/mm/yyyy

*Sex: ☐ Female ☐ Male ☐ Unknown

*Relationship to patient ☐ Mother ☐ Father ☐ Sibling ☐ Other: .....

*Is Family Member 2 affected with the same phenotype as patient? ☐ No ☐ Yes ☐ Partially ☐ Uncertain

## ORDERING HEALTH CARE PROFESSIONAL INFORMATION

(Reporting Address)

*Last Name: ..... *First Name: ..... ***Email:** .....

*Institution: ..... Department: ..... Phone: ..... Fax: .....

*Street Address: ..... *ZIP code: ..... *City: ..... State: .....

***I herewith confirm the correctness of the above given information.**

..... dd/mm/yyyy
Place, Date

.....
Signature of ordering health care professional

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REFERENCE LABORATORY GENETICS

**PATIENT HISTORY**

Required fields are marked with an asterisk (*)

| | |
|---|---|
| *Indication for testing: ☐ Diagnostic ☐ Family history ☐ Other: ..... | ☐ Age of manifestation: ..... ☐ Unaffected |

*Describe the relevant clinical information (attach possible supportive material):

Variants are reported based on the clinical information provided, therefore detailed phenotypic and clinical information increases the likelihood of the diagnosis.

| | |
|---|---|
| *Family history:<br>A. Consanguinity ☐ Yes ☐ No<br>B. Affected siblings ☐ Yes ☐ No<br>C. Please describe other relevant family history: | Pedigree: |
| Previous testing with abnormal results: | Previous testing with normal results: |
| Please specify suspected differential diagnosis (if applicable): | Please specify genes of interest (if applicable): |

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**PLEASE TICK THE APPROPRIATE CLINICAL FEATURES (*)**

Required fields are marked with an asterisk (*)

REFERENCE LABORATORY GENETICS

## METABOLICS

- ☐ Abnormal creatine kinase
- ☐ Abnormal creatine phosphokinase
- ☐ Decreased plasma carnitine
- ☐ Elevated alanine
- ☐ Elevated puryvate
- ☐ Failure to thrive
- ☐ Hypoglycemia
- ☐ Increased CSF lactate
- ☐ Ketosis
- ☐ Lactic acidosis
- ☐ Organic aciduria

## NEUROLOGY

### NEURODEVELOPMENTAL

- ☐ Attention deficit hyperactivity disorder
- ☐ Austism spectrum disorder
- ☐ Delayed language dev.
- ☐ Delayed motor dev.
- ☐ Developmental delay
- ☐ Developmental regression
- ☐ Encephalopathy
- ☐ Fine motor delay
- ☐ Gross motor delay
- ☐ Hearing loss
- ☐ Intellectual disability
- ☐ Learning disability
- ☐ Obsessive-compulsive disorder
- ☐ Psychiatric symptoms
- ☐ Recurrent headache
- ☐ Seizures
- ☐ Speech delay

### NEUROMUSCULAR

- ☐ Ataxia
- ☐ Chorea
- ☐ Dystonia
- ☐ Hyperreflexia
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Muscle weakness
- ☐ Muscular dystrophy
- ☐ Neuropathy
- ☐ Parkinsonism
- ☐ Spasticity

### BEHAVIORAL

- ☐ Attention deficit disorder
- ☐ Psychiatric diseases

### BRAIN MALFORMATIONS AND IMAGING

- ☐ Abnormal cortical gyration
- ☐ Abnormal myelination
- ☐ Abnormalities of basal ganglia
- ☐ Agenesis of the corpus callosum
- ☐ Brain atrophy
- ☐ Cerebellar hypoplasia
- ☐ Cortical dysplasia
- ☐ Hemimegalencephaly
- ☐ Heterotropia
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Hydrocephalus
- ☐ Leukodystrophy
- ☐ Lissencephaly
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Periventricular leukomalacia

### SEIZURES

- ☐ Febrile seizures
- ☐ Focal seizures
- ☐ Generalized seizures

### OTHERS RELATED TO NEUROLOGY

- ☐ Craniosynostosis
- ☐ Dementia
- ☐ Headache
- ☐ Migraine
- ☐ Stroke

## NEPHROLOGY

- ☐ Abnormal renal morphology
- ☐ Abnormal urinary syste
- ☐ Hydronephrosis
- ☐ Kidney malformation
- ☐ Renal agenesis or dysgenesis
- ☐ Renal cyst
- ☐ Renal tubular dysfunction
- ☐ Renal tubulopathy

## CARDIOVASCULAR

- ☐ Angioedema
- ☐ Aortic dilatation
- ☐ Arrhythmia / conduction defect
- ☐ Atrial septal defect
- ☐ Cardiomyopathy
- ☐ Coarctation of aorta
- ☐ Dilated cardiomyopathy
- ☐ Hypertension
- ☐ Hypertrophic cardiomyopathy
- ☐ Hypoplastic left heart
- ☐ Hypotension
- ☐ Lymphedema
- ☐ Malformation of heart and great vessels
- ☐ Myocardial infarction
- ☐ Stroke
- ☐ Tetralogy of Fallot
- ☐ Vasculitis
- ☐ Ventricular septal defect

## OPHTALMOLOGY

- ☐ Abnormal eye movement
- ☐ Abnormal vision
- ☐ Blepharospasm
- ☐ Blindness
- ☐ Cataracts
- ☐ Coloboma
- ☐ Chronic Progressive External Ophthalmoplegia
- ☐ Glaucoma
- ☐ Microphthalmos
- ☐ Nystagmus
- ☐ Ophthalmoplegia
- ☐ Optic atrophy
- ☐ Ptosis
- ☐ Retinitis pigmentosa
- ☐ Retinoblastoma
- ☐ Strabismus
- ☐ Visual impairment

## ENDOCRINOLOGY

- ☐ Diabetes mellitus
- ☐ Hyperparathyroidism
- ☐ Hypoparathyroidism
- ☐ Hyperthyroidism
- ☐ Hypothyroidism
- ☐ Paraganglioma
- ☐ Pheochromocytoma

## GASTROENTEROLOGY

- ☐ Aganglionic megacolon
- ☐ Chronic diarrhea
- ☐ Chronic intestinal pseudo-obstruction
- ☐ Constipation
- ☐ Diarrhea
- ☐ Elevated hepatic transaminase
- ☐ Frequent vomiting
- ☐ Gastroesophageal reflux
- ☐ Gastroschisis
- ☐ Hepatic failure
- ☐ Hepatomegaly
- ☐ Hirschsprung disease
- ☐ Obesity
- ☐ Pyloric stenosis
- ☐ Tracheoesophageal fistula
- ☐ Vomiting

## REPRODUCTIVE SYSTEM

- ☐ Ambiguous genitalia
- ☐ Cryptochidism
- ☐ Hypogonadism
- ☐ Hypospadias
- ☐ Infertility
- ☐ Undescended testis

## DERMATOLOGY

- ☐ Abnormality of the hair
- ☐ Abnormality of the nail
- ☐ Abnormality of skin pigmentation
- ☐ Blistering
- ☐ Connective tissue abnormality
- ☐ Hyperextensible skin
- ☐ Ichthyosis

## HEMATOLOGY AND IMMUNOLOGY

- ☐ Abnormal hemoglobin
- ☐ Abnormality of coagulation
- ☐ Anemia
- ☐ Immunodeficiency
- ☐ Myelofibrosis
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Splenomegaly
- ☐ Thrombocytopenia

## ONCOLOGY

- ☐ Adenomatous colonic polyposis
- ☐ Adenomatous polyposis
- ☐ Brain tumor
- ☐ Breast cancer
- ☐ Colorectal cancer
- ☐ Leukemia
- ☐ Lung cancer
- ☐ Melanoma
- ☐ Myelofibrosis
- ☐ Neoplasm of the lung
- ☐ Neoplasm of the skin
- ☐ Paraganglioma
- ☐ Pheochromocytoma

## SKELETAL, DEVELOPMENT AND PRENATAL

- ☐ Abnormal facial shape
- ☐ Abnormal limb morphology
- ☐ Abnormal vertebral column
- ☐ Abnormality of the skeletal system
- ☐ Cleft lip / palate
- ☐ Club foot / feet
- ☐ Contractures
- ☐ Craniosynostosis
- ☐ Dysmorphic features
- ☐ Ear malformation
- ☐ Fractures
- ☐ Hemihypertrophy
- ☐ Hydrops fetalis
- ☐ Intrauterine growth retardation
- ☐ Joint hypermobility
- ☐ Limb anomaly
- ☐ Multiple joint contractures
- ☐ Oligohydramnios
- ☐ Overgrowth
- ☐ Polydactyly
- ☐ Premature birth
- ☐ Scoliosis
- ☐ Short stature
- ☐ Syndactyly
- ☐ Talipes equinovarus
- ☐ Tall stature
- ☐ Vertebral anomaly

## OTHERS

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# INFORMED CONSENT FOR CONDUCTING GENETIC ANALYSES

Required fields are marked with an asterisk (*)

It is obligatory to make sure that the patient or their legal representative understands and signs the informed consent to conduct genetic analyses. One of the following documents is required by REFERENCE LABORATORY In order to allow them to legally conduct genetic analysis:

1. **Patient's signed informed consent.**
2. **Informed consent signed by the healthcare professional stating that signed consent exists in the files**

**Please make sure that each sample is accompanied by the necessary document. A printout of informed consent must be provided for each person for whom a test is requested.**

## 1. Patient's signed informed consent

By signing this informed consent I (or I as legal representative) confirm that I have been advised by my physician on the genetic basis, purpose, scope, type and implication of the planned genetic analysis and obtainable results, chances of possible prevention and or possible treatment of the potential disease and in view of any possible danger associated with providing the sample necessary for the genetic analysis and the obtaining of the genetic analysis results.

All my doubts have been allayed and I have had sufficient time to allow me to make an informed decision regarding the genetic analysis:

*Genetic analysis:

By signing this informed consent, I certify that I understand the following points (1.1-1.3):

**1.1.** The results of this test could reveal that I and/or my family members have an inherited disease or have an increased chance of being affected by a genetic disease. I am aware that this test might show previously undetected biological relationships including non-paternity.

**1.2**. I am conscious that the results of this test may prove to be uncertain regarding my genetic condition. Some genetic variants are known to generate diseases and others are known to be benign. In addition, there is a number of genetic variants that are found which are of uncertain significance. More extensive testing and or counselling may be recommended for me or family members depending on the results of this test.

**1.3.** I am aware that results from this test may be used with the intention of improving understanding, diagnostics and treatment of similar conditions and presented at meetings, for scientific publications and/or variant databases. I am under the understanding that no identifying information will ever be shown.

By signing this informed consent, I certify that I understand the following points (1.4-1.8):

**1.4. Analysis performance.**

I give permission for Reference Laboratory to conduct the genetic analysis stated above

**1.5. Collection and processing of personal data.**

I certify that I give my permission for my physician and REFERENCE LABORATORY to collect and process my Personal Clinical Data, as far as is necessary in order to conduct the genetic analysis, also including transfers of my Personal Clinical Data between my physician and Reference Laboratory, across international borders. I understand that my Personal Clinical Data includes my personal details (including name and address), family relationships, age and date of birth, ethnicity, nationality, insurance information, symptoms and any other relative medical information, illnesses, any samples with identifiable genetic data and analysis results, and findings.

**1.6. Reporting of the results.**

In cases where REFERENCE LABORATORY is under instruction to do so I give authorisation for them to inform me, my physician or the requesting laboratory regarding the results of the genetic analysis and to provide when requested, the raw data of the genetic analysis to me, my physician or the requesting laboratory.

**1.7. Long term storage and usage of my Personal Clinical Data and (remaining) sample.**

I concede for my Personal Clinical Data and (remaining) sample to be used for verification of analysis results if necessary and aid in further research, improvement and development of diagnostic methods and therapeutic solutions. These measures may offer support, medical advice and guidance related to the diagnosis and treatment of potential diseases for me and my family.

My research data shall be treated as confidential and coded so that my identity shall not be revealed without the key code which will be in possession of Reference Laboratory. The coded research data may be processed within or outside of the European Union and only allowed to be issued for usage by others partaking in the study such as research groups or companies. I hereby give my permission for the usage of the abovementioned research data for the purposes stated here. The data will be stored for twenty years.

By ticking the relevant box underneath, I confirm that:

- I acknowledge that as long as prior consent has been given, REFERENCE LABORATORY will store (1) Personal Clinical Data and results of the genetic analysis I have provided and relevant information on (affected) family members – as long as they have given prior consent and (2) my sample (both the original and processed sample) for a period of up to 20 years and that this data and or remaining samples may be used for internal research, validation of procedures and service and product improvements and developments.
- I am aware that the sample taken for diagnostic purposes may be used for research and that this is voluntary, and that I am able to cancel this and retract my participation at any given moment, even before the study is completed. I am also aware that any data that has been collected up to the date of my decision to withdraw may also be used as part of the research.

# INFORMED CONSENT FOR CONDUCTING GENETIC ANALYSES

Required fields are marked with an asterisk (*)

- I acknowledge that for the purpose of research, both scientific and commercial, and to aid with and contribute to the diagnosis of genetic alterations and diseases of other patients REFERENCE LABORATORY may at any time process my anonymized or pseudonymized Personal Clinical Data, e.g. into its databases and datasets relating to genetic diseases. External physicians, scientists and companies may be given access to pseudonymised or anonymised data for development purposes and so that they are able to carry out research.
- I concede that after twenty years have passed, the anonymity of my Personal Clinical Data and (remaining) sample are preserved and that they will become the property of REFERENCE LABORATORY. Both will then belong to REFERENCE LABORATORY'S archives and will be available for unrestricted use.
- I acknowledge that no compensation will be given for the usage of my Personal Clinical Data or sample by REFERENCE LABORATORY.
- I am aware that data in REFERENCE LABORATORY's databases – once anonymized - cannot be destroyed upon request as it is unidentifiable and untraceable.
- I am aware that, due to its unidentifiable and untraceable nature, data in REFERENCE LABORATORY's databases once anonymized cannot be destroyed upon request.

☐ Yes, I give my consent to use my Personal Clinical Data and long-term storage of the sample as set out in Section 1.7 above.

☐ No, I do not give my consent to use my Personal Clinical Data and long-term storage of the sample as set out in Section 1.7 above.

**1.8. Reporting of secondary/incidental findings.**

Clinical exome, whole exome and genome sequencing (CES, WES and WGS, respectively) are able to analyse thousands of genes simultaneously. As a result, there is the chance that a genetic variant not related to the original reason for which the test was requested is found. Known as incidental or secondary findings, these can provide information that is not linked in any way to your reported clinical symptoms, but could be of medical importance for your future diagnosis and treatment. I am aware of the significance of such findings and give my permission for REFERENCE LABORATORY to report secondary findings in accordance with the "ACMG Recommendations for Reporting of Incidental Findings", or to report non-ACMG recommended incidental findings at its own discretion (because of additional scientific and medical information found on REFERENCE LABORATORY's databases). I am aware that the absence of secondary/incidental findings does not necessarily mean that no other pathogenic variants exist. Upon 28 days of receiving the sample Reference Laboratory will need to receive this consent in order to report any secondary findings. I am aware that my family members are able to decide on their secondary findings regardless of my decision

☐ Yes, I give my consent to the reporting of secondary/incidental findings.

☐ No, I do not give my consent to the reporting of secondary/incidental findings.

I give permission for Reference Laboratory to contact me in reference to further genetic research and/or other genetic services they may consider relevant to me in the future. I may disengage from such contact whenever I so wish.

I am aware that I have the right not to know the results of the genetic analyses as described in the aforementioned written explanation and that I can retract my consent for the future in full or in part at any given time.

If the undersigning is the patient's legal representative, he/she confirms to provide the above consent declarations on behalf of the patient.

| *Date: | *Patient/Legal representative name: | *Patient/Legal representative signature: |
|---|---|---|
| | | |

I am aware that the patient can exercise their right to have their genetic analyses results eliminated at any moment and such request shall be passed on to REFERENCE LABORATORY immediately and without delay. I authorise my own personal data to be stored on REFERENCE LABORATOY's databases for purposes of organisation and invoicing.

I, the undersigned, confirm that consent has been given by the patient or his/her legal guardian and that if the signature is shown above then it is on file. I confirm that the patient or legal representative is capable of giving this consent, correct time frame was given to allow the patient and or legal representative to consider their decision and give consent and up to this moment has not exercised his/her right to find out the results of the genetic analyses.

| *Date: | *Healthcare professional prescriber name: | *Healthcare professional prescriber signature: |
|---|---|---|
| | | |