# Request for genetic examination - Clinical EXOME - Proband

| **Personal data of the examined person (label):** | **Referring physician:** |
|---|---|
| Name and surname:<br>Insurance number:<br>Date of birth:<br>Insurance company: ☐ Self-payer<br>Gender: ☐ male ☐ female<br>Address:<br><br>Diagnosis (ICD):<br>**Provide a valid ORPHA code:**<br>(Newly required **for NGS reimbursement,** a full list is available at https://www.orpha.net/.) | (Name, specialty, establishment ID number, workplace, stamp, signature) |

| **Primary sample:** | **Other material:** |
|---|---|
| ☐ **peripheral blood**, (5ml non-clotting blood in K3EDTA) | ☐ isolated DNA from: |
| ☐ buccal swab (only in case of bone marrow transplantation)* | ☐ cultured cells: |
| ☐ native amniotic fluid (3*10 ml of PB, always collect mother's blood in K3EDTA for comparative analysis) | |
| ☐ native chorionic villi (always collect mother's blood in K3EDTA for comparative analysis) | |
| ☐ tissue of aborted fetus (in physiological solution - DO NOT USE FORMALDEHYDE, always collect mother's blood in K3EDTA to exclude contamination) | |
| ☐ other: | |

| **Date and time of collection:** | **Date and time of indication:** (if different from the collection date) |
|---|---|
| | |

**Clinical data:** **(to be completed by the referring physician)** ☐ **STATIM**

***It is necessary to specify the phenotype which is the reason for the indication in this request form in the form of HPO terms** and to attach a detailed genetic report / ultrasound findings / autopsy findings / and family pedigree. The examination cannot be performed without the phenotype and report!***

Presumed inheritance of genetic disease:

☐ AR ☐ AD (including "de novo") ☐ X-linked

Has the patient undergone bone marrow transplantation?

☐ YES (if yes, please collect 1x blood and 2x buccal swabs)*

☐ NO

Was the patient born after IVF treatment with donated gametes?

☐ YES (if yes, DNA sample of the gamete donor needs to be provided)

☐ NO

***Human Phenotype Ontology; according to the standard http://compbio.charite.de/phenomizer/ (e.g., HP:0004415 Pulmonary artery stenosis)*

**Other PMH** (to be filled in only if not included in the provided genetic report):

**FM** (to be filled in only if not included in the provided genetic report):

**Other family members who are/will be sent for exome sequencing examination (ideally trio - proband and parents)**
(This is only an informative detail for the laboratory, each of them will be sent with a separate request form)

| | | |
|---|---|---|
| Name and surname of the relative: | Year of birth: | Relationship to the proband: |
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**Informed consent* - examined person:**

AGREES
- ☑ with the examination of the sample
- ☐ with the use of the sample for research
- ☐ with sample storage

DISAGREES
- ☐ with sample storage

*) By submitting the request, the referring physician confirms that the patient or legal representative has signed the Informed Consent, which is either stored in the patient's documentation or attached to this request.

**Examination conducted by**: GENNET, s.r.o., GENNET Laboratories, Pekařská 635/6, 158 00 Prague 5 - Jinonice, Tel: 226 231 691

**Laboratory records:**
Date and time of sample/request reception:

Sample/request accepted by: