

Functional Genetic Analysis Request Form

Please complete the form as far as possible. Completed forms can be faxed to 31-10-7043200, or sent as an encrypted PDF to the following email address: loket.klinischegenetica@erasmusmc.nl

For questions or further information, please contact f.verheijen@erasmusmc.nl; m.nellist@erasmusmc.nl; j.saris@erasmusmc.nl

Patient information:

Surname:

First (given) name / initials:

Date of birth (dd-mm-yyyy):

Address:

Medical insurance policy number:

Material (where applicable);
please mark as appropriate

Blood:

Fibroblasts:

Other (please specify):

Date sample was taken (if applicable):

Gene variant information (HGVS nomenclature):

Gene Name	Reference transcript accession number	Nucleotide change (predicted protein variant)

Sender information

Name:

Email:

Department (if applicable):

Institute:

Address:

City:

Country:

Type of analysis requested:

- Diagnostic testing of a variant of uncertain clinical significance (VUS)
- Direct functional assay in patient material (eg. blood, fibroblasts, other)

Functional test requested (please specify):

- Ciliopathies (fibroblasts required)
 - Structural test, tubulin staining for cilia size and number
 - Hedgehog signaling test

DNA-repair defects (fibroblasts required)

- Xeroderma pigmentosum (XP)
- Ataxia telangiectasia (AT)
- Cockayne syndrome (CS)
- Nijmegen Breakage syndrome (NBS)
- Trichothiodystrophy (TTD)

Mechanistic target of rapamycin (mTOR) signaling pathway (*TSC1*, *TSC2*, *DEPDC5*, *NPRL2*, *NPRL3*, *AKT1*, *AKT3*, *TBC1D7*)

- mTOR complex 1 (TORC1) activity assay
- Signaling complex interaction test

Neurofibromatosis type 1

- RAS GTPase activating protein (GAP) assay
- Neurofibromin (NF1) - SPRED1 interaction assay

Lysosomal storage diseases

- GAA variant testing (Pompe disease)
- IDS* variant testing (Hunter disease, MPS II)
- ARSB* variant testing (Maroteaux Lamy, MPS VI)
- ELISA testing for immunoglobulin titers during ERT (Pompe disease, MPS)

Interferonopathies (Aicardi-Goutières syndrome)

- RNA expression profiling (6 gene signature; requires a blood or saliva sample collected in a PAX-tube).

RNA studies (please contact the laboratory for possibilities)

- RNA expression levels
- Investigation of DNA variants predicted to affect mRNA splicing

Fragile X syndrome (hair required)

- Hair root FMRP expression

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