Request form: Molecular genetic analysis
Eye diseases

Senckenberg Centre for Human Genetics

☐ URGENT

Insurance:

Surname, first name:

Date of birth: Sex: ☐female ☐male

Address:

Patient ID/No.:

Insurance/coverage of analysis costs

☐ Invoice to patient
☐ Invoice to institution
☐ Pre-payment
☐ E112 form (EU only)

Ordering physician (stamp, phone, fax, signature)

Clinical diagnosis/differential diagnosis/medical history (ICD-10 code)

Sample: Collection date_____

☐EDTA-blood ☐DNA ☐other material:

Report via:

☐ fax (see fax no. below)
☐ post (see post address below)
☐ other

_____
Signature of physician

Clinical information:

Patient is affected: ☐yes ☐no
Family members affected: ☐yes ☐no
Pregnancy (GW:____): ☐yes ☐ no
Parental consanguinity: ☐yes ☐no
Ethnic origin: _____

Informed consent

The nature, importance and implications of the genetic test/indication detailed below have been explained to you. With your signature, you agree to the performance of the genetic test and to the necessary collection of a blood/tissue sample.
You confirm (delete as appropriate) that

- you have been informed by your physician about the significance and consequences of the genetic test.
- you can withdraw your consent any time to halt the genetic testing procedure (only completed services would be charged).
- recorded data are stored in printed and electronic form and may be used/published in anonymized form for scientific purposes.
- remaining sample material will be available for verification of results, follow-up diagnostic testing requested by your physician, quality controls or scientific purposes.
- the test request may be forwarded to a specialized cooperating medical laboratory.
- the genetic test results may be made available not only to the requesting physician but also to other involved physicians (e.g. in genetic counselling units) as specified: _____.

In particular genome-wide analyses (e.g. whole-exome sequencing/WES, whole-genome sequencing, WGS) may yield incidental findings unrelated to the clinical diagnosis in question, but which may have consequences for you (prevention, therapy) or your family (e.g. disease risk for progeny). Do you wish to be informed about such incidental findings *(If you do not tick any, we will presume that you prefer "no")*?

☐ yes ☐ no

_____
(Genetic analysis: Method, covered diagnoses)

_____
Place, date Signature of the patient or the legal representative

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Please turn →

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Mode of inheritance

☐ autosomal recessive ☐ autosomal dominant ☐ X-linked
☐ unclear ☐ sporadic/simplex ☐ mitochondrial/maternal

For clarity, the list below is only a selection of disorders. If diagnoses you are looking should not be listed, please write them into the free field ("Others") or on the front page of this form. For most of the following indications, we apply NGS analyses taking into account the genes known to be associated with the respective disorder. We are happy to determine the appropriate NGS approach and prioritization of genes on an individual basis according to your needs.

Albinism
- ☐ ocular
- ☐ oculocutaneous
- ☐ syndromic

☐ **Blepharophimosis**

☐ **Duane syndrome**

Glaucoma
- ☐ Glaucoma, congenital
- ☐ Glaucoma, juvenile
- ☐ Glaucoma, others:

Corneal dystrophies
- ☐ Fuchs dystrophy
- ☐ Fabry disease
- ☐ Others: ...

☐ **Kearns-Sayre syndrome**

☐ **Congenital fibrosis of extraocular muscles**

Disorders of the lens
- ☐ Congenital nuclear cataract
- ☐ Galactokinase deficiency
- ☐ Classic galactosemia
- ☐ Cataract, syndromic (please specify): ...
- ☐ Primary lens luxation

☐ **Myopia**

☐ **Neuronal ceroid lipofuscinosis**

Retinal detachment
- ☐ Familial exudative vitreoretinopathy, FEVR
- ☐ Retinoschisis, juvenile, X-linked
- ☐ Stickler syndrome

☐ **Nystagmus, idiopathic**

Ocular malformations, eye development
- ☐ Anophthalmia, Microphthalmia, Coloboma
- ☐ Norrie disease

☐ **Ophthalmoplegia,** progressive external

☐ **Ptosis**

Retinal dystrophies
- ☐ Retinitis pigmentosa (RP)/rod-cone degeneration
- ☐ Congenital stationary night blindness, CSNB
- ☐ Cone/cone-rod/macular dystrophy
- ☐ Early-onset severe retinal dystrophy (EOSRD)
- ☐ Leber congenital amaurosis
- ☐ Stargardt disease
- ☐ Achromatopsia, ACHM
- ☐ Morbus Best
- ☐ Bardet-Biedl syndrome, BBS
- ☐ Joubert syndrome, JBTS
- ☐ Usher syndrome
  - ☐ type 1
  - ☐ type 2
  - ☐ type 3
- ☐ Senior Loken syndrome
- ☐ other: ...

Optic nerve
- ☐ Optic atrophy
- ☐ Leber hereditary optic atrophy, LHON
- ☐ Optic atrophy, syndromic

☐ **Septo-optic dysplasia**

Anterior segment dysgenesis
- ☐ Aniridia
- ☐ Neurofibromatosis
- ☐ Axenfeld-Rieger anomaly
- ☐ Anterior segment mesenchymal dysgenesis
- ☐ Iridogoniodysgenesis
- ☐ Peters/Peters Plus syndrome

☐ **Others:** ...

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