Request form: Molecular genetic analysis
Hearing loss/deafness

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 already 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).

In particular genome-wide analyses (e.g. whole-exome sequencing/WES, whole-genome sequencing, WGS) may yield incidental findings that are not related 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

Age of onset _____ **Progression** ☐ fast ☐ slow ☐ no

Type of deafness

☐ sensorineural ☐ conductive ☐ mixed ☐ central
☐ high frequencies ☐ low frequencies ☐ all frequencies

For clarity, the list below is only a selection of disorders. In case diagnoses you are looking for are not listed, please write them into the free field 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.

☐ **Non-syndromic**

☐ **DFNB1 (*GJB2*-/Connexin-26)**

☐ **Auditory neuropathy**

With eye involvement

☐ Usher syndrome, type 1/2/3
☐ PHARC (polyneuropathy, hearing loss, ataxia, retinits pigmentosa, cataract) syndrome
☐ Deafness with cone-rod dystrophy
☐ Alström syndrome
☐ Refsum syndrome
☐ Heimler syndrome
☐ Wolfram syndrome
☐ Stickler syndrome
☐ Harboyan syndrome (congenital corneal dystrophy and progressive sensorineural hearing loss)
☐ Ehlers-Danlos syndrome type 6
☐ Aniridia and sensorineural hearing loss
☐ Norrie syndrome (oculo-acoustico-cerebral dysplasia)
☐ OFCD syndrome (oculo-facio-cardio-dental syndrome)
☐ Optic atrophy plus (*OPA1*-related)

With endocrinological involvement

☐ Pendred syndrome
☐ Perrault syndrome
☐ DIDMOAD syndrome (diabetes insipidus, diabetes mellitus, optic atrophy, deafness), Wolfram syndrome
☐ Diabetes mellitus, thiamin-responsive megaloblastic anemia and sensorineural hearing loss/Rogers syndrome
☐ Johanson-Blizzard syndrome
☐ Resistance to thyroid hormone and sensorineural hearing loss
☐ HDR syndrome (hypoparathyroidism, deafness and renal disease)
☐ Pituitary hormone deficiency with hearing loss

Malformation/dysmorphic syndromes with deafness

☐ Treacher Collins syndrome
☐ Nager acrofacial dysostosis syndrome
☐ Postaxial acrofacial dysostosis, cupped ears, and conductive hearing loss (Miller syndrome, Genée-Wiedemann syndrome)
☐ Auriculo-condylar syndrome
☐ Towns-Brocks syndrome
☐ LADD (lacrimo-auriculo-dento-digital (syndrome
☐ CHARGE syndrome
☐ Cohen syndrome (incl. microcephaly, cutis verticis gyrata, retinitis pigmentosa, cataract, sensorineural deafness and mental retardation syndrome)
☐ Donnai-Barrow syndrome (facio-oculo-acoustico-renal syndrome, FOARS)
☐ Otopalatodigital syndrome type 1 (OPD1)
☐ Otopalatodigital syndrome type 2 (OPD2)
☐ OSMED (oto-spondylo-megaepiphyseal dysplasia)

With skin involvement

☐ Waardenburg syndrome
☐ Keratitis-ichthyosis-deafness syndrome (KID syndrome)
☐ CHIME (coloboma, heart defects, ichthyosis, mental retardation, ear) syndrome
☐ Björnstad syndrome (pili torti and sensorineural hearing loss)
☐ Woodhouse-Sakati-Syndrome (alopecia, hypogonadism, diabetes mellitus, mental retardation, abnormal ECG and sensorineural hearing loss)

With heart involvement

☐ Jervell and Lange Nielsen syndrome
☐ SANDD syndrome
☐ Kabuki syndrome
☐ Noonan syndrome

With neurological/neuromuscular symptoms

☐ Baraitser-Winter syndrome
☐ Brown-Vialetto-Van Laere syndrome, BVVS (pontobulbar palsy and sensorineural hearing loss)
☐ Cockayne syndrome
☐ Deafness-dystonia syndrome
☐ Chudley-McCullough syndrome
☐ SeSAME syndrome (seizures, sensorineural deafness, ataxia, mental retardation, electrolyte imbalance)
☐ Neurofibromatosis type 2
☐ Mohr-Tranebjaerg syndrome (deafness-dystonia-optic neuropathy syndrome/DDON syndrome)
☐ Autosomal dominant hearing loss with amyotrophy and sensorimotor neuropathy

With renal involvement

☐ Alport syndrome
☐ Branchio-oto-renal syndrome (BOR syndrome)
☐ Nephrotic syndrome with sensorineural deafness
☐ *MYH9*-associated disorders (May-Heggelin-anomaly, Epstein-/Fechtner-/Sebastian-syndrome)
☐ Distal renal tubular acidosis (DRTA) with progressive sensorineural hearing loss
☐ Bartter syndrome

☐ **Deafness, other syndromic:** _____

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