Patient data (please fill out clearly in **block letters**)

Family name

First name

Date of birth

Day Month Year

Pat. ID

Age

☐ female

☐ male

0001 160065780019

MOLECULAR GENETIC ANALYSES

BIOSCIENTIA
HUMAN GENETICS

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Client data

Physician

Sample data

☐ EDTA blood (3-5 ml)
☐ DNA (1-5 µg)
☐ Amniotic fluid
☐ Chorionic villi
☐ Others, please specify

Number of tubes

Sampling date

Time

Suspected diagnosis (for detailed clinical data, please fill in separate field on 2nd page)

Patient / proband is affected ☐ yes ☐ no

Family members affected ☐ yes ☐ no who

Parental consanguinity ☐ yes ☐ no

Ethnic origin

Declaration of Informed Consent

With my signature I declare that I was briefed by my physician: about the nature, importance and implications of the genetic test. With my signature I declare my agreement for the blood/tissue collection and the genetic examinations according to the enclosed request or clarification of the mentioned clinical question.

I have been informed that the recorded data are stored in paper form and/or in electronic form according to legal requirements. I understand that once results have been reported they are subject to the 10-year retention period and cannot be destroyed before their expiry even if requested by the investigated person. I agree that my data will be passed on to a medical clearing house for billing purposes. If necessary, the investigation order can be forwarded to a specialized cooperating laboratory.

I am aware that the diagnosis focuses on those changes that can be directly related to the clinical indication/diagnosis. In case of an extended analysis, I also agree with the reporting of the following findings:

- changes that are not related to the existing symptoms, but that lead to an increased risk of disease and whose knowledge leads to improved treatment and/or prevention (ACMG guideline Kalia et al. 2017; Genet Med). ☐ yes
- changes in genes not previously associated with disease, the variants of which can only be used for clinical decisions after further scientific investigation and confirmation. ☐ yes

I agree that

- the collected data will not be destroyed according to the legal requirements after 10 years, so that they will be available to my family even after my death. ☐ yes
- the collected data is stored and used in a pseudonymised form for scientific and quality assuring purposes. ☐ yes
- any sample material remaining at the end of the analysis is transferred to the laboratory that carried out the analysis and consent to its use for quality assurance and scientific purposes in pseudonymized form. ☐ yes

I am aware that I may withdraw this consent at any time, verbally or in writing, without giving reasons and without this having any adverse consequences for me.

Place and date: Signature of patient or legal guardian:

I have a declaration of consent including all above-mentioned subitems.

Place and date: Signature of attending physician:

specimen material | specimen material | specimen material | specimen material | specimen material | specimen material

This label should be stuck onto the copy attached and kept for your records.

DO NOT SEND TO US.

These labels are for the patient´s tube(s). Those labels not needed - please tear off and discard - do not send to us.

Request and clinical information

We offer a comprehensive spectrum of analyses. For more information please contact us or visit www.bioscientia-humangenetik.de/en

- ☐ **Whole Exome (WES)**
- ☐ **Whole Exome (WES) – Trio** (Use separate forms for each family member)
- ☐ **Multi-Gene panel (according to the suspected diagnosis)**
- ☐ **Customized panel including the following genes:** _____

_____

- ☐ **Single gene / single variant analysis**

Gene: _____

- ☐ Sequencing
- ☐ Deletion / Duplication
- ☐ Single variant: _____
- ☐ Repeat expansion

Array CGH ☐ 180k ☐ 400k (CGH + SNP)

Your clinical information and the patient's history are essential for a targeted and personalized analysis.

Clinical information (copies of medical reports and pedigree data are welcome)

Please checkmark the phenotype(s) presented by your patient:

Neurological abnormality
- ☐ Abnormalities of basal ganglia
- ☐ Agenesis of corpus callosum
- ☐ Ataxia
- ☐ Brain atrophy
- ☐ Chorea
- ☐ Cortical dysplasia
- ☐ Dystonia
- ☐ Hemimegalencephaly
- ☐ Heterotopia
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Leukoencephalopathy
- ☐ Lissencephaly
- ☐ Muscle weakness
- ☐ Muscular dystrophy
- ☐ Neuropathy
- ☐ Spasticity

Neurodevelopmental disorder
- ☐ Autism spectrum disorder
- ☐ Developmental delay
- ☐ Developmental regression
- ☐ Fine motor delay
- ☐ Gross motor delay
- ☐ Psychiatric symptoms
- ☐ Recurrent headache
- ☐ Seizures
- ☐ Speech delay

Metabolism abnormality
- ☐ Abnormal creatine kinase
- ☐ Elevated alanine
- ☐ Elevated pyruvate
- ☐ Failure to thrive
- ☐ Ketosis
- ☐ Lactic acidosis
- ☐ Organic aciduria

Hearing loss
- ☐ conductive
- ☐ sensorineural

Eye abnormality
- ☐ Abnormal eye movement
- ☐ Abnormal vision
- ☐ Blindness
- ☐ Cataract
- ☐ Coloboma
- ☐ Cone rod dystrophies
- ☐ CPEO
- ☐ Optic atrophy
- ☐ Ptosis
- ☐ Retinitis pigmentosa

Dysmorphic features
- ☐ Broad nasal bridge
- ☐ Cleft lip / palate
- ☐ Downslanted palpebral fissures
- ☐ Ear malformation
- ☐ Frontal bossing
- ☐ Hypertelorism
- ☐ Retrognathia
- ☐ Synophrys
- ☐ Upslanted palpebral fissures

Skeletal abnormality
- ☐ Club foot / feet
- ☐ Contractures
- ☐ Craniosynostosis
- ☐ Fractures
- ☐ Limb anomaly
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Overgrowth
- ☐ Polydactyly
- ☐ Short stature
- ☐ Scoliosis
- ☐ Syndactyly
- ☐ Vertebral anomaly

Cardiovascular abnormality
- ☐ Angioedema
- ☐ Aortic dilatation
- ☐ Arrhythmia
- ☐ Atrial septal defect
- ☐ Cardiomyopathy
- ☐ Coarctation of aorta
- ☐ Hypoplastic left heart
- ☐ Stroke
- ☐ Tetralogy of Fallot
- ☐ Ventricular septal defect

Hematologic disease
- ☐ Coagulation disorder
- ☐ Immunodeficiency
- ☐ Leukemia / Lymphoma
- ☐ Myelofibrosis
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Thrombocytopenia
- ☐ Thrombocytosis

Gastrointestinal disease
- ☐ Chronic diarrhea
- ☐ Constipation
- ☐ Cystic liver disease
- ☐ Gastroesophageal reflux
- ☐ Hirschsprung disease
- ☐ Increased transaminases
- ☐ Liver failure
- ☐ Pyloric stenosis
- ☐ Recurrent vomiting

Skin abnormality
- ☐ Blistering
- ☐ Connective tissue abnormality
- ☐ Hair anomaly
- ☐ Ichthyosis
- ☐ Nail anomaly
- ☐ Pigmentation anomaly
- ☐ Skin tumor

Kidney disease
- ☐ Focal segmental glomerulosclerosis
- ☐ Hydronephrosis
- ☐ Kidney dys- / agenesis
- ☐ Polycystic kidneys
- ☐ Proteinuria
- ☐ Renal tubulopathy

Genital anomaly
- ☐ Ambiguous genitalia
- ☐ Cryptorchidism
- ☐ Hypogonadism
- ☐ Hypospadia
- ☐ Infertility

Endocrinologic abnormality
- ☐ Diabetes mellitus
- ☐ Hyperthyroidism
- ☐ Hyperparathyroidism
- ☐ Hypoparathyroidism
- ☐ Hypoth. Hypophyseal disease
- ☐ Hypothyroidism

Prenatal anomaly
- ☐ Cystic hygroma
- ☐ Increased neck fold density
- ☐ Intrauterine growth retardation
- ☐ Oligohydramnios
- ☐ Omphalocele
- ☐ Polyhydramnios
- ☐ Preterm birth

Oncology
- ☐ Brain tumor
- ☐ Breast cancer
- ☐ Colon cancer
- ☐ Gastric cancer
- ☐ Ovarian cancer
- ☐ Lung cancer
- ☐ Renal cancer