MEDICOVER GENETICS

WHOLE EXOME SEQUENCING DECODE&DISCOVER

KLIINISET LISÄTIEDOT LÄHETTEESEEN HPO-TERMEIN

BARCODE

Nimi

Syntymäaika

Sukupuoli (nainen/mies/muu)

POSTNATAL ANALYSES

DEVELOPMENTAL

- ☐ Normal
- ☐ Premature birth (HP:0001622)
- ☐ Intrauterine growth retardation (IUGR, HP:0001511)
- ☐ Polyhydramnios (HP:0001561)
- ☐ Oligohydramnios (HP:0001562)
- ☐ Decreased fetal movement (HP:0001558)
- ☐ Intracranial hemorrhage (HP:0002170)
- ☐ Others:

DEVELOPMENTAL DISABILITIES/DISORDERS

- ☐ Not examined / unknown
- ☐ No developmental defects
- ☐ No intellectual disability
- ☐ Intellectual disability
  ☐ mild, ☐ moderate, ☐ severe
  (HP:0001256, HP:0002342, HP:0010864)
- ☐ Global developmental delay
  ☐ mild, ☐ moderate, ☐ severe
  (HP:0001263, HP:0011343, HP:0011344)
- ☐ Motor delay (HP:0001270)
- ☐ Delayed speech and language development (HP:0000750)
- ☐ Autism (HP:0000717)
- ☐ Developmental regression (HP:0002376)
- ☐ Others:

CRANIOFACIAL ABNORMALITIES

- ☐ Not examined / unknown
- ☐ No craniofacial abnormalities
- ☐ Macrocephaly (HP:000256)
- ☐ Microcephaly (HP:000252)
- ☐ Craniosynostosis (HP:0001363)
- ☐ Broad forehead / Prominent forehead (HP:0000337 / HP:0011220)
- ☐ Oral cleft (HP:0000202)
- ☐ Hypertelorism (HP:0000316)
- ☐ Hypotelorism (HP:0000601)
- ☐ Nasal abnormalities: _____
- ☐ Abnormality of the outer ear (HP:0000356): _____
- ☐ Micrognathia (HP:0000347)
- ☐ Oligodontia (HP:0000677)
- ☐ Others:

BRAIN ABNORMALITIES

- ☐ Not examined / unknown
- ☐ Normal brain MRI
- ☐ Aplasia/Hypoplasia of the corpus callosum (HP:0007370)
- ☐ Agenesis of corpus callosum (HP:0001274)
- ☐ Aplasia/Hypoplasia of the cerebellum (HP:0007360)
- ☐ Aplasia/Hypoplasia of the cerebellar vermis (HP:0006817)
- ☐ Abnormal myelination (HP:0012447)
- ☐ Lissencephaly (HP:0001339)
- ☐ Schizencephaly (HP:0010636)
- ☐ Porencephalic cyst (HP:0002132)
- ☐ Pachygyria (HP:0001302)
- ☐ Polymicrogyria (HP:0002126)
- ☐ Gray matter heterotopia (HP:0002282)
- ☐ Abnormality of the basal ganglia (HP:0002134)
- ☐ Leukoencephalopathy (HP:0002352)
- ☐ Brain atrophy (HP:0012444)
- ☐ Ventriculomegaly (HP:0002119)
- ☐ Hydrocephalus (HP:0000238)
- ☐ Holoprosencephaly (HP:0001360)
- ☐ Others:

RESPIRATORY DIFFICULTIES AND RESPIRATORY SYMPTOMS

- ☐ Not examined / unknown
- ☐ No respiratory abnormalities
- ☐ Respiratory insufficiency (HP:0002093)
- ☐ Respiratory failure (HP:0002878)
- ☐ Recurrent infections (HP:0002719)
- ☐ Bronchiectasis (HP:0002110)
- ☐ Others:

NEUROLOGICAL SYMPTOMS

- ☐ No neurological symptoms
- ☐ Seizures (☐ generalized / ☐ focal)
- ☐ Encephalopathy (HP:0001298)
- ☐ Decreased nerve conduction velocity (HP:0000762)
- ☐ Neuropathy (☐ motor / ☐ sensory)
- ☐ Ataxia (HP:0001251)
- ☐ Tremor (HP:0001337)
- ☐ Dystonia (HP:0001332)
- ☐ Chorea (HP:0002072)
- ☐ Spasticity (HP:0001257)
- ☐ Gait disturbance (HP:0001288)
- ☐ Nystagmus (HP:0000639)
- ☐ Migraine (HP:0002076)
- ☐ Sleep disturbance (HP:0002360)
- ☐ Others:

EYE DEFECTS

- ☐ Not examined / unknown
- ☐ No eye defects
- ☐ Abnormality of vision (HP:0000504):
  _____
  (bilateral? ☐ Yes / ☐ No)
- ☐ Retinopathy (HP:0000488)
- ☐ Anophthalmia (HP:0000528)
  (bilateral? ☐ Yes / ☐ No)
- ☐ Microphthalmos (HP:0007633)
  (bilateral? ☐ Yes / ☐ No)
- ☐ Strabismus (HP:0000486)
  (bilateral? ☐ Yes / ☐ No)
- ☐ Developmental cataract (HP:0000519)
- ☐ Others:

HEARING DEFECTS AND BALANCE DISORDERS

- ☐ Not examined / unknown
- ☐ No hearing defects
- ☐ No balance disorder
- ☐ Sensorineural hearing impairment (HP:0000407)
  (bilateral? ☐ Yes / ☐ No)
- ☐ Conductive hearing impairment (HP:0000405)
- ☐ Vestibular dysfunction: (HP:0001751)
- ☐ Others:

MUSCULOSKELETAL DISORDERS

- ☐ Not examined / unknown
- ☐ No muscular abnormalities
- ☐ No skeletal abnormalities
- ☐ Hypotonia (HP:0001252)
- ☐ Hypertonia (HP:0001276)
- ☐ Elevated circulating creatine kinase concentration (HP:0003236)
- ☐ Ptosis (HP:0000508)
- ☐ Distal arthrogryposis (HP:0005684)
- ☐ Arthrogryposis multiplex congenita (HP:0002804)
- ☐ Short stature (HP:0004322)
- ☐ Skeletal dysplasia (HP:0002652)
- ☐ Tall stature (HP:0000098)
- ☐ Joint hypermobility (HP:0001382)
- ☐ Hand polydactyly / ☐ Foot polydactyly (HP:0001161/HP:0001829)
- ☐ Hand Syndactyly / ☐ Foot Syndactyly, specify: _____
- ☐ Camptodactyly of finger (HP:0100490)
- ☐ Talipes (HP:0001883)
- ☐ Scoliosis (HP:0002650)
- ☐ Pectus carinatum (HP:0000768)
- ☐ Increased bone mineral density (HP:0011001)
- ☐ Osteoporosis (HP:0000939)
- ☐ Delayed skeletal maturation (HP:0002750)
- ☐ Multiple exostoses (HP:0002762)
- ☐ Others:

CARDIOVASCULAR DISEASES

- ☐ Not examined / unknown
- ☐ No cardiovascular abnormalities
- ☐ Atrial septal defect (HP:0001631)
- ☐ Ventricular septal defect (HP:0001629)
- ☐ Pulmonic stenosis (HP:0001642)
- ☐ Heart defect: _____
- ☐ Cardiomyopathy: _____
- ☐ Hypertrophic cardiomyopathy (HP:0001639)
- ☐ Dilated cardiomyopathy (HP:0001644)
- ☐ Arrhythmia (HP:0011675)
- ☐ Aortic aneurysm (HP:0004942)
- ☐ Abnormality of the vasculature (HP:0002597)
- ☐ Pulmonary arterial hypertension (HP:0002092)
- ☐ Others:

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IMMUNOLOGICAL AND HEMATOLOGICAL ABNORMALITIES

- ☐ Not examined / unknown
- ☐ No immunological abnormalities
- ☐ No hematological abnormalities
- ☐ Abnormal inflammatory response (HP:0012647)
- ☐ Immunodeficiency (HP:0002721)
- ☐ Recurrent infections (HP:0002719)
- ☐ Anemia (HP:0001903):_____
- ☐ Neutropenia (HP:0001875)
- ☐ Thrombocytopenia (HP:0001873)
- ☐ Abnormality of coagulation (HP:0001928)
- ☐ Abnormality of circulating enzyme level (Abnormality of iron homeostasis HP:0011021)
- ☐ Others:

ABNORMALITIES OF THE SKIN, NAILS AND HAIR

- ☐ No abnormalities of the skin, nails and hair
- ☐ Multiple cafe-au-lait spots (HP:0007565)
- ☐ Nevus (HP:0003764)
- ☐ Albinism (HP:0001022)
- ☐ Hypopigmentation of the skin (HP:0001010)
- ☐ Hyperpigmentation of the skin (HP:0000953)
- ☐ Eczema (HP:0000964)
- ☐ Ichthyosis (HP:0008064)
- ☐ Nail dysplasia (HP:0002164)
- ☐ Anhidrosis (HP:0000970)
- ☐ Hyperhidrosis (HP:0000975)
- ☐ Alopecia (HP:0001596)
- ☐ Hypertrichosis (HP:0000998)
- ☐ Others:

ABNORMALITIES OF THE KIDNEY AND UROGENITAL TRACT

- ☐ No renal abnormalities
- ☐ No urogenital abnormalities
- ☐ Not examined / unknown
- ☐ Renal agenesis (HP:0000104)
- ☐ Renal dysplasia (HP:0000110)
- ☐ Renal cyst (HP:0000107)
- ☐ Hematuria (HP:0000790)
- ☐ Proteinuria (HP:0000093)
- ☐ Hypospadias (HP:0000047)
- ☐ Cryptorchidism (HP:0000028)
- ☐ Ambiguous genitalia (HP:0000062)
- ☐ Others:

METABOLIC AND ENDOCRINE ABNORMALITIES

- ☐ Not examined / unknown
- ☐ No metabolic abnormalities
- ☐ No endocrine abnormalities
- ☐ Failure to thrive (HP:0001508)
- ☐ Hemihypertrophy (HP:0001528)
- ☐ Obesity (HP:0001513)
- ☐ Abnormality of the mitochondrion (HP:0012103)
- ☐ Lactic acidosis (HP:0003128)
- ☐ Proteinuria (HP:0000093)
- ☐ Hyperglycemia (HP:0003074)
- ☐ Hypoglycemia (HP:0001943)
- ☐ Ketosis (HP:0001946)
- ☐ Diabetes mellitus (HP:0000819)
- ☐ Nephrogenic diabetes insipidus (HP:0009806)
- ☐ Hypothyroidism (HP:0000821)
- ☐ Hypercalcemia (HP:0003072)
- ☐ Hypoparathyroidism (HP:0000829)
- ☐ Exocrine pancreatic insufficiency (HP:0001738)
- ☐ Hypogonadism (HP:0000135)
- ☐ Others:

OTHER ABNORMALITIES

PRENATAL ANALYSES

(please contact us via: exom.support@medizinische genetik.de before ordering a prenatal analysis)

Prenatal trio exomes are only analyzed for variants in known disease-associated genes (clinical exome). The focus of this analysis is on genes that are associated with the abnormalities in the ultrasound and the suspected diagnosis. Only variants that are classified as likely pathogenic (class 4) or pathogenic (class 5) according to ACMG criteria are reported.

- Gestational Age (Week + Day) according to ultrasound / [Add Local Language]: ☐ + ☐
- Number of Fetuses / [Add Local Language]: ☐
- Sex of Fetus / [Add Local Language]: ☐ Female ☐ Male ☐ Unknown
- Gender Information / [Add Local Language] ☐ Yes ☐ No
- Ultrasound Abnormalities (please attach ultrasound findings) / [Add Local Language]:

SKELETAL MALFORMATIONS

- ☐ Skeletal dysplasias
- ☐ Craniosynostoses
- ☐ Limb malformations
- ☐ Others:

BRAIN ABNORMALITIES

- ☐ Holoprosencephaly (HP:0001360)
- ☐ Agenesis of corpus callosum (HP:0001274)
- ☐ Abnormal cortical gyration (HP:0002536)
- ☐ Microcephaly (HP:0000252)
- ☐ Others:

RENAL MALFORMATION

- ☐ Polycystic kidney dysplasia (HP:000113)
- ☐ Renal dysplasia and abnormality of the lower urinary tract (CAKUT) (HP:0000110, HP:0010936)
- ☐ Others:

CARDIOVASCULAR DISEASES

- ☐ Abnormal heart morphology (HP:0001627)
- ☐ Cardiomyopathy (HP:0001638)
- ☐ Arrhythmia (HP:0011675)
- ☐ Others:

- ☐ Hydrops fetalis (HP:0001789)
- ☐ Rasopathy
- ☐ Fetal akinesia
- ☐ Multiple organ malformations
- ☐ Bilateral microphthalmos (HP:0007633) / Anophthalmia (HP:0000528)

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