

Κλινικά στοιχεία ασθενούς:

Παρακαλώ όπως συμπληρώσετε τις παρακάτω κλινικές πληροφορίες για τον ασθενή βασιζόμενη στη βάση δεδομένων Human Phenotype Ontology - HPO.

Η λεπτομερής καταγραφή των κλινικών πληροφοριών αυξάνει την πιθανότητα ανίχνευσης της γενετικής αιτίας του φαινοτύπου του ασθενούς κατά την ανάλυση των δεδομένων από την εξέταση αλληλούχησης επόμενης γενιάς.

Prenatal medical history:

- Normal
- Prematurity
- Intrauterine growth restriction (IUGR)
- Poly- / Oligohydramnios
- Decreased fetal movement
- Other:.....

Developmental disorders

- Intellectual disability
(mild, moderate, severe)
- Global developmental delay
(mild, moderate, severe)
- Delayed motor milestones
- Delayed speech / language development
- Learning difficulties
- Autism spectrum disorder
- Developmental regression
- Other:.....

- No intellectual disability
- No developmental disorder
- Not examined / unknown

Craniofacial anomalies

- Macrocephaly
- Microcephaly
- Craniosynostosis
- Broad forehead
- Cleft lip palate
- Hypertelorism
- Hypotelorism
- Abnormality of the nose
(Please specify:.....)
- Abnormal ears
(Please specify:))
- Micrognathia
- Oligodontia
- Other:.....

- No craniofacial anomalies
- Not examined / unknown

Respiratory symptoms

- Respiratory insufficiency
- Respiratory failure
- Apnea
- Recurrent infections
- Bronchiectasis
- Pneumothorax
- Other:.....

- No respiratory symptoms
- Not examined / unknown

Brain abnormalities

- Lissencephaly
- Schizencephaly
- Porencephaly
- Pachygyria
- Polymicrogyria
- Band heterotopia
- Abnormality of corpus callosum
(Please specify:))

- Hydrocephalus
- Holoprosencephaly
- Abnormality of basal ganglia
- Leukoencephalopathy
- Brain atrophy
- Ventriculomegaly
- Other:.....

- Normal brain MRI
- Not examined / unknown

Neurological symptoms

- Seizures (generalized/ focal)
- Encephalopathy
- Abnormal nerve conduction velocity
- Neuropathy (motor/ sensory)
- Ataxia
- Tremor
- Dystonia
- Chorea
- Spasticity
- Gait disturbances
- Nystagmus
- Mood disturbances
(anxiety, depression, psychosis)
- Migraine, Headaches
- Sleep disturbances
- Unexplained pain
- Other:.....

- No neurological symptoms
- Not examined / unknown

Hearing defects

- Sensorineural hearing impairment
(bilateral? yes/ no)
- Conductive hearing impairment
(bilateral? yes/ no)
- Other:.....

- No hearing defects
- No vestibular abnormalities
- Not examined / unknown

Neuromuscular symptoms

- Ataxia
- Chorea
- Dystonia
- Hypotonia
(generalized, central, muscular)
- Hypertonia
- Muscle weakness
- Muscular dystrophy
- Neuropathy
- Myotonia
- Spasticity
- Elevated creatine kinase
(Please specify:))
- Ptosis
- Other:.....

- No neuromuscular symptoms
- Not examined / unknown

Skeletal symptoms

- Flexion contracture
- Arthrogryposis (congenital? yes/ no)
- Short stature
(skeletal dysplasia? yes/ no)
- Tall stature (overgrowth? yes/ no)
- Joint Hypermobility
- Hand-/ Foot polydactyly
(bilateral? yes/ no)
- Hand-/ Foot syndactyly
(bilateral? yes/ no)
- Camptodactyly of finger
- Clubfoot (congenital? yes/ no)
- Scoliosis
- Kyphosis
- Pectus excavatum
- Pectus carinatum
- Abnormality of bone density
(increased/ decreased)
- Exostosis
- Fractures
- Delayed bone age
- Other:.....

- No skeletal abnormalities
- Not examined / unknown

Cardiovascular defects

- Atrial septal defect
- Ventricular septal defect
- Abnormality of cardiac ventricle
- Tetralogy of Fallot
- Cardiomyopathy
- Arrhythmia
- Aortic aneurysm
- Abnormality of vasculature
(Please specify:)

- Pulmonary arterial hypertension
- Pulmonary arterial stenosis
- Valve abnormalities
- Other:.....

- No cardiac abnormalities
- Not examined / unknown

Skin, nails and hair

- Abnormality of connective tissue
(Please specify:)
- Multiple cafe-au-lait spots
- Port-wine stain
- Albinism
- Progeroid appearance
- Skin lesions
- Eczema
- Edema
- Ichthyosis
- Dysplastic nails
- Anhidrosis
- Hyperhidrosis
- Alopecia
- Hypertrichosis (Where?)
- Other:.....

- No abnormalities of skin, nails and hair
- Not examined / unknown

Other

- Organomegaly (which?)
- Neoplasm / Cancer
- Pancreatitis
- Episodic fever
- Hyperthermia
- Hypothermia
- Constipation, Obstipation
- Diarrhea
- Episodic vomiting
- Other:.....

Notes

.....

.....

.....

.....

.....

.....

Eye defects

- Visual impairment (bilateral? yes / no)
(Please specify:)
- Anophthalmia / Microphthalmia
(bilateral? yes / no)
- Strabismus (bilateral? yes / no)
- Coloboma
- Congenital bilateral cataract
- Blue sclera
- Optic atrophy
- Blindness
- Retinitis pigmentosa
- Other:.....

- No eye defects
- Not examined / unknown

Immunological and hematological abnormalities

- Autoinflammatory disease
- Immunodeficiency
(Please specify:)
- Recurrent infections
- Anemia (Erythrocytes)
- Neutropenia
- Thrombocytopenia
- Abnormal coagulation
- Megaloblastic anemia
- Bone marrow failure
- Leukemia
(Please specify:)
- Hemochromatosis
- Other:.....

- No immunological abnormalities
- No hematological abnormalities
- Not examined / unknown

Hepatic dysfunction

- Liver dysfunction
(Please specify:)
- Recurrent acute liver failure
- Hepatic cysts
- Cholestasis
- Hypercholanemia
- Hepatomegaly
- Other:.....

- No hepatic abnormalities
- Not examined / unknown

Metabolic and endocrine defects

- Failure to thrive
- Obesity
- Suspected mitochondriopathy
- Lactic acidosis
- Proteinuria
- Hyperglycemia
- Hypoglycemia
- Ketosis
- Hypercalcemia
- Diabetes mellitus
- Diabetes insipidus
- Hypothyroidism
- Hypoparathyroidism
- Exocrine pancreatic insufficiency
- Other:.....

- No metabolic abnormalities
- No endocrine abnormalities
- Not examined / unknown
- Copy of laboratory findings attached

Renal and genitourinary tract abnormalities

- Renal cysts
- Renal agenesis
- Horseshoe kidney
- Hypercalciuria
- Hematuria
- Proteinuria
- Hypospadias
- Hydronephrosis
- Hypogonadism
- Cryptorchidism
- Ambiguous genitalia
- Other:.....

- No renal abnormalities
- No genitourinary abnormalities
- Not examined / unknown