MitoMatcher
Phenotype data collection form

mitodiag.fr/documents

Database user agreement:
mitodiag.fr/data/Charte_utilisation_mitomatcher_20211119.pdf
Patient data collection information and agreement:
mitodiag.fr/data/Lettre_information_consentement_mitomatcher_20211119.pdf
Lastest MitoMatcher Data Collection form version:
https://www.mitodiag.fr/data/Mitomatcher-Phenotypic-Data-Collection.pdf

Usage advice: This form is not fillable with Adobe. Use **Master PDF Editor, okular** or other PDF viewer software to display and fill it out. Save the document to your computer under a **meaningful name**, and make sure it is **read only** once it's complete. If you use google chrome to edit and save the file, the fillable fields will be saved in simple flat format and parsing it automatically won't be possible. Consequently, avoid google chrome if possible.

Clinical - Patient overview

Patient pseudonym	
Sample pseudonym	
Date of birth	
Sex	
Age of onset	
Consanguinity	

Sample – Information

Sample ID	
Age at sampling	
Date of collection	22/01/2022
Tissue	
Haplogroup	

Sample – Origin

Reference center	
Sampling center	
Collector's e- mail	

Analysis – Information

Library	
Sequencer	
Mapper/Caller/Pipeline	
Date of analysis	22/01/2022

Clinical information

Leigh syndrome

Leigh Syndrome	☐ (ORPHA:506)
Leigh Syndrome with Cardiomyopathy	☐ (ORPHA:70474)
Leigh Syndrome with Nephrotic Syndrome	☐ (ORPHA:255249)
Leigh Syndrome with Leukodystrophy	☐ (ORPHA:255241)

MELAS, MIDD

MELAS (Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke)	☐ (ORPHA:550)
Maternally-inherited Diabetes And Deafness	☐ (ORPHA:225)

MERRF

MERRF (myoclonus epilepsy with ragged-red fibers)	☐ (ORPHA:551)

NARP

NARP Syndrome	☐ (ORPHA:644)

Leber

Leber Hereditary Optic Neuropathy	☐ (ORPHA:104)

Other

Pearson Syndrome	☐ (ORPHA:699)
Kearns-Sayre Syndrome	☐ (ORPHA:480)
CPEO (Mitochondrial Dna-related Progressive External Ophthalmoplegia)	☐ (ORPHA:663)

Atypical clinic ☐ check if the clinic is atypical, overlapping

Asymptomatic relative ☐ check if asymptomatic

Detailed phenotypes

The ‘HP:number’ corresponds to the Human Phenotype Ontology categorization: https://hpo.jax.org/app/.

- **Normal** *(N)*: phenotype was investigated and was determined to be normal.
- **Abnormal** *(A)*: phenotype was investigated and was determined to be abnormal.
- **Indeterminate** *(I)*: phenotype was investigated and a conclusion could not be made.
- **Unknown**: *(U)*: phenotype is known to not have been investigated.
- Blank *(default)*: absence of information on the phenotype’s status (not know if it was **Not Investigated**, total absence of data).

Biology

3-Methylglutaconic aciduria HP:0003535	☐ N ☐ A ☐ U ☐ I
Metabolic acidosis HP:0001942	☐ N ☐ A ☐ U ☐ I
Methylmalonic acidemia HP:0002912	☐ N ☐ A ☐ U ☐ I
Organic aciduria HP:0001992	☐ N ☐ A ☐ U ☐ I
Abnormality of Krebs cycle metabolism HP:0000816	☐ N ☐ A ☐ U ☐ I
Elevated lactate:pyruvate ratio HP:0032653	☐ N ☐ A ☐ U ☐ I
Hyperalaninemia HP:0003348	☐ N ☐ A ☐ U ☐ I
Elevated circulating creatine kinase concentration HP:0003236	☐ N ☐ A ☐ U ☐ I
Increased serum lactate HP:0002151	☐ N ☐ A ☐ U ☐ I
Increased CSF lactate HP:0002490	☐ N ☐ A ☐ U ☐ I
Malabsorption of Vitamin B12 HP:0200118	☐ N ☐ A ☐ U ☐ I

Cardiology

Abnormal heart morphology HP:0001627	☐ N ☐ A ☐ U ☐ I
Arrhythmia HP:0011675	☐ N ☐ A ☐ U ☐ I
Cardiomyopathy HP:0001638	☐ N ☐ A ☐ U ☐ I
Dilated cardiomyopathy HP:0001644	☐ N ☐ A ☐ U ☐ I
Hypertrophic cardiomyopathy HP:0001639	☐ N ☐ A ☐ U ☐ I
Restrictive cardiomyopathy HP:0001723	☐ N ☐ A ☐ U ☐ I
Congestive heart failure HP:0001635	☐ N ☐ A ☐ U ☐ I
Cardiac conduction abnormality HP:0031546	☐ N ☐ A ☐ U ☐ I
Left ventricular noncompaction HP:0030682	☐ N ☐ A ☐ U ☐ I

Digestive

Cholestasis HP:0001396	☐	N	☐	A	☐	U	☐	I
Elevated hepatic transaminase HP:0002910	☐	N	☐	A	☐	U	☐	I
Macronodular cirrhosis HP:0006577	☐	N	☐	A	☐	U	☐	I
Micronodular cirrhosis HP:0001413	☐	N	☐	A	☐	U	☐	I
Constipation HP:0002019	☐	N	☐	A	☐	U	☐	I
Diarrhea HP:0002014	☐	N	☐	A	☐	U	☐	I
Hepatomegaly HP:0002240	☐	N	☐	A	☐	U	☐	I
Hepatic failure HP:0001399	☐	N	☐	A	☐	U	☐	I
Exocrine pancreatic insufficiency HP:0001738	☐	N	☐	A	☐	U	☐	I
Intestinal pseudo-obstruction HP:0004389	☐	N	☐	A	☐	U	☐	I
Steatorrhea HP:0002570	☐	N	☐	A	☐	U	☐	I
Hepatic steatosis HP:0001397	☐	N	☐	A	☐	U	☐	I

Miscellaneous

Family history HP:0032316	☐	N	☐	A	☐	U	☐	I
Abnormal facial shape HP:0001999	☐	N	☐	A	☐	U	☐	I
Hyperpigmentation of the skin HP:0000953	☐	N	☐	A	☐	U	☐	I
Hydrops fetalis HP:0001789	☐	N	☐	A	☐	U	☐	I
Lipoma HP:0001012	☐	N	☐	A	☐	U	☐	I
Behavioral abnormality HP:0000708	☐	N	☐	A	☐	U	☐	I

Endocrinology

Type I diabetes mellitus HP:0100651 ☐ N ☐ A ☐ U ☐ I
Type II diabetes mellitus HP:0005978 ☐ N ☐ A ☐ U ☐ I
Diabetes mellitus HP:0000819 ☐ N ☐ A ☐ U ☐ I
Decreased response to growth hormone stimulation test HP:0000824 ☐ N ☐ A ☐ U ☐ I
Adrenal insufficiency HP:0000846 ☐ N ☐ A ☐ U ☐ I
Hypoparathyroidism HP:0000829 ☐ N ☐ A ☐ U ☐ I
Hypothyroidism HP:0000821 ☐ N ☐ A ☐ U ☐ I
Intrauterine growth retardation HP:0001511 ☐ N ☐ A ☐ U ☐ I
Failure to thrive HP:0008916 ☐ N ☐ A ☐ U ☐ I

Hematology

Anemia HP:0001903 ☐ N ☐ A ☐ U ☐ I
Abnormality of blood and blood-forming tissues HP:0001871 ☐ N ☐ A ☐ U ☐ I
Neutropenia HP:0001875 ☐ N ☐ A ☐ U ☐ I
Pancytopenia HP:0001876 ☐ N ☐ A ☐ U ☐ I
Thrombocytopenia HP:0001873 ☐ N ☐ A ☐ U ☐ I

Histology

Cytochrome C oxidase-negative muscle fibers HP:0003688 ☐ N ☐ A ☐ U ☐ I
Ragged-red muscle fibers HP:0003200 ☐ N ☐ A ☐ U ☐ I
Increased muscle lipid content HP:0009058 ☐ N ☐ A ☐ U ☐ I

Imagery

Stroke HP:0001297 ☐ N ☐ A ☐ U ☐ I
Abnormal brainstem morphology HP:0002363 ☐ N ☐ A ☐ U ☐ I
Abnormality of thalamus morphology HP:0010663 ☐ N ☐ A ☐ U ☐ I
Aplasia/Hypoplasia of the cerebellar vermis HP:0006817 ☐ N ☐ A ☐ U ☐ I
Abnormal cerebral white matter morphology HP:0002500 ☐ N ☐ A ☐ U ☐ I
Abnormality of the basal ganglia HP:0002134 ☐ N ☐ A ☐ U ☐ I
Abnormal basal ganglia MRI signal intensity HP:0012751 ☐ N ☐ A ☐ U ☐ I
Cerebellar atrophy HP:0001272 ☐ N ☐ A ☐ U ☐ I
Cerebral calcification HP:0002514 ☐ N ☐ A ☐ U ☐ I
Microcephaly HP:0000252 ☐ N ☐ A ☐ U ☐ I
Stroke-like episode HP:0002401 ☐ N ☐ A ☐ U ☐ I

Immunology

Recurrent infections HP:0002719 ☐ N ☐ A ☐ U ☐ I

Muscle

Skeletal muscle atrophy HP:0003202 ☐ N ☐ A ☐ U ☐ I

Distal muscle weakness HP:0002460 ☐ N ☐ A ☐ U ☐ I

Proximal muscle weakness HP:0003701 ☐ N ☐ A ☐ U ☐ I

Muscle weakness HP:0001324 ☐ N ☐ A ☐ U ☐ I

Hypotonia HP:0001252 ☐ N ☐ A ☐ U ☐ I

Muscular hypotonia of the trunk HP:0008936 ☐ N ☐ A ☐ U ☐ I

Exercise intolerance HP:0003546 ☐ N ☐ A ☐ U ☐ I

Myalgia HP:0003326 ☐ N ☐ A ☐ U ☐ I

Ophtalmoplegia (except external ophtalmoplegia) HP:0000602 ☐ N ☐ A ☐ U ☐ I

External ophthalmoplegia HP:0000544 ☐ N ☐ A ☐ U ☐ I

Ptosis HP:0000508 ☐ N ☐ A ☐ U ☐ I

NC

Ataxia HP:0001251 ☐ N ☐ A ☐ U ☐ I

Intellectual disability HP:0001249 ☐ N ☐ A ☐ U ☐ I

Dystonia HP:0001332 ☐ N ☐ A ☐ U ☐ I

Encephalopathy HP:0001298 ☐ N ☐ A ☐ U ☐ I

Seizure HP:0001250 ☐ N ☐ A ☐ U ☐ I

Epilepsia partialis continua HP:0012847 ☐ N ☐ A ☐ U ☐ I

Migraine HP:0002076 ☐ N ☐ A ☐ U ☐ I

Abnormality of movement HP:0100022 ☐ N ☐ A ☐ U ☐ I

Myoclonus HP:0001336 ☐ N ☐ A ☐ U ☐ I

Spastic paraparesis HP:0002313 ☐ N ☐ A ☐ U ☐ I

Global developmental delay HP:0001263 ☐ N ☐ A ☐ U ☐ I

Developmental regression HP:0002376 ☐ N ☐ A ☐ U ☐ I

Parkinsonism HP:0001300 ☐ N ☐ A ☐ U ☐ I

Autistic behavior HP:0000729 ☐ N ☐ A ☐ U ☐ I

Cognitive impairment HP:0100543 ☐ N ☐ A ☐ U ☐ I

Nephrology

Abnormality of the kidney HP:0000077	☐	N	☐	A	☐	U	☐	I
Renal insufficiency HP:0000083	☐	N	☐	A	☐	U	☐	I
Nephrotic syndrome HP:0000100	☐	N	☐	A	☐	U	☐	I
Proximal tubulopathy HP:0000114	☐	N	☐	A	☐	U	☐	I

NP

Motor neuron atrophy HP:0007373	☐	N	☐	A	☐	U	☐	I
Peripheral neuropathy HP:0009830	☐	N	☐	A	☐	U	☐	I
Sensorimotor neuropathy HP:0007141	☐	N	☐	A	☐	U	☐	I
Sensory neuropathy HP:0000763	☐	N	☐	A	☐	U	☐	I
Chronic axonal neuropathy HP:0007267	☐	N	☐	A	☐	U	☐	I
Demyelinating peripheral neuropathy HP:0007108	☐	N	☐	A	☐	U	☐	I
Motor polyneuropathy HP:0007178	☐	N	☐	A	☐	U	☐	I

NS

Optic atrophy HP:0000648	☐	N	☐	A	☐	U	☐	I
Cataract HP:0000518	☐	N	☐	A	☐	U	☐	I
Nystagmus HP:0000639	☐	N	☐	A	☐	U	☐	I
Pigmentary retinopathy HP:0000580	☐	N	☐	A	☐	U	☐	I
Hearing impairment HP:0000365	☐	N	☐	A	☐	U	☐	I
Sensorineural hearing impairment HP:0000407	☐	N	☐	A	☐	U	☐	I
Macular dystrophy HP:0007754	☐	N	☐	A	☐	U	☐	I
Optic neuropathy HP:0001138	☐	N	☐	A	☐	U	☐	I

Cellular phenotype

Abnormal activity of mitochondrial respiratory chain HP:0011922	☐	N	☐	A	☐	U	☐	I
Decreased activity of mitochondrial ATP synthase complex HP:0011925	☐	N	☐	A	☐	U	☐	I
Decreased activity of mitochondrial complex I HP:0011923	☐	N	☐	A	☐	U	☐	I
Decreased activity of mitochondrial complex II HP:0008314	☐	N	☐	A	☐	U	☐	I
Decreased activity of mitochondrial complex III HP:0011924	☐	N	☐	A	☐	U	☐	I
Decreased activity of mitochondrial complex IV HP:0008347	☐	N	☐	A	☐	U	☐	I