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### PATIENT DETAILS

First name(s): ..... Surname: .....  
 Maiden name: ..... Date of birth: \_\_\_\_\_ Gender:  F  M  
 Clinician: .....

**Date and time of sampling:**  
 \_\_\_\_\_ at \_\_\_\_\_ hrs \_\_\_\_\_ min

**Start of treatment (date):**  
 \_\_\_\_\_

### HISTORY-ANAMNESIS

Death amongst siblings  Familial consanguinity

### BEGINNING OF SYMPTOMS

Since birth  During neonatal period  Later (*please specify*): .....

### TREATMENT IN PROGRESS

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 .....

### CLINICAL SYMPTOMS\*

**Morphotype**

- Hypotrophy
- Facial dysmorphia
- Marfanoid appearance
- Other (*please specify*): .....

**Neuromuscular symptoms**

- Hypotonia
- Hypertonia
- Ataxia
- Psychomotor delay
- Psychomotor regression
- Behavioural problems
- Neuropathy
- Convulsions
- Consciousness problems
- Coma
- Macro/microcephaly
- Dyskinesia
- Myopathy
- Other (*please specify*): .....

**Cardiovascular symptoms**

- Hypertrophic/Dilated cardiomyopathy
- Vein and/or arterial thrombosis
- Fibromuscular dysplasia
- Short R-P
- Other (*please specify*): .....

**Digestive symptoms**

- Hepatomegaly
- Splenomegaly
- Nausea
- Eating difficulties
- Inguinal and/or umbilical hernia
- Other (*please specify*): .....

**Dermatological symptoms**

- Angiokeratoma, telangiectasia
- Cutaneous infiltration
- Eczema
- Hair abnormality
- Periarticular node
- Inverted nipples
- Other (*please specify*): .....

**Osteoarticular symptoms**

- Tendon retraction
- Osteoporosis
- Dysostosis multiplex
- Kyphoscoliosis
- Other (*please specify*): .....

**Ophthalmological symptoms**

- Cataract
- Crystalline luxation
- Retinitis Pigmentosa
- Cherry-red stain
- Optic atrophy
- Other (*please specify*): .....

**Renal symptoms**

- Tubulopathy
- Renal failure
- Renal cysts
- Lithiasis
- Other (*please specify*): .....

**Oral symptoms**

- Chronic or recurrent otitis
- Laryngotracheal infiltration
- Sleep Apnea
- Pneumopathy
- Other (*please specify*): .....

\*Please tick box if the symptom is present and add any other useful information that can help diagnosis.

**CLINICAL SYMPTOMS (...)**

Comments *(please specify chronology of symptoms)*:  
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**BIOLOGICAL SYMPTOMS**

- Metabolic acidosis
- Ketonuria
- Hypoglycemia
- Hyperammonemia
- Hyperlactacidemia
- Hepatocellular/cytolysis failure
- Haematological abnormalities *(please specify)*: .....
- Hyperproteinorachy
- Abnormal karyotype
- Other *(please specify)*: .....

**PARACLINICAL SYMPTOMS**

EEG, scan, MRI, echography, potential problems observed: .....

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Histology exams: .....

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Skeletal radiographies: .....

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**SUSPECTED DIAGNOSIS**

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