

## Open questions

- 1/46. What is the major problem that affects your relative's everyday life (quality of life)?  
(Even if it seems unrelated to the genetic condition.)
- 2/46. What is the major behavioral / cognitive problem that affects your relative's and the family life?
- 3/46. Did your relative suffer a major medical problem or discomfort following specific medication?  
(Specify drug and length of treatment.)
- 4/46. What are the major medical problems that occurred until now?  
(Indicate at what age for each)
- 5/46. Please state your relative's strengths

## Weight / Height

- 6/46. Your relative's weight: (kg – lbs / 6m – 1y – 2y – 4y – 6y – 10y – 14y & current value)
- 7/46. Your relative's height: (cm – inches / 6m – 1y – 2y – 4y – 6y – 10y – 14y & current value)
- 8/46. Head circumference: (only if it has been measured by a clinician) (cm – inches / 6m – 1y – 2y – 4y – 6y – 10y – 14y & current value)

## Pregnancy / Early life / Personal care

- 9/46. Was there any problem during pregnancy, labour and/or delivery? (y/n/idk)  
**What words best describe them?** Reduced fetal movement / Lack of fetal movement / Low amniotic fluid / Excessive amniotic fluid / Increased nuchal fold thickness / Abnormal ultrasound results / Caesarean section / Vacuum and/or forceps extraction / Other:
- 10/46. Apgar scores: (only if measured by a professional) (at 1 – 5 – 10min)
- 11/46. Has your relative experienced any health problems in his/her newborn period (first four weeks)? (y/n/idk)  
**What words best describe them?** Signs of anoxia (lack of oxygen) / Jaundice / Hypotonia at birth (muscle weakness) / Hypertonia at birth / Feeding difficulties / Other:  
**What treatment was performed?** Assisted ventilation / Tube feeding / Gastrostomy (tube connecting the stomach) / Jejunostomy (tube connecting the small intestine) / Other:  
**How long was the newborn hospitalized?** (in days)
- 12/46. Developmental milestones: please mark any milestone that your relative has achieved stating his/her age (if achieved)  
Sitting without support / Standing independently / Walking independently / First words (in months)
- 13/46. Personal care:  
**Your relative can:** Actively assist with personal care? Wash, brush her/his teeth? Dress her/himself? Manage shoelaces? Use potty/toilet for urine (daytime) / Use potty/toilet for urine (night-time) / Use potty/toilet for faeces (daytime) / Use potty/toilet for faeces (night-time)  
**How much help does s/he need during the day? How much help does s/he need during the night?**

## Intellectual disability and Autism aspects

- 14/46. Communication ability:  
**Does your relative communicate in any of the following ways?** With speech – Vocal sounds / noises – Gestures – With written words – With symbols / pictures – Signing – None of these choices – Other:  
**At what age did your relative achieve the present level of communication?**  
**Please describe any difference between what your relative understands and what s/he can express:**
- 15/46. Speech ability: (Full correct sentences / Full sentences but often incorrect / No sentence, but single words / Difficult to understand / Not able to speak)  
**Understand what is being said?**  
**Non-verbal communication:** Good expressive non-verbal communication / Poor non-verbal communication / No communication at all  
**Is your relative's speech likely to be understood by:** The family / Others who meet your child for the first time / Neither the family, nor others
- 16/46. Reading ability: (Good / Poor / Non reading)  
**Has been diagnosed with dyslexia or dyscalculia?**
- 17/46. Writing ability: (Good / Poor / Not able to write)
- 18/46. Education:  
**Does your relative has a statement of special educational need (or equivalent)?**  
**What in summary does it say?**  
**What learning support does your relative receive?**
- 19/46. Diagnosis of intellectual disability? (y/n/idk)  
**What is the degree of this intellectual disability?** Mild / Moderate / Severe / Profound
- 20/46. Has an IQ test been performed? (y/n/idk)  
**Which test?**  
**Test result:**
- 21/46. Diagnosis of autism? (autism spectrum disorder) (y/n/idk)
- 22/46. Has an Autism test been performed? (y/n/idk)  
**Which test?**  
**Test result:**
- 23/46. Behavior problems: (y/n/idk)  
**What words best describe them? (mild / moderate / major / nonexistent)** Aggressive / Self aggressiveness (self mutilation) / Impulsivity / Hyperactive / Attention deficit / Shy / Anxious / Depressive tendencies / Restricted interests / Repetitive behavior / stereotypes / Obsessions / Phobias / Diagnosed as schizophrenia / Other?
- 24/46. Feeding problems: (y/n/idk)  
**What words best describe them?** Eats too much / Craves for food / Eats only very restricted food / Does not want to eat (anorexia) / Other:  
**How do you evaluate its (or their) seriousness?** mild / moderate / major
- 25/46. How would you describe your relative's sociability?  
**In each of these four cases: With familiar adults / With unfamiliar adults / With familiar children / With unfamiliar children** (Very sociable / Average sociability / Little sociability / No interaction)

## Physical and neurological problems

- 26/46. Walking problems: (y/n/idk)  
**What words best describe them?** Never learned to walk / Learned to walk but stopped walking / Walk for a limited period, on a limited distance / Walk with cane or other aid / Unstable walk / Other:  
**How do you evaluate its (or their) seriousness?** mild / moderate / major
- 27/46. Tremor: (y/n/idk)  
**How do you evaluate its (or their) seriousness?** mild / moderate / major
- 28/46. Other movement disorders: (y/n/idk)  
**What words best describe them?** Abnormal movements or tics / Fine motor skills problems / Spastic paraplegia / Ataxia / Other: (No / Mild / Moderate / Major / idk)

- 29/46. Epilepsy: has your relative had any seizures (fits)? (y/n/idk)  
**Does this epilepsy require constant medication?** (y/n)  
**What is the name of the drug / molecule?**  
**What is (or was) the effectiveness of the drug?** No effect / Very good / Good / Little effect / Not well tolerated / Major or significant adverse effect / Other:  
**Is this epilepsy pharmaco-resistant?**  
**What were the drug tested?**  
**Please describe the type(s) of epilepsy and its seriousness: (No / Mild / Moderate / Major / idk)** Infantile spasms / Febrile convulsions (fever fit) / Absence seizures / "Grand mal" seizures (Tonic-clonic seizures) / Atonic (drop) seizures / Simple partial seizures (focal seizures) / Complex partial seizures (focal seizures) / Nocturnal seizures / Lennox-Gastaut syndrome / Unknown type of seizures
- 30/46. Has your child had any imaging or other investigations of the head / brain? (brain scanner / MRI / etc.) (y/n/idk)  
**Type of scan / analysis and results**
- Sensory problems**
- 31/46. Vision problems: (y/n/idk)  
**Please describe the type(s) of problem:** (y/n/idk) Far-sighted (hypermetropia, problems with near vision) / Short-sighted (myopia, problems with far vision) / Squint / cross eyed (strabismus) / Cataract / Nystagmus (involuntary eye movement) / Other  
**Does your relative need glasses?** (y/n/idk) Far-sighted lens diopter (+) / Short-sighted lens diopter (-):
- 32/46. Hearing problems: (y/n/idk)  
**What words best describe them?** Deafness (no devices) / Recurrent ears infections / Ear malformation / Other:  
**How are the hearing problems managed?** (y/n/idk) Hearing aids / Hearing tubes (ear or tympanostomy tubes) / Other:
- 33/46. Sense of smell problems: (y/n/idk)  
**How do you evaluate its (or their) seriousness?** mild / moderate / major
- Other problems**
- 34/46. Sleeping disorders: (y/n/idk)  
**What words best describe them?** Sleep apnea / Other:  
**How do you evaluate its (or their) seriousness?** mild / moderate / major
- 35/46. Dental anomalies: (y/n/idk)  
**What words best describe them?** Too many teeth / Too few teeth / Frequent dental caries / Complex orthodontics / Other:  
**How do you evaluate its (or their) seriousness?** mild / moderate / major
- 36/46. Oral / buccal problems: (y/n/idk)  
**What words best describe them?** Swallowing difficulties / Hypersalivation / Cleft lip / Cleft palate / Other:  
**How do you evaluate its (or their) seriousness?** mild / moderate / major
- 37/46. Cardiac problems: (y/n/idk)  
**What words best describe them?** Cardiomyopathy / Cardiac rhythms problems (abnormal EKG) / Cardiac or cardiovascular malformation / Atrial septal defect (ASD) / Ventricular septal defect (VSD) / Patent ductus arteriosus (PDA) / Pulmonary stenosis / Tetralogy of Fallot (TOF) / Other  
**What treatment/surgery (if any) has been needed or is anticipated?**
- 38/46. Vascular problems: (y/n/idk)  
**What words best describe them?** Hypertension / Hypotension / Coagulation problems / Other:  
**How do you evaluate its (or their) seriousness?** mild / moderate / major
- 39/46. Respiratory and pulmonary problems: (y/n/idk)  
**What words best describe them?** Asthma / Hay fever (Allergic rhinitis) / Respiratory rate problem / Pulmonary function problem / Laryngomalacia / Tracheomalacia / Other:  
**How do you evaluate its (or their) seriousness?** mild / moderate / major
- 40/46. Digestive problems: (y/n/idk)  
**What words best describe them?** Regurgitation / Constipation / Diarrhea / Repeated vomiting episodes / Intestinal obstruction / other (No / Mild / Moderate / Major / idk)
- 41/46. Renal/Kidney, bladder and urogenital system problems: (y/n/idk)  
**What words best describe them?** Renal malformation (e.g. horseshoe kidney) / Insufficient renal function / Urinary system malformation / Frequent urinary tract infection / Vesico-ureteral reflux (VUR) / Undescended testicles (cryptorchidism) / other  
**Regularly followed by a specialist:** (y/n)  
**Underwent surgery:** (y/n)  
**Takes medication:** (y/n)  
**What is the name of the drug / molecule?**
- 42/46. Musculo-skeletal problems: (y/n/idk)  
**What words best describe them?** (y/n/idk) Malformation of the vertebral column / Scoliosis / Kyphosis (Hunchback / hump back) / Fragility (ex: broken bones) / Malformation of the upper limb / Malformation of the lower limb / Skull anomaly or malformation / Pectus excavatum (hollowed chest) / Pectus carinatum (pigeon chest) / Hip dysplasia (deformation or misalignment of the hip joint) / Pes planus (flat feet) / Pes cavum (a high arch of the feet) / Club foot (congenital talipes equinovarus CTEV) / Contractures (permanent shortening of a muscle or joint) / Joint laxity (ligamentous laxity, looseness) / other  
**Did your relative went through any surgery?**
- 43/46. Skin, nails and hair problems: (y/n/idk)  
**What words best describe them?** High number of moles / Skin pigmentation disorder (e.g. hypopigmentation) / Eczema, psoriasis or other skin inflammatory problems / Non-inflammatory anomalies of the skin (scar, photosensitivity, etc.) / Abnormal nails (soft, brittle, unusual shape, etc.) / Hair color different from parents and siblings / Fine wispy hair (unlike parents or siblings) / Thick hair (unlike parents or siblings) / Other:  
**How do you evaluate its (or their) seriousness?** mild / moderate / major
- 44/46. Endocrine and metabolic systems problems: (y/n/idk)  
**What words best describe them?** Hyperthyroidism (over-active thyroid) / Hypothyroidism (under-active thyroid) / Growth hormone deficiency / Other:  
**Has your relative been diagnosed with any disorder of the endocrine or metabolic systems?**  
**If applicable, what was the age at first menstruation?**
- 45/46. Blood and immune system problems: (y/n/idk or Yes, but only ear/nose/throat infections)  
**Has your relative been diagnosed with any disorder of the blood or immune system?**
- 46/46. Cancer development: (y/n/idk)

Note: All "yes / no / I don't know" initial questions without sub-questions allow to specify the problem in writing