

## GenIDA – Main questionnaire

### OPEN QUESTIONS

**1/46. What is the major problem that affects your affected relative's everyday life (quality of life)?**  
(Even if it seems unrelated to the genetic condition)

**2/46. What are the major behavioural problems, cognitive problem that affects the life of your relative and the family life?**

**3/46. Did your affected 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 affected relative's weight:**

*unit: kilograms or pounds*

Birth weight - 6m -1y - 2y - 4y - 6y - 10y - 14y & current value

**7/46. Your affected relative's height:**

*unit: centimetres or inches*

Height at birth - 6m -1y - 2y - 4y - 6y - 10y - 14y & current value

**8/46. Head circumference: (only if it has been measured by a clinician)**

*unit: centimetres*

Head circumference at birth - 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?**

Yes / no / I don't know (*unique choice*)

What words best describe them?

- Reduced foetal movements
- Lack of foetal movement
- Low amniotic fluid
- Excessive amniotic fluid
- Increased nuchal fold thickness
- Abnormal ultrasound results
- Caesarean section
- Vacuum and/or forceps extraction
- Other: (*specify*)

**10/46. Apgar scores: (only if measured by a professional)**

1 min:

5 min:

10 min:

**11/46. Has your relative experienced any health problems in his/her newborn period (first four weeks)?**

Yes / no / I don't know (*unique choice*)

What words best describe them?

- Signs of anoxia (lack of oxygen)
- Jaundice
- Hypotonia at birth (muscle weakness) >>> How has hypotonia evolved over time?

- Hypertonia at birth
- Feeding difficulties
- Other: (*specify*)

What treatment was performed?

- Assisted ventilation
- Tube feeding
- Gastrostomy (tube connecting the stomach)
- Jejunostomy (tube connecting the small intestine)
- Other: (*specify*)

How long has the new-born been 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: (*in months*)
- Standing independently: (*in months*)
- Walking independently: (*in months*)
- First words: (*in months*)

**13/46. Personal care:**

Age (*by default, current age*)

Is your affected relative able to: Yes / yes, with help / no

- 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 she/he need during the day?

How much help does she/he need during the night?

**INTELLECTUAL DISABILITY AND AUTISM ASPECTS**

**14/46. Communication ability:**

Age (*by default, current age*)

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: (*specify*)

At what age did your affected relative reach her or his present level of communication? (*in years*)

Describe the difference(s) between what your affected relative understands and what she/he can express:

**15/46. Speech ability:**

Age (*by default, current age*)

*Unique choice*

- Full and correct sentences
- Full sentences but often incorrect
- No sentence, but single words
- Difficult to understand (elocution problems)
- Not able to speak

Understand what is being said?

Yes / no / I don't know (*unique choice*)

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:**

Age (*by default, current age*)

*Unique choice*

- Good
- Poor
- Non reading

Has there been a diagnosis of dyslexia or dyscalculia?

Yes / no / I don't know (*unique choice*)

**17/46. Writing ability:**

Age (*by default, current age*)

*Unique choice*

- Good
- Poor
- Not able to write

**18/46. Education**

Does your relative has a statement of special educational needs (or equivalent) ?

Yes / no / I don't know (*unique choice*)

What in summary does it say?

What learning support does your relative receive?

**19/46. Diagnosis of intellectual disability?**

Yes / no / I don't know (*unique choice*)

What is the degree of this intellectual disability?

Mild / Moderate / Severe / Profound

**20/46. Has an IQ test been performed?**

Yes / no / I don't know (*unique choice*)

Date of the test (*by default: today*)

Which test?

Test result:

**21/46. Diagnosis of autism (Autism Spectrum Disorder)?**

Yes / no / I don't know (*unique choice*)

**22/46. Has an Autism test been performed?**

Yes / no / I don't know (*unique choice*)

Date of the test (*by default: today*)

Which test?

Test result:

**23/46. Behavioural problems:**

Age (*by default, current age*)

Yes / no / I don't know (*unique choice*)

What words best describe them? (Nonexistent / Minor / Moderate / Major)

- Aggressive
- Self-aggressiveness (self-mutilation)
- Impulsivity
- Hyperactive
- Attention deficit
- Shy
- Anxious
- Depressive tendencies
- Restricted interests
- Repetitive behaviour / stereotypes
- Obsessions
- Phobias
- Diagnosed of schizophrenia
- Other: (*specify*)

**24/46. Feeding problems:**

Age (*by default, current age*)

Yes / no / I don't know (*unique choice*)

What words best describe them?

- Eats too much
- Craves for food
- Eats only very restrictive food
- Does not want to eat (anorexia)
- Other: (*specify*)

How do you assess its (or their) seriousness?

Minor / Moderate / Major

**25/46. How would you describe your affected relative's sociability?**

Age (*in years, mandatory*)

In each of these four cases: Very sociable / Average sociability / Little sociability / No interaction

- With familiar adults
- With unfamiliar adults
- With familiar children
- With unfamiliar children

**26/46. Walking problems:**

Age (*by default, current age*)

Yes / no / I don't know (*unique choice*)

What words best describe them?

- Never learned to walk
- Learned to walk but stopped walking
- Walks for a limited period, on a limited distance
- Walks with cane or other aid
- Unstable walk
- Other: (*specify*)

How do you evaluate its (or their) seriousness?

Mild / Moderate / Major

**27/46. Tremors:**

Age (*by default, current age*)

(*unique choice*)

- Yes (*specify*)
- No
- I don't know

How do you assess the seriousness of this (these) problem(s)?

Minor / Moderate / Major

**28/46. Other movement disorders:**

Age (*by default, current age*)

Yes / no / I don't know (*unique choice*)

What words best describe them? No / Minor / Moderate / Major / I don't know

- Abnormal movements or tics
- Fine motor problems, clumsiness
- Spastic paraplegia
- Ataxia
- Other: (*specify*)

**29/46. Epilepsy: Has your relative had epileptic seizures (fits)?**

Yes / no / I don't know (*unique choice*)

Date (*by default: today*)

End of period (*optional: fill only to define a period*)

Does this epilepsy require constant medication? yes / no

What is the name of the drug(s)/molecule(s)? *(If other drugs have been previously tested, please answer this question again and adjust the period of treatment)*

What is (or was) the effectiveness of the drug(s)/molecule(s)? No effect / Very good / Good / Little effect / Not very well tolerated / Major or significant adverse effect / Other: *(specify)*

Is this epilepsy pharmaco-resistant? Yes / no

Please describe the type(s) of epilepsy and its seriousness: No / Mild / Moderate / Major / I don't know

- 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
- Other: *(specify)*

**30/46. Has your child had any imaging or other investigations of the head / brain? (brain scanner / MRI / etc.)**

Age *(by default, current age)*

Yes / no / I don't know *(unique choice)*

Type of scan / analysis and results:

#### **SENSORY PROBLEMS**

**31/46. Vision problems:**

Age *(by default, current age)*

Yes / no / I don't know *(unique choice)*

Describe the type(s) of problem(s): Yes / no / I don't know

- Far-sighted (hypermetropia, problems with near vision)
- Short-sighted (myopia, problems with far vision)
- Squint / cross eyed (strabismus)
- Cataract
- Nystagmus (involuntary eye movement)
- Other: *(specify)*

Does your affected relative need glasses?

Yes / no / I don't know

Far-sighted lens diopter (+):

Short-sighted lens diopter (-):

**32/46. Hearing problems:**

Age *(by default, current age)*

Yes / no / I don't know (*unique choice*)

What words best describe them?

- Deafness (no devices)
- Recurrent ear infections
- Ear malformation
- Other: (*specify*)

What is the seriousness of the hearing impairment? Mild (1) / Moderate (2) / Severe (3) / Profound (4)

1. Mild: Troubles in noisy situations.
2. Moderate: Can hear and understand speech only in quiet situations.
3. Severe: Cannot hear a speech without the use of amplification.
4. Profound: Cannot hear most environmental sounds without the use of amplification.

How are hearing problems managed? Yes / No / I don't know

- Hearing aids
- Hearing tubes (ear or tympanostomy tubes)
- Other: (*specify*)

**33/46. Sense of smell problems:**

Age (*by default, current age*)

(*unique choice*)

- Yes (*specify*)
- No
- I don't know

How do you evaluate its (or their) seriousness? Major / Moderate / Mild

**OTHER PROBLEMS**

**34/46. Sleeping disorders:**

Age (*by default, current age*)

Yes / no / I don't know (*unique choice*)

What words best describe them?

- Sleep apnea
- Other: (*specify*)

How do you evaluate its (or their) seriousness? Major / Moderate / Mild

**35/46. Dental abnormalities:**

Age (*by default, current age*)

Yes / no / I don't know (*unique choice*)

What words best describe them?

- Too many teeth
- Too few teeth
- Frequent dental caries
- Complex orthodontics
- Other: (*specify*)

How do you evaluate its (or their) seriousness? Major / Moderate / Mild

**36/46. Oral / buccal problems:**

Age (*by default, current age*)

Yes / no / I don't know (*unique choice*)

What words best describe them?

- Swallowing difficulties
- Hypersalivation
- Cleft lip
- Cleft palate
- Other: (*specify*)

How do you evaluate its (or their) seriousness? Major / Moderate / Mild

**37/46. Cardiac problems:**

Age (*by default, current age*)

Yes / no / I don't know (*unique choice*)

What words best describe them? Yes / no / I don't know

- Cardiomyopathy
- Cardiac rhythm problem (abnormal EKG)
- Cardiac or cardiovascular malformation
- Atrial septal defect (ASD)
- Ventricular septal defect (VSD)
- Patent ductus arteriosus (PDA)
- Pulmonary stenosis
- Tetralogy of Fallot (TOF)
- Bicuspid aortic valves (BAV)
- Aortic aneurysm (dilation / broadening)
- Other: (*specify*)

What treatment / surgery (if any) have been needed or is anticipated?

**38/46. Vascular problems:**

Age (*by default, current age*)

Yes / no / I don't know (*unique choice*)

What words best describe them?

- Hypertension
- Hypotension
- Coagulation problems
- Other: (*specify*)

How do you evaluate its (or their) seriousness? Major / Moderate / Mild

**39/46. Respiratory and pulmonary problems:**

Age (*by default, current age*)

Yes / no / I don't know (*unique choice*)

What words best describe them? Yes / no / I don't know

- Asthma
- Hay fever (allergic rhinitis)
- Respiratory rate problem



- Pulmonary function problem
- Laryngomalacia
- Tracheomalacia
- Other: *(specify)*

How do you evaluate its (or their) seriousness? Major / Moderate / Mild

**40/46. Digestive problems:**

Age *(by default, current age)*

Yes / no / I don't know *(unique choice)*

What words best describe them? No / Mild / Moderate / Major / I don't know

- Regurgitation
- Constipation
- Diarrhea
- Repeated vomiting episodes
- Intestinal obstruction
- Other: *(specify)*

**41/46. Renal / Kidney, bladder and urogenital system problems:**

Age *(by default, current age)*

Yes / no / I don't know *(unique choice)*

What words best describe them? Yes / no / I don't know

- Renal malformation (e.g., horseshoe kidney)
- Insufficient renal function
- Urinary system malformation
- Frequent urinary tract infection
- Vesico-ureteral reflux (VUR)
- Undescended testicles (Cryptorchidism)
- Other: *(specify)*

Regularly followed by a specialist: Yes / no

Underwent surgery: Yes / no

Takes medication: Yes / no

What is the name of the drug / molecule? *(If other drugs have been previously tested, please answer this question again and adjust the period of treatment)*

**42/46. Musculo-skeletal problems:**

Age *(by default, current age)*

Yes / no / I don't know *(unique choice)*

What words best describe them? Yes / no / I don't know

- Malformation of the vertebral column
- Scoliosis
- Kyphosis (Hunchback / hump back)
- Fragility (e.g., broken bones)
- Malformation in the upper limbs
- Malformation in the lower limbs
- 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: (*specify*)

Did your relative went through any surgery? (*if yes, specify*)

**43/46. Skin, nails and hair problems:**

Age (*by default, current age*)

What words best describe them? Yes / no / I don't know

- 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: (*specify*)

How do you evaluate its (or their) seriousness? Major / Moderate / Mild

**44/46. Endocrine and metabolic systems problems:**

Age (*by default, current age*)

What words best describe them?

- Hypothyroidism (over-active thyroid)
- Hyperthyroidism (under-active thyroid)
- Growth hormone deficiency
- Other: (*specify*)

Has your relative been diagnosed with any disorder of the endocrine or metabolic systems? (*if yes, specify*)

If applicable, what was the age at first menstruation? (*in years*)

**45/46. Blood and immune system problems:**

Age (*by default, current age*)

Does / did your relative have recurrent infections?

- Yes, but only ear / nose / throat infections
- Yes (*specify*)
- No
- I don't know

Has your relative been diagnosed with any disorder of the blood or immune system? (*if yes, specify*)

**46/46. Cancer development:**

Age (*by default, current age*)

(*unique choice*)

- Yes (*specify*)
- No
- I don't know