

Potchefstroom Laboratory for Inborn Errors of Metabolism

Clinical information form (3 pages)

This form could also be completed and submitted on our website: www.pliem.co.za

Or faxed / e-mailed to: Fax 018 299 2316 / 086 463 7094

E-mail: ansie.mienie@nwu.ac.za

Patient Information

Name of Patient: _____ Surname of Patient: _____

Date of Birth: _____ MALE FEMALE

Patient referred by: _____

Patient's clinical history

Please mark applicable information, in order to assist with the analyses and interpretation of the results:

Age at onset of symptoms : _____

Head / Neurologic

- | | | |
|---|--|--|
| <input type="checkbox"/> Aggressive behaviour | <input type="checkbox"/> Abnormal ears (e.g. red) | <input type="checkbox"/> Abnormal face |
| <input type="checkbox"/> Ataxia | <input type="checkbox"/> Alzheimer's in family | <input type="checkbox"/> Amnesia in family |
| <input type="checkbox"/> Behaviour abnormalities | <input type="checkbox"/> Aneurysms (bleeding in brain) | <input type="checkbox"/> Autism in family |
| <input type="checkbox"/> Dementia in family | <input type="checkbox"/> Dark circles under the eyes | <input type="checkbox"/> Deafness |
| <input type="checkbox"/> Down's syndrome | <input type="checkbox"/> Depression | <input type="checkbox"/> Destructive behaviour |
| <input type="checkbox"/> Flushed cheeks / face | <input type="checkbox"/> Dysmorphic features | <input type="checkbox"/> Epicanthic folds (ears) |
| <input type="checkbox"/> Insomnia | <input type="checkbox"/> Hyperactive | <input type="checkbox"/> Hyper-acusis (hearing) |
| <input type="checkbox"/> Learning disability | <input type="checkbox"/> Irritability (severe) | <input type="checkbox"/> Lack of eye contact |
| <input type="checkbox"/> Mental retardation | <input type="checkbox"/> Lethargy / coma | <input type="checkbox"/> Loss of speech |
| | <input type="checkbox"/> Migraine / Headache (severe) | <input type="checkbox"/> Obsessive-compulsive disorder |
| <input type="checkbox"/> Parkinson's in family | <input type="checkbox"/> PDD (pervasive developmental delay) | |
| <input type="checkbox"/> Photophobia (sensitivity to light) | <input type="checkbox"/> Poor concentration | <input type="checkbox"/> Restless (severe) |
| <input type="checkbox"/> Schizophrenia in family | <input type="checkbox"/> Seizures | <input type="checkbox"/> Self-mutilation / self-injury |
| <input type="checkbox"/> Speech delay | <input type="checkbox"/> Unexpected throat clearing | <input type="checkbox"/> White blotches on face |
| <input type="checkbox"/> Withdrawn (severe) | | |

MOTOR / MUSCULAR

- Hypertonia (toe walkers)
- Hypotonia (muscle weakness)
- Loss of meaningful hand use
- Motor neuron disease
- Movement disorders
- Multiple sclerosis
- Psychomotor delays
- Spasticity

ABDOMEN

- Coeliac disease
- Constipation (severe)
- Diarrhoea
- Eating disorders
- Enlarged liver
- Enlarged spleen
- Feeding problems
- "Leaks" sour milk as baby
- Poor sucking reflex
- Mother vegetarian during breast feeding
- Vomiting (projectile or otherwise)

SKIN / HAIR / NAILS

- Abnormal hair growth
- Acne (severe)
- Café o'lait markings
- Dermatitis
- Diminishing pigmentation
- Dry, brittle hair
- Eczema
- Hives / itching (severe)
- Impaired wound healing
- Increased pigmentation
- Loss of hair
- Photosensitive
- Skin rashes

EYES

- Eye abnormalities
- Optic neuritis
- Recurrent otitis media

IMMUNITY

- Increased bacterial infections (e.g. repeated ear, nose, throat infections)
- Melanomas
- Hyper IgE syndrome
- Increased fungal infections
- Increased viral infections

BLOOD / HEART

- Anaemic
- Arteriosclerosis
- Bruises easily
- Congenital heart defect
- Family history of heart disease
- Haemophiliacs (bleeders)
- History of Thrombosis
- Sleep apnoea (breathing stops while asleep)

MOUTH

- Abnormal teeth
- Abnormal tongue (includes sores in mouth and on tongue)
- Cleft palate
- Excessive drooling
- Halitosis (very bad breath / acetone on breath)
- High palate
- Low oral tone (Mouth hangs open)

SKELETAL

- Abnormal skeleton
- Growth retardation
- Rheumatoid arthritis
- Skeletal degeneration
- Weak motor integration

ENDOCRINE

- Diabetes type 1
- Diabetes type 2
- Under- / overactive thyroid

SYNDACTYLY

- Webbing between fingers
- Webbing between toes
- Webbing between fingers & toes

MISCELLANEOUS

- Sudden Infant Death Syndrome in family
- Fever attacks
- Sleep disturbances
- Strange body odour observed
- Sudden weight changes
- Other (please specify: _____)

Is there anything that precipitates the behaviour of the person referred (e.g. colds, food, tiredness, etc.)?

How many siblings show the same clinical symptoms as the patient?

Medication / supplements the patient is using: _____
