Potchefstroom Laboratory for Inborn Errors of Metabolism
Clinical information form (3 pages)

This form could also be completed and submitted on our website:
pliem.co.za/test-request.php

Or e-mailed to: ansie.mienie@nwu.ac.za / marli.dercksen@nwu.ac.za

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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

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- ☐ Withdrawn (severe)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

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

SYNDACTLY

☐ 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: _____

_____