Tel#: 018 2312 / 018 285 2652
E-mail address: ansie.mienie@nwu.ac.za

Centre for Human Metabolomics
PLIEM / NBS / CRS sites, 11 Hoffmann street, Building F3, Office Lab G19, ground floor (new building)
Faculty of Natural and Agricultural Science, Biochemistry, North-West University, 2520 Potchefstroom Campus, RSA

Potchefstroom Laboratory for Inborn Errors of Metabolism
Clinical information form

Clinical- and **routine pathology test results** may give an indication of metabolic disorders present and should be correlated with results found during metabolic testing. Additional tests may also be recommended if the clinical profile suggests a clinical correlation.

Please complete the patient information section and tick applicable clinical presentation, in order to assist with the analyses to be done and interpretation of all the results.

Please **e-mail** the **completed form** to **ansie.mienie@nwu.ac.za.**

PATIENT INFORMATION	
First name(s) of patient	
Surname of patient	
Date of birth	
Gender	☐ MALE ☐ FEMALE
Referring clinician	
Symptoms present since	

Head / Neurologic

☐ Abnormal ears (e.g. red) ☐ Abnormal face ☐ Aggressive behaviour ☐ Alzheimer's in family

☐ Amnesia in family ☐ Aneurysms ☐ Ataxia ☐ Autism in family

☐ Behaviour abnormalities ☐ Deafness ☐ Dark circles under the eyes ☐ Dementia in family

☐ Destructive behaviour ☐ Depression ☐ Down's syndrome ☐ Dysmorphic features

☐ Epicanthic folds (ears) ☐ Flushed cheeks/face ☐ Hyperactive ☐ Insomnia

☐ Irritability (severe) ☐ Lack of eye contact ☐ Hyper-accuses (hearing) ☐ Learning disability

☐ Lethargy / coma ☐ Loss of speech ☐ Mental retardation ☐ Migraine / Headache

☐ OCD (Obsessive-compulsive disorder) ☐ Parkinson's in family ☐ Seizures

☐ PDD (pervasive developmental delay) ☐ Photophobia (sensitivity to light)

☐ Poor concentration ☐ Restless ☐ Schizophrenia in family ☐ Speech delay

☐ Self-mutilation / self-injury ☐ Unexpected throat clearing ☐ White blotches on face ☐ Withdrawn

☐ MRI brain done (results _____) ☐ EEG done (results _____)

Motor / Muscular

- ☐ Hypertonia (toe walkers)
- ☐ Motor neuron disease
- ☐ Multiple sclerosis
- ☐ Psychomotor delays
- ☐ Loss of meaningful hand use
- ☐ Spasticity
- ☐ Hypotonia (muscle weakness)
- ☐ Motor-vocal tics (Tourette's syndrome)
- ☐ Movement disorder (please describe): _____

Abdomen

- ☐ Coeliac disease
- ☐ Constipation (severe)
- ☐ Diarrhoea
- ☐ Eating disorders
- ☐ Enlarged liver
- ☐ Enlarged spleen
- ☐ Poor sucking reflex
- ☐ Vomiting (projectile or otherwise)
- ☐ Feeding problems (please describe): _____

Skin / Hair / Nails

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

Eyes

- ☐ Eye abnormalities (please describe): _____
- ☐ Optic neuritis
- ☐ Recurrent otitis media

Immunity

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

Blood / Heart

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

Mouth

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

Skeletal

☐ Abnormal skeleton ☐ Growth retardation ☐ Rheumatoid arthritis ☐ Skeletal degeneration
☐ Weak motor integration ☐ Abnormal gait

Endocrine

☐ Diabetes type 1 ☐ Diabetes type 2 ☐ Underactive thyroid function ☐ Overactive thyroid function

Syndactyly

☐ Syndactyly fingers ☐ Syndactyly toes ☐ Syndactyly fingers and toes ☐ Webbing between fingers or toes

Miscellaneous

_____ Indicate how many **female** siblings _____ Indicate how many **male** siblings

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

☐ Sudden Infant Death Syndrome in family ☐ Previous baby / babies died of unknown causes (this include relatives as well)

☐ Fever attacks ☐ Sleep disturbance (please describe): _____

☐ Strange body odour observed (describe odour): _____

☐ Sudden weight gain ☐ Sudden weight loss

☐ Sample collection for testing done when the **symptoms were most prominent**

☐ Sample collection for testing done when the **symptoms were NOT most prominent**

☐ **Medication / Supplements / treatment the patient was on during sample collection**:

_____

☐ **OTHER relevant information**: _____

_____

_____