

REFERRING CLINICAN INFORMATION

Name: _____
Address: _____

E-Mail: _____
Phone: _____

Stamp

PATIENT INFORMATION

First Name: _____
Family Name: _____
Date of birth : (dd/mm/yyyy) _____
Geographic origin of patient: _____
Cresnet ID: _____

ANTHROMOMETRIC DATA

Date of anamnesis (dd/mm/yyyy): _____
Birth weight (g): _____
Birth length (cm): _____
Gestation week: _____
Weight (kg): _____
Height (cm): _____

Sex: Male
 Female

Onset of puberty Not yet
 Hypogonadism
 Yes, spontaneous at age of _____

Menarche Not yet
 Yes, spontaneous, age: _____

DIAGNOSES

Here we ask you to provide detailed information about the health profile of the patient. Does one or more of the following clinical features were diagnosed by a medical professional?

SGA (Small for Gestational Age)

 Y N

SGA with catch-up growth up to 4 years

 Y N

SGA without catch-up growth up to 4 years

 Y N

Silver-Russel Syndrome

 Y N

Diagnoses affecting the Anterior Pituitary
 Y N

IGHD (Isolated Growth Hormone Deficiency)

 Y N

 affected Hormone: **GH**

Hormone Level:

 normal
 diminished
 n.d.

n.d. not determined

MPHD (Multiple Pituitary Hormone Deficiency)

 Y N

 affected Hormone: **TSH**

Hormone Level:

 normal
 diminished
 n.d.

 affected Hormone: **PRL**

Hormone Level:

 normal
 diminished
 n.d.

 affected Hormone: **Cortisol**

Hormone Level:

 normal
 diminished
 n.d.

 affected Hormone: **ACTH**

Hormone Level:

 normal
 diminished
 n.d.

 affected Hormone: **LH/FSH**

Hormone Level:

 normal
 diminished
 n.d.

Diagnoses affecting the Posterior Pituitary
 Y N

 affected Hormone: **ADH**

Hormone Level:

 normal
 diminished
 n.d.

CLINICAL FEATURES

Age at Clinical Diagnosis (years):

Symptoms leading to diagnosis: *(Please tick as appropriate)*

Growth hormone deficiency

- Growth retardation
- Truncal obesity
- Acromicria
- Frontal bossing
- Late fontanelle closure
- Micropenis
- Prolonged Jaundice
- Hypoglycemia

Panhypopituitarism

- Short Neck
- Limited neck rotation
- Makroglossia
- Late dentition
- Dry Skin
- Umbilical hernia
- Septo-optic dysplasia

Unspecific clinical signs

- Neurological deficits
- Microcephaly
- Triangular face
- Cleft palate
- high pitch voice
- Hypothyroidism
- Cardiac malformations
- Clinodactyly
- Brachydactyly

Further abnormalities:

Morphological Abnormalities

Imagines of the pituitary region performed: Yes No

Pituitary Size small normal enlarged
 Location of the anterior pituitary in situ ectopic
 Location of posterior pituitary in situ ectopic

Growth Chart

Please enclose a growth chart with indication of bone age and start and stop of hormone replacement therapy.

Family

Please give a pedigree drawing below, sign all affected family members and when possible give data for height and weight.

Consanguinity of parents:

Y	N	U
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 (U=Unknown)

= male, female not affected

= male, female affected

Hormone Replacement

Please give information about hormone replacement therapy. For example start, stop and dose of hydrocortisone

Materials enclosed with this form are:

Blood sample (please provide a minimum of 2 ml EDTA – do not freeze)

DNA sample (please provide a minimum of 50µg)

CHECKLIST

Blood or DNA sample

Clinical data provided

Consent Form (NGS)

Überweisungsschein (for German public health insurance)

Please send the samples to the following address:

**Pädiatisches Forschungszentrum
Labor Prof. Pfäffle
3. Floor, Room 3010
Liebigstraße 19, Haus C
04103 Leipzig
Germany**