

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, stope 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
“Laboranforderungsschein” completed and signed by the patient
Consent Form (NGS or WES)
for German senders (Überweisungsschein)

Please send the samples to the following address:

Prof. Dr. R. Pfäffle
University Hospital for Children and
Adolescents
3. Floor, Room 3010
Liebigstraße 19
04103 Leipzig