SOTOS SYNDROME : NSD1 SCREENING

Institut de Pathologie et de Génétique, Asbl
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Website : www.ipg.be
Centre of Human Genetics

Patient name : .....

Date of birth : .....

Sex : ☐ F ☐ M

Home address : .....

Telephone : .....

Billing address : .....

Local ID : .....

To be filled in by DNA-laboratory employees :

Material
☐ BL ☐ CV ☐ FI ☐ AMN ☐ DNA ☐ Biopsy ...
Quantity : .....

Date sample received : .....

DNA-number : .....

(The next fields are mandatory) Please fill in :

Referring doctor :
.....
.....
.....
.....

Date of sampling : .....

Hospital : .....
.....

Department : .....
.....

Address : .....
.....
.....

Telephone / tracer / e-mail : .....
.....

Material :
Per investigation we need 20 ml EDTA blood samples (small children 5-10 ml) or 50 µg of DNA. Clearly mention the **NAME, DATE OF BIRTH and SEX** on the blood samples. Prenatal and other materials only after consultation with laboratory staffs (+ 32 71 44 71 81).
Sending :
Do not freeze the samples. Send at room temperature. Ec countries : from Monday to Wednesday by regular post, after Wednesday send by express. Send prenatal material on the same day with a courier. Non EC countries : Send with a courier.

PURPOSE :
☐ Carriership / exclusion of a diagnosis
☐ Confirmation of clinical diagnosis
(with respect to a known gene defect in the family)
☐ Presymptomatic testing
☐ Partner testing
☐ Prenatal testing
☐ Archiving (for possible future diagnosis)
☐ Research

Mutation known in the family ? ☐ **Yes** ☐ **No**
☞ If yes : .....

Have family members been referred to our lab before ? ☐ **Yes** ☐ **No**

Name :	Date of birth :	Relation to patient :
.....	.....	.....
.....	.....	.....
.....	.....	.....

Analysis won't be realized without an informed consent (see page 7) and clinical information.

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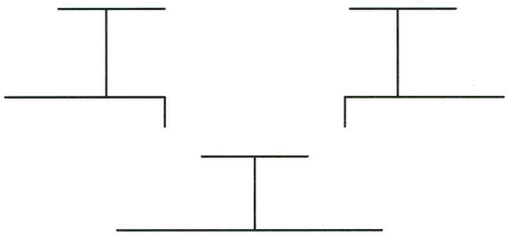
FAMILIAL INFORMATION

Consanguinity in the family ? ☐ **Yes (Indicate in genealogy)** ☐ **No**

Genealogy :

Mark the person to be investigated with an arrow (↗).
Affected persons : full shade (■), carriers semi shade (◧).



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CLINICAL INFORMATION

Date of clinical exam : ..... **Age at clinical exam :** .....

1. Facial appearance :

	Yes	No	Unknown
Frontotemporal hair sparsity	☐	☐	☐
High bossed forehead	☐	☐	☐
Downslanting palpebral fissure	☐	☐	☐
Long narrow face	☐	☐	☐
Prominent narrow face	☐	☐	☐
Malar flushing	☐	☐	☐
Dolicocephaly	☐	☐	☐

Other : .....

2. Overgrowth :

		Yes	No	Unknown
Prenatal onset overgrowth		☐	☐	☐
Postnatal onset overgrowth		☐	☐	☐
Advanced bone age		☐	☐	☐
Bone age : ..... years ..... months for chronologic age : ..... years ..... months.				
Hemihypertrophy		☐	☐	☐
Height ..... cm (..... SD)	> +2SD	☐	☐	☐
Weight ..... cm (..... SD)	> +2SD	☐	☐	☐
Head circumference ..... cm (..... SD)	> +2SD	☐	☐	☐

Paternal height : ..... cm (..... SD) ; head circumference ..... cm (..... SD)
Maternal height : ..... cm (..... SD) ; head circumference ..... cm (..... SD)

3. Learning disability :

	Yes	No	Unknown
Developmental delay	☐	☐	☐

☞ if yes : *Mild* ☐ *Moderate* ☐ *Severe* ☐

Sitting position ..... months
Walk ..... months
Development quotient .....

	Yes	No	Unknown
Speech delay	☐	☐	☐
Cognitive impairment	☐	☐	☐

☞ if yes : *Mild* ☐ *Moderate* ☐ *Severe* ☐

Intellectual quotient .....

4. Behavioral problems :

Yes	No	Unknown
☐	☐	☐

Description : .....

5. Seizures :

Yes	No	Unknown
☐	☐	☐

Description : .....

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6. Cranial MRI/CT abnormalities :

	Yes	No	Unknown
Ventricular dilatation	☐	☐	☐
Hypoplasia or agenesis of the corpus callosum	☐	☐	☐
Mega cisterna magna	☐	☐	☐
Wide/cavum septum pellucidum	☐	☐	☐
Cerebral atrophy	☐	☐	☐
Small cerebellar vermis	☐	☐	☐

Other : .....

7. Pregnancy complication :

	Yes	No	Unknown
Preclampsia	☐	☐	☐

Other : .....

8. Neonatal complications :

	Yes	No	Unknown
Jaundice	☐	☐	☐
Hypotonia	☐	☐	☐
Poor feeding	☐	☐	☐
Neonatal hypoglycemia	☐	☐	☐
Hypocalcemia	☐	☐	☐

Other : .....

9. Cardiac abnormalities :

Yes	No	Unknown
☐	☐	☐

☞ If yes, description :

.....

.....

10. Renal abnormalities :

	Yes	No	Unknown
Tumors	☐	☐	☐

☞ If yes, description :

.....

.....

11. Skeletal abnormalities :

	Yes	No	Unknown
Joint hyperlaxity	☐	☐	☐
Pes planus	☐	☐	☐
Scoliosis	☐	☐	☐
Craniosynostosis	☐	☐	☐
Pectus excavatum	☐	☐	☐
Talipes equinovarus	☐	☐	☐
Vertebral anomalies	☐	☐	☐
2/3 toe syndactyly	☐	☐	☐
Contractures	☐	☐	☐

Other : .....

12. Otologic problems :

	Yes	No	Unknown
Chronic otits media	☐	☐	☐
Conductive hearing loss	☐	☐	☐
Perceptive hearing loss	☐	☐	☐

Other : .....

13. Ophtalmologic abnormalities :

	Yes	No	Unknown
Astigmatism	☐	☐	☐
Cataract	☐	☐	☐
Nystagmus	☐	☐	☐
Myopia	☐	☐	☐
Hypermetropia	☐	☐	☐
Strabismus	☐	☐	☐

Other : .....

14. Digestive problems :

	Yes	No	Unknown
Constipation	☐	☐	☐
Cholesteatoma	☐	☐	☐
Gastrooesophageal reflux	☐	☐	☐
Umbilical hernia	☐	☐	☐

Other : .....

15. Tumors : ☐

Yes	No	Unknown
☐	☐	☐

☞ If yes, description :

.....

.....

16. Various other features :

	Yes	No	Unknown
Cryptorchidism	☐	☐	☐
Hydrocele	☐	☐	☐
Phimosis	☐	☐	☐
Hemangioma	☐	☐	☐
Hypodontia	☐	☐	☐
Hypoplastic nails	☐	☐	☐
Hypothyroidism	☐	☐	☐
Skin hyperpigmentation	☐	☐	☐
Skin hypopigmentation	☐	☐	☐

Other : .....

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PROF. C. VERELLEN – DUMOULIN
DR A. DESTRÉE
DR C. FOURNEAU
DR I. MAYSTADT
DR P. RIBAÏ

INFORMED CONSENT

PATIENT : LAST NAME : .....
FIRST NAME : .....
DATE OF BIRTH : .....
ADDRESS : .....

REFERING DOCTOR : .....

GENETIC TESTING REQUESTED FOR : .....
.....

1. I have been informed about the purpose of this genetic test.
2. I have received an explanation of the limitations of this genetic test. I know that genetic analysis may be long (sometimes many years) and does not always succeed.
3. I have discussed the benefits and risks of this genetic test with my physician and/or other health care. I understand the meaning of possible test results and have been informed how I will receive the result.
5. I have been informed who may have access to my biological sample, and that any leftover sample may be retained by the laboratory.
6. I have been informed who may have access to my genetic test result, which is part of my confidential medical record.

I consent to have a sample taken for genetic testing on the above-named patient for the condition(s) listed above.

Date : .....

Signature of Patient or Authorized Designee:

Circle one: **Self** **Parent(s)** **Legal Guardian Care**

Print Name of Physician or Authorized Delegee explaining the above information:

Date : .....

Signature of Physician or Authorized Delegee :