Information on the procedure at the USE

Dear advice seeker,

For your information, we would like to briefly introduce the procedure at the University Centre for Rare Diseases (USE) in Dresden.

The USE does not offer consultation hours in the conventional sense, and no treatment appointments are made. In order to use the USE, you first need a brief statement from your attending specialist on the necessity of an assessment by us, a referral slip to the USE, as well as all meaningful findings on the symptoms. Furthermore, the patient questionnaire must be completed and signed by the patient.

Patients, relatives and doctors who would like to contact us are asked to have these questionnaires filled out first. They serve as an initial assessment of the case. You can download the questionnaire online from our website or have it sent to you after contacting our coordinator.

You can find the questionnaire on the Internet at: **http://www.uniklinikum-dresden.de/use**

You can reach our coordinator
Tues. and Thurs. 9:30-11:30
☎ +49(0)351 458 5608

By sending us the questionnaires and the findings, you agree that they may be examined by our interdisciplinary team of doctors. This team is made up of experts who have specialised in both organic and mental illnesses. We will use your documents exclusively for the assessment in the context of your request to the USE. After receipt and review, we will decide whether a presentation at an existing centre of excellence (in Dresden or at another location) is suitable for you or whether your case should first be discussed in our interdisciplinary case conference. In this case conference, specialists from several departments will discuss the most suitable course of action for you. Both you and your referring doctor will be informed in writing about the results of this conference. Due to the large number of requests to the USE, we ask for your patience if it may take some time to process your request. The nature of the result is also open, for example, further diagnostic or therapeutic steps or concrete links to special outpatient clinics may result. However, it is also possible that no new findings can be obtained.

The first step is to submit:
- **Specialist's questionnaire**, completed in full
- **Patient's questionnaire**, completed in full and signed
- **Referral slip** from the specialist
- **Copy of all significant findings** (doctor's letters, laboratory results, genetic findings)
- If applicable, **informative photos** of externally visible changes in the skin/hair/ mucous membranes/eyes
- If applicable, informative photos of the face, hands, feet, ears
- Copy of the **vaccination certificate**

If you agree with this procedure, please send your request to the following address:

Universitätsklinikum Carl Gustav Carus
an der Technischen Universität Dresden
UniversitätsCentrum für Seltene Erkrankungen
z.H. Tanita Kretschmer
Fetscherstraße 74
01307 Dresden

Please do not use staples to staple your documents. Refrain from sending additional documents as they will not be processed. After the assessment of your case in the USE has been completed, your documents will be digitised and the originals destroyed in accordance with data protection regulations. They will not be returned. Therefore, please submit only copies of the meaningful findings.

Yours sincerely,

Your USE team

Specialist questionnaire on medical history

Universitätsklinikum Carl Gustav Carus

Dear colleague,

you or your patient have contacted the USE because he/she is suspected of having a rare disease. In order to be able to help as promptly and meaningfully as possible, we need specific information about the patient, in particular a justification for the need for an assessment by a Centre for Rare Diseases and not, for example, by a specialised university outpatient clinic or a specialist. Please understand that we can only accept documents with a corresponding explanation by the treating specialist.

Patient information

Name, surname of the patient:

Date of birth of the patient:

Address of the patient:

Medical information on the disease

Suspicion of a rare disease from which group of forms?

brief explanation of the suspicion:

Why should the patient be assessed in the Rare Disease Centre and not in a specialised outpatient clinic?

Date, signature, stamp of the medical specialist

Medical History Questionnaire -Children

Universitätsklinikum
Carl Gustav Carus

Dear Advice Seeker,
You have contacted the USE because your child has or is suspected of having a rare disease. In order to be able to help you promptly and specifically, we need the following information detailed and complete information about your child and his or her symptoms. Therefore, please fill out the form completely and consult your pediatrician if you have any questions. We also ask you to enclose copies of all findings and medical and doctor's letters that are relevant to your request.

Presentation of the child's symptoms:

Please briefly summarize the child's medical condition in your own words:

1. basic data

1.1 Information on the patient

Date of birth: |__|__| / |__|__|__|__| (month/year)

Is the place of birth in Germany?

☐ Yes ☐ No, but: _____

Postal code (PLZ) of the place of birth (first 3 digits): |__|__|__|

Address: _____

Sex of patient: ☐ female ☐ male

nationality: _____

current height : |__|__|__|cm

current weight : |__|__|__|kg

current head circumference : |__|__|cm

1.2 Information on the patient's biological parents and grandparents

Mother's date of birth:|__|__| / |__|__|__|__| (month/year)

Father's date of birth: |__|__| / |__|__|__|__| (month/year)

Country of origin of parents and grandparents:

(Note: This question is asked because some rare diseases are more common in individual countries of origin).

Father: _____ Mother: _____

Maternal Grandmother: _____

Maternal Grandfather: _____

Paternal Grandmother: _____

Paternal Grandfather: _____

2. pedagogical support

2.1 Facilities

Does your child attend kindergarten? ☐ Yes, since: _____ ☐ No

Does your child attend school? ☐ Yes, since: _____ ☐ No

Does your child live in an institution? ☐ Yes, which one: _____ ☐ No

2.2 Funding requirements

Does your child have special needs?

☐ No ☐ Physiotherapy ☐ Occupational therapy ☐ Speech therapy
☐ Other forms of support: _____

3 Data on the birth of the child
(to be taken from the maternity passport or the child's yellow booklet)

3.1. Birth

In which week of pregnancy was the child born?
|__|__|+ |__| (WOP and day)

Birth weight: |__|__|__|__ g ☐ unknown
Body length: |__|__,|__| cm ☐ unknown
Head circumference: |__|__,|__| cm ☐ unknown

Childbirth:
☐ spontaneous
☐ planned caesarean section
☐ emergency cesarean section
☐ forceps delivery
☐ birth with suction cup
☐ unknown

Malformations noted at U1?
Yes, _____ ☐ No ☐ unknown

Malformations noted at U2?
Yes, _____ ☐ No ☐ unknown

4. pregnancy information

4.1. Conception

Did the pregnancy occur spontaneously? ☐ Yes ☐ No

Occurrence of pregnancy as a direct result of infertility treatment?
☐ Yes ☐ No

If Yes, after which?
Hormone treatment ☐ IVF ☐ ICSI
☐ other infertility treatment:_____

4.2 Prenatal diagnostics

(to be taken from the maternity record, WOP = week of pregnancy).

First trimester screening:
☐ inconspicuous ☐ not done ☐ unknown
☐ conspicuous: _____

Triple-Test:
☐ inconspicuous ☐ not done ☐ unknown
☐ conspicuous: _____

Ultrasound examinations/fine diagnostics abnormal?
☐ Yes ☐ No ☐ not done ☐ unknown
If Yes, what abnormalities and at what week of pregnancy?
1. _____
2. _____
3. _____

Has a chorionic villus sampling been performed?
☐ Yes ☐ No
Reason for examination:_____
Result:_____

Was an amniocentesis performed?
☐ Yes ☐ No
Reason for examination:_____
Result: _____

Was an umbilical cord puncture performed?
☐ Yes ☐ No
Reason for examination: _____
Result: _____

Was there a multiple pregnancy?
(incl. terminations during pregnancy)
☐ No
☐ Yes, specify if necessary (number of fetuses, singleness/twoness, deliveries during pregnancy with indication of WOP): _____

4.3. course of pregnancy

Chronic diseases of the mother before/during pregnancy?

☐ Yes, which:_____ ☐ No

Were any medications taken during pregnancy?

☐ No ☐ Yes, which ones?
(Preparation/dose/period of use; incl. vitamin preparations/nutritional supplements)
_____
_____

Eating habits during pregnancy

☐ normal ☐ vegan ☐ vegetarian ☐ gluten-free

5. child development

5.1 Developmental milestones

(as best as you can remember)
Purposeful grasping at |__|__| months
Turning from supine to prone at |__|__| months
Sitting freely at |__|__| months
Crawling at |__|__| months
Walking independently at |__|__| months
First 2 to 3 words at |__|__| months
First two-word sentences at |__|__| months
Clean and dry during the day at |__|__| months
Currently counting safely to: _____

5.2 Behaviour

Were there any neonatal adjustment problems?
Yes (which): _____ ☐ No

Are there/were there problems in kindergarten or school?
Yes (which): _____ ☐ No

Are there behavioural problems at home?
Yes (which): _____ ☐ No

Have you been approached by supervisors (educators/teachers) about your child's behaviour?
Yes: _____ No

6. information on family history

Have there been miscarriages in other pregnancies of the mother?

☐ Yes, number: |__| Cause/diagnosis: _____

☐ No

Are the families of the child's mother and father related?

☐ Yes ☐ No ☐ unknown

If Yes, how? _____

Are there any malformations or chronic diseases in family members?

☐ No

☐ Yes (which):

_____

_____

_____

7. the child's medical history

7.1. Hospital

Has your child ever had an operation?

☐ No

☐ Yes, operation/year:_____

Were there any other inpatient hospital stays?

☐ No

☐ Yes, disease / year: _____

_____

7.2 Course of the disease

Please indicate your child's current chief complaint:

_____

_____

_____

_____

_____

At what age did the symptoms that led to the suspicion of a rare disease start?

Symptom	Complaints since (birth/age of child):

With which symptoms did the disease start?

_____

_____

Has a diagnosis already been made?
☐ No ☐ Yes (what diagnosis, when and by whom?): ☐
_____

Has a rare disease been suspected?
☐ No
☐ Yes (what diagnosis, when and by whom?): _____

7.3 Preliminary findings

Which specialists have you already seen with the patient?

☐ Allergist	☐ General	☐ Practitioner	☐ Angiologist
☐ Anaesthetist	☐ Ophthalmologist	☐ Chiropractor	☐ Surgeon
☐ Diabetologist	☐ Dermatologist	☐ Endocrinologist	☐ Gastroenterologist
☐ Gynaecologist	☐ Haematologist	☐ ENT specialist	☐ Homeopath
☐ Human geneticist	☐ Immunologist	☐ Internist	☐ Cardiologist
☐ Lung specialist	☐ Renal specialist	☐ Neurologist	☐ Neurosurgeon
☐ Oncologist	☐ Orthopaedist	☐ Pathologist	☐ Psychiatrist
☐ Psychologist	☐ Psychosomatist	☐ Rheumatologist	☐ Radiologist
☐ Environmental physician	☐ Urologist	☐ Dentist	

☐ Others: _____
☐ Specialised paediatrician (which one):_____

Is your child currently receiving treatment?

☐ No
☐ Yes (since when/with whom?):_____

Were there any abnormalities in the course of the initial examinations?
(Preventive medical check-ups, to be taken from the yellow booklet)
☐ No
☐ Yes (which one and when?): _____

Do you already have laboratory test results, especially genetic tests?
☐ No
☐ Yes (please enclose)

7.4.Medication/allergies/intolerances/aids

Is your child taking medication?
☐ No
☐ Yes (which one/since when/dosage):
_____
_____

Does your child have allergies/other intolerances?
☐ No
☐ Yes: _____

Does your child need any aids? (e.g. glasses, aids etc.)
☐ No
☐ Yes: _____

☐

8. Contact with the Centre for Rare Diseases

Are you interested in participating in a study / inclusion in an (anonymous) patient register of patients with rare diseases?

☐ Yes ☐ No ☐ don´t now
☐ further information requested

Have you already had contact with a centre for rare diseases?
☐ No
☐ Yes (which one? when?):_____

I revocably agree that:

my disease-related data (images, recorded interviews and texts) are stored for research, my data provided as well as those resulting from further recommendations and examinations will be passed on to doctors treating me and to the staff of the Centre for Rare Diseases used in encrypted (pseudonymised) form for scientific purposes and published anonymously in specialist journals, my data is passed on to doctors who should be involved in the treatment.

I hereby confirm the accuracy of the information I have provided and my consent to.

_____ _____

Date, Place Signature, name in block letters