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 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 -Adults

Universitätsklinikum Carl Gustav Carus

Dear advice seeker,

you have contacted the USE because you have or are suspected to having a rare disease. In order to be able to help you promptly and in a targeted manner, we need as much detailed and complete information as possible about you and your symptoms. Therefore, please fill out the form completely and consult your GP if you have any questions. We also ask you to enclose copies of all findings and doctor's letters that are relevant to your case.

General patient data:

name, surname	
date of birth *(DD/MM/YYYY)*	
gender	☐ **male** ☐ **female** ☐ **divers**
address	
telephone number	
E-Mail	
Insurance	☐ **public health insurance** ☐ **private**
Gainful employment /study/ apprenticeship etc.	
sickness leave since	
highest level of education	
Family status/children	
Nationality	
Weight in kg	
Body height in cm	
Name, address and telephone number of the treating general practitioner or specialist	

When were you last abroad, especially before the onset of the disease, and where?

What is your relationship with the patient?	
☐ I am the patient myself	☐ I am not the patient, but: Contact details:

What is the main reason for contacting the USE (multiple answers possible)?	
☐ Diagnosis	☐ Expert opinion
☐ Information	☐ Second Opinion
Other reason:	

Please formulate your exact request:

Disease progression

Q **1: At what age did the complaints start?**			
☐ Since birth		☐ Since childhood	
☐ First complaints at the age of _____			
Which organ systems are affected?			
☐ Eyes	☐ Reproductive organs	☐ Circulation	☐ Adrenal glands
☐ Pancreas	☐ Throat	☐ Liver	☐ Nerves
☐ Blood (formation)	☐ Skin	☐ Lungs	☐ Kidneys
☐ Intestine	☐ Heart	☐ Stomach	☐ Ears
☐ Gall bladder	☐ Hormone system	☐ Spleen	☐ Thyroid
☐ Brain	☐ Immune system	☐ Muscles	☐ Digestive system
☐ Joints	☐ Bones	☐ Nose	☐ Teeth
☐ Other:			

Q **2: Did you experience the following complaints:**		
Weight loss	☐ Yes, kg:	☐ No
Night sweats	☐ Yes	☐ No
Fever attacks	☐ Yes	☐ No
Bleeding tendency	☐ Yes	☐ No

Q **3: Please indicate your current main complaints and their intensity (from 1-10)** **(1= hardly/slightly present, 10= maximum present):**										
	1	2	3	4	5	6	7	8	9	10
1.										
2.										
3.										
4.										
5.										

Q **4: With which symptoms did the disease start?**
Are there any other complaints?

Q **5: Have you already been diagnosed for your current complaints?**	
☐ Yes:	☐ No
☐ Suspected diagnoses only, namely: Who made these diagnoses/suspected diagnoses:	

Q **6: Is there any doubt about this diagnosis?**	
☐ Yes	☐ No
By whom were these doubts expressed and why?	

Q **7: Has a supervising doctor expressed a specific suspicion of a rare disease?**	
☐ Yes, suspicion of:	☐ No
Have you ever been to Dresden University Hospital because of your complaints?	
☐ Yes Date: Department/treating doctor:	☐ No

Q **8: Did similar complaints occur in your family/blood relatives?**		
☐ Yes	☐ No	☐ No statement possible

If yes, with whom?		
☐ Parents	☐ Siblings	☐ Children
☐ Grandparents	☐ Great-Grandparents	☐ Other:

Q **9: Please indicate the complaints and (existing) diagnoses per affected family member:**

Q **10: Did other diseases occur more frequently in your family/blood relatives or are genetic diseases known in your family?**	
☐ Yes:	☐ No
☐ No statement possible	

Q **11: When were you first treated by a doctor for your complaints? And with whom?**

Q **12: Where have you already made representations?**			
☐ Allergist	☐ General Practitioner	☐ Angiologist	☐ Anaesthetist
☐ Ophthalmologist	☐ Chiropractor	☐ Surgeon	☐ Diabetologist

☐ Dermatologist	☐ Endocrinologist	☐ Gastroenterologist	☐ Gynaecologist
☐ Haematologist	☐ ENT specialist	☐ Homeopath	☐ Human geneticist
☐ Immunologist	☐ Internist	☐ Cardiologist	☐ Paediatrician
☐ Lung specialist	☐ Kidney specialist	☐ Neurologist	☐ Neurosurgeon
☐ Oncologist	☐ Orthopaedist	☐ Pathologist	☐ Psychiatrist
☐ Psychologist	☐ Psychosomatist	☐ Rheumatologist	☐ Radiologist
☐ Environmental physician	☐ Urologist	☐ Vein specialist	☐ Dentist
☐ Tropical medicine	☐ Other:		

Q 13: Gynaecological history (only to be filled in if applicable):		
Abnormalities during menstruation:		☐ None
Pain during menstruation:	☐ Yes:	☐ No
Thromboses that have occurred:	☐ Yes, trigger (if known):	☐ No
Miscarriages	☐ Yes, in the year:	☐ No

Q 14: Do you have pets?	
☐ Yes	☐ No

Q 15: Do you have any special dietary habits (e.g. meat-free diet, vegan diet, etc.)?

Q 16: Do you have any other illnesses and if so, which ones?	
	Name of the disease
☐ Heart disease	
☐ Circulatory and vascular diseases	
☐ Lung and respiratory diseases	
☐ Blood diseases	
☐ Liver diseases	
☐ Kidney and urinary tract diseases	
☐ Diseases of the digestive tract	
☐ Metabolic diseases	
☐ Thyroid diseases	
☐ Eye diseases	
☐ Nervous disorders	
☐ Mood disorders	
☐ Allergies/intolerances	
☐ Diseases of the skeletal system	
☐ Muscular diseases	
☐ Other	
Are there any foreign objects on or in your body (piercings, prostheses, pacemakers, etc.)? ☐ Yes ☐ No If yes, what and where?:	

Q **17: Which operations have you had so far and at what time?**

Q **18: Please tick whether you have the following symptoms or events:**			
Joint stiffness in the morning	☐ no	☐ yes, for _____ minutes	
	☐ no	☐ yes, in the year: ☐ with skin rash ☐ with antibiotics	
Night's rest disturbed by pain	☐ no	☐ rarely	☐ often
Back pain	☐ no	☐ yes	☐ also at night
Back pain radiating into a leg	☐ no	☐ left	☐ right
Back pain radiating into an arm	☐ no	☐ left	☐ right
Painful whitening, subsequent blueness of the fingers when cold	☐ no	☐ yes, in the year:	
Inflammation or redness of the eyes	☐ no	☐ yes, since (year)	
Dryness of eyes/mucous membranes (also mouth, genital area)	☐ no	☐ yes, in the year:	
Other changes to the skin and mucous membranes (also mouth, genital area)	☐ no	☐ yes, in the year:	
Pain during urination	☐ no	☐ yes, in the year:	
Diarrhoea	☐ no	☐ yes, bloody	☐ yes, not bloody
Chronic inflammatory bowel disease in yourself or in family	☐ no	☐ yes, in the year	
Osteoporosis	☐ no	☐ yes, in the year	
Shortness of breath when climbing stairs	☐ no	☐ yes, after _____ floors	
Stroke	☐ no	☐ yes, since	
Tuberculosis in you or in the family	☐ no	☐ yes, since:	
Rheumatic diseases in the family	☐ no	☐ yes:	

Q **19: Please tick whether you have the following symptoms or events:**	
	If yes, since when?
☐ Pneumonia	
☐ Bone inflammation	
☐ Blood poisoning	

☐ Chronic viral infections	
☐ Chronic skin or genital warts	
☐ Chronic fungal infections	
☐ Chronic swelling of the lymph nodes	
☐ Enlarged spleen	

Q **20: Which medicines (incl. food supplements) do you currently take, in which dosage and since when?**	
Medication / food supplement, since when	*Dosage*

Q **21: Dealing with physical complaints**					
	never	rarely	sometimes	often	very often
I think that my physical complaints signs of a serious illness.	☐	☐	☐	☐	☐
I am very worried about my health.	☐	☐	☐	☐	☐
My health worries hinder me in my everyday life.	☐	☐	☐	☐	☐
I am convinced of the seriousness of my physical complaints.	☐	☐	☐	☐	☐
My physical complaints frighten me.	☐	☐	☐	☐	☐
My physical complaints occupy me most of the day.	☐	☐	☐	☐	☐
Others tell me that my physical complaints are not bad.	☐	☐	☐	☐	☐
I am worried that my physical complaints will never stop.	☐	☐	☐	☐	☐
The worries about my health rob me of energy.	☐	☐	☐	☐	☐
I think that the doctors do not take my physical complaints seriously.	☐	☐	☐	☐	☐
I am worried that I will continue to be affected by my physical complaints in the future.	☐	☐	☐	☐	☐
My physical complaints make it difficult for me to concentrate on other things.	☐	☐	☐	☐	☐

Q **22: Have you had an anxiety attack (sudden feeling of fear or panic) in the last 4 weeks?**	
☐ Yes	☐ No

Q **23: During the last two weeks, how often did you feel affected by the following complaints?**				
	Not at all	**On individu-al days**	**On more than half of the days**	**Almost every day**
Being nervous, anxious or tense	☐	☐	☐	☐
Not being able to stop or control worrying	☐	☐	☐	☐
Excessive worry about various matters	☐	☐	☐	☐
Difficulty relaxing	☐	☐	☐	☐
Restlessness, making it difficult to sit still	☐	☐	☐	☐
Quick temper or irritability	☐	☐	☐	☐
Feeling anxious, as if something bad is going to happen	☐	☐	☐	☐

Q **24: How much did you feel affected by the following complaints in the last 4 weeks?**			
	Not affected	**Little affected**	**Strongly affected**
Worry about your health	☐	☐	☐
Worries about your weight or appearance	☐	☐	☐
Difficulties with your spouse, significant other, boyfriend/girlfriend	☐	☐	☐
Burden of caring for children, parents or other family members	☐	☐	☐
Stress at work or school	☐	☐	☐
Financial problems or worries	☐	☐	☐
Having no one to discuss problems with can discuss problems with	☐	☐	☐
Something bad that happened recently.	☐	☐	☐
Thoughts of terrible events from the past, or dreams about it - e.g. the destruction of one's home, a serious accident, physical violence, or a sexual act under duress	☐	☐	☐

Q **25: During the last two weeks, how often did you feel affected by the following complaints? Please underline where applicable.**				
	Not at all	**on single days**	**On more than half of the days**	**almost every day**
Little interest or pleasure in your activities	☐	☐		☐
Low mood, melancholy or hopelessness	☐	☐	☐	☐
Difficulty falling asleep or staying asleep, or increased Sleep	☐	☐	☐	☐
Fatigue or feeling of having no energy	☐	☐	☐	☐
Decreased appetite or excessive need to eat.	☐	☐	☐	☐
Poor opinion of oneself; feeling that one has failed or having disappointed the family	☐	☐	☐	☐
Difficulty concentrating on something, e.g. reading the newspaper or watching television.	☐	☐	☐	☐
Were your movements or speech so slowed down that others would notice it? Or, on the contrary, were you " fidgety" or restless, that it made you have a stronger urge to move than usual?	☐	☐	☐	☐

Q **26: Do you take or have you ever taken medication for anxiety, depression or stress?**	
☐ Yes	☐ No
Which?	

Declaration of consent

I hereby give my consent that the data submitted in the questionnaire and the findings presented may be inspected by doctors of the USE for the purpose of evaluating my case and recommending the further course of action. This also includes the inspection of genetic diagnostic findings which comply with the Gene Diagnostics Act. Medical confidentiality according to §203 StGB and data secrecy according to §6 Sächsisches
Saxon Data Protection Act remain unaffected. I can revoke this declaration of consent in writing or verbally at any time without giving reasons.

I consent to the use and evaluation of the data in anonymised form for scientific purposes.

☐ Yes
☐ No

_____ _____

place, date signature of the patient, if applicable of the caregiver