



## 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 -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:

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

<b>When were you last abroad, especially before the onset of the disease, and where?</b>

<b>What is your relationship with the patient?</b>	
<input type="checkbox"/> I am the patient myself	<input type="checkbox"/> I am not the patient, but: Contact details:

<b>What is the main reason for contacting the USE (multiple answers possible)?</b>	
<input type="checkbox"/> Diagnosis	<input type="checkbox"/> Expert opinion
<input type="checkbox"/> Information	<input type="checkbox"/> 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?**

<input type="checkbox"/> Eyes	<input type="checkbox"/> Reproductive organs	<input type="checkbox"/> Circulation	<input type="checkbox"/> Adrenal glands
<input type="checkbox"/> Pancreas	<input type="checkbox"/> Throat	<input type="checkbox"/> Liver	<input type="checkbox"/> Nerves
<input type="checkbox"/> Blood (formation)	<input type="checkbox"/> Skin	<input type="checkbox"/> Lungs	<input type="checkbox"/> Kidneys
<input type="checkbox"/> Intestine	<input type="checkbox"/> Heart	<input type="checkbox"/> Stomach	<input type="checkbox"/> Ears
<input type="checkbox"/> Gall bladder	<input type="checkbox"/> Hormone system	<input type="checkbox"/> Spleen	<input type="checkbox"/> Thyroid
<input type="checkbox"/> Brain	<input type="checkbox"/> Immune system	<input type="checkbox"/> Muscles	<input type="checkbox"/> Digestive system
<input type="checkbox"/> Joints	<input type="checkbox"/> Bones	<input type="checkbox"/> Nose	<input type="checkbox"/> Teeth
<input type="checkbox"/> Other:			

**Q 2: Did you experience the following complaints:**

Weight loss	<input type="checkbox"/> Yes, kg:	<input type="checkbox"/> No
Night sweats	<input type="checkbox"/> Yes	<input type="checkbox"/> No
Fever attacks	<input type="checkbox"/> Yes	<input type="checkbox"/> No
Bleeding tendency	<input type="checkbox"/> Yes	<input type="checkbox"/> 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?



<b>Q 5: Have you already been diagnosed for your current complaints?</b>	
<input type="checkbox"/> Yes:	<input type="checkbox"/> No
<input type="checkbox"/> Suspected diagnoses only, namely: Who made these diagnoses/suspected diagnoses:	

<b>Q 6: Is there any doubt about this diagnosis?</b>	
<input type="checkbox"/> Yes	<input type="checkbox"/> No
By whom were these doubts expressed and why?	

<b>Q 7: Has a supervising doctor expressed a specific suspicion of a rare disease?</b>	
<input type="checkbox"/> Yes, suspicion of:	<input type="checkbox"/> No
Have you ever been to Dresden University Hospital because of your complaints?	
<input type="checkbox"/> Yes Date: Department/treating doctor:	<input type="checkbox"/> No

<b>Q 8: Did similar complaints occur in your family/blood relatives?</b>		
<input type="checkbox"/> Yes	<input type="checkbox"/> No	<input type="checkbox"/> No statement possible

<b>If yes, with whom?</b>		
<input type="checkbox"/> Parents	<input type="checkbox"/> Siblings	<input type="checkbox"/> Children
<input type="checkbox"/> Grandparents	<input type="checkbox"/> Great-Grandparents	<input type="checkbox"/> Other:

<b>Q 9: Please indicate the complaints and (existing) diagnoses per affected family member:</b>

<b>Q 10: Did other diseases occur more frequently in your family/blood relatives or are genetic diseases known in your family?</b>	
<input type="checkbox"/> Yes:	<input type="checkbox"/> No
<input type="checkbox"/> No statement possible	

<b>Q 11: When were you first treated by a doctor for your complaints? And with whom?</b>

<b>Q 12: Where have you already made representations?</b>			
<input type="checkbox"/> Allergist	<input type="checkbox"/> General Practitioner	<input type="checkbox"/> Angiologist	<input type="checkbox"/> Anaesthetist
<input type="checkbox"/> Ophthalmologist	<input type="checkbox"/> Chiropractor	<input type="checkbox"/> Surgeon	<input type="checkbox"/> Diabetologist



<input type="checkbox"/> Dermatologist	<input type="checkbox"/> Endocrinologist	<input type="checkbox"/> Gastroenterologist	<input type="checkbox"/> Gynaecologist
<input type="checkbox"/> Haematologist	<input type="checkbox"/> ENT specialist	<input type="checkbox"/> Homeopath	<input type="checkbox"/> Human geneticist
<input type="checkbox"/> Immunologist	<input type="checkbox"/> Internist	<input type="checkbox"/> Cardiologist	<input type="checkbox"/> Paediatrician
<input type="checkbox"/> Lung specialist	<input type="checkbox"/> Kidney specialist	<input type="checkbox"/> Neurologist	<input type="checkbox"/> Neurosurgeon
<input type="checkbox"/> Oncologist	<input type="checkbox"/> Orthopaedist	<input type="checkbox"/> Pathologist	<input type="checkbox"/> Psychiatrist
<input type="checkbox"/> Psychologist	<input type="checkbox"/> Psychosomatist	<input type="checkbox"/> Rheumatologist	<input type="checkbox"/> Radiologist
<input type="checkbox"/> Environmental physician	<input type="checkbox"/> Urologist	<input type="checkbox"/> Vein specialist	<input type="checkbox"/> Dentist
<input type="checkbox"/> Tropical medicine	<input type="checkbox"/> Other:		

<b>Q 13: Gynaecological history (only to be filled in if applicable):</b>		
Abnormalities during menstruation:		<input type="checkbox"/> None
Pain during menstruation:	<input type="checkbox"/> Yes:	<input type="checkbox"/> No
Thromboses that have occurred:	<input type="checkbox"/> Yes, trigger (if known):	<input type="checkbox"/> No
Miscarriages	<input type="checkbox"/> Yes, in the year:	<input type="checkbox"/> No

<b>Q 14: Do you have pets?</b>	
<input type="checkbox"/> Yes	<input type="checkbox"/> No

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

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



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

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**Q 18: Please tick whether you have the following symptoms or events:**

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

**Q 19: Please tick whether you have the following symptoms or events:**

	If yes, since when?
<input type="checkbox"/> Pneumonia	
<input type="checkbox"/> Bone inflammation	
<input type="checkbox"/> Blood poisoning	



<input type="checkbox"/> Chronic viral infections	
<input type="checkbox"/> Chronic skin or genital warts	
<input type="checkbox"/> Chronic fungal infections	
<input type="checkbox"/> Chronic swelling of the lymph nodes	
<input type="checkbox"/> Enlarged spleen	

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

<i>Medication / food supplement, since when</i>	<i>Dosage</i>

**Q 21: Dealing with physical complaints**

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

**Q 22: Have you had an anxiety attack (sudden feeling of fear or panic) in the last 4 weeks?**

<input type="checkbox"/> Yes	<input type="checkbox"/> No
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<b>Q 23: During the last two weeks, how often did you feel affected by the following complaints?</b>				
	<b>Not at all</b>	<b>On individual days</b>	<b>On more than half of the days</b>	<b>Almost every day</b>
Being nervous, anxious or tense	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Not being able to stop or control worrying	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Excessive worry about various matters	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Difficulty relaxing	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Restlessness, making it difficult to sit still	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Quick temper or irritability	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Feeling anxious, as if something bad is going to happen	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

<b>Q 24: How much did you feel affected by the following complaints in the last 4 weeks?</b>			
	<b>Not affected</b>	<b>Little affected</b>	<b>Strongly affected</b>
Worry about your health	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Worries about your weight or appearance	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Difficulties with your spouse, significant other, boyfriend/girlfriend	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Burden of caring for children, parents or other family members	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Stress at work or school	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Financial problems or worries	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Having no one to discuss problems with can discuss problems with	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Something bad that happened recently.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
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	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>



<b>Q 25: During the last two weeks, how often did you feel affected by the following complaints?</b>				
<b>Please underline where applicable.</b>				
	Not at all	on single days	On more than half of the days	almost every day
Little interest or pleasure in your activities	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>
Low mood, melancholy or hopelessness	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Difficulty falling asleep or staying asleep, or increased Sleep	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Fatigue or feeling of having no energy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Decreased appetite or excessive need to eat.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Poor opinion of oneself; feeling that one has failed or having disappointed the family	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Difficulty concentrating on something, e.g. reading the newspaper or watching television.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
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?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

<b>Q 26: Do you take or have you ever taken medication for anxiety, depression or stress?</b>	
<input type="checkbox"/> Yes	<input type="checkbox"/> 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.
<input type="checkbox"/> Yes
<input type="checkbox"/> No

\_\_\_\_\_  
place, date

\_\_\_\_\_  
signature of the patient, if applicable of the caregiver

