

## Transition consultation

An important concern for us is also the support of patients with rare genetic diseases in the **transition from pediatric to adult medicine**. For this purpose, we offer a transition consultation together with the Clinical Genomics Department of the Institute for Human Genetics and Genomic Medicine at Uniklinik RWTH Aachen. If you are interested, please contact the **ZSEA** using the contact details provided.

## Clinical genetic diagnostics

The conclusion of selective contracts with different health insurance companies (AOK, BKK, vdek et al.) enable us to offer patients in suitable cases to perform **comprehensive molecular diagnostics**.

If you are interested, please contact the **ZSEA** or the **Secretariat of the Institute of Human Genetics and Genome Medicine (www.humangenetik.ukaachen.de)**. The medical contact person is **Univ.-Prof. Dr. med. Miriam Elbracht**.



European  
Reference  
Networks



**PARTNER**  
im Virtuellen  
Krankenhaus NRW

## Central Numbers

phone: +49 241 888 07 152  
Fax: +49 241 80 33 87021  
zsea@ukaachen.de

### Office hours by telephone

Tuesday and Wednesday: 10:00 – 11:00  
Thursday: 13:00 – 15:00

[www.ZSEA.ukaachen.de](http://www.ZSEA.ukaachen.de)

## Contact

### Executive Board Spokesman

**Univ.-Prof. Dr. med. Martin Mücke**  
**Uniklinik RWTH Aachen**  
**Institute of Digital General Medicine and**  
**Center for Rare Diseases (ZSEA)**

### Postal address (ZSEA)

Pauwelsstraße 30  
52074 Aachen

### Visiting address (ZSEA)

Bahnhofstraße 18  
52064 Aachen

### Contact persons

#### Dr. med. Julia Fleck

Medical patient guide

#### Dr. phil. Michaela Hesse

Social scientist

#### Kyros Mani

Medical Director

#### Dr. rer. nat. Christopher Schippers

Coordinating Managing Director

#### Ira Verhoeven

Non-medical patient guide

[zsea@ukaachen.de](mailto:zsea@ukaachen.de)

## How you can help us

If you would like to support our work, please consider making a donation! Your donations are very welcome, and **100 percent** of the amount given goes towards our work for patients with rare diseases.

### Sparkasse Aachen

IBAN: DE27 3905 0000 0013 0040 15  
BIC: AACSD33

**Please give "Donation IA 225040" as the purpose of payment** so that your donation can be allocated to the Center for Rare Diseases in Aachen. You will automatically receive a donation receipt if you also give your address.

**UNIKLINIK**  
**RWTHAACHEN**

Center for Rare Diseases  
Aachen (ZSEA)



## Center for Rare Diseases (ZSEA)

**Univ.-Prof. Dr. med. Martin Mücke**  
Executive Board Spokesman

**Univ.-Prof. Dr. med. Miriam Elbracht**  
Deputy Board Spokesman

**Kyros Mani**  
Medical Director



Univ.-Prof. Dr. med.  
Martin Mücke



Univ.-Prof. Dr. med.  
Miriam Elbracht

## Dear Patients, Dear Colleagues, Ladies and Gentlemen,

Those affected who are suspected or diagnosed with a rare disease present us all with special challenges. We therefore founded the Center for Rare Diseases (ZSEA) at Uniklinik RWTH Aachen in 2014. In addition to an office with a patient guide function, which supports in particular the diagnosis finding, eleven treatment and research centers in different indication areas, in which experts advance care and support accordingly, are networked with each other.

We see ourselves explicitly as a partner who wants to provide competent medical contacts.

The main goals are to accompany patients on their way to the correct diagnosis and to provide them with care and support for the rare diseases that we treat where necessary. We will gladly support you with our expertise, which also includes psycho-social counseling.

Yours sincerely

Univ.-Prof. Dr. med.  
Martin Mücke

Univ.-Prof. Dr. med.  
Miriam Elbracht

## Definition

A rare disease is one that occurs in no more than one in two thousand people. It is estimated that there are up to 8,000 rare diseases worldwide, probably affecting around four million people in Germany alone.

## Tasks and goals

- Optimization of **patient care** and improvement of quality of life of persons afflicted and their relatives by multidisciplinary expert teams
- Intensifying and focusing **research** to secure a fast knowledge transfer into practice
- Optimization of **training and further education** in the health sector and raising public awareness of rare diseases
- **Cooperation** with patient organizations and self-help groups to respond better to patients' needs

## Focal areas

- Rare neurological diseases
- Rare hematological diseases
- Rare liver and gastrointestinal diseases
- Facial and skeletal malformations
- Rare kidney diseases in adults
- Rare inflammatory diseases of infancy and adolescence
- Retinopathies and retinopathy syndromes
- Syndromal diseases and infantile breathing regulatory disorder
- Rare allergies and skin diseases
- Rare hereditary tumor diseases
- Arrhythmogenic right ventricular cardiomyopathies and keratinopathies (research only)

## What's important for us

We ask **patients** and their relatives to contact us only after consulting a doctor as to whether it might make sense to contact the ZSEA (further information at [www.ZSEA.ukaachen.de](http://www.ZSEA.ukaachen.de)). In the event of a **confirmed diagnosis**, please get in touch with the contact persons in the relevant treatment and research centers (see ZSEA homepage). **Kyros Mani** and **Dr. med. Julia Fleck** are happy to help if the **illness symptoms are unclear** and for all other inquiries.



The ZSEA is a member of the North Rhine-Westphalian Centers for Rare Diseases ([www.nrw-zse.de](http://www.nrw-zse.de)).