



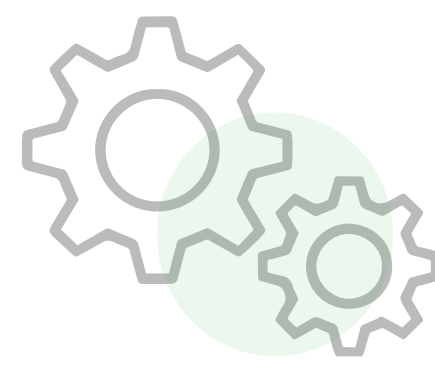
RESEARCH FOR RARE

German Networks on Rare Diseases



WHAT IS A RARE DISEASE?

A disease is considered rare if fewer than five in 10,000 people are affected. However, since around 8,000 rare diseases are known by now, the impact of all rare diseases taken together on the population is high: Four million people with a rare disease live in Germany, 30 million in Europe. 70 percent of these mostly chronic diseases manifest in childhood.



SPECIAL CHALLENGES

Most rare diseases are of genetic origin (80 percent), but their causes are often unexplored. The relatively small numbers of affected people, experts and suitable medications complicate diagnosis and appropriate therapies. As a result, those affected go through an odyssey of medical visits, while the course of the disease can worsen irreversibly. In order to avoid those developments, patient pathways have to be defined and specialist knowledge is needed.



RESEARCH OBJECTIVES

Research focuses on genetic causes, underlying disease mechanisms, improved diagnostics and new treatments to be developed. For these purposes, the Federal Ministry of Education and Research (BMBF) has been funding translation-oriented research networks on various rare diseases since 2003.



MEANING OF COLLABORATION

Networking at national and international level is of great value to advance new methods and technologies. In Europe, researchers are actively involved in the establishment of European Reference Networks (ERN) for rare diseases. Additionally, the intensive collaboration between patients, patient organisations and physicians is very important. This is the basis for carrying out clinical studies and establishing patient registries and biobanks.

RESEARCH NETWORKS AT A GLANCE:

ADDResS

Translational Research for Persons with Abnormal DNA Damage Response
Prof. Dr. med. Christian Kratz - Hannover Medical School

CONNECT-GENERATE

German Network for Research on Autoimmune Encephalitis
PD Dr. med. Frank Leypoldt - University Hospital Schleswig-Holstein, Kiel

GAIN

German Genetic Multi-Organ Auto-Immunity Network
Prof. Dr. med. Bodo Grimbacher - Medical Center – University of Freiburg

HiChol

Network for Hereditary Intrahepatic Cholestasis
Prof. Dr. med. Verena Keitel-Anselmino - University Hospital Magdeburg

MyPred

Network for Young Individuals with Syndromes Predisposing to Myeloid Malignancies
Dr. med. univ. Miriam Erlacher, PhD - Medical Center – University of Freiburg

NEOCYST

Network for Early Onset Cystic Kidney Disease
Dr. Jens König - University Hospital Münster

STOP-FSGS

Network for Speed Translation-Oriented Progress to Treat FSGS
Prof. Dr. med. Tobias B. Huber - University Medical Center Hamburg-Eppendorf

TreatHSP.net

Network for Translational Research on Hereditary Spastic Paraplegias
Prof. Dr. med. Rebecca Schüle - Heidelberg University Hospital

Treat-ION

Network for New Therapies for Neurological Ion Channel and Transporter Disorders
Prof. Dr. med. Holger Lerche - University Hospital Tübingen



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