RESEARCH FOR RARE

Translational Network for Sarcoma (TranSaRNet)

The Network aims to improve cure rates in high-risk malignant sarcoma. To achieve this goal, novel non-cross-resistant therapies are needed that efficiently target dormant and chemo-resistant residual tumor cells and overcome tumor cell resistance.

Autoinflammatory Disorders in Children and Adolescents (AID-NET)

The primary research goal of this consortium is to elucidate the pathophysiology of autoinflammatory syndromes. Scientific projects focusing on secretory pathways and novel effector mechanisms of innate immunity have been combined with translational approaches to identify genetic and serological markers.

Cellular Approaches for Rare Pulmonary Diseases (CARPuD2)

Only limited or no therapeutic options are available for the rare pulmonary diseases α1-antitrypsin deficiency, cystic fibrosis, and surfactant deficiencies. The generation of induced pluripotent stem cells opens new avenues for the exploration of novel cellular therapies based on genetically corrected patient-specific stem cells.

Systemic Amyloid Light-Chain (AL) Amyloidosis (GERAMY)

Systemic amyloid light-chain amyloidosis is a rare but fatal protein folding disorder. It is the aim of the consortium to increase knowledge of the pathogenesis of AL amyloidosis and enable new developments to improve the diagnosis and prognosis of, and therapy for, affected patients.

Imprinting Diseases

Errors in imprint erasure, establishment and maintenance lead to aberrant gene expression and disease. Using locus-specific and genome wide genetic and epigenetic studies, the consortium will identify patients who have single or multiple imprinting defects and search for genetic variants in cis-regulatory elements and transacting factors involved in genomic imprinting.

Netzwerk Imprinting-Erkrankungen

Hannover
Münster
Essen
Duisburg
Heidelberg
Tübingen
Ulm
Freiburg
Munich

Hereditary Retinal Disorders (HOPE)

Hereditary retinal disorders represent a very heterogeneous group of blinding diseases affecting about 30.000 people in Germany. HOPE aims at improving and refining the diagnostics of hereditary retinal disorders and at developing new therapeutic strategies close to human application.

Primary immundeficiency disorders (PID-NET)

Primary immundeficiency disorders represent rare inborn errors of the immune system. PID-NET scientists implement a national platform for patients with defined primary immundeficiency disorders in order to improve diagnosis and therapy as well as to develop innovative genetherapeutic strategies.

Neurological and ophthalmological Ion Channel Disorders(IonNeurONet)

The goal of IonNeurONet is to create a clinical and research network for neurological and ophthalmological ion channel disorders. Collaboration of clinicians, geneticists and physiologists will be ideal to identify novel disease mechanisms as a basis for future therapies.

Mitochondrial Diseases (mitoNET)

Mitochondrial disorders are caused by genetically induced biochemical defects of the cellular energy production. Through the national network with its biobanks, patient registries and scientific projects mitoNET aims to improve health care for patients with mitochondrial diseases.

Craniofacial Disorders (FACE)

Very often the causes of craniofacial disorders are still unclear. The goal of the consortium is to study craniofacial disorders in order to identify aberrant gene regulatory mechanisms relevant for classification of new disease entities and new therapeutic approaches.

Motoneuron Diseases (MNDnet)

The concept of this consortium is a combination of patient's interests, clinical research and basic research. MND-Net aims to build up a German register in order to develop the basis for therapeutical interventional studies and analyzes the psychosocial consequences of these rare orphan diseases.

Genetic Modification of the CFTR-Gene for Cystic Fibrosis (GALENUS)

Patients with cystic fibrosis suffer from chronic lung diseases and malfunction of the pancreas, life expectancy is still not more than 40 years. The consortium aims to achieve new results through research and long-term studies in animal models in order to develop inventive strategies for affected patients.

Rare diseases are not so rare: It is estimated that there are approximately 7000-8000 rare diseases. Their prevalence however varies greatly and more than 4 million people in Germany alone are affected. About 80% of rare diseases present themselves during infancy. Many of them are still not curable.
The German Ministry for Education and Research has supported research activities devoted to rare diseases since 2003. Researchers from all over Germany have joined together to investigate certain disease groups. Currently, 12 such research networks are being funded. The aim of the research networks, in which both researchers and medical specialists are involved, is to find the underlying causes of currently incurable diseases and to explore and develop new therapeutic approaches. The latest technologies such as next generation sequencing for genome research are available.
Close collaboration of research networks located all over Germany and specialised centers for rare diseases allows for the integration of research results into patient care. The networks also cooperate with patient advocacy groups, who bring their expertise to the table.
Our alumni networks still actively engage in our research network for rare diseases. For further information please visit our website: www.research4rare.de

SPONSORED BY THE

Federal Ministry of Education and Research

Contact

Coordinating Center for the Research Networks on Rare Diseases
University Hospital Munich; Dr. von Hauner Children's Hospital

Spokesman: Prof. Dr. Christoph Klein

Lindwurmstraße 4 • D-80337 Munich
T +49. (0)89. 51 60 - 51 26 • F +49. (0)89. 51 60 - 77 02
info@research4rare.de • www.research4rare.de