RESEARCH FOR RARE

Forschung für seltene Erkrankungen

Neuronal Ceroid Lipofuscinoses (NCL2TREAT)
NCL2TREAT wants to enhance the understanding of the biogenesis and role of lysosomes in cellular homeostasis, identify and characterise novel lysosomal components, generate cell and mouse models of lysosomal diseases, develop novel therapies and acquire clinical data for natural history descriptions for NCLs.
Coordinator: Prof. Dr. Thomas Braulke, University Medical Center Hamburg-Eppendorf

Autoinflammatory Disorders in Children and Adolescents (AID-Net)
Innate autoinflammatory diseases are characterized by recurrent, self-limited episodes of fever and inflammation. The primary research goal of AID-net is to elucidate and modulate the pathophysiology of autoinflammatory syndromes by using a translational approach.
Coordinator:
Prof. Dr. Johannes Roth, University Hospital Muenster

Early Onset Cystic Kidney Disease (NEOCYST)
Hereditary cystic kidney diseases are among the most important causes of chronic renal failure in childhood. The primary objective of NEOCYST is to improve the life of patients and their families by increasing the knowledge of the epidemiology, genetics, molecular pathophysiology and long-term outcomes of pediatric cystic kidney disease with and without extrarenal manifestations.
Coordinator: Prof. Dr. Martin Konrad, University Hospital Münster

Imprinting-Erkrankungen (IMPRINTING)
Errors in imprint erasure, establishment and maintenance lead to aberrant gene expression and disease. Using locusspecic 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.
Coordinator: Prof. Dr. Bernhard Horsthemke, University Hospital Essen

Idiopathic FSGS (STOP-FSGS)
Idiopathic FSGS (focal segmental glomerulosclerosis) is a rare disease representing a major cause for irreversible loss of renal function and life-long need for dialysis. STOP-FSGS wants to address several key questions regarding the pathogenesis, diagnosis and novel therapies of FSGS.
Coordinator:
Prof. Dr. Marcus Möller, University Hospital Aachen

RASopathies (GeNeRare)
RASopathies constitute a heterogeneous group of disorders with a common molecular pathogenesis and overlapping phenotypic patterns. GeNeRare addresses the pathophysiology and molecular pathogenesis that is critical for long-term outcome but will also study novel aspects of RASopathies.
Coordinator:
Prof. Dr. Martin Zenker, University Hospital Magdeburg

Charcot-Marie-Tooth (CMT-NET)
Charcot-Marie-Tooth disease is an inherited disease of the peripheral nervous system without known cure. Affected patients suffer from sensory and motor impairments in various degrees. CMT-NET aims to identify genetic and non-genetic risk factors as well as novel therapeutic targets.
Coordinator:
Prof. Dr. Michael Sereda, University Medical Centre Göttingen

Dystonias (DysTract)
Dystonias comprise a heterogeneous group of idiopathic and incurable movement disorders.
The consortium combines human clinical and experimental animal research to describe the entire disease path of dystonia from a molecular level to brain network abnormalities. Patients will immediately benefit through standardizing the diagnosis and treatment approach to dystonia and establishing a reference database for all groups involved in dystonia care.
Coordinator: Prof. Dr. Jens Volkmann, University Hospital Würzburg

Cognitive impairment disorders with defective chromatin (CHROMATIN-Net)
CHROMATIN-Net explores phenotypic and molecular overlap of various rare syndromes associated with abnormal chromatin dynamics. These disorders include Coffin-Siris, Cornelia de Lange, and Nicolaides-Baraitser syndrome which show an overlapping phenotypic presentation with a broad spectrum of dysmorphic features and variable degrees of cognitive impairment.
Coordinator:
Prof. Dr. André Reis, University Hospital Erlangen

Primary immundeficiency disorders (PID-NET)
Primary immunodeficiency disorders (PID) represent rare inborn errors of the immune system predisposing to recurrent infections, autoimmunity, allergy, and cancer. PID-NET plays a substantial role in coordinating primary immunodeficiency research in Germany and thus helps to improve diagnosis and therapy of patients with rare inherited immunodeficiency syndromes.
Coordinator:
Prof. Dr. Christoph Klein, University Hospital Munich

Research for rare diseases – Together toward Diagnosis and Treatment

Despite huge efforts and initiatives from both researchers and medical specialists, the underlying causes for many rare diseases are not yet understood. For most of the rare diseases curative therapies are not available.

In order to strengthen clinical research on rare diseases, the German Ministry for Education and Research (BMBF) supports ten networks for rare diseases (funding period 2015/16-2018). Within these networks, about 50 research projects work on specific disease groups in order to develop innovative approaches for diagnosis and therapy. Close interaction of preclinical research and clinical practice can contribute decisively to the improvement of the situation of patients with a rare disease.

For more information, please visit our website: www.research4rare.de

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