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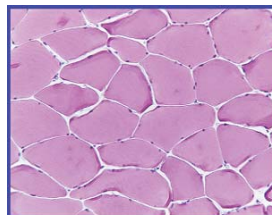
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**Muscular Dystrophy (MD-NET)**

Muscular dystrophies are a group of more than 30 hereditary diseases that are characterized by progressive wasting of skeletal muscle frequently involving the heart and other organs. The MD-NET unites expert physicians and scientists throughout Germany who – among other projects – carry out double-blind, placebo-controlled therapeutic trials for Duchenne and limb girdle muscular dystrophy.



muscle section

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**Epidermolysis bullosa**

Epidermolysis bullosa (EB) is a group of hereditary skin diseases in which little mechanical strain already causes skin blisters and wounds. This reduces the quality of life and seriously impairs the socio-economic situation of the patients and their relatives. Until this day, EB cannot be healed. Only a better understanding of the disease will enable the development of molecular treatments and especially of gene therapeutic approaches.



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**Skeletal Dysplasias (SKELNET)**

Skeletal dysplasias are hereditary disorders of bone development and encompass several hundred diseases with symptoms like dwarfism, disproportionate anatomy, joint problems or severe premature degenerative processes. Each particular disease of this large group of disorders is a rare condition by itself and many of them are not well studied, yet. Running on an innovative internet platform, SKELNET will contribute to the coordination of research activities in this field and provide better support and treatment for patients.



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**Hereditary Metabolic Disorders (METABNET)**

Some of the many occurring hereditary metabolic disorders are diagnosed in the course of newborn screening. Most of these diseases result in acute and chronic brain dysfunction and have a high mortality rate if not treated. The often required life-long dietetic or drug therapy is complicated, expensive and very burdensome for the patients and their families. One of the projects within the framework of METABNET is an international cross-section study concerning glutaric aciduria which is carried out in 15 countries.

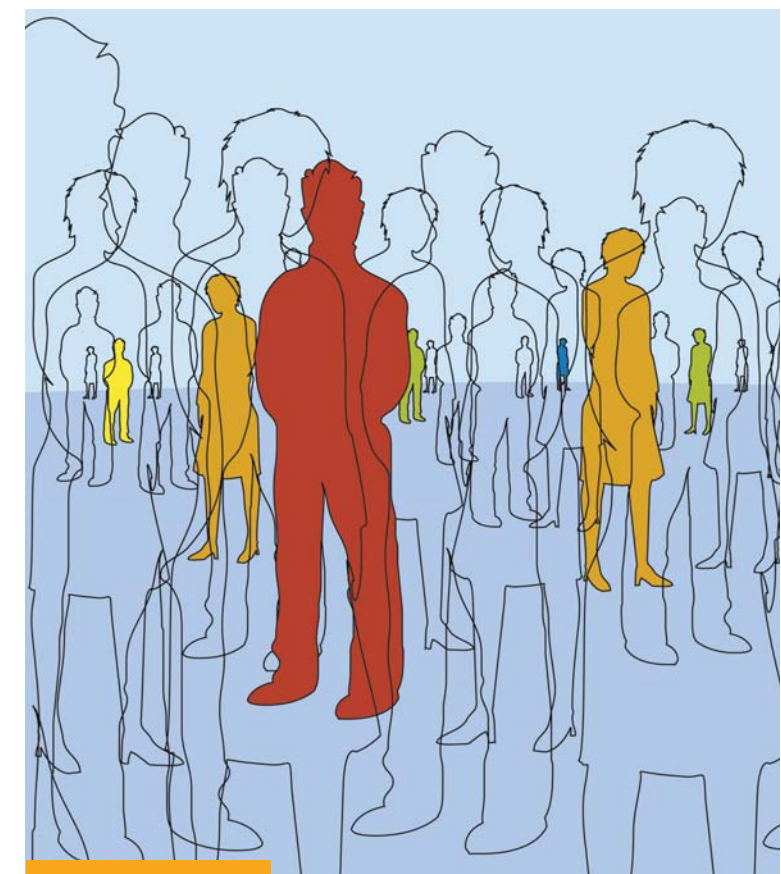


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Federal Ministry  
 of Education  
 and Research

**Rare Diseases –  
 Millions of Patients**



RESEARCH

# Rare Diseases – Millions of Patients

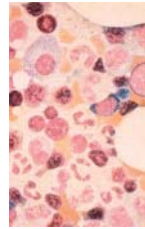
More than 7,000 of the approx. 30,000 known diseases are classified as “rare diseases”. A disease is considered “rare” if less than one in 2,000 people suffers from it. However, taken together these diseases are not a rare phenomenon – several million persons are affected in Germany alone.

Most frequently, rare diseases are caused by genetic defects. Patients often suffer from severe symptoms which require expensive and complex interdisciplinary treatment and care. The diseases are a heavy burden for the patients and their families and sometimes lead to death at a very young age. The rareness of the individual disease poses a serious challenge for research and patient care.

Therefore the Federal Ministry of Education and Research (BMBF) has been funding ten disease-specific networks for rare diseases since 2003. A funding period of 5 years with a total budget of 25 million Euros is planned. The networks will foster the nationwide cooperation of basic and clinical research for these diseases. Their common objectives include the systematic study of disease causes and progression, dissemination of knowledge, improvement of diagnosis and better treatment of patients. Patient associations are tightly integrated in this collaboration. The national funding program forms the basis for an increased international networking in this research field.

## Congenital Bone Marrow Failure Syndromes

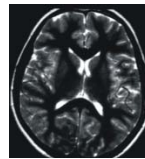
The aim of the network is to improve the diagnosis and treatment of rare failures in blood formation (bone marrow failure syndromes, bmfs). These include hereditary disorders of the white blood cells (neutropenias), the red blood cells (Blackfan-Diamond anemia and dyserythrocytic anemia), the platelets (thrombocytopenias) as well as pancytopenia (Fanconi’s anemia). The varying risk to develop a secondary malignancy (such as a secondary leukemia) or a bone marrow aplasia represents a major problem for patients suffering from these diseases.



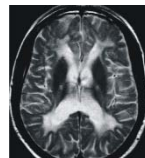
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## Leukodystrophies (Leukonet)

The white substance of the nervous system (myelin) ensures that nerve impulses are passed on at high speed. Some rare congenital diseases cause the disintegration of myelin and the progressive loss of physical and mental abilities. These incurable diseases are called leukodystrophies. The close cooperation between clinical and basic research in Leukonet is essential for the identification of disease genes.



MRI of a normal brain.  
Myelin appears black.



Leukodystrophy: degenerating myelin appears white.

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## Intersexuality

The network focuses on atypical somatosexual differentiation and intersexuality and studies the causes of discrepancies between chromosomal sex and the internal and external genitalia. In a clinical evaluation study previous treatments and experiences made with the medical care are documented and the patients are asked about their mental health as well as their health-related quality of life. It is accompanied by projects evaluating the molecular and clinical basis of gonadal dysgenesis and androgen biosynthesis.



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## Ichthyoses and related Keratinization disorders (NIRK)

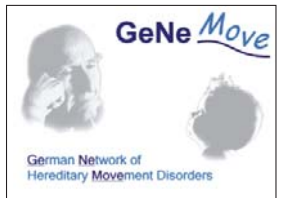
Ichthyoses are a group of rare, generalized genetic keratinization disorders which are associated with marked scaling and often a severe inflammation of the skin. These symptoms result in severe physical and psychological handicaps and have dramatic impacts on the quality of life. One study of NIRK explores the possibility of an externally applicable enzyme substitution therapy for one form of lamellar Ichthyosis to improve treatment of this disease.



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## Hereditary Movement Disorders (GeNeMove)

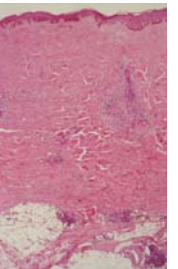
Hereditary Movement Disorders are characterized by progressive ataxia, spasticity, coordination disorders, paralysis or lacking motor control - with up to now symptomatic therapies at best. The cause and progression of the disorders are only partially known. GeNeMove studies the genetic and molecular causes of these neurological diseases, develops scales and disease markers to follow disease progression and initiates treatment studies. Among other things, this work is based on the establishment of a national gene bank.



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## Systemic Scleroderma (DNSS)

Induration and thickening of the skin as a result of increased deposition of connective tissue is characteristic for scleroderma. In systemic scleroderma, the disease spreads to the inner organs (such as the lung and the gastrointestinal tract) where it causes irreversible damage. Drawing on epidemiological data gathered from more than 1,000 patients in 31 hospitals so far, specialist centers for organ involvement and basic research cooperate in DNSS to study the causes of the disease and advance diagnostic and therapeutic procedures.



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