Treatment and Research Centre of Rare Diseases established in Tübingen

On 22nd January 2010, the first German Treatment and Research Centre of Rare Diseases (ZSE) was officially opened in Tübingen. Interdisciplinary teams will from now on be developing new therapies under one roof for around three to four million patients suffering from rare diseases in Germany. The centre in Tübingen will offer optimal patient treatment, coordinate the cooperation between international specialists, and provide competent advice and information. In addition, the centre will also establish a central biomaterial database and a register.

People with rare diseases often suffer two-fold – from the disease itself and from the fact that their disease is barely known. Patients and their relatives often experience a true odyssey; despite consulting many doctors at many different hospitals, they still do not receive the help they are hoping for. Those who are affected talk about false diagnoses and false treatments, helpless physicians and last not least the lack of social acceptance of their allegedly “exotic” disease. In addition, there are few adequate drugs to treat such diseases; the number of sufferers is often too small to motivate pharmaceutical companies to invest in the expensive development of a special drug.

On the other hand, all rare diseases taken together are anything but rare as Dr. Jörg Richstein, Chairman of the Alliance of Chronic Rare Diseases (ACHSE) highlighted at the official opening of the Tübingen-based ZSE. “Taken together, rare diseases can be seen as a common disease,” said Richstein. According to a European definition, rare diseases are of very low prevalence; low prevalence is defined as less than one in 2,000 people having a specific disease. Prof. Dr. Olaf Rieß is director of the Department of Medical Genetics at the University Hospital in Tübingen (UKT) and spokesperson of the new centre. Rieß also provided figures on the number of rare diseases. “On the global level, there are around 8,000 rare diseases. In Germany alone, around four million people suffer from rare diseases.

One in 20 Germans suffers from a rare disease
Rieß also spoke about the deficiencies connected with rare diseases and highlighted the qualitative and quantitative undersupply in Germany associated with delayed access to treatment. All speakers therefore rated the establishment of the ZSE as very positive, including Wolfgang Zöller, Patient Representative of the German Government, and Klaus Tappeser, Head of Department of the Baden-Württemberg Ministry of Science, Research and the Arts. Eva Luise Köhler, wife of the German Federal President and ACHSE patron, opened the ceremony. She called the Tübingen ZSE concept groundbreaking and referred to the Centre as a point of departure, providing comprehensive care, carrying out research and working to obtain greater public awareness.

With its already existing clinical structures, Tübingen has ideal prerequisites for the establishment of a superordinate centre. The UKT is already home to six centres focusing on the treatment and research of rare diseases, all of which are now being integrated into the ZSE. There are plans to integrate further centres, some of which are currently being established. Representatives of the six existing centres presented their goals and activities; they also provided examples that showed the urgent need for action.

The following, previously existing six centres have become part of the new ZSE:
- Centre for Rare Neurological Diseases and Developmental Disorders
- Centre for Rare Eye Diseases
- Interdisciplinary Cystic Fibrosis Centre Stuttgart-Tübingen
- Centre for Rare Skin Diseases
- Centre for Rare Congenital Infectious Diseases
- Centre for Rare Genital Malformations in Women

Translational and transnational research

Despite their considerable differences, rare diseases still have some things in common. They are often very severe and most of them are caused by gene defects. Rare diseases include diseases, which, despite being rare, are well known for their striking symptoms, for example Huntington’s disease. Tübingen is a leader in the development of Huntington’s disease models. Another example is cystic fibrosis, a common hereditary disease whose underlying gene defect was not discovered until 1989. It was possible to increase the average life expectancy of cystic fibrosis sufferers from 18 to 40 years due to the intensive research carried out over the last thirty years. The ZSE’s goal is to carry out focused basic research and clinical studies and increase cystic fibrosis patients’ life expectancy to 60 years of age.
It is envisaged that the combined research activities will help knowledge to be transferred rapidly into clinical application. The translational research competence is one of the new ZSE’s strengths. In addition, existing networks will be expanded across national borders. The expansion of transnational research is required because, in the case of rare diseases, the international cooperation of researchers and physicians is the only way of collecting sufficient patient data to have statistically reliable results. The international exchange of scientists and physicians will also be strengthened in the ZSE’s education and training activities. The ZSE will also work closely with self-help groups, as part of the expansion of the centre’s public relations work, amongst other things.

Benefit goes far beyond individual therapies

Prof. Dr. Herbert Müther, Prorector of the University of Tübingen, pointed out that rare diseases should not be investigated on a case-by-case basis, but that this type of research should be integrated into a common environment. He also pointed out that there are often marginal phenomena that lead to a paradigm change in medicine. He thus linked to the general benefit of the research into rare diseases for the entire field of medicine. Prof. Dr. Michael Bamberg, Managing Medical Director and Chairman of the UKT Board of Directors referred to a political debate when he stated that the establishment of the ZSE also implements a decision of the EU which sets out to improve the recognition and visibility of rare diseases, to support national plans for rare diseases in the EU Member States and to strengthen cooperation and coordination for rare diseases at the European level.

At the end of the official opening ceremony, Prof. Dr. Ingo Autenrieth, Dean of the Medical Faculty, handed over the founding documents and also highlighted the unexpected support from the University and the University Hospital, which will provide 100,000 euros in initial financing. The money will be used to establish ZSE’s structures. Autenrieth emphasised that structured processes are important to implement the ZSE’s plans. He also mentioned that comprehensive quality control measures will be implemented in order to be able to offer best possible diagnostics, therapy and preventive measures.

Further information:

Treatment and Research Centre of Rare Diseases (ZSE Tübingen)
Prof. Dr. Olaf Rieß, Spokesperson
Calwerstr. 7
72076 Tübingen
Tel.: +49 (0)7071 29-85170
E-mail: info(at)zse-tuebingen.de