

## Twofold distinction for the Europe-wide registry on alpha1-antitrypsin deficiency-related liver disease

Aachen, 26th of May 2017 – Alpha1-antitrypsin deficiency (AAT deficiency) is a frequently overlooked metabolic disorder. Apart from lung disease, AAT deficiency leads to liver disease in up to 50% of patients. Although involvement of the liver is the second most common cause of decreased quality of life and life expectancy in Alpha1 patients, no preventative care plan, like that one implemented to avoid lung involvement, has yet been drawn up. To close this gap, the Clinic for Gastroenterology and the Center for rare liver diseases of the University Hospital Aachen (Germany) under the leadership of Dr. Pavel Strnad initiated a multicenter registry study. This study has now been distinguished by two European organizations: the development of this registry is supported financially and politically by the European Reference Networks „RARE-LIVER“ and the „European Association for the Study of the Liver (EASL)“.

The development of liver disease in patients with AAT deficiency is massively underappreciated. Unfortunately, chronic liver disease is often detected late even if AAT deficiency is already known. This alarming shortcoming is facilitated by the fact that the affected patients generally have only unspecific symptoms if any at all. Moreover, routine diagnostic measurements (e.g. liver function tests) often reveal no abnormalities. In the case of a late diagnosis, the diverse complications of liver disease can no longer be effectively prevented. Because of insufficient data, there is no well-established preventative care plan for AAT deficiency-related liver involvement.. Additionally, many questions about the natural history of this disease are still unknown. "This is exactly the main reason for the establishment of our registry." says Prof. Christian Trautwein, director of the Clinic for Gastroenterology. Dr. Strnad, principal investigator of the registry, adds: "There are clear indications that not just patients with severe AAT deficiency but although carriers of the mutated gene have a higher risk for liver disease. Therefore, AAT mutations increases the susceptibility to liver disease in a big population group of 2-4 % of all Europeans."

In March 2017, the European commission has established 24 European Reference Networks for the research and treatment of rare diseases. Those networks have the aim to improve the quality of treatment of patients with rare diseases in Europe. The University Hospital Aachen is involved in two networks. One of them is the RARE-LIVER European Reference Network on Hepatological Diseases, which includes 28 teams of experts on rare liver diseases from 11 countries. Within the network, Aachen is the Europe-wide coordinating center for AAT deficiency.

The Europe-wide study group with its operational headquarters in Aachen is collaborating with various patient support groups and other hospitals specialized in AAT deficiency-related lung disease. "Because only together with our patients and other caregivers we can improve the current situation and achieve sustainable results.", says Dr. Karim Hamesch, clinician scientist from the Clinic for Gastroenterology of the University Hospital Aachen.

Experts from ten European countries are currently working together to collect important clinical data from patients with AAT deficiency. "With the aid of this registry and follow-up examinations, we can better understand this disease and thereby

establish preventative care plans as well as treatment strategies", Dr. Hamesch continues.

The project will now be supported not only by the European commission but although by EASL. "We are really pleased about the extraordinary high amount of contribution", says Prof. Trautwein. "The promoted registry is an outstanding example of valuable scientific and clinical collaboration on an international level. We are highly motivated to improve patient-centered care and push diagnostic and therapeutic progress."

Up to date, about 800 persons from five European countries have been recruited into the registry. "The establishment of the European Reference Network for rare liver diseases has been the first step. The realization of this European registry is the second one. We are excited about the positive feedback", says Dr. Strnad. "But there are still many goals that remain to be accomplished."

Further information about the topic: [www.alpha1-liver.eu](http://www.alpha1-liver.eu)

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#### **Über die Uniklinik RWTH Aachen (AöR)**

Die Uniklinik RWTH Aachen verbindet als Supramaximalversorger patientenorientierte Medizin und Pflege, Lehre sowie Forschung auf internationalem Niveau. Mit 34 Fachkliniken, 25 Instituten und fünf fachübergreifenden Einheiten deckt die Uniklinik das gesamte medizinische Spektrum ab. Hervorragend qualifizierte Teams aus Ärzten, Pflegern und Wissenschaftlern setzen sich kompetent für die Gesundheit der Patienten ein. Die Bündelung von Krankenversorgung, Forschung und Lehre in einem Zentralgebäude bietet beste Voraussetzungen für einen intensiven interdisziplinären Austausch und eine enge klinische und wissenschaftliche Vernetzung. Rund 7.000 Mitarbeiterinnen und Mitarbeiter sorgen für patientenorientierte Medizin und eine Pflege nach anerkannten Qualitätsstandards. Die Uniklinik versorgt mit 1.400 Betten rund 48.000 stationäre und 183.000 ambulante Fälle im Jahr.