Medizinische Universität Graz

Rector's Office
Auenbruggerplatz 2, A-8036 Graz

Thomas Edlinger, BA
Public relations and event management

thomas.edlinger@medunigraz.at
Tel. +43 / 316 / 385-72055
Fax +43 / 316 / 385-72030

Press release
For immediate publication

Rare Disease Day: Genetic diagnostics at Med Uni Graz

Graz, 25 January 2022: Since 2008, Europe and Canada have celebrated Rare Disease Day at the end of February—on 29 February in leap years and on 28 February in all other years. The day was proclaimed by EURORDIS, an alliance of patient organizations, in order to raise awareness of the special challenges in treating and researching rare diseases. Research on these rare and rarest diseases is also conducted at Med Uni Graz, where those affected receive clinical care and a diagnosis. The Diagnostic and Research Institute of Human Genetics assumes an important role in this process.

Anything but everyday life

Inflammation, fracture, wear and tear or even cancer: Most diseases and injuries with which the majority of people will be confronted over the course of their life are well studied and treatable. Yet there are diseases that affect so few people that they remain undiagnosed for a long time and have only a few known treatments. Some maladies affect only a handful of people worldwide. A disease that affects fewer than 5 in 10,000 people is considered to be a rare disease. Correct diagnosis and treatment of these diseases are a great challenge—above all in poorer regions of the world. Due to their rarity, there are usually no large studies, nor do medical professionals have much experience with their treatment: The treating physicians face great challenges.

Diagnostics and research at the limit

There are rare diseases in all medical fields. Rare diseases may be the reason for organ abnormalities and developmental disorders in children, familial eye disease, heart disease, disease of the nervous or skeletal systems or cancers as well as prenatal ultrasound abnormalities during pregnancy. Many rare diseases—around 70%—have a genetic cause, which is why the Diagnostic and Research Institute of Human Genetics plays a particularly important role in diagnosis. "There are several reasons why the diagnosis of rare genetic diseases can be very important for the families affected. The first step is always to understand what exactly the disease is. Then the progression of the disease can be estimated as well as the risks that exist and whether special health screening is necessary. Targeted therapies are already available for a smaller number of rare diseases. The families affected are also concerned about a potential risk for other family members. In the case of severe developmental disorders and organ abnormalities above all, the parents of a child with a rare disease would like to know whether their other children are also at risk. One of our most important responsibilities as medical geneticists is to make a diagnosis using a variety of examination methods followed by the family consultation, where we provide information and indicate treatment options," explains Sarah Verheyen, a specialist at the Diagnostic and Research Institute of Human Genetics. Research on rare diseases is also being pursued at Med Uni Graz. A new publication resulting from a

collaboration with Indian and German researchers discusses a rare form of mucopolysaccharidosis (MSP).

Mucopolysaccharidosis simply explained

Mucopolysaccharidoses are diseases in which the breakdown and storage of sugar molecules are disrupted. "We have now discovered that alterations in the ARSK gene, which previously had not be assigned to any disease, lead to a new subtype of mucopolysaccharidosis, which we have called MPS X. MPS X is a storage disease in which certain building blocks in the body, the glycosaminoglycans, cannot be broken down fully. As a result, they are stored in the cells of different tissues and lead to health problems in several organ systems," says Sarah Verheyen about the rare disease.

Search for patients worldwide

A genetic analysis at Med Uni Graz determined that one possible cause for a skeletal system disease affecting two siblings was a gene with the name ARSK. This gene had previously not been assigned to any human disease. Using a global database, a second family with two children with genetic alterations in the same gene was found in India. The research is based on the exact clinical characterization of this new rare disease and functional studies that confirm the effect of changes in the ARSK gene. Its findings should help to better identify the disease in the future. Another project on MPS aims to determine the consequences of frequently occurring inflammatory reactions in children's bodies.

Inflammation in MPS: Special blood vessel cells should clarify the cause

Inflammatory processes that may favor cardiovascular disease often occur in MPS patients. A new project at the Department of Paediatrics and Adolescent Medicine, supported by the Austria MPS Society, is concerned with the causes of this inflammation and its effects on the blood vessel cells of those affected. For project leader Silvija Tokic, the primary focus is on endothelial cells, which have been largely ignored in prior research on MPS. Endothelial cells are essential to the correct functioning of blood vessels. They form an important barrier between the blood vessel and the adjacent tissue. "Inflammatory processes can greatly affect the endothelial cells and thus disrupt the barrier function, which in turn may lead to a permanently pathological dysfunction of the layer of cells," explains the researcher, describing the problem. Together with her team, she would like to obtain patient-specific blood vessel cells from the blood, characterize the inflammation status of those affected and analyze the reason for the changed barrier function.

Contact and further information:
Sarah Verheyen
Medical University of Graz
Diagnostic and Research Institute of Human Genetics
Tel.: +43 316 385 73816
Email: sarah.verheyen@medunigraz.at

Contact for information regarding the project:
Silvija Tokic, PhD
Medical University Graz
Division of Pediatrics
Tel.: +43 316 385 30752
E-Mail: silvija.tokic@medunigraz.at

Profile: Sarah Verheyen

Sarah Verheyen has worked at the Diagnostic and Research Institute of Human Genetics in Graz since 2014. In a team of 11 other physicians, she cares for patients at the institute's clinic under the direction of Professor Michael Speicher. Genetic counseling and consultations are conducted by the team of physicians in other divisions as well, e.g., Ophthalmology, Oncology, Gynaecology and Cardiology. A competence center for rare genetic diseases in children/of childhood has been established with Barbara Plecko and the Department of Paediatrics and Adolescent Medicine.