Training and information in rare diseases are scarce and the need for increased awareness and improved diagnosis leading to developed patient care is crucial. The Recordati Rare Diseases Foundation was created with the mission to fill the educational gaps and to increase awareness in this area.

Each year the Foundation organises advanced independent, professional education and teaching courses, which are highly specialised in the area of rare diseases. Since the year 2000, the year the former "Orphan Europe Academy" was created, we have trained over 2 000 healthcare professionals world-wide. The events help to create a network of specialists around the world who can make an important contribution to the identification, recognition and management of rare diseases and in finding new treatments in the field.

In Europe, a disease or disorder is defined as rare when it affects less than 1 in 2000 citizens.

The Foundation aims to increase collaboration with patient associations, to contribute in reaching out, not only to healthcare professionals but also to patients and the general public.

With this objective in mind, the Foundation

→ supports the **1st Patient & Expert Meet** in Prague on Rare Rare Disease Day, 29th of February 2016. This meeting is organised by an Australian patient association and a European reference network dedicated to a rare disease called Homocystinuria.

→ continues to create greater education on Rare Disease Day through its 2 day advanced teaching course to healthcare professionals from across the world on Homocystinuria and methylation defects. The objective is increased diagnosis through newborn screening and improved patient care to prevent irreversible complications.

In Europe, people suffering from one of the over 6000 different identified rare diseases is estimated at over 30 million.

Individuals with homocystinurias, methylation defects and folate defects have an enormous need for improved medical awareness, optimisation of the diagnostic process and therapy, and improved networking between healthcare professionals and patients. The Patient & Expert Meet will bring together key stakeholders from across the world to strengthen their interaction. The stakeholders include patients, clinicians and researchers. The main goal of the meet is to enhance the exchange of information and ultimately promote improved health outcomes for patients affected by these disorders.

Questions to Tara Morrison, Director and Chairperson of HCU Network Australia:

How did you become the president of the HCU Network Australia?

When our two sons were diagnosed with Homocystinuria we were left feeling isolated and with many unanswered questions about this devastating disease. Our initial desire was to want to change this experience for others. This was further prompted, when soon after diagnosis, our eldest son saw an advert about childhood cancer. He asked "why does everyone know about what Charlotte had and nobody knows what I have?" His sister is a survivor of childhood cancer. Our desire was consolidated and in 2014 came the inception of HCU Network Australia.

What is homocystinuria?

Homocystinuria is a rare disease caused by an inability to metabolize certain amino acids, which if untreated leads to an abnormal accumulation of homocysteine, and in some cases methionine, and ultimately disorders of multiple organ systems and reduced life expectancy.

What does the disease mean to you daily?

Our sons require life long medical and dietary management to avoid any further complications associated with the disease. As a child this translates to daily medications, an unpalatable supplement drink and adhering to a strict and measured low protein diet. As a parent it means coming to terms with the knowledge that had your child's disease been detected and treated at birth, they would not now carry the burden of disability.

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How do patient associations help in rare diseases?

A patient organisation has a role in connecting and supporting individuals and families affected by disease. It can provide education and information to those faced with an unimaginable diagnosis. Lastly, it can be a platform to raise funds to support and encourage medical expertise.

If you had 1 message to communicate on rare disease day, what would it be?

World Rare Disease Day is an opportunity to raise awareness. For me it is about medical awareness. So we can close the gap between onset of symptoms and a definitive diagnosis. So a treatable disease does not go undiagnosed, or is misdiagnosed, despite the best efforts of the patient, the family and their health care professionals.

Tara Morrison and her family

Questions to Prof. Viktor Kožich, M.D., Ph.D., Professor of Medical Genetics and Head of Department at the Institute of Inherited Metabolic Disorders, Charles University in Prague-First Faculty of Medicine and General University Hospital in Prague:

How do patient associations help the healthcare professional?

There are several ways how healthcare professionals benefit from collaboration with patient associations. The patient associations provide a unique feedback on quality of life, which helps the healthcare professionals in understanding patient needs. The enthusiasm of many members of patient associations virtually infuses energy into the professionals and stimulates the development of better diagnostic procedures and novel therapeutic options for rare diseases. Moreover, the patients associations represent a strong voice in negotiations of healthcare professional with healthcare payers as well as with payers of the social services. Collaboration between patient associations and healthcare professionals is a bi-directional continuous process, which may enrich and benefit both sides.

What does the collaboration with the Recordati Rare Diseases foundation mean to you?

As part of the scientific organising committee of the course on homocystinurias and methylation defects, I enjoyed the opportunity to invite excellent international experts in the field. Such a high-quality meeting will enable disseminating knowledge on homocystinurias and on practical diagnostic and therapeutic solutions to physicians and laboratory specialists from nearly 30 countries world-wide. The course will offer a unique opportunity for the participants to meet colleagues from other countries, to share experiences and to meet renowned experts in the field with whom they may discuss various topics and cases quite freely. Such high level learning experiences and intense networking possibilities are scarce in the rare disease field and it is important that the Recordati Rare Diseases Foundation makes such courses possible.

For more information on:

- → *Rare Disease Day:* *www.rarediseaseday.org*
- → *HCU Network Australia:* *www.hcunetworkaustralia.org.au*
- → *European Network and Registry for Homocystinurias and Methylation Defects (E-HOD):* *www.e-hod.org*
- → *Recordati Rare Diseases Foundation:* *www.rrd-foundation.org*

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