



Den vzácných onemocnění



February 29, 2016 is the Day of rare diseases

Press release

Correct diagnosis can save the life of people suffering from rare diseases

There are thousands of insufficiently known diseases that only a small number of patients suffer from. They often do not receive correct treatment as their physician has never seen their disease and they themselves often do not know they can suffer from such a disease. This fact is also emphasized on February 29 by this year's Day of rare diseases with the slogan: [Join us, we want to be heard](#).

If there is only one patient in a particular district that suffers from the respective disease, it is no surprise that the general practitioner cannot recognize his/her diagnosis or that he/she sends him/her to the incorrect specialist. However, if these patients go to trained specialists, their quality of life can be significantly improved or their life can even be saved. "As the first step to a successful diagnosis, it is important to think about rare diseases", says Prof. Milan Macek, Head Physician of the Institute of Biology and Medical Genetics at the University Hospital Motol, who specialises in rare diseases.

Consulting e-mail helps in complex cases

Also for this reason, it is important that a patient with long-term medical problems that are not successfully treated knows that his/her problem could be caused by a certain rare disease. "The Czech Association of Rare Diseases ("Česká asociace pro vzácná onemocnění" - ČAVO) operates a consulting e-mail help@vzacna-onemocneni.cz that physicians as well as patients can use in complex cases", notes René Břečťan, Vice-Chairman of ČAVO. The helpline is run under the professional auspices of the National Coordination Centre for Rare Diseases in Motol and directs patients to the right physicians.

Rare diseases should be treated at specialised centres

For certain rare diseases, there are already specialised centres: for example, for cystic fibrosis, butterfly skin disease, hereditary metabolic defects, or oncologic diseases. ČAVO seeks to expand the number of such centres so that for every rare diagnosis (a group of 20 to 30 similar diseases), there is a specialised workplace. The existence of such centres is necessary also with regard to cooperation within the European Union: they connect to networks sharing professional experience as well as treatment capacities if necessary.

Stories of people suffering from rare diseases on the website vzacni.cz

Insufficient knowledge of rare diseases is often linked to a lack of understanding of people around the patient, no interest, worries, creating barriers between people. “We would like to see as few such barriers as possible. On the website vzacni.cz, we describe the rare diseases one after another. We show the diseases as they are. We ask people what life with a rare disease means to them, how they handle it, and what helps them. For each disease, we search for additional information in the professional literature as well as links to other sources available on Internet”, says Chairwoman of ČAVO, Anna Arellanesová.

Rare disease

Rare diseases are seen as such diseases occurring in fewer than five cases per 10 thousand. The rarity of a disease is a handicap for patients. It mainly concerns insufficiently known and little investigated diseases. More than 6 thousand diagnoses of rare diseases are described; however, efficient causal treatment is available only for several dozens of them. Determining the correct diagnosis is a big problem: physicians see such diseases only rarely and determining a diagnosis can take years. This, of course, reduces the chances to effectively use available means of treatment.

The Day of rare diseases

The Day of rare diseases takes place every year on the last day of February. The whole event focuses on showing the general public as well as those who participate in making decisions on issues of health and social care what rare diseases are and what they mean for the life of patients.

At the international level, the event is coordinated by EURORDIS. In individual countries, individual associations of patients as well as their national associations take part in it.

The first Day of rare diseases took place in 2008. Since that time, more than a thousand events have been held in 80 countries around the world.

About ČAVO

ČAVO associates organizations of patients suffering from rare diseases as well as individual patients, represents and enforces their interests, and seeks to increase knowledge about

specific issues of rare diseases among specialists in healthcare systems, representatives of government as well as international institutions and the lay public. At the international level, it is an associate member of EURORDIS, the European organization representing 695 organizations of patients from 63 countries and covering approximately 4,000 diseases.

EURORDIS – the European organization for rare diseases

The European Organisation for Rare Diseases is the European organization representing organizations of patients as well as individuals engaged in the area of rare diseases. It was created following an initiative by patients and its mission is to improve the quality of life of all people suffering from rare diseases living in Europe. EURORDIS represents 695 organizations of patients suffering from rare diseases from 63 countries in the world and covers more than 4,000 individual diagnoses. It thus represents the voices of 30 million patients suffering from rare diseases throughout Europe.



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