

Press release

UNITING TO BE HEARD - THE PRORARIS CHALLENGE ***FORMATION OF THE SWISS RARE DISEASE ALLIANCE***

The Swiss Rare Disease Alliance will be launched on the 3rd international rare disease day, February 28, 2010, under the name of ProRaris. This alliance will make it possible to group together the one hundred or so rare disease associations that exist in Switzerland, in order to release patients from their isolation and sensitize the public at large, the political authorities and the health institutions to a phenomenon that appears to concern nearly 500,000 people in Switzerland.

Geneva, February 22, 2010 - Being ill is an ordeal. Not knowing what one is suffering from or how to be cured merely adds suffering to pain. It is the lot shared by all those who are victims of a rare disease and it's the beginning of a long process that will take you from doctor to doctor, from specialist to specialist, from diagnostic error to lack of medical observation. A real diagnostic obstacle course amplified by isolation and lack of information.

The perception error that each of us may make with regard to this global phenomenon is due to the way it has been labelled. Everything that is rare seems exceptional, or even impossible to locate. Yet the scourge we are talking about concerns no less than 30 million people in Europe alone, suffering from one of the 7,000 rare diseases inventoried worldwide. When extrapolated to the scale of Switzerland, it is conceivable that 500,000 people are concerned in our country, that is, 6.5% of the population.

Such figures make it possible to relativize the notion of rareness and to justify the action taken by several heads of organizations concerned in founding **ProRaris**, the Rare Disease Alliance in Switzerland. Because although there are already about a hundred Swiss associations, the representativeness of patients suffering from rare diseases was impaired by the absence of an umbrella organization capable of being a credible and influential lobby with the authorities. *"In Europe, many countries have already taken specific, large-scale measures, such as adopting national plans for combating rare diseases. Little is known about this subject among the general public in Switzerland even today. It was therefore important to bring together all the people concerned in order to improve the visibility and understanding of this problem in our country"*, explained **Loredana D'Amato Sizonenko**, Coordinator of **Orphanet Switzerland** and co-founder of ProRaris.

A symbolic date was therefore chosen for the official announcement of the creation of the **ProRaris** alliance - February 28, 2010, International Rare Disease Day. Unlike most of the European countries, where this type of alliance exists already, Switzerland did not have the benefit of any representative body at the national level.

ProRaris has many objectives. The priority is to become the spokesperson of all the Swiss rare disease associations but also of isolated patients so as to represent them in dealings with the various public agencies, to expand networking among health professionals, to gather the expertise necessary to innovate in public health and to promote medical research. Furthermore, in order to enhance its effectiveness and benefit

from the power of an international network, **ProRaris** will have a close link with the European organization **Eurordis**, which it will join as soon as it is established.

"We are at last going to get the patients out of obscurity, defend their joint interests and make their voices heard at the highest level to release them from their isolation and distress. That is our driving force and the justification for the creation of ProRaris", explained **Bhira Meyer**, Vice-president of the **Association Enfance and Maladies Orphelines** (AEMO), co-founder of the alliance.

What is a rare disease?

A rare disease is defined as an incapacitating or life-threatening disorder that affects less than one person out of 2,000 and requires a special joint effort to treat it. While it is of genetic origin in 80% of the cases, the other causes are infectious, environmental, auto-immune, degenerative or tumoral. These diseases are often chronic, progressive, degenerative and cause disabilities that impair the quality of life. All areas of medical specialization are concerned. The recurrent problems that patients have to cope with are diagnostic delays and errors, lack of information, psychological suffering due to isolation and lack of therapeutic hope, as well as the absence of practical support in daily life.

Charlotte, age 6, suffering from Sanfilippo syndrome

"Incurable!". The word has been uttered and a whole world collapses. Charlotte was four years old when her parents, who live in Geneva, finally learned that their daughter was suffering from an incurable and devastating orphan disease, Sanfilippo syndrome, after spending two years having a string of consultations with different specialists but without obtaining a diagnosis. Charlotte's life expectancy is reduced and that's where the struggle starts. *"One cannot accept the foretold death of one's child"*, testified Frédéric, Charlotte's father, *"it's impossible. When the diagnosis was announced, I was simply advised to get information from the web site about whether a French association existed..."*. For this rare disease, as for many others, research is done only because some parents refuse to accept the foretold death of their children and fight to raise funds and finance researchers. *"Unfortunately, it's only a first stage. Later on, you still have to manage to convince a pharmaceutical company to follow up on the researchers' work"*, explained Frédéric. And it's at this level in particular that the creation of a Swiss alliance will be useful. *"ProRaris will at last give us the help we were waiting for in the French-speaking part of Switzerland to put pressure on the authorities at the highest level, so that this cause is heard more clearly and supported more effectively"*, concluded Frédéric.

Useful links

- Orphanet, the reference portal for information on rare diseases and orphan drugs in Europe: www.orphanet.ch
- European Organisation for Rare Diseases: www.eurordis.org
- Rare Disease Day: www.rarediseaseday.org
- Association Enfance et Maladies Orphelines: www.aemo.ch

Contact persons

Orphanet Switzerland
Service de Médecine Génétique - HUG
Dre Loredana D'Amato Sizonenko
Tél: 022 379 56 96
E-mail: loredana.damatosizonenko@orpha-net.ch

Association Enfance et
Maladies Orphelines (AEMO)
Mme Bhira Meyer
Tél: 024 473 20 10
E-mail: info@aemo.ch