Dear Europe, dear friend,

I am a citizen in a bit of a hurry, reaching at the last minute for my computer to give you my unconditional support for the challenge you have so courageously decided to confront – that of rare diseases.

As a father of a little girl, Léa Rose, afflicted with a rare disease, I was forced to face up to this same challenge, alongside my wonderful wife and my son, then aged three. Once we had recovered from the shock of the diagnosis, we all found ourselves in the same arena. At its centre, there was a thornless rose which had just bloomed (three months) and around her prowled an enormous and as yet undefeated beast – spinal amyotrophy type I. The diagnosis was rapidly followed by the prognosis: a life expectancy of a few months to a year, two years at most.

I have described the backdrop and if it seems dramatic, this is not to lend weight to my words, but rather to allow them to convey the situation of a father of a young sufferer of a rare disease.

I will try to answer as many of the questions as possible and will stop a few minutes before midnight so as not to miss the consultation's deadline.

Question 1: Is the current EU definition of a rare disease satisfactory?

This definition has been well established for very many years and is the most commonly used. Changing a definition or even talking about changing it creates confusion in people's minds. The challenge lies not in the definition but in how to react to the problem. Let's not waste time and energy rephrasing questions, the struggle is already so unequal and resources so limited.

Question 2: Do you agree that there is a pressing need to improve coding and classification in this area?

Giving the disease a precise name has allowed us to understand it more easily. Before fighting an enemy, one has to learn to get to know it. The idea that we could have been faced, as is so often the case with rare diseases, with a "nameless beast" sends shivers down my spine.

The more we manage to codify and classify each of these diseases, the more we can give to patients and all the other people involved the vocabulary they need to share their experiences and so enhance their knowledge of the disease in question.

Question 3: Can a European inventory of rare diseases help your national/regional system to better deal with RD?

The use of the same name for the disease saved us time with regard to measures requiring the involvement of other countries' health systems. Given the magnitude of the task, there is great added value in establishing a European inventory of rare diseases which the Member States could use as a basis for their work. Transnational cooperation at European level and beyond also requires a common vocabulary for the purposes of exchanging information.

Question 4: Should the European Reference Networks privilege the transfer of knowledge? The mobility of patients? Both? How?

The first question one asks oneself when confronted with the diagnosis of a rare disease is whether it would be possible to obtain better treatment elsewhere. This question was particularly pertinent to us, since we had just moved to Luxembourg, a small country which we hardly knew and which is not as internationally famed for its medical research as it is for its banking services.

We therefore looked in our own countries of origin and neighbouring countries to find the best hospital. We even contacted a team of researchers in Hong Kong which seemed to be making progress in the field and was offering clinical trials!

We very quickly came to the realisation that for our little girl the distance between the hospital and the house a few kilometres away would be quite enough – the risk of moving her far outweighed the lure of clinical trials abroad.

In this situation, one rarely knows if one has made the right choice because the disease progresses and there is no second chance under the same circumstances.

We were lucky enough to be assisted by a team of doctors who did not claim to be experts in the field and did not hesitate to contact colleagues outside their own clinic and in other countries (this open-mindedness meant that the help we received included the collaboration of geneticists and doctors from neighbouring countries). They were even open to working with the research centre in Hong Kong, provided that the centre operated within a defined framework and that all the necessary precautionary measures were taken (no recklessness).

To return to your question, in my view, your networks should facilitate the transfer of knowledge. This sharing of knowledge is essential and allows patients to avoid the risks involved in being moved from one hospital to another. Transferring sick people means uprooting them and disrupting the comfort they derive from proximity to their families, friends and local doctors. In extreme cases, this solution may be the only option, but it should not be encouraged without good reason.

Rather than moving the patients, shouldn't we promote the mobility of doctors and their expertise in order to care for patients?

It is sensible to establish reference centres to gather expertise, but this excellence can only spread if this knowledge is not only cultivated but above all shared between the centres and with hospitals, i.e. closer to the patient.

Question 5: Should on-line and electronic tools be implemented in this area?

Once the diagnosis was given, the first reaction was to consult the information available on the internet. The best information is given on American websites, which is what we found first. Orphanet also really helped us to understand the disease.

We naturally devoured the information on the websites on therapeutic research in search of the magic potion. However, we soon had to accept that research in the field would help tomorrow's children but not today's.

The disease progressed so rapidly that we did not really have the time to turn to a patients' association via the internet.

There is so much to learn from other people's experiences, particularly as regards rare diseases. On-line and electronic tools are and will be increasingly vital to effectively combating these diseases. The technology exists and has been tried and tested. Many models for sharing information exist and could be used as a basis for creating even better versions. There is a community of patients suffering from rare diseases with the will and ability to look after themselves. This community consists of extraordinary men and women who are the best ambassadors for the European project, since they know better than anyone the extent to which the future of humanity lies in people's ability to approach each other, to share and to achieve solutions together.

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Given the life expectancy of our daughter, we became accustomed to celebrating her birthday every week. It was at the age of 20 that this great lady passed away. Her big eyes looked up one last time at her mother, who had just played her a lullaby on the violin. A tear rolled down her cheek – not a tear of suffering or sadness but a tear of love for all these shared moments. It snowed that day in Luxembourg.

Two years and two days after Léa passed away, her mother, her big brother, her father and now her little sister are all still filled with the expression in her eyes, her love and her message of hope. We set up the Léa Rose fund (www.learose.info) to support research and provide assistance to patients suffering from rare diseases. Her mother plays concerts and records CDs to raise public awareness and fundraise. This is of course a modest contribution in the face of the challenge of rare diseases but with each passing day we find reasons – not least this public consultation – to remain hopeful with regard to the community of patients suffering from rare diseases. Thank you, dear Europe, and all those who enable you to function. You are WONDERFUL and should know it!

On behalf of Léa Rose and all children of today and tomorrow, we wish you a good night under your sky blue flag sprinkled with stars. Many dreams such as these will gently engulf you. Tomorrow is a big day for all of us, as it is the first day after the consultation, the first day of action. If ever you felt alone in this great challenge on which you have embarked, know that there are many of us, very many of us here to support you and stand up for your values.

Axel van der Mersch,

Father of Léa Rose

Luxembourg, 14 February 2008

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www.learose.info