ELECTIONS TO THE EURORDIS BOARD OF DIRECTORS

GENERAL ASSEMBLY 23 MAY 2012, BRUSSELS

PRESENTATION OF CANDIDATES

AFM- TELETHON, FRANCE, REPRESENTED BY BÉATRICE DE MONTLEAU

My name is Beatrice de Montleau, and I am the mother of 3 children, 2 young adults and a teenager. I discovered the world of rare diseases in 2001, when my youngest child, Axel, was diagnosed with Duchenne Muscular Dystrophy.

Like all of you, at this moment when discovering the disease, my life changed radically. After 36 years of happiness and success, after being strong and self-confident, we became weak and lost. My first family was in fact small.

But after a while, I realised that we are stronger being with other families, being together instead of being alone with the disease.

So, I met AFM-Telethon, the French associations against muscular dystrophies. At this stage, my family became "national".

And since then, I got more and more involved in the area of rare diseases, as a volunteer.

In 2003, I became member of the AFM Financial Committee, as I was working in a bank. In 2006, I was elected as Board member, Secretary General and then Treasurer, which mandate I still have.

In between, I took the responsibility of the International Commission of AFM Board members.

As you know, the aim of AFM is to help families and to cure diseases through proofs of concept. So, the 36 on-going clinical trials and the 500 scientific projects financed every year are dealing not only with muscular dystrophies but also with all rare diseases. Our laboratories have good results using gene therapies, stem cells or pharmacology.

And this is logically why I have been elected to Eurordis Board in 2009, representing AFM in this wonderful organisation. AFM is supporting Eurordis since the beginning, being convinced that rare disease have to be one strong voice in Europe.

After 3 years, I can now say that I know Eurordis quite well and I am so honoured of representing all of you for taking decisions regarding Eurordis strategy and main actions. At this stage, my family became European.

Eurordis, thanks to you, to its team, and to all its professional network of patients representative, so successful in giving power to patient voice and making rare disease recognised through regulations and networking.

Since last year, I also represent Eurordis in a new international consortium, IRDiRC, with the aim of launching 200 new therapies for rare diseases and means to diagnose most rare diseases by the year 2020.When I am sitting in the Executive Committee, I keep in mind all your 30 million pair of eyes, and I try to find the best way to serve you all. My family is now worldwide.

I would hope to go on contributing to Eurordis Board by serving its mission, which is :

- to build a strong community of patients
- to be their voice at European level
- to fight against the impact of rare diseases in their life.

My single wish is to see all of the patients you represent to be cured and happy.
That is why I would be honoured if you elect me for a new mandate. »

Association Française Lesch-Nyhan Action, France, represented by Alain Donnart

I am the grandfather of 8 year old Baptiste, who suffers with Lesch-Nyhan Syndrome (LNS). It is on his behalf that I have worked as Treasurer/Administrator of the French Association Lesch-Nyhan Action (LNA) since 2007. Being closely involved with my grandson's care means I have a personal understanding of the effect on families and an insight into their needs. I have the wonderful experience of being closely involved with LNA where we raise money for research, support families, distribute information, bring together specialists and facilitate the sharing of knowledge. I strive to be a strong advocate for Lesch-Nyhan and would be proud to do the same for the Rare Diseases in Eurordis. I am convinced that my experience will contribute, through a convergence of other Associations, to implement and succeed with vital research projects at a European level.

Croatian Society for Rare Diseases, Croatia, represented by Vlasta Zmazek

My name is Vlasta Zmazek and I'm mother of two sons, Marko & Matija. Matija is 29 and has Epidermolysis bullosa.

I am president of DEBRA Croatia since 1996, from the beginning and at the same time support and „Big mama" of DEBRA Slovenia, DEBRA Bosnia and DEBRA Serbia. For third mandate I am member of DEBRA International Executive Committee.

Today in Croatia we have a dedicated team of specialist for EB, we assure that costs of daily care are covered by Health insurance and Debra Centre is providing 24 hour service for its members. Since 2007 I am active in EURORDIS & Croatian society for Rare Diseases. In 2010 I was elected President of The Croatian Society for Rare Diseases. Since then I am involved in the organizing of National conferences and preparation of National Plan for Rare Diseases in cooperation with the Ministry of Health.

With my experience with Rare Diseases and patients organizations and having faith we can really improve the quality of life for people with Rare Diseases, I believe, I can contribute to the achievement of EURORDIS mission.

Genetic & Rare Disorders Organisation, Ireland, represented by Avril Daly

I have worked for the Patient Irish Charity Fighting Blindness since June 2000 firstly as Head of Public Affairs and now as CEO. In my current role I am responsible for developing the organisations portfolio of research, developing our support and outreach services and raising awareness of genetic

and rare retinal diseases such as RP, LCA, Stargrdt's Disease and Ushers Syndrome among the general public, health care professionals and policy makers.
For four years I have been the Chair of GRDO – The Genetic and Rare Disorders Organisation, which is the Irish national alliance supporting families affected by genetic and rare disorders. I represent GRDO on the national Steering Committee established by the Irish Minister for Health in 2011 working on the development of a framework for a National Plan for Rare Diseases.
I am also a member of both The Medical Research Charities Group (MRCG) and The Irish Platform for Patient Organisations Science and Industry (IPPOSI) who are working closely with GRDO the campaign for the establishment of a National Plan for Rare Diseases in Ireland in the shortest possible time frame. I represent Retina International and Retina Europe on the Board of The European Patients Forum.
My background is in communications and media and I have worked on film and television production and contribute to local and national radio as a commentator. Following graduation I worked with a Publishing company for over three years and was responsible for the editorial content and promotion of a monthly Communications magazine. In that time I also devised and successfully launched a title dedicated to the telecoms sector in Ireland.
As a student I taught drama to children and young adults with special needs and worked as a volunteer with the Irish Wheelchair Association.
I was diagnosed with Retinitis Pigmentosa in 1998 and am registered as Vision Impaired.
I am married and live in Dublin.

SELBSTHILFE ICHTHYOSE E.V.,FERMANY, REPRESENTED BY GESKE WEHR

I was born in Berlin-Wilmersdorf in 1967 and moved to the Rhineland in 1972, where I have since lived in and around the Cologne area. I have the support of a loving husband, and we have three children, two sons and a 28-year old daughter who has blessed us with two grandchildren. My eldest son who was born in 1996 suffers from an autosomal recessive congenital Ichthyosis, and my younger son, born in 1998, has a mild form of atopic eczema.

My eldest son's Ichthyosis is a genetic keratinisation disorder with a prevalence of 1 in a 100 000. Ichthyosis skin does not renew in the same way as healthy skin. Either the production of skin cells is much too fast or the dead cells do not fall off fast enough. In both cases there is something like a traffic jam in the horny layer which leads to very thick and dry skin all over the body.
Fortunately this condition is not life-threatening, but good therapy and ongoing care is very necessary and very time consuming. It's a visible disease with associated social problems which include, lack of self-esteem and a markedly reduced quality of life because people with Ichthyosis often look dirty, the scales on the skin are dark, with raised edges that look like sunburn.

We were very fortunate, because in 1996 after contacting the German Health Authorities we were sent to the German "Children Network" who directed us to the very young patienorganization Selbsthilfe Ichthyose e.V. They gave us the address of the most knowledgeable specialist in Germany who we first met in 1997. At Selbsthilfe Ichthyose it was indescribable to meet people with the same problems as my family and who could answer my many, many questions. I have not missed a meeting since 1999, and since 2000 I co-organize the annual

meetings. Between 2001 and 2006 I was part of the Board of Directors as the minute-taker, and currently I support our members in asserting their claims to the authorities and health insurances, from the first application up to the sometimes necessary lawsuit.

Between 2008 and 2010 I was part of the ACHSE e.V. Board of Directors, the German Alliance for Rare Diseases.
In 2002 my knowledge of the English language led me to the EURORDIS conference, and in 2004 I started the process of founding a European Network for Ichthyosis (eni). We legalized eni in 2010, and I am currently its CEO, on a voluntary basis.
I would very much like to offer my skills and knowledge to support the EURORDIS aims and targets, and I believe I can help this organization move policy to improve the quality of life for all people in the Rare Disease community

UNIAMO, ITALY, REPRESENTED BY SIMONA BELLAGAMBI

Simona is the aunt of a fourteen years old girl with Tuberous Sclerosis. Her involvement in RD issues began at her niece Alice's birth with the wish to help her and her sister to find some answers to their needs. In the following years her commitment has first expanded to the members of the Italian TSC Association and then to RD patients at national and international level with her collaboration with UNIAMO and EURORDIS .

From 1999 to 2009 she served the TS Italian Association as national secretary and representative in the International network. Besides her collaboration in the associative activities, she was in charge of the dedicated help line service and contributed to the set up of the Italian network of Centres of reference for TSC. Firmly believing in a global approach as unique possibility to face the common needs of all RD patients and families she offered her collaboration to the Italian Federation for Rare Disease for which she became the representative in the Council of National Alliances of EURORDIS in 2005. She has been the promoter and coordinator in Italy of international awareness events such as Rare Disease Day and Play Decide sessions within the POLKA project as well as workshop on Centres of Expertise for the Rapsody project. She has been speaker on RD issues at several national and international conferences and was appointed as EURORDIS Advisor in the EUROPLAN project with the supervision of the Spanish, Greek and Italian Conferences.

After years of activity with EURORDIS her conviction on the potentialities and capacity of this great organization in giving voice to the needs and rights of millions of patients and families , increased steadily as well as her desire to contribute further as member of the Board of Directors and most of all as part of the team.

VAINCRE LES MALADIES LYSOSOMALES, FRANCE, REPRESENTED BY ANNE-SOPHIE LAPOINTE

My husband and I have had five children. Two of them have Hunter's disease (MPS2). Vianney died in January 2006 and Théophile has been treated by Enzyme Replacement Therapy (ERT) since in December 2006. We have experienced the advances in research in the field of rare diseases.

I have been in VML (Vaincre les Maladies Lysosomales), an organization with more than 50 rare diseases, since 1998 as member and since 2004 in a family committee which concern family's support. For 4 years I have been elected in the board as vice-president and treasurer and now as president.

I believe in the action of patient's organization to give comfort and means to recover dignity for the patient and his surround. Through our international networks (Mucopolysaccharidoses, Niemann-Pick, Batten disease), I know the importance to work all together for the same interest: the family's care and its quality of life.

Last year I attended a master in ethics of health and now I start a PhD about ethical issues for patient organizations in clinical trials. I participate in an international ethics committee for an European program called Leukotreat (LKT). We work on database and informed consent before clinical trials. There's lot of sense for a patient organization to work there.
