Preface

In 2010, after contemplating the matter for years, I finally established the BLACKSWAN Foundation in Porza, Switzerland (www.blackswanfoundation.ch), with the ambition to support research on orphan and rare diseases. Needless to say, it was not easy, and it still is not. However, I have been witnessing gestures of kindness and support from so many people that, even though I feel that I am at the beginning of what I set forth to accomplish, I feel I have already gone a long way.

The medical community does not see rare diseases as a field for priority action. In fact, poor characterization of the natural causes, pathologies and low numbers of cases make diagnosis difficult, often resulting in a real ordeal for patients and their families, who find themselves on a seemingly endless odyssey from one consultant to another. Rare diseases offer persons affected little hope of survival – and an even lower chance of leading a normal life. These diseases go hand in hand with discrimination, isolation and lack of understanding by others. Paradoxically, administrative and bureaucratic obstacles along the way aggravate the problem, as standards and norms cause these diseases to fall through the safety net. I have dedicated my life to studying rare diseases and have had to focus my attention on the lack of public funding made available for research in this sector, especially in a country like Switzerland where investments in pharmaceutical research and development are considerable.

A problem that the BLACKSWAN Foundation wishes to redress is the idea to support active advanced research into rare diseases by means of a nonprofit structure in

order to fill the gap, where possible, caused by the chronic lack of public and private funding in this area. The goal is to find a solution to this very important public health problem. In fact, there are already 8,000 rare diseases listed in the scientific literature, and 5 new diseases are added each week. In other words, this means that some 6–8% of the world population suffer from rare diseases: 500 million around the world. The BLACKSWAN Foundation channels all donations towards preclinical and clinical research into rare diseases. A committee of recognized international experts is responsible for approving projects for foundation financing.

The BLACKSWAN Foundation organized the first global symposium, the RE(ACT) Congress (www.react-congress.org), dedicated to research on orphan and rare diseases, which took place in February 2012, in Basel, Switzerland. The aim of this symposium was and will be to promote research on rare diseases and also to increase awareness of the field among industry, general public and policymakers. The congress' name, 'RE(ACT)', symbolizes the need to react to present health industry approaches by patients and their families.

I believe that progress on information, diagnosis and care for patients suffering from rare disease relies on all stakeholders. Cures evolve from science; yet, the translation of scientific advances into benefits for patients requires international cooperation beyond the walls of the laboratory. Such initiatives aim to lift rare diseases from obscurity, destroying the veil of ignorance surrounding these, much too often, life-threatening disorders.

Now, my dream is to establish a platform where research scientists meet on an ongoing basis, so that if a patient wishes to verify the state-of-the-art in terms of research development on any given disease, his enquiry need not be more than an e-mail away. It is necessary, however, to convene research scientists from around the globe and motivate all involved in the RE(ACT) Community (www.react-community.org) to move beyond research and lectures and to make a concrete contribution to the further development of their research projects by taking them out of the university laboratories and pushing them into the business arena.

In conclusion, through the creation of the RE(ACT) Community, and thanks to the interaction of its 5 member groups (research scientists, patient organizations,

specific venture capital investors, pharmaceutical industries and regulators), I am convinced that we can change the traditional business approach towards new avenues of therapy by bringing medical solution to rare disease patients and interesting returns to investors.

I would like to thank all researchers working on rare and orphan diseases and especially those who agreed to contribute to and be part of this themed issue of *Molecular Syndromology*. I would also like to thank Michael Schmid, Chief Editor, who had the brilliant idea to focus on rare diseases with a special issue and who chose me as guest editor. Many thanks from the bottom of my heart; I loved this challenge!

Dr. Olivier Menzel, Porza, 2012

E-mail: olivier.menzel@blackswanfoundation.ch