INSIGHT

Dr Stephen Groft
Senior adviser to the director of the National Centre for Advancing Translational Science, National Institute for Health in the US

Turning point for rare disease research

Decades of innovation and collaboration are now coming together to advance research on rare diseases, but we must keep momentum going to ensure rare diseases are recognised as a public health and research priority.

"Today we have greater knowledge, there are more products in the pipeline, a growing number of patient advocacy groups and greater public awareness," confirms Dr Stephen Groft, Senior Adviser to the Director of the National Center for Advancing Translational Science at the National Institute for Health in the US.

International collaboration has undoubtedly played a vital role in bringing expertise together to advance research in rare diseases. Everyone from governments, regulators to bio-pharma companies has been involved. One area in particular that has led to major breakthroughs has been the growth of patient advocacy groups.

To date, one of the major stumbling blocks to advancing research had been the lack of critical mass of research investigators to carry out studies. Social media changed all that, as individuals across the globe with similar diseases have been able to connect.

Perhaps one of the greatest challenges continues to be funding, although, as Groft explains, progress is being made. "Governments have been more willing to invest and the biopharmaceutical companies can see now a viable business model. Crowdsourcing is also growing and projects such as the online RE(ACT) Community should be encouraged, particularly to fund smaller trials."

Amongst other developments, 2015 saw new drugs approved to treat multiple myeloma, melanoma and pancreatic cancer and pulmonary arterial hypertension, alongside additional breakthroughs for cystic fibrosis. There's certainly momentum on rare disease research, however much more international attention is needed to push forward research and increase prevention, diagnosis and treatments for rare disease patients.

Healthcare and treatment for rare diseases is a human rights issue and a global strategy to ensure rare diseases are recognised as a public health and research priority is certainly needed. The scientific community and patient organisations have a central role to play in advising policy-makers on the most urgent points that need public action. In this sense, the BLACKSWAN Foundation, in collaboration with Rare Diseases International (RDI) and Eurordis, is planning the launch of a petition in support of research (blackswanfoundation.ch/en/petition/) during the International Congress on Research of Rare and Orphan Diseases (Barcelona, 9 -12 March). ■

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Patients must be at the centre of research into rare diseases

More than 30 million people across Europe suffer from a rare disease. Whilst scientific developments have led to improvements in diagnostics and treatments, patient care must remain a priority to ensure continued progress

By Kate Sharma

Rare disease is a big issue. At the moment, more than 6,000 rare diseases have been identified impacting an astonishing 30 million people across Europe alone. Yann Le Cam, CEO of EURORDIS, the European Organisation for Rare Diseases, reminds us that it's important to look behind the numbers. "Above everything else we need to recognise that every life has worth," he says.

Diagnostic odyssey

Every patient has his or her own story, but so many are characterised by what is known as the 'diagnostic odyssey.' Countless referrals and inconclusive investigations typify many individuals' struggle to find a diagnosis.

"So often, it's only when a crisis of health occurs that a diagnosis can be made," confirms Le Cam. "There is a lot of pressure on front-line healthcare staff. It's not realistic to expect them to be specialists in everything, which is why we need to define patient-centred healthcare pathways, including centres of expertise."

Pan-European collaboration is proving successful in developing