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EDITORIAL

10 Years of Rare Disease Day

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Rare Disease Day is a global awareness day celebrated on the last day of February each year, which in 2017 reaches its 10th edition. The Day aims to make the general public, stakeholders and decision-makers aware of these 6–8000 diseases, which are mostly inherited, affect 6% of the population and consume a disproportionate part of Health budgets. Advanced gene and cell therapies have provided the first treatments for these diseases, albeit priced at very high levels, and will undoubtedly bring further therapies for many rare diseases. Successful clinical development of potential treatments requires involvement of all stakeholders to deliver effective, safe and affordable treatments in a form that is acceptable to the people affected.

I find it very fitting that my first editorial for *Gene Therapy*, following my recent appointment as Editor-in-Chief, is on the topic of RDD. Rare diseases fall squarely within the remit of gene and cell therapies, and are at the core of my research interests (see http://agctlab.org). (The outgoing editors Joe Glorioso and Nick Lemoine, who have steered *Gene Therapy* for the last two decades and cemented the journal's excellent reputation, formally stepped down last December, but have very kindly provided a smooth transition for the incoming Editor-in-Chief. Joe and Nick will write a farewell editorial to be included in the *Spinraza* special issue next April. Rafael wants to acknowledge their outstanding mentoring and ongoing support, and on behalf of the journal, thank them for their leadership and contributions).

A disease is defined as rare if it affects fewer than 5:10 000 people (in the European Union) or fewer than 200 000 US Americans at any given time. There are 6–8000 rare diseases, of which about 80% are genetic. Taken together, they affect 6% of the world's population (it would be good to collect more data in support of this much-quoted figure) and take a disproportionate share of the health budget, estimated at 20%. Half or more of rare diseases affect children. In some rare diseases, only a handful of individuals are known to be affected, while in other disorders it can be hundreds of thousands. It is not unusual for diagnosis to take 5 years, or in many cases the person remains undiagnosed because the disease may not have been characterised, so-called syndromes without a name. Rare diseases have a tremendous human, social and financial cost.

All these figures are important, but sometimes they fail to convey the message. When I do outreach talks on rare diseases, I typically use two examples to try to illustrate how common rare diseases are, and how many rare diseases there are. Six per cent of people being affected means that in your street, on the train that you use for your daily commute, in your child's school and at the gym where you train, there will be several people affected. At the universities or research organisations where we teach and research, hundreds of people will be affected. If you look around yourself, you may see them, giving an example of endurance and determination in their struggle with daily activities that we take for granted. And often, with a smile on their faces (see *Same but Different* at http://www.samebutdifferentcic.org.uk). My second example is the length of time it may take to read out loud the list of names of the known rare diseases: I estimate it at 12 h.

RDD is a global awareness day coordinated by EURORDIS, a non-governmental alliance of patient organisations and individuals active in the field of rare diseases, dedicated to improving the quality of life of all people living with rare diseases in Europe. RDD is celebrated on the last day of February because in a leap year it falls on a rare day, the 29th of February. The last few years over 80 countries have taken part and the events are showcased at RDD's homepage, http://www.rarediseaseday.org. The main objective is to raise awareness among the general public and decision-makers about rare diseases and their impact on patients' lives. Policy makers, public authorities, industry representatives, researchers, health professionals and anyone who has a genuine interest in rare diseases can be involved.

This year marks the 10th edition of RDD. These 10 years have been intense, exciting and full of milestones, but we are only just beginning to deliver the promise of genetics and stem cell research. Only a handful of medicinal products that mediate or are produced through genetic modification have received clearance. The market approval of *H101* (an oncolytic adenovirus) in China in 2005 was followed in this decade by those of Glybera (adeno-associated virus vector, approved in 2012 but used only once), Strimvelis (retroviral vector-transduced CD34+ hematopoietic stem cells) and Zalmoxis (retroviral vector-transduced allogeneic T-cells) in the European Union, and Eteplirsen and *Spinraza* (antisense oligonucleotides) in the United States. The only product currently approved in both European Union and United States is Imlygic (oncolytic HSV-1). Some of these approvals were controversial, as are their huge price tags. On the research front, the development of human induced pluripotent stem cells and genome editing technology, particularly CRISPR/Cas, have been tremendous advances, quickly progressing towards clinical development. In 2011, the International Rare Diseases Research Consortium was launched, with the ambitious goals of, by 2020, developing diagnostics for most rare diseases and new treatments for 200 of them. Repurposing of existing drugs for application to rare diseases has been quite successful, providing some treatments in record time. Politically, in Europe, since 2008, rare diseases are a priority area for action in public health programmes, and national strategies have been produced.

Most of these developments are very encouraging, but much remains to be done. Learning from painful experience, we must ensure that all stakeholders of relevance, from patients, their families and associations to other funders, researchers, pharmaceutical and ancillary companies, clinical community and political authorities work in a coordinated manner to deliver effective, safe and affordable treatments for rare diseases. There is plenty of work to be done, and much to learn in the process—normal day at the office for us.

AUTHOR BIOGRAPHY

RJY-M is a Reader in advanced therapy and Director of Planning and Resources at the School of Biological Sciences, Royal Holloway, University of London, UK (http://www.royalholloway.ac.uk). Rafael received his BSc and PhD in Biochemistry and Molecular Biology from the Autonomous University of Madrid, Spain. He leads the advanced gene and cell therapy lab (http://AGCTlab.org) at Royal Holloway. Rafael is a member of the Board of the British Society for Gene and Cell Therapy (http://www.bsgct.org) and currently its Treasurer. He believes it is very important to engage with the wider society, and is a trustee of the Genetic Alliance UK (http://www.geneticalliance.org.uk) and organises a yearly outreach event on RDD (http://www.royalholloway.ac.uk/rdd). He is delighted to have been appointed as Editor-in-Chief of *Gene Therapy* and will set out his vision for the journal in an editorial to be published on 20th April, in a special issue devoted to *Spinraza* and more generally to the importance of involving all stakeholders for the successful development and marketing of advanced therapies.

CONFLICT OF INTEREST

The authors declare no conflicts of interest.

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RJ Yáñez-Muñoz
AGCTlab.org, Centre for Biomedical Sciences, School of Biological Sciences, Royal Holloway, University of London, Egham, UK
E-mail: rafael.yanez@royalholloway.ac.uk or ryanez-gt-editor@royalholloway.ac.uk