AN INSIGHT INTO RARE DISEASES

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A rare disease is defined as one that affects no more than 1 in 2000 people,[1] and with over 6000 rare diseases affecting at least 3.5–5.9% of the global population, this works out at a conservative estimate of 18–30 million people in the European Union and 263–446 million people worldwide.[2]

Unmet needs

As each rare disease affects a small number of people who are likely to be geographically separated, scientific knowledge is scarce and fragmented. Furthermore, due to the clinical heterogeneity of these unpreventable, chronic diseases, as well as a lack of effective treatments, there is a vast unmet medical need.[3] Consequently, rare diseases are an emerging global public health priority[2] and it is now widely recognised that international collaboration is vital to improving diagnoses and discovering treatments.[3]

A diagnostic odyssey

Many rare diseases manifest themselves in childhood and often lead to a dependency on care throughout patients' lives; it is imperative to recognise that the significant suffering experienced is not only limited to the patients but also extends to their families. The previously mentioned lack of rare disease knowledge is often further complicated by the presence of relatively common symptoms masking the underlying cause. Patients and their families undergo a 'diagnostic odyssey' – the period from initial symptom presentation to a final diagnosis, usually comprising of the patient enduring multiple referrals and a series of unnecessary, often invasive, investigations.[4]

A 2015 report by Rare Disease UK surveyed 1203 patients who represented over 450 rare diseases. The report found that 45% of all respondents waited over 1 year to receive a diagnosis, with 25% having to wait over 5 years. The report highlights that the average patient with a rare disease consults five doctors, receives three misdiagnoses and waits an average of 4 years for their diagnosis.[5]

Incredible efforts in incredible time

The International Rare Diseases Research Consortium (IRDiRC) was established in 2011 by the European Commission and the US National Institutes of Health to promote and organise international efforts with the aim of accelerating medical breakthroughs for people with rare diseases.[1] As such, 200 new therapies were developed for rare diseases in 6 years, 3 years ahead of schedule, showing an evident eagerness within the international rare diseases research community to collaborate across borders, with their patients in the forefront of their mind. Now, the IRDiRC vision is to: *"Enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention."*[6]

Increasing and sharing our knowledge

Currently, patients and their families are given very little information about their condition and often resort to independent research in an attempt to make informed decisions about their care and treatment. Technological developments enable patients with rare diseases to access patient organisations, thereby providing patients with vital support and information surrounding patient registries and possible clinical research.[5] Many patients are keen to partake in research – not only can clinical trials potentially offer a cure or improve diagnosis, for future generations if not for themselves, but patients can provide researchers and healthcare professionals with valuable insights into living with a rare disease.[5]

With genetic technologies rapidly advancing, the number of rare disease classifications are decreasing by linking disparate phenotypes to unifying genetic diagnoses.[2] These successful advancements, however, require the awareness of rare diseases to be raised to incentivise the compensation of rare disease treatments for the small market size, as well as achieving equitable access to health systems that meet the needs of all those affected by rare diseases around the world.[7]

Now is the time to build new bridges – the general public, policy makers, researchers and healthcare professionals need to raise the bar for rare diseases research worldwide.

Ten take-home messages about rare diseases:

1. A rare disease is defined as a disease that affects no more than 1 in 2000 people.
2. At least 263–446 million people worldwide are affected by a rare disease.
3. Rare diseases are often unpreventable, chronic diseases, with many resulting in early death.
4. Rare diseases are an emerging global public health priority.
5. The average patient with a rare disease consults five doctors, receives three misdiagnoses and waits an average of 4 years for a final diagnosis.
6. Information is critical in empowering patients to make informed decisions about their care and treatment.
7. The collaborative international effort of the IRDiRC has resulted in the development of 200 new therapies for rare diseases in 6 years.
8. Researchers should engage with relevant patient organisations to ensure recent results are relayed to patients in non-specialised language.
9. Rare Disease Day® takes place on the last day of February each year to raise awareness about rare diseases and their impact on patients' lives.
10. More progress is needed – all patients with rare diseases should have fair access to appropriate care and treatment, with no one left behind.

1. European Commission. Rare diseases. 2021. https://ec.europa.eu/info/research-and-innovation/research-area/health-research-and-innovation/rare-diseases_en. Accessed 7 June 2021; **2.** Wakap SN, Lambert DM, Olry A, *et al*. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet* 2020;28(2):165–173; **3.** Publications Office of the European Union. Collaboration. A key to unlock the challenges of rare diseases research. 5 March 2021. https://op.europa.eu/en/publication-detail/-/publication/2ab5235e-7fbe-11eb-9ac9-01aa75ed71a1/language-en/format-PDF/source-193764078. Accessed 7 June 2021; **4.** Black N, Martineau F, Manacorda T. Diagnostic odyssey for rare diseases: exploration of potential indicators. 2015. https://piru.ac.uk/assets/files/Rare%20diseases%20Final%20report.pdf. Accessed 7 June 2021; **5.** Rare Disease UK. The Rare Reality – an insight into the patient and family experience of rare disease. January 2016. https://www.raredisease.org.uk/media/1588/the-rare-reality-an-insight-into-the-patient-and-family-experience-of-rare-disease.pdf. Accessed 7 June 2021; **6.** International Rare Diseases Research Consortium (IRDiRC). Vision & goals. 2021. https://irdirc.org/about-us/vision-goals/. Accessed 7 June 2021; **7.** Rare Disease Day®. What is a rare disease? 2021. https://www.rarediseaseday.org/article/what-is-a-rare-disease. Accessed 7 June 2021