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Seeing Is Believing: Invisibility Exacerbates Inequality for Patients Living with Rare Disease

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AT the 11th European Conference on Rare Diseases (ECRD), held virtually between 27th June and 1st July as an official event of the 2022 French Presidency of the Council of the European Union (EU), experts discussed how invisibility in rare disease acts as a roadblock to reducing inequalities. The session, chaired by Ana Rath, Orphanet, French National Institute for Health and Medical Research (Inserm), France, provided insight into the inequalities, inequities, and injustice that people living with rare disease (PLWRD) face, referencing the United Nations (UN) Resolution on PLWRD 2021[1] and the UN Sustainable Development Goals (SDG).[2] The panel also highlighted how we can act at the local, regional, national, and global levels to bring rare diseases into focus, affect change, and start to bridge the inequality gaps.

SUSTAINABLE DEVELOPMENT GOALS: THE IMPACT OF LIVING WITH RARE DISEASE

Alongside the physical symptoms PLWRD experience, the functional, educational, financial, occupational, and social impacts have been chronically overlooked due to rare disease invisibility.

Rath discussed how lack of data on the functional and social impacts of rare diseases exacerbates the inequities and inequalities that PLWRD experience. This was addressed further by Flaminia Macchia, Rare Diseases International (RDI), who discussed how rare disease impacts all aspects of a person's life. To start, Macchia highlighted the UN Resolution on PLWRD 2021,[1] which focuses on "Recognising the need to promote and protect the human rights of all persons, including the estimated 300 million PLWRD worldwide," and stated that this resolution interlinks several of the UN SDGs.[2]

Elaborating further, Macchia explained how the entire family of PLWRD can experience poverty as a result of reduced occupational opportunities for PLWRD and their caregivers, impacting SDG 1 (no poverty).[2] This links to SDG 5[2] on gender equality, where Macchia reported that testimonies demonstrate that females are disproportionately discriminated against as either the patient or the primary caregiver, frequently having to reduce or stop paid work to support their family member. In terms of SDG 3[2] (good health and wellbeing), unnamed rare diseases often have no available treatment and there is little knowledge around them, which leads to poorer health outcomes as well as social isolation and exclusion. Quality education (SDG 4)[2] is affected as PLWRD often experience difficulties integrating into mainstream education systems due to the lack of knowledge and support, leading to inequalities from an early age. Macchia also reported the impact of rare disease on SDG 8[2] (decent work and economic growth) as PLWRD experience

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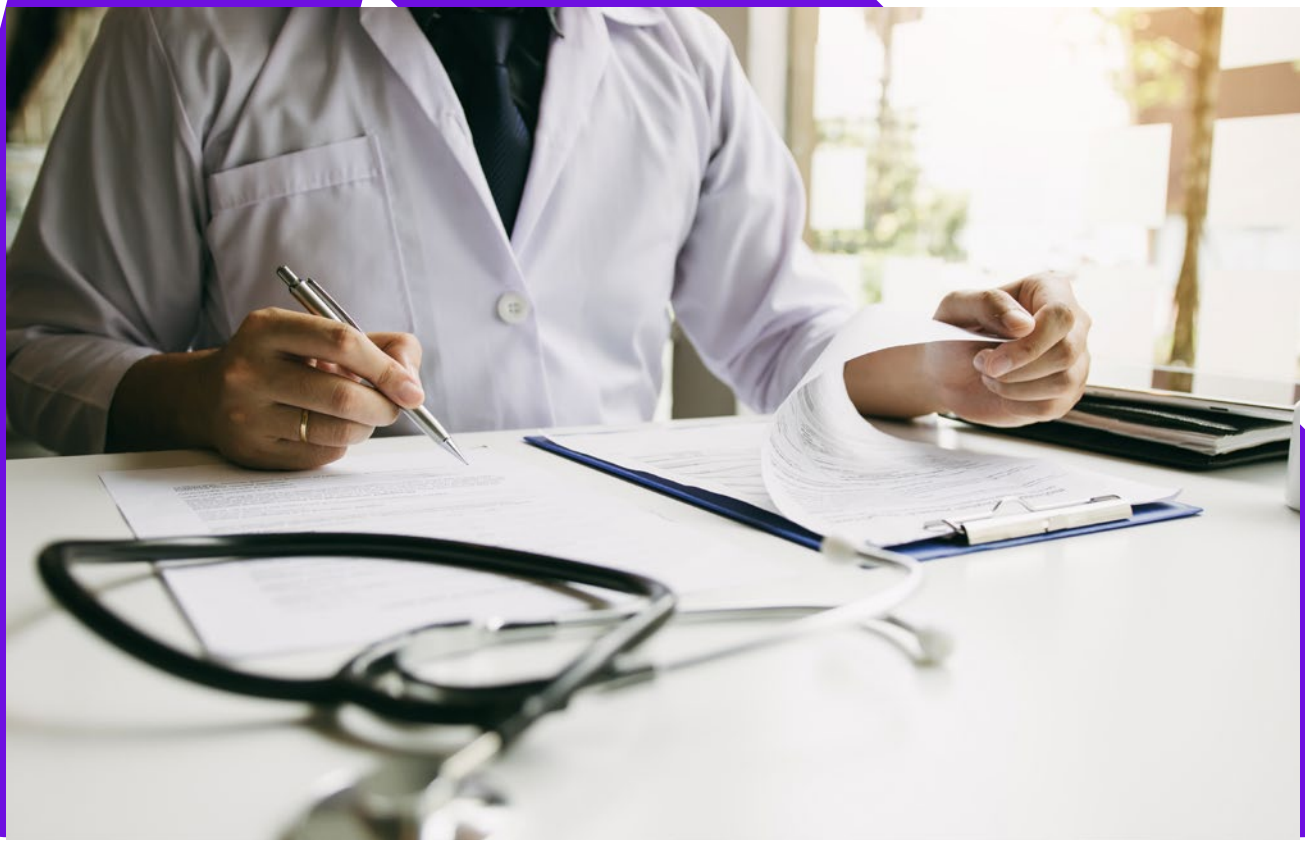
challenges with finding, keeping, and returning to work. All these factors reveal how rare disease affects all aspects of life and is a barrier to achieving SDG 10, reducing inequalities.[2]

Inmaculada Placencia Porrero, Directorate General for Employment, Social Affairs and Inclusion, European Commission (EC), spotlighted the statistics on the inequalities experienced by persons with disabilities, including PLWRD. Linking to SDGs 8,3,4, and 1,[2] respectively, the EC identified that 50% of people with disabilities have concerns about employment, report unmet health needs at a rate four-times higher than people without disabilities, are more likely to leave education earlier and have lower educational attainments, and experience a 28% risk of poverty compared to 18% for people without disabilities. Furthermore, 52% of people with disabilities felt discriminated against.

Elvira Martinez, Advocacy and International Relations, Spanish Federation of Rare Diseases (FEDER), discussed the conclusions from the second ENSERio study[3] evaluating the health and social needs of PLWRD in Spain. They found that families living with rare disease often waited >4 years for diagnosis, with one-fifth waiting for more than a decade to obtain a diagnosis. Of concern, the study showed that just 34% of PLWRD have access to treatment and 40% were not satisfied with the healthcare they receive. Furthermore, financial cover for essential products was not provided for 50% of PLWRD, resulting in financial burden to these families.



WHAT FACTORS PERPETUATE INVISIBILITY?

Because rare diseases are rare by definition, there is a paucity of knowledge surrounding these conditions, which has led to a lack of policy and ultimately an unjust system for PLWRD. A lack of knowledge leads to difficulty in obtaining a diagnosis, acquiring access to appropriate healthcare and social services, political commitment to addressing the inequalities, and incentivising research. Without knowledge and research data, policy makers are unaware of the needs of PLWRD feeding into a self-perpetuating invisibility cycle. Anne-Sophie Lapointe, Rare Diseases Project at The French Ministry of Health and Solidarity, pointedly commented: "We need action to improve the knowledge of rare disease."

Macchia commented: "From a global perspective, it is very clear that invisibility is indeed the first barrier towards reducing inequalities for persons living with the rare disease and their families. The invisibility of the rare disease community is everywhere, in most, if not all healthcare and social systems," and continued that it is "always very easy to ignore the needs of a population that is invisible. No visibility means no understanding of the specific needs to target the support and restore equality."

Androulla Eleftheriou, Thalassaemia International Federation (TIF), a non-government, non-profit, patient organisation, spoke on TIFs vision, work, and progress in unveiling the unmet needs of persons living with thalassaemia. However, Eleftheriou also discussed how there is a lack of funding and research evaluating the epidemiology of rare disease and the needs of those with rare disease diagnoses. Therefore, ongoing research into identification of the needs and inequalities experienced by PLWRD in order to develop enforceable policies to affect change and improve quality of life and outcomes is required.

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Furthermore, many of the studies that encompass rare diseases tend to group them together into a generic umbrella term, rather than indexing them as individual diseases. This was stressed by Juanita Haagsma, Department of Public Health, Erasmus University Medical Centre (Erasmus MC), Rotterdam, the Netherlands, who explained how global burden of disease studies combine data on multiple variables including the functional impact of disease, to generate a burden of disease unit, known as disability-adjusted life years (DALY). DALYs enable comparison between diseases with different characteristics. Whilst this work is pivotal in providing the information required for agenda and priority setting, the main limitation is that rare diseases are often grouped together, rather than indexed individually and thus missing true insight into the functional impact of each individual disease.

TACKLING THE PROBLEM: HOW CAN WE UNMASK THE MASKED?

The UN pertinently sought to address the inequalities and inequities PLWRD face daily, with the UN Resolution PLWRD 2021[1] and SDG 10,[2] which focus on reducing discrimination and working towards achieving the SDGs. One of the key factors that will play a role in achieving this is to make the hidden visible, which will require efforts at local, national, and global levels. The UN Resolution and SDGs provide addressable action points for member states and a start point to begin bridging the inequality gaps.

Globally reliable geographic data are scarce, and Rath stated that "we don't have data because we cannot identify where patients are," further compounding the invisibility of rare diseases and resulting inequalities. Eleftheriou highlighted the need for epidemiological research and commented that whilst there have been highly important and valuable molecular studies into thalassaemia, "cornerstone research areas" such as epidemiology and screening are lacking.

Patient organisations will be key in tackling invisibility across all levels. Eleftheriou discussed the mission of TIF, which is the "promotion of disease-specific national programmes that should be co-ordinated and funded by healthcare systems." TIF has identified that, within Europe and across the globe, PLWRD experience significant inequalities in the extent and quality of services available, which is most stark in lower income countries. In fact, even within Europe, less than 20% of patients are receiving the appropriate care for thalassaemia.

At the local level, Lapointe suggested that "training by patient organisation and patients" can help to raise awareness and increase visibility, and discussed how at national and international levels. European Reference Networks (ERN) "should become an extension of national healthcare systems" for the provision of shared knowledge and resources to improve care across Europe and that international information sharing should include medical and non-medical specialities to address all aspects of disease burden. This point was further highlighted by a direct quote from Rath: "Listen to patients; they are experts."

The importance of robust, well-designed epidemiological research in promoting rare disease awareness and reducing inequalities was highlighted by Eleftheriou, who stated that "there is a great need to improve national registries and upgrade epidemiological work, as well as a great need to actually design new epidemiological work."

Rath introduced the Orphanet disability project, its aim to measure the impact of rare disease on daily life, and explained that information collated from the project could be used by health and social care systems to anticipate challenges and develop strategies and solutions to improve quality of life for PLWRD. Following on from this, Martinez presented the second ENSERio study,[3] which was conducted by FEDER to implement policy and improve outcomes for PLWRD. The data they obtained provided crucial insight into the unmet needs of PLWRD in Spain. Martinez explained that data from these studies can raise awareness and be used as an advocacy lever to encourage decision makers to implement local policy change and that robust data collected at the local level can drive changes at the national level.

The successes of previous research could be utilised, up scaled, and applied at the global level to raise awareness, collaboratively collect data, and affect change by informing policy and developing assessment/needs frameworks.

KEY RECOMMENDATIONS

The key recommendations that came from this discussion included collaborative work between organisations and federations to reinforce the work being performed at regional, national, and global levels, training by patient organisations and patients themselves as rare disease experts, incentivising research, and development of strong policies and assessment frameworks with adequate funding. ●

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