



to the United Nations Special Rapporteur on the Rights of People with Disabilities for the study on “*the right of persons with disabilities to the highest attainable standard of health*” to be presented at the 73rd session of the General Assembly (October 2018).

Word count: 3,684

On behalf of the NGO Committee for Rare Diseasesⁱ, a substantive committee established under the umbrella of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO) to share knowledge about rare diseases (RDs) and raise their visibility on the political stage globally, we welcome the opportunity to provide input from the RD perspective on the right of persons with disabilities to the highest attainable standard of health.

The governance of the NGO Committee for Rare Diseases is led by the current members of the Inception Executive Board: [Ågrenska](#), [EURORDIS-Rare Diseases Europe](#), [International Alliance of Patients' Organizations](#), [International Alliance of Women](#), [International Federation for Spina Bifida and Hydrocephalus](#), [World Federation of Hemophilia](#), the global patient voice being represented through [Rare Diseases International](#) (RDI), the global alliance of people living with a RD of all nationalities across all RDs.

Most people living with a rare disease are living with disability

The WHO's definition of 'disability'¹, which reflects into the International Classification of Functioning and Disability, states that as an umbrella term, it covers 'impairments (problems in body function or structure), activity limitations (difficulties in executing a task or action), and participation restrictions (problems experienced by an individual in involvement in life situations)'.

Disability is a universal human experience in which everyone can be placed in a continuum of functioning and can be vulnerable to experiencing disability over the course of their lives². This rethinking of disability is the key to mainstreaming disability within the public discourse.

RDs are often chronic, highly complex, progressive and **severely disabling**, frequently affecting life expectancy and generating specific care needs³. Therefore, in line with the three WHO core principles of disability it is possible to explicitly highlight how **RDs are highly disabling and thus most people living with RDs are also living with disability**.

ⁱ For further information, please refer to www.ngocommitteerareddiseases.org.

www.ngocommitteerareddiseases.org/members/

www.ngocommitteerareddiseases.org/members/executive-board/ for details of the Committee's members and of our Inception Executive Board formed 11 Nov. 2017.

Persons with RDs represent a **marginalised and invisible, yet statistically significant population** of nearly 300 million individuals worldwide. The confrontation with a RD is a life changing and often devastating event for people living with a RD and their families.

A high percentage of people with a RD are affected by motor, sensorineural or intellectual impairments, which can occur simultaneously: 72% of people living with a RD involved in a European-wide survey on the impact of RDs on daily life carried in 2017 through the RareBarometer programme declared having difficulties with motor or sensorial functioning⁴.

RDs are often invisible and may vary in time due to fluctuation of symptoms. They generate significant daily life and care burdenⁱⁱ : people with RDs find it difficult to carry a routine and face misunderstanding about the disease development. Dealing with one's disease is a time burden. Consequently, participation of people with RDs in the social and professional life can be jeopardised due to absenteeism.

Evidence suggests that people with RDs face:

- Significant limitations in Activities of Daily Living (ADLs): e.g. 77% of people living with a RD experience difficulties in daily activities and tasks;
- Difficulties with the recognition and compensation of their disability, when generated by e.g. invisible impairments, symptoms' variation over time and significant care and time burden⁵.

These limitations in ADLs and the significant care and time burden have serious consequences on the social and employment inclusion of people living with a RD and their carers⁶.

The right to health for people living with a rare disease

Most people living with a RD are living with a disability. The condition can be invisible, it can generate extensive health care needs, as well as a significant time burden to deal with its everyday consequences and, in addition, it can affect work, social life and participation can be restricted⁷.

With the advancement of therapeutic/healthcare progress, people with a RD have a longer life expectancy, higher functioning and greater expectations towards an autonomous and fulfilling life. In 2008, the European Rare Disease Task Force put the spotlight on the significant impact of RDs on patients' life expectancy and disability. It concluded that RDs substantially affected patients' life expectancy and accounted for life-long disabilities in the European population⁸.

RDs and disabilities have cumulative effects in terms of social exclusion⁹. The rights approach is thus key: **people with RDs must not be seen just as patients, but as human beings with rights to health and well-being**. Fully implementing these rights will give people with RDs a chance to fulfill social functions and contribute to common welfare.

ⁱⁱ For specific figures and references, please consult the annex "Factsheet: Most People Living with a Rare Disease are Living with Disability" and Castro R, Senecat J, de Chalendar M, Vajda I, Dan D, Boncz B and EURORDIS Social Policy Advisory Group (2017) "Bridging the gap between health and social care for rare diseases: key issues and innovative solutions", in Posada de la Paz M., Taruscio D., Groft S. (Eds.) Rare Diseases Epidemiology: Update and Overview , Springer International Publishing <http://www.springer.com/gp/book/9783319671420>

A number of European and International binding instruments enshrine the right to access to healthcare and the fight against discrimination:

- **The International Covenant on Economic, Social and Cultural Rights¹⁰ (1996)** enshrines "the right of everyone to the enjoyment of the highest attainable standard of physical and mental health".
- **The UN Convention on the Rights of Persons with Disabilities¹¹ (UNCRPD) (2006)** has given the organisations of persons with disabilities, including those of people living with disabling health conditions such as RDs, a strong legal framework to advocate for their rights.
- **The EU Treaty** enshrines a high level of human health protection in all Union policies and activities and the promotion of employment, improved living and working conditions, proper social protection, and combating of exclusion.
- **The European Charter Fundamental Rights¹²** recalls fundamental rights including respect and protection of human dignity, integrity of the person, non-discrimination, integration of people with disabilities, social security and social assistance, healthcare.
- **The EU Disability Strategy (2010-2020)** does not define disability but makes reference to the approach taken by the UNCRPD to consider disability an 'evolving concept' and supports the implementation of the UNCRPD.
- **The EU chronic / non-communicable diseases strategy** involves an integrated response with efforts to strengthen health systems. In the area of RDs, the EU helps pool scarce and fragmented resources, share expertise and information across borders¹³.
- **The UN Sustainable Development Goals (SDGs)** put inclusion at the core of the 2030 Agenda with its bearing principle 'to leave no one behind' but also to 'reach the furthest behind, first'¹⁴. The SDGs provide a renewed framework to implement the UNCRPD¹⁵.

Regrettably, these dispositions are still far from being put in practice and responding to the situation. A vast majority of people with RDs remain discriminated.

Disability assessments must be made flexible enough to take into account the disabilities faced by persons with RDs, ensuring no one is left behind. In the recent EURORDIS Rare Barometer survey, 34% of people living with a RD who have been submitted to a disability assessment found that the percentage of disability assigned to them to be fairly low or far too low¹⁶. It is therefore important to have frameworks that engage people living with disabling health conditions in the design of health, social and disability policies and services.

Answers to the Questionnaire

As a community of people with severely disabling health conditions and with specific challenges in access to healthcare, we hereby share the experiences and data from our community. We hope these will support the case to bring forward the specific health care access needs of the community of persons with disabilities at large.

Answers to Questions 2 and 3

2. Please provide any information and statistical data (including surveys, censuses, administrative data, literature, reports, and studies) related to the exercise of the right to health of persons with disabilities in general,
3. Please provide information on discrimination against persons with disabilities in the provision of healthcare, health insurance and/or life insurance by public or private service providers.

People with RDs face barriers to high standards of physical and mental health. These barriers still need to be overcome before people with RDs are fully included in society.

- **Accessibility**

- **Access to an accurate and timely diagnosis** is the first hurdle experienced by people with RDs¹⁷. This is a troublesome experience that may take years, often causing deleterious consequences for families and patients. A late diagnosis delays the beginning of adapted treatment and can have severe direct consequences on the state of health of the affected individual, as well indirect consequences on the mental health of those around him. This can be explained by a lack of knowledge about rare conditions among health care and service providers, organizational shortcomings and lack of cooperation on all levels¹⁸.
- **Proper and timely access to treatment, health services and care** is also problematic: the European Union, one of the pioneers of RD policy worldwide, stated that patients suffering from rare conditions should be entitled to the same quality of treatment as other patients¹⁹. However, EURORDIS 2017 Rare Barometer 'Access to Treatment' survey²⁰ revealed that 44% of the respondents reported a worsening of their access to care over the last two years. It also revealed that 47% of the respondents had difficulties in accessing treatment due to the organisation of care (including long waiting time, difficulty to find a specialised doctor, long distance to visit the doctor, understandable information about an individual's own condition and to an accurate timely diagnosis). These figures echo the findings of the EurordisCare3 Survey²¹ of 2009, meaning that little progress has been made in almost a decade.
- **Access to infrastructure:** RDs generate additional social support needs. Due to different constraints in various countries, people with RDs and their families might find themselves without an appropriate and inclusive social support framework or without access to the existing social benefits. Social issues as a whole have been constantly reported as a main concern among people with RDs and families, therefore becoming a concern of patient organisations and decision makers²².

In the 'Impact on daily life' Rare Barometer survey²³, 65% of the respondents had to visit different health, social and local services in a short period of time, 67% felt that these services communicated badly between each other about the person living with a RD, treatments, the consequences of the disease and other relevant information. 7 in 10 found that organising care was time-consuming; 6 in 10 found it hard to manage.

Along the same line, a survey conducted in 2016 by the Swedish National Agency for Rare Diseases (NFSD)²⁴ investigating the experiences of 528 people living with a RD also revealed that each family had to deal with more than 40 contacts from the primary healthcare, social insurance, school, social services and employment institutions creating a considerable burden. Among the respondents, 40% had a coordinating contact person but only 45% of those said that it met their needs. In addition, among those respondents with experience with habilitation, 40% expressed it did not meet their needs. If the situation is precarious in a high-income country like Sweden, well

known for its progressive social policies, we can only expect it to be worse in the rest of the world.

- **Availability**

- New treatments for an immense majority of the RDs known to date are to be invented.
- **Problems of availability and access to existing medicines:** 24% of respondents to the 'Access to Care' survey²⁵ revealed that during the past 12 months they did not get the medical treatment they needed because it was not available in their country (this is 17% higher than the general population) and 15% of the respondents said they did not get the treatment because they could not afford it. This represents a difference of 9 percentage points compared to the general population. 19% were prevented from accessing the treatment they needed because the waiting list was too long.

- **Marginalisation of people with RDs**

- Due to the difficulties in having access to a proper diagnosis, treatment, appropriate care, recognition of their disabilities, access to adequate services and support, people with RDs are invisible and are stigmatised. EurordisCare3 survey (2009)²⁶ revealed that **nearly one in 5 patients experienced rejection by a healthcare professional**, often due to the complexity of the disease.
- **People with RDs still have uncovered needs regarding access to disability benefits:** in the 'impact on daily life' study, 50% stated having uncovered needs regarding the access to disability benefits and 28% stated their disability benefits were not enough to cover their needs²⁷.
- **People with RDs are badly informed about their rights:** 70% do not feel well informed about their social rights or the help they could be entitled to²⁸. Lack of information has been reported referring to the rights related to the consequences of the disease, financial help and relevant social services that can help them.
- Because of a non-supportive and marginalising environment, **people with RDs and their carers have difficulties accessing the labour market or remaining at work:** the same survey²⁹ showed that 58% of carers assisting people living with a RD can be absent from work over 15 days/year, 21% had been absent more than 90 days in the last 12 months and that 70% had to halt or reduce their professional activity due to the disease. **This leads to precarious financial situations** (69% of the participants in the EURORDIS Rare Barometer survey stated a decrease in income) and insurances, be it private or public, do not take this into account.
- **Gender is an extra layer of complexity** as women and girls living with a RD suffer from multiple exclusions. The EurordisCare3 survey found that women face more rejection in looking for a diagnosis³⁰, meaning treatment is delayed. In addition, women are at the forefront of the caring activities (in the 'impact of daily life' survey, 64% of the sample declared the mother organised and coordinated care in the household³¹). In fact, mothers of children with a rare disease have reported high parental stress and high physical and emotional strain³².

5. Please describe to what extent and how are persons with disabilities and their representative organizations involved in the design, planning, implementation and evaluation of health policies, programmes and services.

To put people living with disabling health conditions, including RDs, first and leave no-one behind, policy makers need the guidance, expertise and real-life evidence coming from patient organisations to produce sound policies. For instance, in Europe, for many years, EURORDIS-Rare Diseases Europe, founding member of the NGO Committee for Rare Diseases, has been a prime partner in moving the RD policy agenda forward at EU level. This was encapsulated in '20 Years of Achievements in the Rare Disease Community' (2017)³³.

The existing gaps in access to healthcare are similar to people with RDs and people with disabilities. Civil society organisations for people with RDs across the world have been working closely with all stakeholders for the last two decades, to ensure the involvement of people with RDs in the design, implementation and evaluation of health policies, programmes and services. Some important steps have been achieved and can be uptaken by other organisations.

As they face common hurdles and needs, cooperation between RDs and disability organisations is highly inspirational and mutually rewarding. Sharing best practices and joining forces are key to find answers to their common challenges.

Thus, the experience of EURORDIS-Rare Diseases Europe (Board of the NGO Committee for Rare Diseases) in involving people with RDs can be shared:

- **Advocacy from civil society representing people living with a RD** is based on the **knowledge and real life experience of people living with a RD and their carers**, generated through the [Rare Barometer survey programme](#) bringing their perspective to the forefront during research, therapy development and policy making.
- **Rare Disease Day** is always an opportunity for people living with a RD and their carers to raise awareness about the disease to a large public
- **EURORDIS Open Academy** promotes capacity-building trainings to prepare people living with a RD to engage in research, therapy development, care design and implementation as well as decision making.
- **Connecting people living with a RD with the medical, scientific and policy communities** has been supported through networks such as [RareConnect](#), the [Council of National alliances](#) and the [Council of European Federations](#).
- **Engaging people living with a RD** at all relevant levels to ensure the effective **implementation of new policies and legislation** relevant to patients' needs (research, therapy development, Healthcare pathways), e.g. at the [European Medicines Agency](#).

European reference networks³⁴

In 2017, the European Commission approved the first [European Reference Networks \(ERN\)](#). ERNs allow knowledge sharing and care coordination across the EU to improve access to diagnosis, treatment and provision of high-quality healthcare for people with RDs. They are networks of centres of expertise and healthcare providers. For the first time, ERNs provide clinicians the opportunity to work cross border in Europe in healthcare in order to tackle the RD challenge. They

are a good example of how complex disabling health needs can be addressed through the transfer of specialised expertise.

The involvement of patients in their development was key through advocating for their formation, but also in the design and everyday operations of the networks. This is why EURORDIS established a '[European Patient Advocacy Group](#)' for each ERN disease grouping. In addition, as it is important to empower people with RDs with the necessary knowledge to effectively benefit from the ERNs, the organisation is also implementing a leadership capacity-building programme which will give patient representatives the knowledge and skills they need to be able to effectively participate in ERN activities.

Endnotes

¹ <http://www.who.int/topics/disabilities/en/>

² Cieza et al., Rethinking Disability, BMC Medicine(2018)16:14. Available at: <https://bmcmmedicine.biomedcentral.com/track/pdf/10.1186/s12916-017-1002-6?site=bmcmmedicine.biomedcentral.com>

³ European Commission Expert Group on Rare Diseases, 2016: Recommendations to support the incorporation of Rare Diseases into social policies and services, https://ec.europa.eu/health/sites/health/files/rare_diseases/docs/recommendations_socialservices_policies_en.pdf

⁴ '[Juggling care and daily life: The balancing act of the rare disease community](#)'. 2017 - European-wide survey with over 3000 respondents, conducted via [Rare Barometer Voices](#) in the scope of the EU-funded [INNOVCare](#) project.

⁵ '[Juggling care and daily life: The balancing act of the rare disease community](#)'. 2017 - European-wide survey with over 3000 respondents, conducted via [Rare Barometer Voices](#) in the scope of the EU-funded [INNOVCare](#) project.

⁶ European Commission Expert Group on Rare Diseases, 2016: Recommendations to support the incorporation of Rare Diseases into social policies and services, https://ec.europa.eu/health/sites/health/files/rare_diseases/docs/recommendations_socialservices_policies_en.pdf

⁷ Castro R. et al. (2017) Bridging the Gap between Health and Social Care for Rare Diseases: Key Issues and Innovative Solutions. In: Posada de la Paz M., Taruscio D., Groft S. (Eds.) Rare Diseases Epidemiology: Update and Overview. Advances in Experimental Medicine and Biology, vol 1031. Springer, Cham <https://www.ncbi.nlm.nih.gov/pubmed/29214594>

⁸ Rare Diseases Task Force (2008) Health indicators for rare diseases: state of the art and future directions. http://www.eucerd.eu/?post_type=document&p=1207 . Accessed 8 July 2016

⁹ Castro R. et al. (2017) Bridging the Gap between Health and Social Care for Rare Diseases: Key Issues and Innovative Solutions. In: Posada de la Paz M., Taruscio D., Groft S. (Eds.) Rare Diseases Epidemiology: Update and Overview. Advances in Experimental Medicine and Biology, vol 1031. Springer, Cham <https://www.ncbi.nlm.nih.gov/pubmed/29214594>

¹⁰ <http://www.ohchr.org/Documents/ProfessionalInterest/cescr.pdf>

¹¹ <https://www.un.org/development/desa/disabilities/convention-on-the-rights-of-persons-with-disabilities/convention-on-the-rights-of-persons-with-disabilities-2.html>

¹² <http://eur-lex.europa.eu/legal-content/EN/TXT/HTML/?uri=CELEX:12012P/TXT&from=EN>

¹³ https://ec.europa.eu/health/rare_diseases/policy_en

¹⁴ A/RES/70/UN General Assembly p3/35

¹⁵ EDF 'European Human Rights Report Issue 2 – 2018 – The 2030 Agenda and the Sustainable Development Goals: A European Perspective to respect, protect and fulfil the UN CRPD'. Available at: http://www.edf-feph.org/sites/default/files/edf_-_sdgs_human_rights_report_final_accessible.pdf

¹⁶ '[Juggling care and daily life: The balancing act of the rare disease community](#)'. 2017 - European-wide survey with over 3000 respondents, conducted via [Rare Barometer Voices](#) in the scope of the EU-funded [INNOVCare](#) project.

¹⁷ "The Voice of 12,000 Patients: Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe", EURORDIS-Rare Diseases Europe, 2009, 324 p. www.eurordis.org/publication/voice-12000-patients

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- ¹⁸ 'Participation in society for people with a rare diagnosis', G. Jaeger, AC. Røjvik, B. Berglund, Disability and Health Journal 8 (2015) 44-50, <https://www.ncbi.nlm.nih.gov/pubmed/25164983>
- ¹⁹ European Union Orphan Drug regulation (1999) https://ec.europa.eu/health/sites/health/files/files/eudralex/vol-1/reg_2000_141_cons-2009-07/reg_2000_141_cons-2009-07_en.pdf
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- ²⁴ Nationella Funktionerna Sällsynta Diagnoser (NFSD), Riksförbundet Sällsynta diagnoser (RFSd) and Neuroförbundet (NeuroF) (2016) "Sammanställning av resultat från enkätundersökning om samhällets stöd rörande personer som lever med sällsynta diagnoser och deras närstående - Nationell nivå", more information: <https://www.nfsd.se>
- ²⁵ Access to treatment: Unequal care for European rare disease patients, EURORDIS - Rare Barometer survey, 2017, 3 p. https://www.eurordis.org/sites/default/files/2017_02_17_Access%20to%20treatment_Analysis_Final.pdf
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- ²⁹ 'Juggling care and daily life: The balancing act of the rare disease community". 2017 - European-wide survey with over 3000 respondents, conducted via Rare Barometer Voices in the scope of the EU-funded INNOVCare project.
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- ³⁴ https://www.eurordis.org/content/about-european-reference-networks#About_ERNs