Rare2030

Foresight in Rare Disease Policy

A knowledge-base summary:

PATIENT PARTNERSHIPS FOR RARE DISEASES

https://www.rare2030.eu/our-work

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1. INTRODUCTION TO THE TOPIC

It is now well-recognised that people living with rare diseases (RD) and their families are experts on the diseases that affect them and have a valuable contribution to make to shaping meaningful RD research, policies and services. 'Patient partnership' can be defined as a mutual relationship between researchers, policy makers and health and social care providers (on the one hand) and people and families affected by rare diseases, which improves the quality of service and benefits both sets of stakeholders. It implies a relationship of trust and the active engagement of people or family members living with the disease in shaping decisions at the direct care, organizational and system level. Patient partnership implies patients as equal partners and encompasses patient empowerment and engagement. Patient engagement is "a process through which individuals and communities are able to express their needs, present their concerns, devise strategies for involvement in decision-making, and take political, social, and cultural action to meet those needs" (EPF 2015).

The term patient empowerment is often used interchangeably with others such as patient involvement and patient-centred care. The same 2015 EPF position paper uses the following definition of patient empowerment:

> Empowerment is "a multidimensional process that helps people gain control over their own lives and increases their capacity to act on issues that they themselves define as important." Collective empowerment is "a process through which individuals and communities are able to express their needs, present their concerns, devise strategies for involvement in decision-making, and take political, social, and cultural action to meet those needs."

Advocacy by patient organisations is recognised as an important element in defining policies on rare diseases. The need to fully engage and empower patients in all issues relating to rare diseases is emphasized in the 2009 Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02. Paragraph (20) states that "*The WHO defined empowerment of patients as a 'prerequisite for health' and encouraged a 'proactive partnership and patient self-care strategy to improve health outcomes and quality of life among the chronically ill'. In this sense, the role of independent patient groups is crucial both in terms of direct support to individuals living with the disease and in terms of the collective work they carry out to improve conditions for the community of rare disease patients as a whole and for the next generations.*" It proceeded to recommend (paragraph 21) that "*Member States should aim to involve patients and patients' representatives in the policy process and seek to promote the activities of patient groups*".

Section VI of the document is entitled EMPOWERMENT OF PATIENT ORGANISATIONS, and Member States are explicitly asked to

- Consult patients and patients' representatives on the policies in the field of rare diseases and facilitate patient access to updated information on rare diseases.
- Promote the activities performed by patient organisations, such as awareness-raising, capacity-building and training, exchange of information and best practices, networking and outreach to very isolated patients.

At the EU level, a remarkable example of the adoption of the latter principle is the 2015 Addendum to EUCERD Recommendations on European Reference Networks (ERNs) for Rare Diseases (31 January 2013) whereby "Patients and patient representatives should play an integral role in the decision and opinion making process in RD ERNs and be involved in structural and clinical network activities. It is recommended that RD ERNs demonstrate meaningful patient involvement, patient-centredness and empowerment through recognition of the role of patients, as experts by experience and co-producers of knowledge, in RD ERN structural and clinical activities and therefore demonstrate meeting the legal requirements in the Delegated Acts".

2. PATIENT ADVOCACY, ORGANISATIONS, AND SUPPORT GROUPS

Rare disease patient organisations and support groups initially emerged in the United States in the 1980s, to support patients and their families. Since these beginnings, disease-specific patient support groups have multiplied, as have umbrella organisations for rare diseases in a number of countries across the world and international umbrella organisations linking them all together.

With this increased number of patient advocates and their matured organisation in the form of support groups, their role as advocates has also expanded to include active roles in research, research planning and development of new therapies.

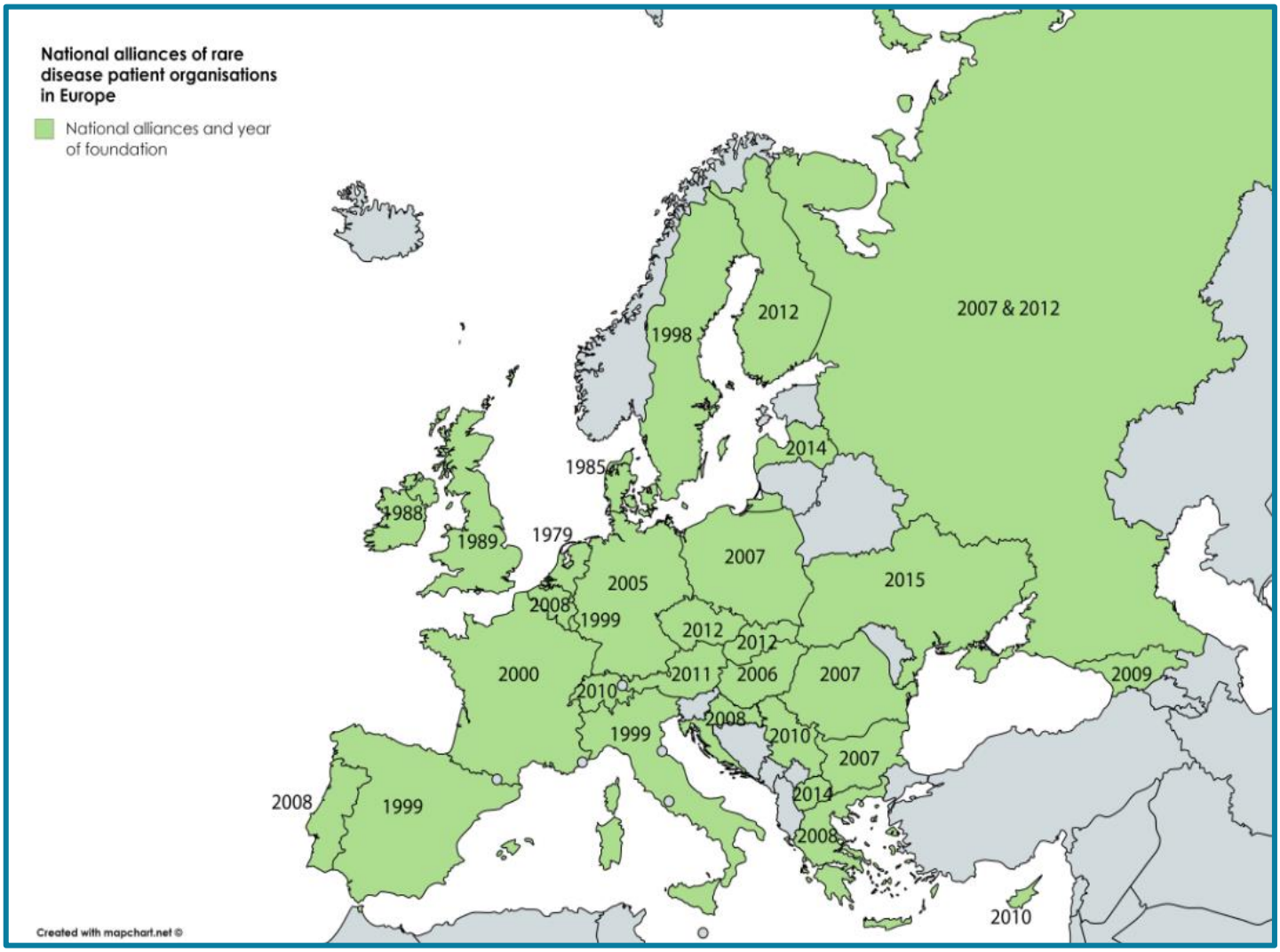
Patient participation or engagement is increasingly viewed as an innovative and viable approach to ensuring appropriate care in the current environment constrained by limited resources. Shared decision-making is the process by which a clinician and a patient jointly make a health decision after discussing options, potential benefits and harms, and considering the patient's values and preferences. The key is the recognition that in every medical or health decision, there are at least two sources of expertise: the clinician has had lengthy and extensive medical training; but only the patient knows his/her attitude to risk, how the illness is experienced in his/her particular social circumstances, and his/her values and preferences.

2.1. National Alliances of patient organisations

National alliances of rare diseases patient organisations play a key role in advocacy and governance: they provide patients with a common 'home' (essential for very rare conditions, which may not otherwise be able to set-up dedicated patient organisations) and enable stakeholders to impact national policies by speaking with a single cohesive 'voice'. Many national alliances have played (or are playing) key roles in elaborating the national plans or strategies for rare diseases under development or already in place.

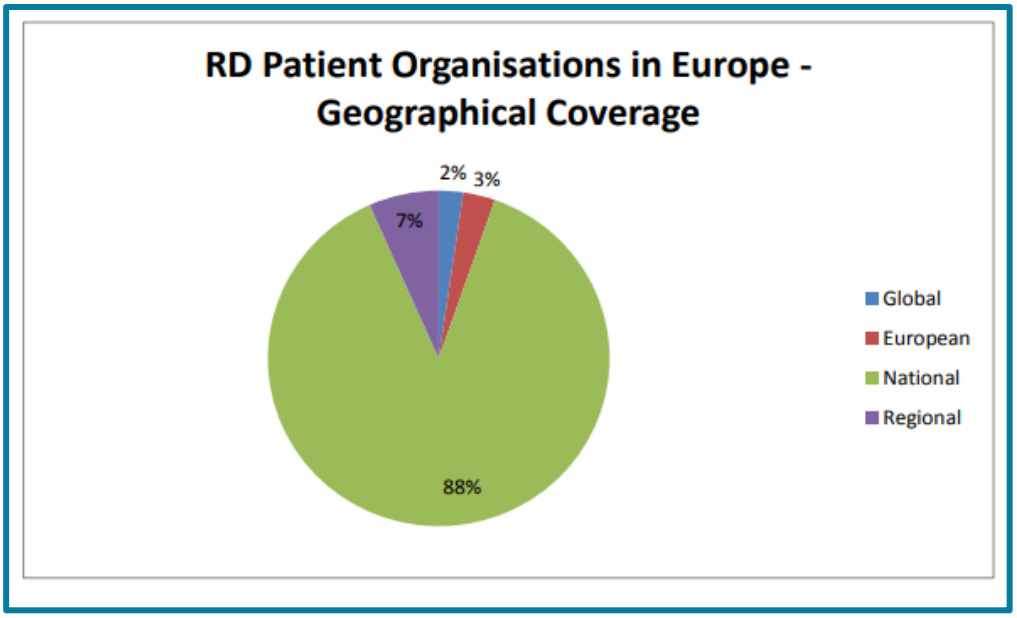
Europe has many national alliances for rare diseases: 29 of these are members of the European Network of National Alliances for Rare Diseases. This network is governed by a EURORDIS-established Council of National Alliances for rare disease patient organisations, which unites the majority of Alliances in Europe with Alliances in the USA and Canada. The Council allows national representatives of rare diseases to work together on common European and international actions, for instance Rare Disease Day.

The map below shows the founding dates of individual National Alliances in Europe:



The table below shows patterns in terms of number of disease-specific patient organisations (POs) registered in the Orphanet database between 2011 and the present day (POs were able to select more than one option)

	No. of disease-specific POs registered in Orphanet in 2011	No. of disease-specific POs registered in Orphanet in 2018	No, of disease-specific POs registered in Orphanet in 2019
National level	1885	2078	2078
Regional level	122	181	181
European level	70	83	83
Global level	45	63	63
Total No. of distinct POs registered	2376	2405	2405



2.2. Patient Advocates Supporting Rare Diseases as a Global Health Priority

Following the success of rare disease patient movements in the United States, Europe, Japan, Canada and Australia, EURORDIS initiated **Rare Diseases International**, which today stands along as the global alliance of people living with a rare disease of all nationalities across all rare diseases.

RDI's mission is to be a strong common voice on behalf of rare disease patients around the world, to advocate for rare diseases as an international public health priority and to represent its members and enhance their capacities. RDI brings together national and regional rare disease patient alliances from around the world as well as international rare disease-specific federations to create the global alliance of rare disease patients and families. RDI has more than 50 member organisations from over 30 countries, which in turn represent rare disease patient groups in more than 100 countries worldwide.

Following this international expansion, **the NGO Committee for Rare Diseases** was initiated by the Ågrenska Foundation and EURORDIS, with a view to bringing greater political recognition of the challenges of rare diseases at the global level. Its formation was approved by a vote of 27 CoNGO (Conference of NGOs in Consultative Relationship with the United Nations) member organisations in April 2014, and its inception meeting as a Substantive Committee within CoNGO took place in October 2015 in New York.

The NGO Committee for Rare Diseases aims to promote multi-stakeholder collaboration and actions for rare diseases within the United Nations system. It is established under the umbrella of the CoNGO and acts as a forum of interested parties such as NGOs from the field of rare diseases and beyond; United Nations bodies and agencies; as well as individual experts. The NGO Committee for Rare Diseases is a multi-stakeholder, inclusive, global ecosystem focused on rare diseases, which aims:

- To increase visibility of rare diseases at the global level

- To extend and share knowledge about rare diseases and their unmet needs
- To connect NGOs interested in rare diseases and their partners within a global platform
- To promote international, multi-stakeholder collaboration and actions for rare diseases
- To align rare diseases as a global priority in public health, research and medical and social care policies

3. BUILDING CAPACITY FOR PATIENT ENGAGEMENT - TRAINING AND DEVELOPMENT INITIATIVES

Improving health literacy and education not only empowers patients but also contributes to the sustainability of healthcare systems. Health literacy is a dynamic, interactive process that encompasses capacity-building and aims to influence individual lifestyle decisions, but also raises awareness of the determinants of health, and encourages actions which may lead to a modification of these determinants. Education and training can be for all stakeholders: patients, health professionals and institutions. It allows to promote innovative and high-quality, truly patient-centred, sustainable health systems of the future. Patient organisations often fulfil the task of ensuring education for patients and healthcare professionals through helplines, information and ad hoc trainings. Due to the lack of knowledge about most rare diseases, patients are often experts on their diseases and have a valuable contribution in shaping meaningful rare disease research, policies and services.

By providing training, patient advocacy groups empower patients and ensure they have the confidence and knowledge needed to bring their expertise to discussions on **leadership, digital health, health care, research and medicines development** with policy makers, industry and scientists.

Examples of such trainings at the European and International level include:

1. EURORDIS - Rare Diseases Europe Open Academy
2. European Patients Academy (EUPATI)
3. Patient Centred Outcomes Research Institute (PCORI) Training for Rare Disease Patient Advocates
4. Numerous patient trainings by national or disease-specific patient organisations

EURORDIS identifies and supports rare disease patient representatives for participation in:

- Patients' representatives involved in EMA scientific committees and working parties
- Protocol assistance
- Scientific Advisory Groups (SAG) at the Committee for Human Medicinal Products
- Other meetings such as discussions on guidelines and risk management programmes

EURORDIS also provides the link between its trained alumni and research, regulatory and healthcare provision by:

- nominating patient representatives to the European Medicines Agency (EMA), where trained patients actively engage in scientific committees and working parties, protocol assistance, Scientific Advisory Groups (SAG) at the Committee for Human Medicinal Products, other meetings such as discussions on guidelines and risk management programmes
- creating the European Patient Advocacy Groups (ePAGs) in every European Reference Network to promote a patient-centric approach in both delivery of clinical care, service improvement and strategic development and decision-making
- representing patient needs alongside 13 international organisations on the International Rare Disease Research Consortium (IRDiRC) Patient Advocates Constituent Committee (PACC)

With the growing recognition that patients can and should be more involved in the medicines development process, a multi-stakeholder effort to develop a framework for structured, effective, meaningful and ethical patient engagement supporting the integration of patient perspectives into drug development is underway via the landmark PARADIGM IMI project. To build capacity for patient engagement it is necessary to increase the capabilities at the organisational level across the relevant stakeholder groups (health care industry, patient organisations, regulatory and HTA bodies, payers). PARADIGM proposes – via its Recommendations on the required capabilities for patient engagement- that the capability requirements for all stakeholders to implement patient engagement in the context of medicines research and development should be defined as:

> *"The competencies (understood as knowledge, skills and behaviours) and resources that each stakeholder type should aspire to have in place in order to be able to undertake the planning, implementation and reflection of effective, ethical and sustainable patient engagement activities across the medicines lifecycle".*

In addition to the capability framework, PARADIGM will develop a set of tools to help operationalize patient engagement including a specific code of conduct on patient engagement, a monitoring and evaluation framework to measure the 'return on the engagement' and show the value of PE activities and guidance on relevant topics (e.g. management of competing interests, community advisory boards (CABs), etc.). These tools build on those developed by existing and recognised initiatives in the field of patient engagement in medicines development:

- Patient-Focused Medicines Development
- EUPATI
- National Health Council Fair Market Value calculator
- WECAN Reasonable agreements between patient advocates and pharmaceutical companies
- The Patient Preferences in Benefit-Risk Assessments during the Drug Life Cycle (PREFER) project

3.1. Role of rare disease patients in data collection

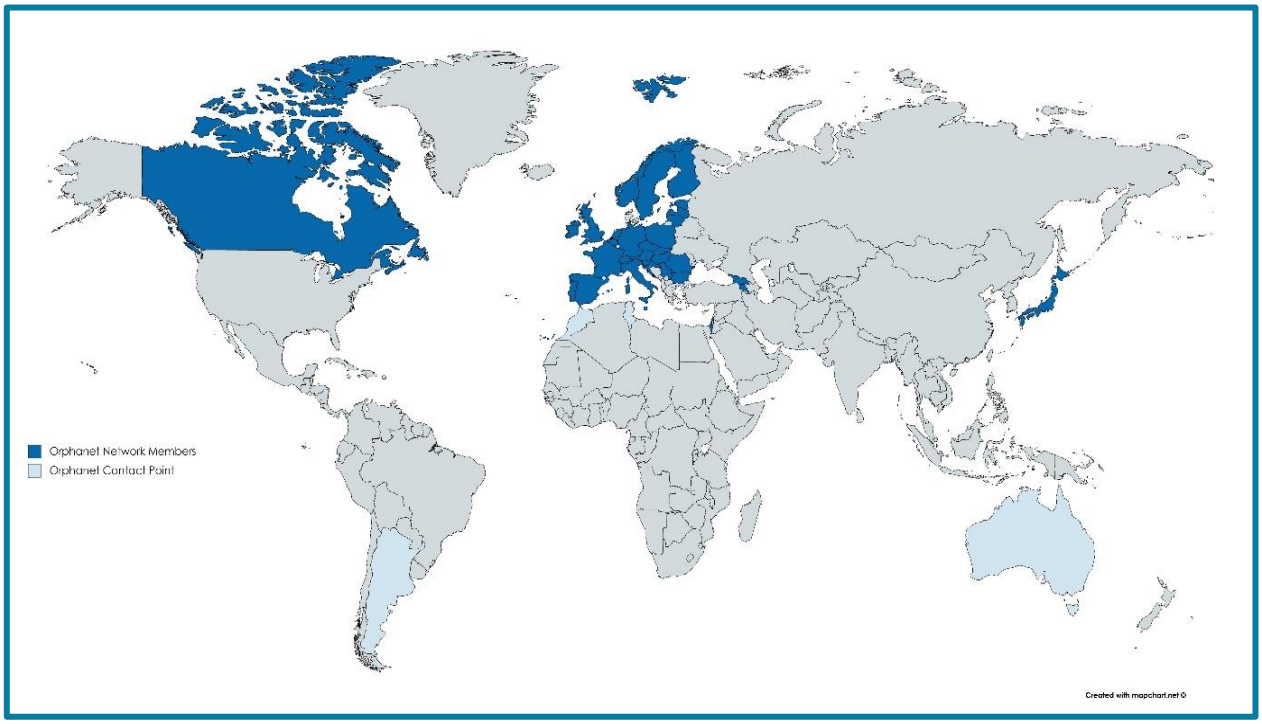
Historically, the involvement of patients in public health research consisted solely of subject-based participation. By encouraging patient input in the development, design and distribution of the surveys and treating them as *de facto* experts on their respective diseases and actors of their research, patient advocacy organisations like EURORDIS have successfully achieved a paradigm shift in the role of patients in the generation of quantitative data on their own health, as well as on the provision of healthcare services through programs and activities such as:

- Eurobarometer
- Rare Barometer Voices
- Patient-led/initiated registries
- The inclusion of Patient Centred Outcome/Experience Measures in data collection initiatives

4. INFORMATION SERVICES AND RESOURCES ON RARE DISEASES

Orphanet (www.orpha.net), the rare disease and orphan drug database, has been delivering information to, and for, patients since 1997. Orphanet aims to provide expert-reviewed and accessible free information on rare diseases to all audiences, including patients. Indeed, around a quarter of Orphanet's users are patients and their families. The multilingual approach of Orphanet means that information is available now in 8 languages; information is only accessible to patients when it is in their national language, so efforts are being made when the budget exists to include more languages. Orphanet produces the Orphanet nomenclature of rare diseases, an essential resource to improve the visibility of rare disease patients in health information systems.

The wide range of information available via the Orphanet website can help empower patients, in many ways. They are able to learn more about a disease through the quality texts on their disease (including texts geared to patients, and emergency/clinical practice guidelines and handicap factsheets). They can find expert resources (centres of expertise and European Reference Networks, for example) in their country to orientate them, and they can find support through the directory of patient organisations for specific rare diseases in 35 countries across the world, as well as alliances and federations of patient organisations and helplines in each country. Orphanet thus also provides increased visibility to patient organisations. Orphanet also provides cross-linking to the Genetic and Rare Disease Information Center (USA's NIH) website, which provides information dedicated to patients, so as to provide additional resources. In order to incorporate the patient perspective in the determination of Orphanet's strategy, a patient representative is part of Orphanet's International Advisory Board.

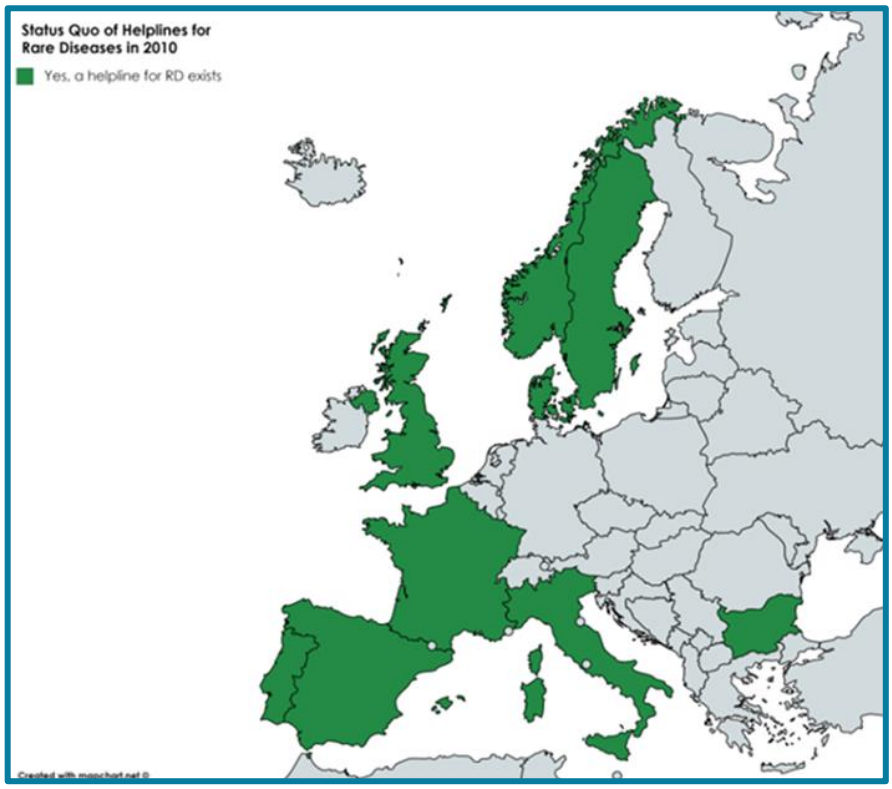


Map courtesy of Orphanet

An important information resource, in many countries, are **helplines dedicated to rare diseases**.

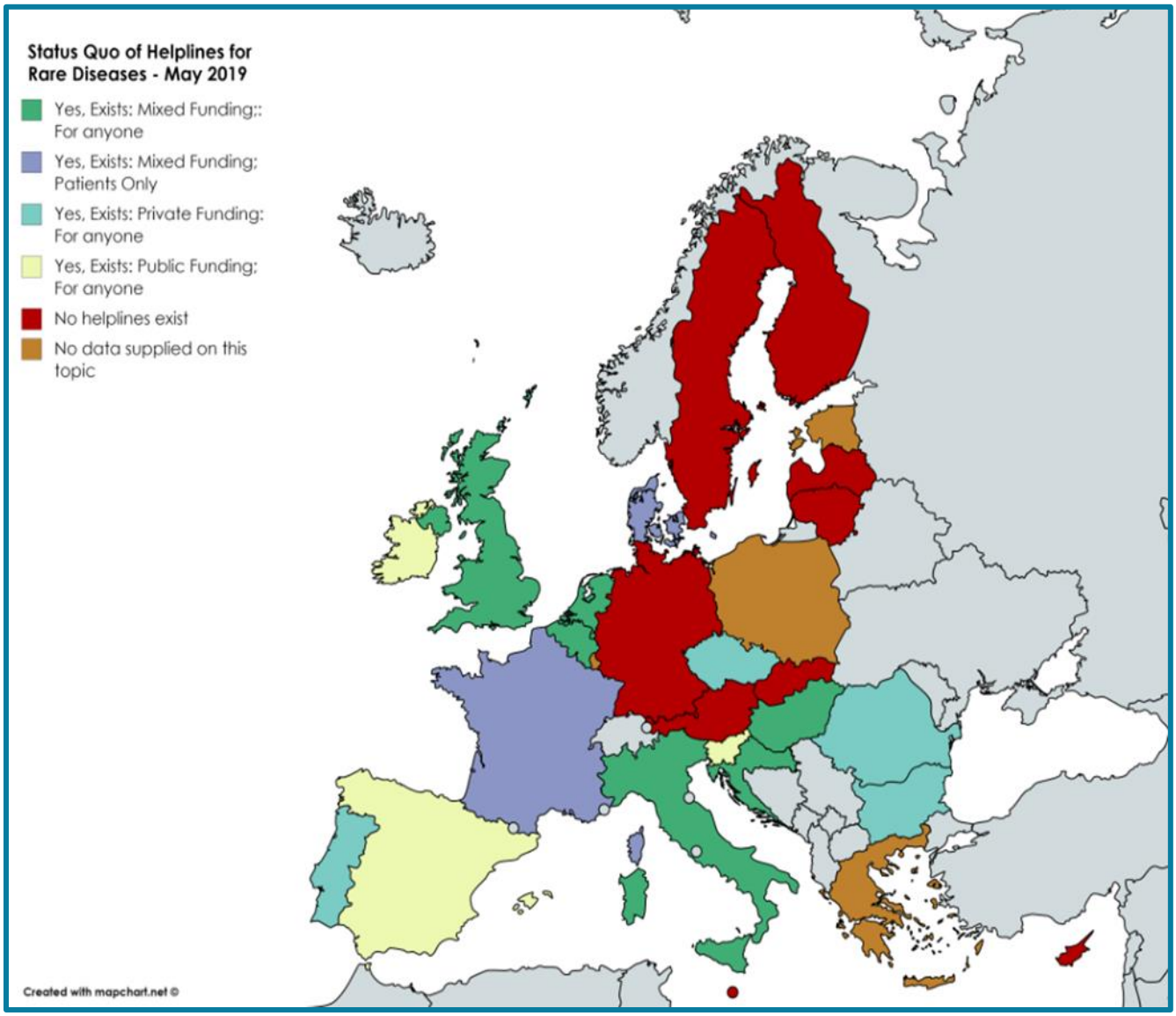
The *Resource on the State of the Art of Rare Disease Activities in Europe* (SotAR) collects data from all EU Member States (MS), through Data Contributing Committees composed of representatives of the following: National Competent Authority; Orphanet national team; national alliance of RD patient organisations.

The Committees provide data in response to strategic questions designed to allow MS to share information on their national activities in accordance with the EUCERD *Recommendations on Core Indicators for Rare Disease National Plans and Strategies*.



The map above records the status quo back in 2010, when only 8 MS (plus Norway) reported the existence of national helplines (however, these were identified as *official* helplines).

From 2015 onwards, countries were asked more specific questions on their helplines, in accordance with the EUCERD Recommendations on Core Indicators: where helplines exist, the Recommendations asked countries to specify the audience of these helplines (just patients, just professionals, or anyone?) and the means of support (private funding, public funding, or a mixed model?). The map below is based upon data received through the SotAR as of May 2019 (please note that this data will change as countries clarify and update their responses).



In 2019, as per the map above, **15 EU MS reported the existence of helplines for RD** (note however the greater number of options in terms of reporting *types* of helpline – it is likely that patient organisation-run helplines existed in 2010 but were not counted as official by the CNA representative providing the information)

5. RESULTS OF THE RARE DISEASE LITERATURE REVIEW*

*The earlier sections of this document were elaborated via research, partner expertise, and data stemming from the Resource on the State of the Art of Rare Disease activities in Europe. This final section is a summary of the results of a literature review performed by INSERM Orphanet, and is designed to highlight peer-reviewed publications which may suggest trends in this broad topic.

Recently, **a paradigm shift may be observed when considering the place, role and attention directed towards the patient**. Indeed, it seems as if an underlying change in patient management and public health structures is emerging in certain parts of the world. The patient is no longer attributed a *de facto* passive role but becomes **an actor in the health system realm**. The model also tends to **place the patient, as an individual with specificities, and personal experience, at the centre of the model** (23). This results in the design of increasingly **personalised healthcare pathways** (7; 29) which implies that healthcare systems take more responsibility for patient management and necessitates optimised **information provision for patients to be able to make informed choices** (21). Care is becoming less focused on the disease and the treatment but rather **takes into account the patient's experience and perspective for modelling a system** (4). Hence, a distinctive trend is the **change in the relationship between the patient and the healthcare professional towards a more collaborative relationship** (21). Improved communication is one of the key attributes of this system model which requires **constant and free flow of information between all participants** including patients, caregivers and parents of the patient in the case of paediatric care.

With the accumulation of knowledge, thanks in part to the ease with which knowledge is disseminated and the hurdles faced by rare diseases patients in navigating the healthcare system, **patients and caregivers tend to become experts in their diseases** (14; 28). Consequently, this **empowers them to organise, become agents of their own treatment odyssey and shape their own clinical pathway**, although this is often due to the absence of other available options. As such, a number of patient organisations have arisen from personal initiatives. These mobilisation efforts have produced significant results in building **tightly-knit and active communities driving policy**, helping to **break the isolation** of the patients and producing and providing valuable information (2; 8; 9; 16; 30). Over time, **these organisations have gained a strong position and voice in the rare disease community** and now act as representatives for patients on a global scale: EURORDIS at the European level, for instance, and Rare Disease International at the international level (8; 16).

Part of the patient empowerment process is built on the **provision of information to patients,** who are also in demand of such knowledge, as well as capacity building measures. In particular, social networks, such as PatientsLikeMe or RareConnect, specific to rare diseases, have taken up the challenge to respond to the growing request for knowledge provision and sharing, with the support evolution of digital technologies that allow sharing and co-creation of knowledge. Patients are **solicited in their role as experts of their disease to provide data, evaluation and feedback on their experience**. It prompts the emergence of **two-sided informational pathways,** where both patient or experiential knowledge and scientific or medical information are equally valued (29).

In this schema, patients become also **generators of knowledge and data,** informing research, clinical care and treatment. A direct manifestation of this trend is the development of **patient reported outcomes**

measures which are valuable reports directly obtained from the patient about their health status or treatment without being interpreted by an intermediary. These instruments make the **patients' voices central to clinical decision-making** (24). This is also the case for research in which **patients' capacity to provide data, and their experiences and views, are increasingly taken into account** as a way to determine research design, to foster patient recruitment, adaptation of patient intervention, dissemination and vulgarisation of research results. **Patients then reach a status of co-researchers** with the establishment of a bi-directional engagement and a productive and mutually beneficial working relationship (10; 11; 21; 22; 31; 32). For instance, patients and patient organisations are increasingly consulted for biobanking or the setting-up of registries (3). In terms of publication of research results, patient advocates and organisations are increasingly co-publishing research results. Some peer-reviewed journals such as the British Medical Journal (BMJ) has even established a Patient and Public Partnership strategy designed to promote co-production of content and help advance the global debate on patient and public involvement.

As a result, it is becoming more **common for multidisciplinary groups of stakeholders to collaborate in order to either formulate new policies, conduct research or improve treatment options and healthcare systems** (1; 32). An area in which patient public and private collaboration is documented to be particularly beneficial is orphan drug development (18; 19; 33; 34), as patient involvement is claimed to be fundamental for improving the likeliness of a drug to complete the orphan drug lifecycle. As a matter of fact, guidelines informed by both patient and pharmaceutical representatives for best practices in terms of patient and industry interactions are also reported (25). Moreover, **patients are increasingly included in strategic, multi-stakeholder committees, expert groups, boards of various organisations** such as the RDTF, EUCERD, the Commission Expert Group on RD, ERNs, IRDiRC and research consortia (12; 17; 20; 32).

The patient empowerment process also occurs thanks to the information dissemination generated by a diversity of sources and means. Over the years, in addition to the development of **Orphanet** resources**,** a number of **helplines for patients to obtain up-to-date quality information have been established (see above):** these lines are able to respond to enquiries on anything related to their condition or status (15). Use of web searches and online tools is also steadily increasing – improvements have been noted, in terms of ability to allow patients to establish their own diagnosis with relatively good results (27), thus completing their expertise.

Another development which forcefully places the patients at the centre of the arena is the range of possibilities afforded by **the use of social media**; not only can social media be used for information and advocacy purposes with undeniable success (29), but it is also a **core resource for seeking, producing, mining and sharing health data** (6; 26).

An additional observed trend concerns **patients' capacity to travel** and choose the location of their treatment centre. On the virtual side, the development of **ePrescription** possibilities and the current efforts to standardise **cross-border patient summary services** allows for the travel of patient *data*, facilitating the solicitation of extraterritorial healthcare services (cf. CEN and HL7 Patient Summary Standards - ehealth standards).

REFERENCES FROM THE RARE DISEASE LITERATURE REVIEW

FULL LIST OF ARTICLES / PUBLICATIONS FOUND IN THE LITERATURE REVIEW:

- https://docs.google.com/spreadsheets/d/1SRXASsFiD9sdQz286SVo860XdTpGaOIncyjlhGphULI/edit?usp=sharing

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Rare2030

Foresight in Rare Disease Policy

The health of 30 million people living with a rare disease in Europe should not be left to luck or chance. The Rare 2030 foresight study prepares a better future for people living with a rare disease in Europe by gathering the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations.

Since the adoption of the Council Recommendation on European Action in the field of Rare Diseases in 2009, the European Union has fostered tremendous progress to improve the lives of people living with rare diseases. Rare2030 will guide a reflection on rare disease policy in Europe through the next ten years and beyond.

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