Journal of Community Genetics (2021) 12:241–246
https://doi.org/10.1007/s12687-021-00524-5

ORIGINAL ARTICLE

# Our greatest untapped resource: our patients

Matt Bolz-Johnson[1] · Tom Kenny[2] · Yann Le Cam[1] · Ines Hernando[1]

Received: 10 November 2020 / Accepted: 23 March 2021 / Published online: 21 April 2021


**Abstract**
Healthcare is continually evolving to meet the changing needs of twenty-first century populations whilst striving to keeping pace with medical and technological advancements. Patients and clinicians remain the constants in this evolving environment, sitting at the cutting edge of new evidence and innovation and at the coalface of clinical services which need to address the increasingly challenging health priorities we face as a society. Patients and clinicians, positioned centre stage in this changing world, must adjust their relationships and partnerships to reduce the burden of illness and ensure that multifaceted care needs are all properly addressed. In rare diseases, this relationship between patients and professionals demands a new model of care, in which patients are active, valued partners in their own care and function not as 'enlightened self-interested' individuals but as experts by experience. The unique characteristics of rare diseases demand that care evolves beyond multidisciplinary team care to 'Networked-care', in which care is prescribed based upon the body of experience and expertise of a community of experts and patients (who are experts by experience). Healthcare models are being redrawn around a new norm of clinical practice based on true patient-clinical partnerships in care. A partnership with patients, when supported by proper investment, is a collaborative relationship that aligns both the medical and clinical perspectives of professionals with a holistic perspective of patients' life experiences. Such partnerships can (i) ensure that decisions around care and design of services are needs-led, (ii) reduce the fog of uncertainty that surrounds rare diseases, (iii) amplify the success of new discoveries, and (iv) create breakthrough innovations: in these ways, patient-clinical partnerships increase the efficiency and effectiveness of our work and build a more sustainable future for our healthcare services.

## Introduction

The year 2020 ushered in a new decade, full of expectation and renewed hope. This vantage point offers us the opportunity to start afresh, break away from old practices and behaviour, make progress in our work and research, and build on our knowledge to enrich the wealth of evidence in the field of rare diseases. What breakthroughs will we witness and shape, as we are continuing our quest to serve our most vulnerable patient communities? Will we see new scientific discoveries, new ground-breaking treatment and innovation or witness the evolution of a new model of healthcare, one driven by enhancement in our knowledge and expansion of new horizons in evidence for rare diseases? When addressing rare diseases, the smaller the evidence base, the greater the opportunity for both patients and professionals to share their expertise based on their collective experience, because experience is the driving force behind new knowledge. But with new knowledge comes change: change in service competencies and change in our professional roles, requiring us to redraw our old concepts of patient-centred care in a new world full of new norms which frame our daily practice; new innovation to prescribe and dispense; and a new, closer relationships between patients and professionals. The diversity and complexity of rare diseases actually creates major potential for the emergence of new innovations and scientific discoveries, which can address the needs of people living with a rare disease but also impact on the burden of care for more common conditions, like dementia, cancer, and a range of chronic diseases.

The characteristics of healthcare systems today have been forged by the trends of previous decades, namely the increasing burden of care from an ageing population, growing economic pressures, and ever-increasing healthcare costs. The dawn of a new decade brings very real choices about how

This article is part of the Topical Collection on *Rare Diseases*

✉ Matt Bolz-Johnson
matt.johnson@eurordis.org

[1] EURORDIS — Rare Diseases Europe, Paris, France

[2] Square Root Thinking, Cologne, Germany

people live and the true costs (and impact) of fundamental decisions. How do we re-address the tension between seeking to contain spiralling healthcare costs whilst ensuring access to innovations, and ensuring more equitable access to high-quality healthcare in a climate of greater rationing of free-at-the-point-of-need services? Whilst the political and economic environment will continue to ebb and flow, the needs of people suffering with a rare disease will still remain. However, the role played by people living with a rare disease and the nature of their involvement in the healthcare system *will* continue to evolve, as these individuals occupy a central place in service design and delivery — as care receivers, as experts by experience, and as shared leaders. This role is of central importance in the drive for greater service efficiency and efficacy, aimed at targeting patient needs.

Today, there is a clear and present need to invest precious health resources into evidence-based care and best practices (both acknowledged and developing), which relieve the disease burden for people by addressing both short-term and long-term needs. Rare diseases, due to their unique characteristics and complexities, require new competencies for patients, clinicians and hospitals to provide 'networked care' beyond individual hospitals and countries. In this paradigm shift from multidisciplinary team (MDT) care to network-care, we see a forging of new partnerships between patients and professionals: these new partnerships, in which care is informed by the wealth of experience of both patients and professionals, redefine our concept of patient-centred care. In such relationships, rare disease patients play a central role as experts by experience in care provision and as navigators of complex healthcare pathways.

Beyond forging stronger partnerships, more widespread strategic reforms of the healthcare systems in Europe are necessary: this reform is starting today with rare diseases but will benefit tomorrow the management of all chronic diseases, whilst also leading to optimal health in ageing. Care of the future must address the holistic, 360° needs of people living with rare diseases, through greater integration of medical and social care: it must move away from the mantra of simply 'treating illness' to embrace a more integrated and strategic approach which merges the prevention of co-morbidities, a timely diagnosis, and coordinated care for co-morbidities which treats the person as a whole. At the same time, the health literacy of patients and their families must be increased, empowering people to understand and manage their own care and provide feedback on their experiences, for optimal outcomes. Patients must be supported to manage their data across their lifetimes, providing continuity of medical and social history and mitigating the impact of the ever-increasing mobility of our society.

The rare disease community can be confident of achieving its ambition, namely, of improving the lives of every one of the >300 million people living globally with a rare disease, as in fact it has already laid the foundation for this future over the past few decades. During a decade of advocacy — from 2000 to 2010 — for the rare disease community in Europe, spearheaded by a solidarity movement led by EURORDIS (Rare Diseases Europe), 'Rare Diseases' was recognized as a public health priority in the EU and its Member States. Now, through greater global recognition — driven by Rare Diseases International — the United Nations (UN) has initiated groundbreaking policy action by including rare diseases in the WHO Universal Health Coverage. As a recognized public health priority, rare diseases have galvanized political support for cross-border collaboration, and *global* cooperation and action in healthcare and research has begun. Countries around the world have the opportunity to tackle the public health needs of rare diseases once and for all, positioning rare diseases as a proof of concept and success story to demonstrate that global action can address other health prioritizes and emergencies, offering hope for all.

From 2010 to 2020, the infrastructure to tackle the public health needs of rare diseases was established, to incentivize therapeutic development, enable research and build healthcare networks. The new paradigm of 'networked-care' for highly specialized healthcare was established, with the launch of European Reference Networks (ERNs). Now is the critical, make-or-break time for the ERN system. The networks need to be supported and nourished and given the space to experiment in their new partnership approach, in order to grow, whilst continuing to support the existing healthcare systems. The success of the ERN model is now gaining traction globally and propelling an expansion in the vision of networked-care, where rare disease expert centres across the globe will be connected under a global network for rare diseases. With each new decade, we make a step forward and celebrate our achievements, such as the launch the ERNs, the European Joint Programme for Rare Disease research (EJP RD), the UN Committee for Rare Diseases. Now, in a new decade, we need to set out new goals and our ambition and emphasize how patient partnerships will make this a reality.

## Rare diseases/highly specialized healthcare

With rare, comes rarity: rarity of patients, of experts and of knowledge, with a limited evidence base and lack of available treatments. These factors have a significant impact on the quality of life and life-expectancy of people living with a rare disease, as many diseases are progressive, degenerative, disabling and life-threatening. Whilst each rare disease is unique, there are commonalities that unite individual, isolated, and often invisible cases, to create a critical mass that cannot and should not be ignored. Rare diseases offer great opportunities to advance medicine and healthcare for the many, not just for people directly affected by these diseases. To enable this,

clinicians and patients are developing a new set of competencies that can unlock the potential that patient-professional partnerships and shared leadership offer.

Due to their rarity, people affected with a rare disease are often isolated and lost in the day-to-day demands placed on local healthcare services (EURORDIS 2017). It is normal for general practitioners and even specialists to only see one case of an individual rare disease in their career. The attention of healthcare systems is weighted towards the ever-growing volume of demands for urgent care, oncology and management of chronic diseases. It is easy to understand why the needs of the 'one in a million' patients are overlooked, in the daily orchestration of hospital services. However, whilst individual people affected with a specific rare disease are 'few and far between', the collective needs of all people affected with rare diseases cannot be ignored, as they constitute a heavy burden for hospital systems and healthcare budgets. The specificities of rare diseases mean that the needs and preferences of the patient population are critical in informing both individual care and the design and delivery of highly specialized services. Addressing these needs and preferences requires the reporting of health outcomes relevant for patients and the collection of real-world evidence: this enables the patient to become the central actor in the expanded model of networked-care, which goes beyond the provision of traditional hospital services.

The rare disease community cannot afford to wait for new evidence to be published in the traditional way, as time is precious and finite. The wealth of clinical experience and size of the evidence base for rare diseases is at best, limited, and will remain so. There is a direct relationship between the prevalence of a disease, the level of complexity and needs, and the amount of evidence and expertise available. We will never be afforded large volumes of published evidence, due to the nature of low-prevalence diseases. However, there is room for other type of knowledge, based on expert experience. Drawing only on the clinical experience and emerging best practices is a self-limiting strategy in rare diseases, as in fact the pool of knowledge for each and every rare disease is considerably larger than that held by the medical teams. People living with a rare disease have experiential knowledge of their condition, viewed through a life-lens as opposed to the clinic-lens. The potential to drive forward new innovative approaches and elucidate best practice can be enhanced by drawing on the total sum of the experience and knowledge of both professionals and patients.

Adherence to evidence-based care for a given disease is higher amongst clinicians and hospitals that see a higher number of cases of that disease. This adherence leads to significant improvements in health outcomes and quality of life of patients, with improved mortality rates and reduced morbidity. However, patient care remains fragmented across the healthcare systems, and patients are often only referred to an expert centre after years of being undiagnosed or even misdiagnosed, potentially receiving inappropriate and ineffective treatment. Patients are often the best people to assess what their needs are and what outcomes are important to them, to inform shared decision-making and balance the different benefits and side-effects of treatments. However, patients in healthcare tend to remain as passive subjects of discussion and treatment: often patient involvement remains a good intention and aspiration, rather than a reality. A cornerstone of modern-day healthcare is patient-centred care, which requires patients to be empowered to share their needs, experiences, and knowledge and to be active in their own care whilst also involved in the design and development of healthcare services.

There is a growing body of evidence that active and meaningful patient involvement improves the outcomes and efficacy of interventions, as well as the quality and safety of services, whilst driving service efficiencies (Epstein and Street 2008; Coulter and Ellins 2007; Charmel and Frampton 2008). Patient experience has been demonstrated to be the single most important element in assessing the quality of healthcare, as it has been shown to be a predictor of survival rates (Gupta et al. 2013; Gupta et al. 2014). Studies have also evidenced the fact that patient involvement in healthcare services and their evaluation and designation improves both the relevance and quality of the healthcare services as well as their ability to meet patient needs (Brett et al. 2014; Cornman and White 2017; Fønhus et al. 2018; Sharma et al. 2017; Tokhi et al. 2018). However, experience and appreciation of patients as valued partners varies considerably from clinician to clinician, hospital to hospital, and between countries.

The legitimacy of patients and affected individuals to be involved in their own care or in service design stems from their experience and knowledge of living with a rare disease. Patients with a rare disease manage their symptoms and day-to-day challenges, which enables them to build their knowledge and expertise about their disease; develop an expert view, from their first-hand experience; and gain insight into the natural history of the disease and its impact on their daily lives (Menon et al. 2015; Armstrong et al. 2017). Many draw upon a lifetime of experience and holistic understanding, which is broader than the medical perspective of a rare disease held by a typical clinic or hospital setting. People living with a rare disease, and their families, are required to extend their role and secure additional competences in healthcare literacy, in order to case manage their own condition and navigate complex healthcare systems and arrangements, between multi-clinic teams and numerous hospitals, and sometimes across borders. Rare diseases therefore bring an additional level of complexity for both patients and clinicians alike, which impacts upon the role and function of both, demanding a deeper, closer relationship and collaboration. The rarer the disease area, the fewer the experts and scarcer the knowledge. People affected with a rare disease should be recognized as experts of their own disease.

For rare diseases, therefore, we must reframe the concept of a patient-centred approach, moving away from mere consultation and involvement towards a model of partnership and shared leadership (Carman Kristin et al. 2013). Our new concept of patient-centred healthcare, in which the patient is at the centre of their care, is a member of the multi-professional team, and is simultaneously a key and valued partner in developing evidence and designing services, requires patients with rare diseases to develop unique competencies: *Enlightened 'self-interest' individuals* embody willingness, commitment and a degree of altruism, investing their time and expertise — beyond personal gain — for the greater good and for those who follow. Whereas, *experts by experience* hold deep personal experience, knowledge and expertise, gained from a lifetime of knowledge of the disease and its natural history. The most important of these two competencies is patient's deep personal knowledge and experience. The degree of experience and knowledge of the disease increases with the complexity and severity of a disease and the length of time a patient has lived with the condition. Their ability and potential to contribute as a partner in shaping rare disease care are also related to the size of the existing evidence base: the smaller the evidence base (and number of experts), the greater the opportunity (and need) for patients to share their expertise and experience with the clinicians.

## Examples of patient and advocate achievements

### Rare disease patients as leaders

Whilst each individual rare disease is rare, the global population affected with one of the +6000 rare diseases equates to a significant population of over 300 million people. The rare disease community has progressively come together to act with one voice and built a movement to advocate for the needs of all rare diseases. Patients and their representatives have played (and continue to play) a critical role in securing and building political commitment to tackle the public health needs (2000–2010), informed expert recommendations (EUCERD, 2012–2015[1]; RD-Action, 2015–2018[2]); and now model future policy scenarios in a foresight study (RARE 2030 Foresight Study, 2019–2020[3]). Without the solidarity and coordinated action of the community as a whole, the political support and investment into community activities would not have delivered the results we are seeing today. Many other public health priority areas have been less successful than rare diseases in harnessing political declarations. The strength of the rare disease community comes from its solidarity and the legitimacy it has accrued as a respected global actor in healthcare, research and medicines' development.

### Rare disease patients as experts by experience and partners in decision-making

Rare disease patients play an active role in the design and delivery of European infrastructures and services, particularly in the European Medicines Agency (EMA) therapeutic development and licencing processes, in the development of ERNs, and in the European Joint Programme for Rare Disease Research. Patient representatives are co-chairs of working groups in ERNs, for research, training and education and guidelines, as well as being voting members of ERN Network Boards and playing a key role in the Networks' advisory groups, to shape Network workplans and strategies.

Patient representatives are recognized for their expertise and contribution in these European initiatives, as experts by experience: shaping the design of clinical trials, registries, endpoints and real-world data collection for new medicines, and sharing their needs along their patient journey (Bolz-Johnson M. et al. 2019), to design clinical pathways and author new guidelines and consensus statements.

Although patient involvement is generally perceived as beneficial and recommended in developing clinical services, there is no clear consensus on the most effective approach. The involvement of patients in healthcare service design has evolved over recent decades. Patients have moved from being consulted on the changes proposed, to being central actors shaping the development of services and medicines, from the very beginning of the process — from mapping their needs against the current service configuration to enabling transformational change to be driven from the patient perspective, patients are increasingly involved in the whole lifecycle of projects, medicine development and service improvement. Without meaningful involvement of patients throughout the lifecycle, the baseline needs cannot be established and used as a reference point for change.

### Rare disease patients involved in their own care

People affected with a rare disease, and their families, are now often actively involved in their care and treatment, due to the disease burden and impact on their lives. With limited treatments available, and given the high cost of many interventions — which can generate significant side-effects and risks of complications — patients' preferences and opinions to weigh up the benefits and harms of treatment and inform shared decision-making is essential. In addition, patients' involvement in their own care is important to report their experiences,

[1] A summary of the EUCERD Joint Action and its achievements can be found in Lynn et al. (2017) and Hedley et al. (2018)
[2] See http://www.rd-action.eu/
[3] See https://www.rare2030.eu/

outcomes and real-world evidence of treatment efficacy, which can aid further research and increase treatment options for themselves and others.

## Barriers for patient involvement and partnership

To optimize patient involvement in rare diseases, we need to address the perceptions and behaviour barriers, for both patients and clinicians. Some patients may be reluctant to provide input and some clinicians can be resistant to receiving their input. Patients can lack health literacy, in terms of terminology and content, for instance, and have an unwillingness to contribute if they do not feel confident enough to express themselves. They may also feel they do not have time to provide input, or may have concerns around clinician resistance to their opinion, which can be a barrier to their input (Armstrong et al. 2017). However, these concerns can be easily overcome with capacity building and mentoring to (i) understand the technical aspects of developing standards of care and guidelines and informing service redesign, (ii) enhance mutual understanding regarding a patient's role, and (iii) ensure smooth collaboration and optimize group dynamics. Approaches such as involving facilitators can also help mitigate patients' reluctance and maximize engagement. Patient representatives may also require support to ensure they understand the aims of an exercise and their role in the process, and have the educational capacity to provide meaningful input. There is a major need for clear communication and information provision from the outset, to ensure patients understand the goals and their role in the process, and to enable representatives to be well-equipped and possess the capacity to provide meaningful input.

Published evidence indicates a need to bring clinicians on board more effectively, as they may initially be resistant to patient input. Their perceptions and behaviour may also impact on the degree a patient inputs to any given process or activity (Armstrong et al. 2017). For instance, research has highlighted the fact that when it comes to guideline development, clinicians may initially feel patient input will stifle debate, as they would need to use lay language. However, this perception has been reported to change over time, and by the end of the process in question, clinicians had a positive perception of the value of the patients' contribution to the discussions (Chalmers et al. 2017).

## Conclusion

Partnership with patients is a valued relationship that we need to invest in, in order to align the medical and clinical perspectives of professionals with a holistic perspective of life experiences. This partnership will enhance our opportunity to achieve our shared ambitions, amplify the success of new discoveries and create breakthrough innovations. Both clinicians and patients share a common goal, but partnership enables us to shine a spotlight on the issues and challenges we face from different perspectives: aligning our efforts will maximize the results of our work and bring benefits for both parties.

We, the rare disease community, call for the hallmark of the new decade to be patient and clinician co-design: working as equal partners in healthcare and research, collaborating in groups to identify the barriers and drivers in the journey of the patients, analysing patient reported experience and patient reported health and social outcomes, and creating ideation and shaping solutions together in the organization of care, in the best practice of care, and in therapeutic education. Our newly forged partnership needs to be balanced and truly equitable, respectful of different though complementary experiences and responsibilities in the patient's journey as well as in the healthcare system, without one overshadowing another — as true partners. Redefining the patient-centred approach in the field of rare diseases, to hold at its heart the values of a true patient-professional partnership, ensures decisions around care and design of services are needs-led, reducing the fog of uncertainty that surrounds rare diseases, and strengthening the efficiency and effectiveness of our work and the resources we invest, building a more sustainable future.

**Funding** The perspectives shared in this article do not originate from any particular project or funding opportunity. EURORDIS was/is a partner in the following projects (which are mentioned in the manuscript)

- Rare 2030 PP-1-2-2018-Rare 2030, co-funded by the European Union Pilot Projects and Preparatory Actions Programme
- RD-ACTION: 'Data and Policies for Rare diseases' 677024 which received funding from the European Union's Third Health Program
- EUCERD Joint Action: Working for Rare Diseases (No. 2011 22 01) which received funding from the European Union's Second Health Programme

## Declarations

**Ethical approval** This article does not discuss or contain any studies with human participants or animals performed by any of the authors.

**Conflict of interest** The authors declare no competing interest.

## References

Armstrong MJ, Mullins CD, Gronseth GS, Gagliardi AR (2017) Recommendations for patient engagement in guideline development panels: a qualitative focus group study of guideline-naïve patients. PLoS One 12(3):e0174329. https://doi.org/10.1371/journal.pone.0174329

Brett J, Staniszewska S, Mockford C, Herron-Marx S, Hughes J, Tysall C, Suleman R (2014) Mapping the impact of patient and public

involvement on health and social care research: a systematic review. Health Expect 17:637–650. https://doi.org/10.1111/j.1369-7625.2012.00795

Carman Kristin L, Dardess P, Maurer M, Sofaer S, Adams K, Bechtel C, Sweeney J (2013) Patient and family engagement: a framework for understanding the elements and developing interventions and policies, Health Affairs Journal Vol. 32, No. 2: New Era of Patient Engagement https://doi.org/10.1377/hlthaff.2012.1133

Chalmers JD, Timothy A, Polverino E, Almagro M, Ruddy T, Powell P, Boyd J (2017) Patient participation in ERS guidelines and research projects: the EMBARC experience. Breathe Sheff Engl 13(3):194–207. https://doi.org/10.1183/20734735.009517

Charmel PA, Frampton SB (2008) Building the business case for patient centered care. Healthc Financ Manage 62(3):80–85

Cornman DH, White CM (2017) Discerning the perception and impact of patients involved in evidence-based practice center key informant interviews [Internet]. Agency for Healthcare Research and Quality (US); Report No.: 17-EHC032-EF. AHRQ Methods for Effective Health Care

Coulter A, Ellins J (2007) Effectiveness of strategies for informing, educating, and involving patients. BMJ. 335(7609):24–27

Epstein R, Street R (2008) Patient-centered care for the 21st century: physicians' roles, health systems and patients' preferences. Philadelphia (PA). ABIM Foundation

EURORDIS (2017) Juggling care and daily life: The balancing act of the rare disease community. http://download2.eurordis.org.s3.amazonaws.com/rbv/2017_05_09_Social%20survey%20leaflet%20final.pdf. Accessed October 2020

Fønhus MS, Dalsbø TK, Johansen M, Fretheim A, Skirbekk H, Flottorp SA (2018) Patient-mediated interventions to improve professional practice. Cochrane Database Syst Rev. CD012472. doi: https://doi.org/10.1002/14651858.CD012472.pub2

Gupta D, Lis CG, Rodeghier M (2013) Can patient experience with service quality predict survival in colorectal cancer? J Healthc Qual 35:37–43

Gupta D, Rodeghier M, Lis CG (2014) Patient satisfaction with service quality as a predictor of survival outcomes in breast cancer. Support Care Cancer 22:129–134

Hedley V, Aymé S, Murray H, Rodwell C (2018) 2018 report on the state of the art of rare disease activities in Europe http://www.rd-action.eu/wp-content/uploads/2018/09/Final-Overview-Report-State-of-the-Art-2018-version.pdf Accessed 15 Nov 2020

Lynn S, Hedley V, Atalaia A, Evangelista T, Bushby K (2017) How the EUCERD joint action supported initiatives on rare diseases. European Journal of Medical Genetics 60(3):185–189. https://doi.org/10.1016/j.ejmg.2017.01.002

Menon D, Stafinski T, Dunn A, Short H (2015) Involving patients in reducing decision uncertainties around orphan and ultra-orphan drugs: a rare opportunity? The Patient - Patient-Centered Outcomes Research 8(1):29–39

Sharma AE, Knox M, Mleczko VL, Olayiwola JN (2017) The impact of patient advisors on healthcare outcomes: a systematic review. BMC Health Serv Res 17(1):693. https://doi.org/10.1186/s12913-017-2630-4

Tokhi M, Comrie-Thomson L, Davis J, Portela A, Chersich M, Luchters S (2018) Involving men to improve maternal and newborn health: a systematic review of the effectiveness of interventions. PLoS One 13(1):e0191620. https://doi.org/10.1371/journal.pone.0191620