

VIEWES AND OPINIONS

PRORARIS



Alliance Maladies Rares – Suisse
Allianz Seltener Krankheiten – Schweiz
Alleanza Malattie Rare – Svizzera



PROMOTING INCLUSIVE, PATIENT-ORIENTED RESEARCH: A RARE DISEASE PATIENT PERSPECTIVE

Authors: **Therese Stutz Steiger**^{1,2} and **Olivier Menzel**^{1,3}

Affiliations: ¹ProRaris Swiss Alliance; ²Swiss Osteogenesis Imperfecta Association (SVOI), former president; and ³BLACKSWAN Foundation for research on orphan diseases, chairman and founder

Patients and members of the public contribute valuable information and perspectives to research projects because they have experienced a disease themselves or are close to someone affected by an illness. Patients with rare diseases are incredibly motivated to participate in research projects. Any advancement of scientific or medical knowledge or favourable political decisions can increase the possibility of a cure – or at least a treatment – that can stop or slow the progression of their disease and improve their quality of life. Patients' lived experience may help researchers fill gaps in understanding conditions that they know mainly from theory. In this article, the authors address the need for more focus on patient-oriented clinical and public health research, the importance of bringing patients' perspectives into research, issues around patient involvement, and areas for future research on the topic.

Rare diseases severely impact people's everyday lives and the lives of those around them: they often face overwhelming time, health, and social costs. For support, patients and their caregivers are often involved in patient organisations such as [ProRaris](#), an umbrella organisation that advocates

for patient groups and patients in Switzerland living with a rare disease. Patients and patient organisations can help provide researchers with an insider's perspective on their health conditions, thus improving research related to a cure or treatment (FOPH 2014).

FOCUS ON PUBLIC HEALTH RESEARCH

According to the Organisation for Economic Co-operation and Development's (OECD's) 2001 definition, biomedical research encompasses the full spectrum of health- and disease-related research, from basic research to public health research through to clinical research (OECD n.d.). However, the understanding of biomedical research is now commonly restricted to discovery research and thus does not cover the full range of health-related research. Public health research (Egger et al. 2021), by nature observational, focuses on the evaluation of population health and well-being and patient experiences (Moreau et al. 2021). This research typically happens outside of the controlled clinical setting of experimental or interventional research and aims to improve the physical and mental aspects of the

human condition through longitudinal, interdisciplinary, multi-methodology research. The well-funded biomedical approach to research is oriented towards developing technological solutions that can be commercialised. An excellent recent example is the development of SARS-CoV-2 vaccines. Public health research is often neglected, yet an increased focus on such research could address many patient-oriented topics, including:

- patients' criteria for quality of care
- how patients perceive interdisciplinary and inter-professional collaboration
- patient self-management
- patient empowerment
- life-course and transition challenges.

PATIENT INVOLVEMENT IN RESEARCH: "NOTHING ABOUT ME WITHOUT ME!"¹

Patient and public involvement (PPI) in research (biomedical or public health) means that a research project is carried out "with/by" patients or the public.² Patients' opinions must have a more significant influence on the decisions that affect them. Care that is respectful of and responsive to their preferences cannot be provided without patients' participation in both their own healthcare decisions and the research that informs such decisions.

It is important to emphasise that patients do not all share the same experiences and skills, and they are not all willing to be involved to the same degree in research. Moreover, different studies may have different needs for patient involvement. What is certain is that all patients can contribute to some degree and make a difference at every stage of research (see **Box 1** on the next page).

¹ From Valerie Billingham during session 356 of the Salzburg Global Seminar in 1998 entitled Through the Patient's Eyes.

² Adapted from the National Institute for Health Research (NIHR) INVOLVE's supporting statement (<https://www.invo.org.uk>).

Box 1: Degrees of patient expertise and involvement

Level of expertise	Description
Lay patients	<ul style="list-style-type: none"> • Have a passive role in their healthcare and no experience with PPI • May be looking to become a member of a patient organisation • Have not had medical training or expertise in any profession linked to medicine or the life sciences
Patient actors/partners	<ul style="list-style-type: none"> • Participate in their healthcare (on an individual level) and act as advocates more globally (on a collective level) • Are likely a member of a patient organisation • Do not necessarily have experience with PPI or in the medical field
Patient experts	<ul style="list-style-type: none"> • Have experience with PPI or play an active role in a patient organisation (Stutz Steiger 2016) • May be members of a patient organisation's committee/board and thus have relevant skills and experience • Can take on multiple roles with or without other patients, can partner with health professionals, and can act as a trainer or research partner • Frequently have medical training or are in a profession linked to medicine or the life sciences

PPI CONSIDERATIONS: COMPENSATION AND SELF-MANAGEMENT

PPI depends on patients sharing their expertise. Rare disease patient expertise is in increasingly high demand for various reasons, including a lack of understanding of many rare diseases, lobbying with drug agencies, patient recruitment, and fundraising (Halsbeck et al. 2016). The PPI aspects of a research project and patient experts' contributions require a considerable time investment. To attract enough PPI contributors, they must be adequately compensated financially, beyond any non-material benefits they may experience by contributing. This argument favours having more PPI contributors considered active members of a project team (Pomey et al. 2021) and on the project's payroll.

Self-management refers to how people who are affected by a chronic condition manage their health and themselves (FOPH 2018). Self-management requires striking a balance between enabling patients to manage their health, not putting them under too much pressure to do so, and not setting unrealistic self-management goals. For patients with low self-management, actively participating in a research project can be a source of additional stress. Patients with high levels of self-management are excellent candidates for PPI in research projects, primarily due to their ability to critically reflect on their health situation. Including their perspectives on self-management and other patient-relevant topics can significantly improve participative research study designs.

PPI IN FUTURE RESEARCH

Comprehensive and practical PPI can be challenging to achieve (NHS Health Research Authority n.d.). The first step is involving patients. Involving the wider public raises additional methodological questions but might be particularly important for preventive (e.g. vaccination) and screening interventions targeting non-diseased persons. Biomedical and public health researchers should remember that the main objectives of their research are to prevent disease, promote its early detection where

appropriate, cure illness where possible, or improve treatment and thus improve patients' quality of life. PPI can be a powerful tool in helping retain the focus on these objectives (see PPI action points in **Box 2** on the next page). In light of all these considerations, the research agenda for the next ten years must include PPI. In Switzerland, a dedicated national research program addressing ways to organise PPI and, more generally, citizen science is overdue.

Box 2: PPI action points

- Introduce incentives for patients and public involvement in research
- Emphasise that knowledge of disease pathogenesis is essential but patients are not just research subjects
- Encourage patients to participate in health literacy and become active members of research teams
- Increase interdisciplinarity with patient inclusion to improve patient recruitment and participation
- Expand Switzerland's national concept on rare diseases to include the interaction between action and research (see the Federal Office of Public Health's current [National Rare Disease Policy](#) (in German))

ACKNOWLEDGEMENTS

The authors would like to thank the following people for their input on and revision of this manuscript: Matthias Egger and Marcel Zwahlen at the Institute of Social and Preventative Medicine (ISPM) in Bern, Jacqueline De Sá at ProRaris, and Nadine Stoffel-Kurth and Alberto Marcacci at the Federal Office of Public Health (FOPH).

REFERENCES

- Egger M, Razum O, and Rieder A, eds. (2021) Public Health Kompakt, 4th edition, pp. 1–2; 9–24. Berlin/Boston: Walter de Gruyter GmbH. (in German)
- Federal Office of Public Health FOPH (2014) National Rare Disease Policy. Accessed 2 July 2021. <https://www.bag.admin.ch/bag/en/home/strategie-und-politik/politische-auftraege-und-aktionsplaene/nationales-konzept-seltene-krankheiten.html> (in German)
- Federal Office of Public Health FOPH (2018) Referenzrahmen: Selbstmanagement-Förderung bei chronischen Krankheiten und Sucht. Accessed 2 July 2021. <https://www.bag.admin.ch/bag/de/home/strategie-und-politik/nationale-gesundheitsstrategien/strategie-nicht-uebertragbare-krankheiten/praevention-in-der-gesundheitsversorgung/selbstmanagement-foerderung-chronische-krankheiten-und-sucht/referenzrahmen-selbstmanagement-foerderung.html> (in German)
- Haslbeck J, Mohylova M, Zanoni S, and Stutz Steiger T (2016) Patienten und Angehörige beteiligen. Einbezug von Betroffenen bei Akademie-Projekten im Bereich «Gesundheit» – Bericht zuhanden der Schweizerischen Akademie der Medizinischen Wissenschaften (Vol. 11), p. 8. Swiss Academies of Medical Sciences. https://www.samw.ch/dam/jcr:76073096-395a-4640-8d83-c68480164047/bericht_samw_patienten_beteiligen.pdf (in German)
- Moreau KA et al. (2021 Jan) Patient involvement in medical education research: Results from an international survey of medical education researchers. *Journal of Patient Experience*. doi: <https://doi.org/10.1177%2F2374373520981484>
- NHS Health Research Authority (n.d.) Planning & improving research [web page]. Accessed 2 July 2021. <https://www.hra.nhs.uk/planning-and-improving-research/>
- Organisation for Economic Co-operation and Development OECD (n.d.) OECD glossary of statistical terms [online version]: Biomedical research. Last updated 4 January 2006. Accessed 27 September 2021. <https://stats.oecd.org/glossary/detail.asp?ID=217>
- Pomey M-P et al. (2021) Assessing and promoting partnership between patients and health-care professionals: Co-construction of the CADICEE tool for patients and their relatives. *Health Expectations* 24(4):1230–1241. doi: <https://doi.org/10.1111/hex.13253>
- Stutz Steiger T (2016) Gesunde Bewältigung einer chronischen Krankheit: Austausch unter seinesgleichen. Researchgate. doi: [10.13140/RG.2.1.1965.9124](https://doi.org/10.13140/RG.2.1.1965.9124) (in German)