Review Article

Shared decision-making in rare diseases: an overview

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Abstract

Patients with rare diseases often face difficulties in clinical care due to the low prevalence of their diseases and the resulting healthcare providers' lack of expertise. Valid and standardized guidelines for clinical management are also lacking due to the scarcity of research and the variability of the clinical expressivity within each disease. Clinical decision-making in an uncertainty context should take advantage of involving patients in deeper informational process to promote valid shared decision-making between patients/caregivers and healthcare professionals. This process of patient/caregiver empowerment is a priority in the context of rare diseases, as it encourages acquisition of information that will help improving patient-healthcare professional's interaction, and building a collaborative relationship. It is also a chance for healthcare professionals to learn about rare diseases from the perspective of patients.
The aim of this article is to conduct an overview of existing studies focused on promoting patients/caregivers empowerment and shared decision-making (using or not decision aids) in the area of rare diseases.

Key words

Caregivers, decision aids, empowerment, patients, rare diseases, shared decision-making.

Background

Rare diseases (RDs) are defined in Europe as chronic life-threatening or debilitating conditions affecting no more than 5 in 10,000 people, characterized by difficulties in diagnosing and, for most of them, by having no effective therapy [1]. Though the accurate incidence and prevalence of most RDs are still unknown [2], their low magnitudes limits both the growth of clinical experience and research activities to improve the availability of valid knowledge on diagnostic and therapeutic procedures [3]. Besides, different constraints to research in RDs such as funding limitations, limited commercial interest and logistic barriers for patient recruitment and engagement have also contributed to the gap of valid scientific knowledge [4]. Additional limitations are the regulatory burdens, fragmented infrastructure, inconsistent databases, and the lack of proper communication between researchers, healthcare professionals, and patients [5].

All these circumstances, together with the scarcity and limited effectiveness of available diagnostic and therapeutic tools, maintain a high uncertainty and anxiety among healthcare professionals, patients and families, explaining the existing rates of errors and delays to get an appropriate diagnosis and treatment [6]. Because often there is not a single or 'best' option based on scientific evidence, it is necessary to inform patients and incorporate their values and preferences in the process of decision-making [7]. In this context, patient and caregiver empowerment and participation in decision-making along their care processes became an important aspect to improve the quality of healthcare in RDs.

Empowerment of people affected by RDs is relevant to support effective participation in decision-making along the interaction with healthcare professionals mainly when diagnostic or therapeutic options are complex or supported by limited evidence of efficacy and/or safety. At individual level, the term 'empowerment' involves

an assessment of one's knowledge and learning acquired through the personal experience of living with the disease, in addition to the knowledge acquired from biomedical sources. It also encompasses action toward self-management of the disease, which requires creating the necessary capabilities. Empowerment of patients and their organizations are one of the main aims of the European Council Recommendation in the field of RDs (2009/C151/02), and World Health Organization (Statement 2010), considering it an essential concept of health promotion and disease management.

In this sense, the RARE-Bestpractices project (www.rarebestpractices.eu) is developing a platform to enhance clinical management and to reduce healthcare inequalities for RDs patients by improving the exchange of knowledge and reliable information on RDs. The European Commission (European Union Seventh Framework Programme) funded this four-year study (until December 2016) and its main goal is to promote communication on the management of RDs. It provides mechanisms to identify and prioritize clinical RDs research needs, taking into consideration both patients' and healthcare professionals' needs and interests. In addition, it addresses patients' and caregivers' demand for updated and evidence-based clinical practice guidelines on RDs [8-10], and facilitates timely, effective and efficient translation of research results for general audiences and patients.

Other actions from the European Commission also increase patient involvement in research as active contributors on decision-making, not just as cases of study [11]. The access of patients with capacity to make decisions to the Committee for Orphan Medicinal Products, or the creation of a working group with patient representatives in response to the first European Union Public Health Programme (2003-2008) are some examples. The participation of patient organizations in research [12-14] or the incorporation of patients' perspective in the study designs [15-18] is also noteworthy.

Despite the advances described, neither the healthcare systems nor the healthcare professional organizations in the European Union are doing clear or powerful efforts to inform, sensitize and train healthcare professionals to work with patients in a needed scenario of informed and participatory decision-making. This report provides an overview of the literature to identify published studies focused on promoting empowerment of RDs patients/caregivers and shared decision-making in the clinical encounter, either evaluating basic variables for its implementation or using specific decision aids.

Shared decision-making and patient decision aids

Promoting patient participation in healthcare is considered an ethical imperative, and the Salzburg Declaration [19] states it, where healthcare professionals and patients are invited to become co-responsible for healthcare management. The 'Shared Decision-Making' model (SDM) [20] involves a two way process where the healthcare professional provides the technical expertise and the patient brings their values, preferences and concerns regarding the interventions to choose [21]. The mutual exchange of information and acceptance of the decision may not occur in other decision models, such as the paternalistic or informed models; however, this aspect is a prerequisite for SDM, although mutual acceptance does not always indicate the existence of a SDM [22]. In the paternalistic model, patients play a passive role. Then, physicians suppose that they know the best option to choose in the decisional process and inform patients about it, although they must give their informed consent. In the informed model, information exchange is also from physicians to patients, but the decision is taken by patients alone, needing the physician's agreement to implement the preferred choice [23].

To support the SDM process, patient decision aids (PtDAs) have been developed [24]. They are instruments that help make specific and deliberate decisions, providing information about the available options and their expected results. PtDAs can be presented in different formats (print, video, CD-ROM, Web) and with different levels of informative detail, but they always include information on the potential risks and benefits of each option [25, 26]. These tools differ from educational materials and the informed consent to the extent that they help elicit patients' preferences and values regarding the different options. They can be used during or outside consultation but, although they represent a facilitator of communication between patients and healthcare professionals in the clinical encounter, they must not be considered as a substitute of professionals' advice. PtDAs are useful in very specific contexts, such as RDs, where many healthcare professionals may lack the necessary knowledge about the therapeutic options and their effects. Thus, having PtDAs might guarantee the access of patients, families and professionals to scientifically valid and adapted information to promote SDM [22].

Currently a PtDAs inventory with more than 600 tools is available, of which more than a half are accessible on the Ottawa Health Research Institute's website (https://decisionaid.ohri.ca/). Academic institutions have developed some of these PtDAs, whereas others have been created by organizations that are specialized in the dissemination of healthcare information. Some of the main institutions devoted to the development and assessment of PtDAs are Healthwise/Informed Medical Decisions Foundation (www.healthwise.org; www.imdfoundation.org), Knowledge and Encounter Research Unit (Mayo Clinic) (http://www.mayo.edu/research/labs/knowledge-evaluation-research-unit) or Cardiff University (Decision Laboratory: www.decisionlaboratory.com).

Owing to the large variability of available PtDAs for dif-

ferent medical conditions, the International Patient Decision Aids Standards (www.ipdas.ohri.ca) were established to assess the quality of these tools taking into account three dimensions: content, development process and effectiveness assessment [27]. To establish the effectiveness of PtDAs it is necesary to evaluate the characteristics and quality of the decision-making process and the quality of the choice made [28]. In the first case, measures should explore if PtDAs help patients to: a) recognize that a decision needs to be made; 2) feel informed about the options (including risks, benefits, and consequences); 3) be clear about what matters most to them; 4) discuss goals, concerns, and preferences with their healthcare professionals; 5) be involved in decision-making. Considering the quality of the final choice implies to evaluate the extent to which patients are informed and receive treatments that are concordant with their goals and treatment preferences. In this sense, is useful to measure the patient's understanding of the information and the use of exercises to elicit preferences.

Clinical trials assessing the effectiveness of these tools versus standard practice have shown that PtDAs increase patients' knowledge, the proportion of people with accurate risk perceptions, the proportion of patients choosing an option congruent with their values, reducing decisional conflict related to feeling uninformed and feeling unclear about personal values, as well as the proportion of people who were passive in decision-making and who remained undecided post-intervention. PtDAs have also a positive effect on patient-healthcare professional communication, increasing the satisfaction with the decision-making process. It has also been observed that when patients are adequately informed about different therapeutic procedures with comparable effectiveness, they tend to choose the least invasive procedures and tend to start treatment earlier [25].

Empirical evidence for shared decision-making in rare diseases

Empirical studies about SDM or PtDAs interventions are still very scarce in the field of RDs. Hossler et al [29], in a pre-post uncontrolled study, analysed the acceptability of an interactive computerized decision aid to help engage patients with amyotrophic lateral sclerosis (ALS) in effective advance care planning. ALS patients perceived quantity and quality of information very positively, as it was the overall satisfaction with the PtDA and its accuracy in reflecting patients' wishes. The intervention prompted patients to discuss advance care planning with their families and to share their advance care directives generated by the software with their physicians. In other study, De Abreu et al [30] assessed the responses of patients with lupus nephritis and their physicians to a PtDA describing the treatment options and their potential benefits and risks. A significantly higher rate of physicians selected oral treatment options (96% vs 68%, $p < 0.001$). Decision justification was different for patients and clinicians in each group; risk of side effects and the risk/benefit trade-offs were more relevant for physicians, whereas risk potential and absence of prior joint involvement were the variables that predicted patients' choice of the oral option. Finally, Yazdani et al [31] published the preliminary results (meta-analyses of effectiveness results and focus groups) of the development of a PtDA for racial/ethnic minorities with lupus nephritis. The use of this tool is expected to facilitate patient-centred care in these cases.

In addition, some qualitative studies explored patients' views about SDM and their involvement in healthcare. Hanneman-Weber et al [32] published a multi-level empirical study protocol that uses a three phases' mixed method. The aim of the study was to assess the contribution of communication processes and SDM among healthcare teams, in order to improve the satisfaction of patients affected by ALS, Marfan's syndrome, Wilson's disease, epidermolysis bullosa, Duchenne muscular dystrophy, and neurodegeneration with brain iron accumulation. A year later, the first results were also published [33], analysing interaction experiences among 107 patients affected by the six mentioned RDs. Using semi-structured interviews, four interaction patterns were identified: paternalistic, collaborative, led by the patient and confrontational. This analysis also showed that professionals' lack of knowledge becomes a handicap that creates uncertainty and dissent within the highly specialized treatment process that any RD demands. In such cases, the patient becomes the expert in a way that he may lead the interaction with the professional. Regarding the willingness to change roles, this research and others suggest a higher resistance to SDM from professionals than from patients [34].

Patient satisfaction with the healthcare system might be a relevant variable for an adequate implementation of SDM in the field of RDs. Despite some studies provide information on expectations and satisfaction with healthcare services by patients affected by ALS [35, 36], there is a lack of valid knowledge explaining the basic psychosocial processes which support the way in which people with RDs cooperate or get involved with the healthcare providers. The systematic review carried out by Foley et al [37] concluded that ALS patients are often dissatisfied with healthcare services and have unmet expectations of their care. In order to shed light on this knowledge gap, a subsequent study by the same author [38] carried out in-depth interviews with 34 people affected by ALS, reporting that older participants had a wider acceptance of the disease and the idea of death than young or middle-aged patients. This study also observed that families play a relevant role in participants' commitment to healthcare services, as well as in decisions taken at different stages of life.

Discussion and conclusions

People with RDs, their caregivers and support organizations are among the groups with the greatest activity within the healthcare sector, mainly because of their own struggle looking for acknowledgement and answers that may improve their healthcare. Thus, it is crucial that health policy makers and professionals acknowledge the active and collaborative role of these groups, fostering SDM during the clinical process. Yet providing patients with a certain level of control to approach their disease might call for a change in traditional roles in healthcare services, as well as having adapted materials available to the users.

According to the revised literature, the field of RDs is indeed expanding, indicating political and social acknowledgement towards research and resources development that empower the patients involved. Nevertheless, despite studies that highlight the contribution of patients´ organizations and explore outcome measures that are essential for an adequate implementation of SDM, no specific PtDAs have been designed so far. Because of the available scientific evidence about the effectiveness of these tools, especially in the uncertain context that surrounds RDs, it is advisable to allocate resources to design and assess these PtDAs.

Recognition of the need of valid, reliable and accessible educational materials also requires designing, developing and assessing specific PtDAs for RDs, focused on the SDM between healthcare professionals and patients. The European Commission is promoting virtual communities or the provision of resources that support patient empowerment and shared decision-making. In this sense, the RareConnect project (www.rareconnect.org) gives support to patients´ communities with specific RDs, and has moderators that supervise the uploaded contents on the website. Other projects, such as the EURORDIS Summer School, offer face to face training programmes, but also offer access to freely available on-line modules, webcasts and slide presentations (www.eurordis.org).

In Europe, some webs could become a valid option to host informative resources and PtDAs. Orphanet (www.orpha.net) offers information about several RDs, available treatments and experts location. PatientsLikeMe (www.patientslikeme.com) is another example of health data-sharing platform where patients can share and learn from their experiences and outcome-based health data, and aligning patient and industry interests through data-sharing partnerships. Likewise, the web platform PyDeSalud (www.pydesalud.com), though currently deals mainly with high prevalence diseases, is also planning the development of PtDAs for Spanish speakers people affected by RDs. This website, whose contents are obtained from rigorous research and are supervised by a scientific experts committee, promotes the exchange of informative material which facilitate SDM and PtDAs. It also contains patients' experiences and encourages the participation of expert patients in determining and prioritizing research needs in healthcare agendas. In this way, narrative based medicine [39] helps SDM, acting as a bridge between the clinical knowledge of the physician and the patients' subjective experiences, and supporting the various stakeholders toward the improvement of knowledge and the shared management of the disease [40].

Consequently, PtDAs are expected to reduce the current level of uncertainty at the same time that support SDM, lightening the difficulties due to the lack of specialization required in the medical encounter with patients affected by RDs. Additionally, the creation of virtual communities of practice which host shared experiences coming both from clinical practice and patients, may contribute to improve the clinical and therapeutic data of this group of diseases [41].

In conclusion, it is advisable to increase support for actions aiming at the empowerment of people affected by RDs and their organizations, as well as allocating resources for research in SDM and the creation of PtDAs targeting these patients.

References

1. Taruscio D, Gentile AE, Evangelista T, Frazzica RG, Bushby K, Montserrat AM. Centres of Expertise and European Reference Networks: key issues in the field of rare diseases. The EUCERD Recommendations. Blood Transfus 2014;12(3):s621-5. Available from: http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4044812/
2. Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europes' Challenges, (2009/C 151/02). Available from: http://ec.europa.eu/health/rare_diseases/docs/2014_rarediseases_implementationreport_en.pdf
3. Griggs RC, Batshaw M, Dunkle M, Gopal-Srivastava R, Kaye E, Krischer J, et al. Clinical research for rare disease: opportunities, challenges, and solutions. Mol Genet Metab 2009;96(1):20-6. doi: 10.1016/j.ymgme.2008.10.003
4. Gagne J, Thompson L, O'Keefe K, Kesselheim A. Innovative research methods for studying treatments for rare diseases: methodological review. BMJ 2014;349:g6802. doi: http://dx.doi.org/10.1136/bmj.g6802
5. Légaré F, Ratté S, Gravel K, Graham ID. Barriers and facilitators to implementing shared decision-making in clinical practice: update of a systematic review of health professionals' perceptions. Patient Educ Couns 2008;73(3):526-35. doi: 10.1016/j.pec.2008.07.018
6. Faurisson F. Survey of the delay in diagnosis for 8 rare diseases in Europe: EurordisCare2. Available from: http://www.eurordis.org/IMG/pdf/Fact_Sheet_Eurordiscare2.pdf
7. Weston WW. Informed and shared decision-making: the crux of patient-centred care. CMAJ 2001;165(4):438-9. Available from: http://www.ncbi.nlm.nih.gov/pmc/articles/PMC81370/
8. Taruscio D, Morciano C, Laricchiuta P, Mincarone P, Palazzo F, Leo CG, et al. RARE-Bestpractices: a platform for sharing best practices for the management of rare diseases. Rare Dis Orphan Drugs 2014;1(1):5-8. Available from: http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4249596/
9. Hilton-Boon M, Ritchie K, Manson J, Taruscio D, Auld J, Uk J, et al. Improving the retrieval and dissemination of rare

disease guidelines and research recommendations: a RARE-Bestpractices initiative. Rare Dis Orphan Drugs 2014;1(1):20-9. Available from: http://rarejournal.org/rarejournal/article/view/33

10. Sejersen T, Del Giovane C, Filippini G, Leo CG, Meerpohl JJ, Mincarone P, et al. Methodology for production of best practice guidelines for rare diseases. Rare Dis Orphan Drugs 2014;1(1):10-9.
11. Marcus AD. Patients with rare diseases work to jump-start research; advocacy groups create their own tissue banks to aid in drug treatment. Wall Street Journal (East Ed) 2006;11:D1, D2. Available from: http://www.wsj.com/articles/SB115257685637802872
12. Barr PJ, Scholl I, Bravo P, Faber MJ, Elwyn G, McAllister M. Assessment of patient empowerment: a systematic review of measures. PLoS ONE 2015;10(5):e0126553. doi: 10.1371/journal.pone.0126553
13. Bravo P, Edwards A, Barr PJ, Scholl I, Elwyn G, McAllister M, the Cochrane Healthcare Quality Research Group, Cardiff University. Conceptualising patient empowerment: a mixed methods study. BMC Health Serv Res 2015;15:252. Available from: http://www.biomedcentral.com/1472-6963/15/252
14. Linertová R, Serrano-Aguilar P, Posada-de-la-Paz M, Hens-Pérez M, Kanavos P, Taruscio D, et al. Delphi approach to select rare diseases for a European representative survey. The BURQOL-RD study. Health Policy 2012;108(1):19-26. doi: 10.1016/j.healthpol.2012.08.001
15. Serrano-Aguilar P, Trujillo-Martin MDM, Pérez de la Rosa A, Cuellar-Pompa L, Saavedra-Medina H, Linertova R, et al. Patient participation in a clinical guideline development for systemic lupus erythematosus. Patient Educ Couns 2015;98(9):1156-63. doi: http://dx.doi.org/10.1016/j.pec.2015.05.022
16. Serrano-Aguilar P, Trujillo-Martín MM, Ramos-Goñi JM, Mahtani-Chugani V, Perestelo-Pérez L, Posada-de la Paz M. Patient involvement in health research: a contribution to a systematic review on the effectiveness of treatments for degenerative ataxias. Soc Sci Med 2009;69(6):920-5. doi: 10.1016/j.socscimed.2009.07.005.
17. Caron-Flinterman JF, Broerse JE, Bunders JF. The experiential knowledge of patients: a new resource for biomedical research? Soc Sci Med 2005;60: 2575-84. Available from: http://www.ncbi.nlm.nih.gov/pubmed/15814182
18. Crawford MJ, Rutter D, Manley C, Weaver T, Bhui K, Fulop N, et al. Systematic review of involving patients in the planning and development of health care. BMJ 2002;325:1263-67. Available from: http://www.ncbi.nlm.nih.gov/pubmed/12458240
19. Salzburg statement on shared decision making. BMJ 2011;342:d1745. doi: http://dx.doi.org/10.1136/bmj.d1745
20. Braddock CH. The emerging importance and relevance of shared decision making to clinical practice. Med Decis Making 2010;30(5):5S-7S. doi: 10.1177/0272989X10381344
21. Gafni A, Charles C, Whelan T. The physician-patient encounter: the physician as a perfect agent for the patient versus the informed treatment decision-making model. Soc Sci Med 1998;47(3):347-54.
22. Joseph-Williams N, Elwyn G, Edwards A. Knowledge is not power for patients: a systematic review and thematic synthesis of patient-reported barriers and facilitators to shared decision making. Patient Educ Couns 2014;94(3):291-309. doi: 10.1016/j.pec.2013.10.031
23. Charles C, Gafni A, Whelan T. Decision-making in the physician-patient encounter: revisiting the shared treatment decision-making model. Soc Sci Med 1999; 49(5):651-661. http://www.ncbi.nlm.nih.gov/pubmed/10452420
24. Coulter A, Stilwell D, Kryworuchko J, Mullen PD, Jenn C, van der Weijden T. A systematic development process for patient decision aids. BMC Med Inform Decis Mak 2013;13(Suppl 2):S2. doi: 10.1186/1472-6947-13-S2-S2
25. Stacey D, Légaré F, Col NF, Bennett CL, Barry MJ, Eden KB, et al. Decision aids for people facing health treatment or screening decisions. Cochrane Database Syst Rev 2014; 1:CD001431. Available from: http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD001431/abstract;jsessionid=B7B4B25864312A510DC3749B0BCFA651.f02t03
26. Elwyn G, Frosch D, Rollnick S. Dual equipoise shared decision making: definitions for decision and behaviour support interventions. Implement Sci 2009;4:75. doi:10.1186/1748-5908-4-75
27. Elwyn G, O'Connor A, Stacey D, Volk R, Edwards A, Coulter A, et al. Developing a quality criteria framework for patient decision aids: online international Delphi consensus process. BMJ 2006;333:417. doi: http://dx.doi.org/10.1136/bmj.38926.629329.AE
28. Sepucha K, Borkhoff C, Lally J, Levin C, Matlock D, Jenn C, et al. Establishing the effectiveness of patient decision aids: key constructs and measurement instruments. BMC Med Inform Decis Mak 2013;13(Suppl 2):S12. doi:10.1186/1472-6947-13-S2-S12
29. Hossler C, Levi BH, Simmons Z, Green MJ. Advance care planning for patients with ALS: feasibility of an interactive computer program. Amyotroph Lateral Scler 2011;12(3):172-7. doi: 10.3109/17482968.2010.509865
30. De Abreu MM, Gafni A, Ferraz MB. The use of a decision board to elicit Brazilian patients' and physicians' preferences for treatment: the case of lupus nephritis. Value Health 2011;14(5):S141-6. doi: 10.1016/j.jval.2011.05.015
31. Yazdany J, Feldman CH, Liu J, Ward MM, Fischer MA, Costenbader KH. Quality of care for incident lupus nephritis among Medicaid beneficiaries in the United States. Arthritis Care Res (Hoboken) 2014;66(4):617-24. doi: 10.1002/acr.22182
32. Hannemann-Weber H, Kessel M, Budych K, Schultz C. Shared communication processes within healthcare teams for rare diseases and their influence on healthcare professionals' innovative behavior and patient satisfaction. Implement Science 2011;6:40. doi: 10.1186/1748-5908-6-40
33. Budych K, Helms TM, Schultz C. How do patients with rare diseases experience the medical encounter? Exploring role behavior and its impact on patient-physician interaction. Health Policy 2012;105(2-3):154-64. doi: http://dx.doi.org/10.1016/j.healthpol.2012.02.018
34. Grut L, Kvam MH. Facing ignorance: people with rare disorders and their experiences with public health and welfare services. Scandinavian Journal of Disability Research 2012;15(1):20-32. doi:10.1080/15017419.2011.645870
35. Hughes RA, Sinha A, Higginson I, Down K, Leigh PN. Living with motor neurone disease: lives, experiences of services and suggestions for change. Health Soc Care Community 2005;13(1):64-74. doi: 10.1111/j.1365-2524.2005.00530.x
36. Gruis KL, Wren PA, Huggins JE. Amyotrophic lateral sclerosis patients' self-reported satisfaction with assistive technology. Muscle Nerve 2011;43(5):643-7. doi: 10.1002/mus.21951
37. Foley G, Timonen V, Hardiman O. Patients' perceptions of services and preferences for care in amyotrophic lateral sclerosis: a review. Amyotroph Lateral Scler 2012;13(1):11-24. doi: 10.3109/17482968.2011.607500
38. Foley G, Timonen V, Hardiman O. Acceptance and decision making in amyotrophic lateral sclerosis from a life-course perspective. Qual Health Res 2014;24(1):67-77. doi: 10.1177/1049732313516545

39. Charon R. Narrative medicine: a model for empathy, reflection, profession, and trust. JAMA 2001;286(15):1897-902. Available from: http://jama.jamanetwork.com/article.aspx?articleid=194300

40. Garrino L, Picco E, Finiguerra I, Rossi D, Simone P, Roccatello D. Living with and treating rare diseases: experiences of patients and professional health care providers. Qual Health Res 2015;25(5):636-51. doi: 10.1177/1049732315570116

41. Dwyer AA, Quinton R, Morin D, Pitteloud N. Identifying the unmet health needs of patients with congenital hypogonadotropic hypogonadism using a web-based needs assessment: implications for online interventions and peer-to-peer support. Orphanet J Rare Dis 2014;9:83. doi: 10.1186/1750-1172-9-83