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Becoming a research participant: Decision-making needs of individuals with neuromuscular diseases

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Abstract.
BACKGROUND: Research has shown that some people with neuromuscular diseases may have a lower level of education due to lower socioeconomic status and possibly compromised health literacy. In view of these data, it appears important to document their decision-making needs to ensure better support when faced with the decision to participate or not in research projects.
OBJECTIVES: 1) To document the decision-making needs of individuals with neuromuscular diseases to participate in research; 2) To explore their preferences regarding the format of knowledge translation tools related to research participation.
METHODS: This qualitative study is based on the Ottawa Decision Support Framework. A two-step descriptive study was conducted to capture the decision-making needs of people with neuromuscular diseases related to research participation: 1) Individual semi-directed interviews (with people with neuromuscular diseases) and focus groups (with healthcare professionals); 2) Synthesis of the literature.
RESULTS: The semi-directed interviews ($n = 11$), the two focus groups ($n = 11$) and the literature synthesis ($n = 50$ articles) identified information needs such as learning about ongoing research projects, scientific advances and research results, the potential benefits and risks associated with different types of research projects, and identified values surrounding research participation: helping other generations, trust, obtaining better clinical follow-up, and socialization.
CONCLUSION: This paper provides useful recommendations to support researchers and clinicians in developing material to inform individuals with neuromuscular diseases about research participation.

Keywords: Research participation, neuromuscular diseases, patient needs, patient engagement, shared-decision making

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1. Background

Since the early 2000's, research interest into populations with neuromuscular diseases has increased [1]. Among all the existing rare diseases, neuromuscular diseases are among the most frequent. In Canada, people with neuromuscular diseases are often referred to a neuromuscular or movement disorders university-affiliated clinic. The Saguenay-Lac-Saint-Jean university-affiliated neuromuscular clinic is one of the largest clinics in Canada who follows this population. This region has the highest prevalence of myotonic dystrophy type 1 (DM1) and autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSAC) worldwide and one of the largest cohorts of oculopharyngeal muscular dystrophy (OPMD) affected individuals. In relation to the latter, a university-affiliated research team has grown over the years and set up their research facilities within the *Groupe de recherche interdisciplinaire sur les maladies neuromusculaires*. As a result, an interdisciplinary research program has been ongoing since 2006 with multifaceted research activities (e.g., natural history studies, international registries, biomarkers identification, clinical trials, development and implementation of clinical practice guidelines and interventions, documentation of outcome measures metrological properties) [2]. Research has shown that some people with neuromuscular diseases may have a lower level of education due to lower socioeconomic status [3] and possibly compromised health literacy [4]. In view of these data, it appears important to document the decision-making needs of people with neuromuscular diseases to ensure better support for their decision to participate in research projects.

The objectives of this research are: 1) to document the decision-making needs of adult individuals with neuromuscular diseases in regard to participation in research activities; 2) explore preferences in format of knowledge translation tools related to their participation in research activities.

2. Methods

This study was based on the Ottawa Decision Support Framework [5]. The descriptive qualitative design with individual semi-directed interviews (with individuals with neuromuscular diseases) and focus groups (with healthcare professionnals) was chosen for its flexibility in the topics discussed by participants [6, 8]. For the individual interviews, the three nurse case managers at the *Groupe de recherche interdisciplinaire sur les maladies neuromusculaires* identified potential participants from their caseload. Participants with neuromuscular diseases were selected according to pre-established criteria to obtain a representative sample of affected individuals related to personal characteristics [9] (e.g., diagnosis of neuromuscular disease, age, sex, education) and representativeness of level of exposure to research activities (e.g., questionnaire or biopsy). Individuals who had severe impairments related to cognition or verbal communication, or an inability to consent were excluded. An initial contact was made by their nurse case manager to obtain participant consent to be contacted by the research team.

The formal recruitment was done by a trained research professional. Participants completed a sociodemographic questionnaire. For the focus groups, participants had to have more than two years work experience with neuromuscular disease. In addition, at least three types of healthcare professions had to be represented (e.g., nurse, physiotherapist, neurologist). Interviews and focus groups were approximately one-hour long. Both interview guides had the same content but language was adapted to each group's level of literacy.

A descriptive study was conducted to define decision-making needs (information needs and values) [6]. This design helps to guide the actions that need to be taken when little information exists in a specific context [7]. The study was conducted with patient partners as co-investigators on the research team. Patient partners were trained by our research team and with the Quebec SPOR-Unit, prior to

the project to become familiar with the research process, data collection and interview method. They participated in the creation of the project, in the co-animation of the interviews and in the analysis of the results. The study was carried out in two steps: 1) semi-directed individual interviews (people with neuromuscular disease) and focus groups (healthcare professionals and 2) synthesis of the literature regarding the information needs and values of people with neuromuscular disease related to their participation in research activities.

For step 1, the research team, including patient partners, determined the themes of the interview guide to cover the three main categories of the Ottawa Decision Support Framework: 1) Decisional Needs; 2) Decisional Outcomes; 3) Decision Support [5]. In this study, decisional needs included: decision type/timing, decisional conflict, clinical needs, values, lack of knowledge, support and resources. Decisional outcomes included: quality of the decision based on knowledge of the different options and the values linked to these options. Decision support included knowledge translation tools and significant persons (family, caregivers, healthcare professionals team). All interviews were recorded and transcribed verbatim in order to perform a qualitative inductive interpretive analysis [9]. The themes included: 1) Knowledge about the clinical, teaching, and research missions of the *Groupe de recherche interdisciplinaire sur les maladies neuromusculaires*; 2) Knowledge about the types of research projects and clinical services offered by the clinic; 3) Context of their attendance (e.g., research and care experiences); 4) Knowledge of the issues related to research participation. To help the discussion around knowledge translation tools, we provided the context for the creation of information and decision-making tools before asking questions on the content, format, and use of these tools including: 5) Information decision-making needs; 6) Research value decision-making needs; 7) Tool format. To lessen the interviewer's influence on the participants' point of view, the interviewer used the reformulation technique throughout the interviews. The same process was used for the two focus groups.

After data collection, the themes were entered into the NVivo 11 Software [10]. An extraction grid was constructed based on the themes to proceed to the synthesis of the literature. An in-depth reading of the content of the individual interviews and focus groups was done by three members of the research team. Then, emerging themes were classified according to the three predetermined categories of the Ottawa Decision Support Framework (Decisional Needs, Decisional Outcomes, Decision Support). Subthemes for each category were determined inductively afterwards. The themes were then reviewed by the entire research team and proposals were made regarding the emergence of new themes in relation to the Ottawa Decision Support Framework.

For step 2, the general question that guided the literature synthesis was: *What are the decision-making needs of individuals with neuromuscular disease regarding information needs and values related to their participating in research activities?* Results from step 1 supported the selection of the main concepts for the literature synthesis. An extraction grid was constructed by a research professional validated by the research team before data extraction. The search strategy was divided into three main concepts: 1) Person with a rare genetic disease or neuromuscular disease; 2) Participation in research; 3A) Values associated with participation in research; 3B) Information needs related to participation in research. The principal keywords were rare disease, neuromuscular diseases, patient participation, patient involvement, patient empowerment, patient preference, patient experience, patient values, information, registry, clinical trial, and biobank. This literature synthesis was conducted by one reviewer using the Cochrane method for rapid review and included: a) A comprehensive literature search in databases; b) Pre-defined inclusion and exclusion criteria (i.e., study eligibility criteria); c) Discussion about the limitations of included studies [11]. A comprehensive literature search in four databases was performed (Pubmed, CINAHL, PSYCHInfo, and EMBASE) and bibliographic references were also consulted from selected articles. To be included articles had to be written in French or English within the last 10 years. One member of the research team (the research professional that conducted

the qualitative analysis) carried out a first reading of the abstracts followed by a full reading of the articles if they were included. A second person confirmed article selection (principal investigator).

The project was approved by the *Comité d'éthique de la recherche du Centre intégré universitaire de santé et des services sociaux du Saguenay–Lac-Saint-Jean*. The experiments were undertaken with the understanding and written consent of each subject, and the study conforms with The Code of Ethics of the World Medical Association (Declaration of Helsinki), printed in the British Medical Journal (18 July 1964).

3. Results

This study was conducted among eleven individuals with neuromuscular diseases, four healthcare and social services professionals, and seven research-related professionals. For participants with neuromuscular disease, four individuals had DM1, three had OPMD and four had ARSACS. Six were men and five were women. Six had completed high school. Six had already participated in research activities. Health and social service professionals had significant work experience (more than three years) with people with neuromuscular disease and represented at least three different types of professions (doctor, nurse case manager, social worker). As for research-related professionals, they had at least two years of work experience at the *Groupe de recherche interdisciplinaire sur les maladies neuromusculaires*, significant work experience with individuals with neuromuscular disease, and included researchers, research professionals, master or doctoral students involved in research (see Table 1).

3.1. Step 1: Descriptive qualitative design

The results are presented according to the three main concepts of the Ottawa Decision Support Framework: 1) Decisional Needs; 2) Decisional Outcomes; 3) Decision Support [5].

3.2. Decisional needs

Decisional needs, refer to different levels of information that affected individuals with the same diagnosis have about their own disease. Experience with research activities at the *Groupe de recherche*

Table 1
Participant's characteristics

Code	Sexe	Age (years)	Disease	High school education	Prior research participation
01	W	50–60	ARSAC	No	Yes
02	W	30–40	ARSAC	Yes	Yes
03	W	30–40	ARSAC	Yes	Yes
04	M	60–70	OMPD	No	No
05	M	70–80	OMPD	Yes	No
06	W	50–60	OMPD	Yes	No
07	W	50–60	ARSAC	Yes	No
08	M	50–60	DM1	No	Yes
09	M	30–40	DM1	No	Yes
10	M	30–40	DM1	Yes	Yes
11	M	30–40	DM1	No	Yes

interdisciplinaire sur les maladies neuromusculaires is different according to the neuromuscular disease (DM1, OPMD, ARSACS). Since the early 2000 s, individuals with DM1 have been asked to participate in research activities. Many shared that the more they participated, the more comfortable they felt participating again because they have more information about ongoing research on their disease. However, a structured research program on OPMD and ARSACS has only recently been put in place. One individual said:"*There are diseases where there is no research culture (...). ARSACS or OPMD, there is no research. This is new. They don't know what to expect. It is a culture that is not rooted, compared to other diseases, such as myotonic dystrophy (...). They have been doing intensive research with them since 2002.*" *(Group2Research)*

3.3. Decisional outcomes

Decisional outcomes refer to the values associated with research participation for all affected individuals. All participants strongly emphasized the desire to help others with the disease. For example, many of them talked about helping future generations and being part of the solution (feeling of pride), and not part of the problem because of the genes they passed on to their children (feeling of guilt). Many wanted to give everything to research (e.g., compensation, donation, access to their medical files, test results). Some expressed an interest in participating in projects that address all neuromuscular disease, while others wished to participate only in projects that address their own disease.

> "*To advance research. Yes, they found the gene, but it has to continue. Maybe it's too late for us, but, for the new ones, it will be convenient to know that they can find a pill or medication.*" *(P-ARSACS)*

In terms of decision-making needs related to information needs, some themes need further exploration, including knowledge of scientific advances related to their disease and the need to receive detailed information about their participation in a research project when the level of risk is higher (for example, muscle biopsy).

Information needs differed, namely the need to know their own individual results from their participation in research activities (progression, or in comparison to normative data), the amount of information and level of detail.

> "*In my case, I don't know where the research is at, either. These are research programs. Researchers have results; they sometimes try to tell us how their results work, but it's not clear.*" *(P-DM1)*

3.4. Decision support

Regarding values associated with the decision to participate in research or not, similarities were found, including the importance of having a trust-based relationship with the *Groupe de recherche interdisciplinaire sur les maladies neuromusculaires* healthcare professionals. This trusting relationship helps them feel free to ask questions. They also feel more comfortable knocking on the professional's door when they are welcoming and interested in their wellbeing. The trust between individuals with neuromuscular disease and research professionals, combined with strong collaboration between neuromuscular clinical healthcare professionals, encourages recruitment for research.

> "*The feeling of relation and belonging is very strong (...). They feel that we need them. They feel appreciated. We have always taken care of our patients (...). We call them. They call us (...). We have developed a relationship of trust. That's why you don't have any difficulty recruiting. There is continuity.*" *(Group 1 Clinicians)*

For information comprehension and retention, participants with DM1 expressed the need to have the same information repeated across many knowledge translation tools and more graphical representation

of the concepts associated with the different research activities. Indeed, specific projects are often not clear to them, and they appreciate practical examples to which they can relate to, to get the essence of their participation and goals of the research activities proposed to them. Often, research projects on physical exercise or nutrition are more tangible for them. Individuals with OPMD expressed the need for more specific details in the information provided to them. Various media were discussed for the creation of knowledge translation tools to help the decision-making process. No consensus was reached on any, and media preferences included iPads, videos, websites including social networks like Facebook, posters or leaflets.

> "*There would be an advertisement that would be interesting to do (or) a DVD that would (play in) the waiting room.* '*You have myotonic dystrophy. Do you know that there is such a thing? (We are having) a study.*' *The papers, I don't look at it. But I keep them all in an envelope. If I have a problem, I'll be able to look at them. But I don't read them; I trust you.*" *(P-DM1)*

3.5. Step 2: Synthesis of the literature

As the studies reviewed addressed different research activities, presentation of the results was divided into the four most common research activities in the area of neuromuscular disease: 1) registries ($n = 8$); 2) clinical trials ($n = 17$); 3) biobanks ($n = 13$) and 4) academic research projects ($n = 12$) (see Fig. 1 and Table 2).

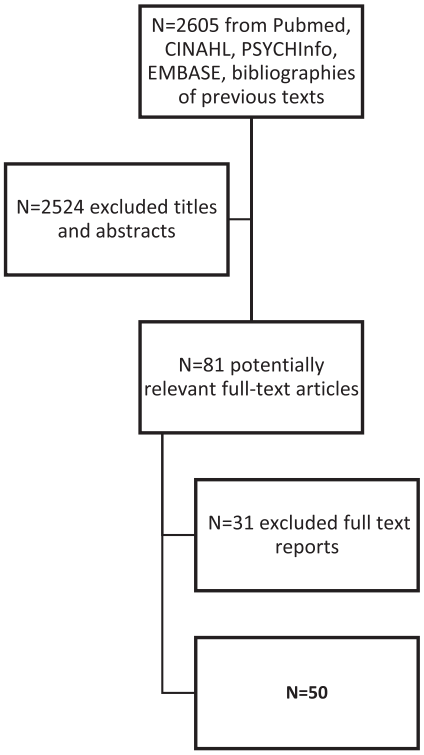

Fig. 1. Results of literature review.

Table 2
Results details of synthesis of the literature ($n = 50$)

	Study			Population			
#	Sources	First author, years, Country	Title	Design study	Age	Sex	Diagnostic
1	PUBMED	Woodward, 2016, (Belgium, France, Italy, Russia, Spain and UK)	An innovative and collaborative partnership between patients with rare diseases and industry-supported registries: the Global aHUS Registry	Quantitative descriptive	Unknown	All	Rare diseases
2	PUBMED	Coathup, 2016, Japan	Using digital technologies to engage with medical research: views of myotonic dystrophy patients in Japan	Quantitative descriptive	18 +	All	Myotonic dystrophy
3	PUBMED	Gupta, 2011, World Organization	Strategies for Improving Identification and Recruitment of Research Participants	Unknown	Unknown	Unknown	Rare Lung disease
4	PUBMED	Kirkpatrick, 2015, USA	GenomeConnect: Match-making Between Patients, Clinical Laboratories, and Researchers to Improve Genomic Knowledge	Unknown	Unknown	Unknown	Genetic disease
5	Previous texts	Workman, 2013, USA	Engaging Patients in Information Sharing and Data Collection: The Role of Patient-Powered Registries and Research Networks	Qualitative	Unknown	All	$Patients_{SEP}$ with many types of diseases
6	Previous texts	EUCERDEMA, 2011, European	Towards a public-private partnership for registries in the field of rare diseases	Qualitative	Unknown	All	Rare diseases
7	Previous texts	Whiddette, 2005, Australia	Patients' attitudes towards sharing their health information	Quantitative descriptive	18 +	All	Adult primary-care patients
8	Previous texts	Schwartz, 2005, USA	A Patient Registry for Cognitive Rehabilitation Research: A Strategy for Balancing Patients' Privacy Rights With Researchers' Need for $Access_{SEP}$	Quantitative descriptive	16 and +	All	Stroke or traumatic brain injury
9	PUBMED	Bardach, 2018, USA	Motivators for Alzheimer's Diseases Clinical Trial Participation	Quantitative descriptive	54.6–89.8	All	Alzheimer's diseases

(*Continued*)

Table 2
(*Continued*)

Study				Population		
#	Sources	First author, years, Country	Title	Design study	Age	Sex	Diagnostic
10	PUBMED	Biedrzycki 2011, USA	Research Information Knowledge, Perceived Adequacy, and Understanding in Cancer Clinical Trial Participants	Quantitative descriptive	18 +	All	Gastrointestinal cancer
11	PUBMED	van der Biessen, 2018, Netherlands,	Understanding how coping strategies and quality of life maintain hope in patients deliberating phase I trial participation	Quantitative descriptive	18 +	All	Incurable cancer
12	PUBMED	Carroll, 2012, USA	Motivations of patients with pulmonary arterial hypertension to participate in randomized clinical trials	Qualitative	All	All	Pulmonary arterial hypertension
13	PUBMED	Godskesen, 2016, Sweden	Differences in trial knowledge and motives for participation among cancer patients in phase 3 clinical trials	Quantitative descriptive	18 +	All	Cancer patients
14	PUBMED	Godskesen, 2014, Sweden	Hope for a cure and altruism are the main motives behind participation in phase 3 clinical cancer trials	Quantitative descriptive	18 +	All	Cancer patients
15	PUBMED	Grill, 2013, USA	Risk disclosure and preclinical Alzheimer's diseases clinical trial enrollment	Quantitative random-ized	+ 46	All	Cognitive normal
16	PUBMED	Henrard, 2015, Belgium	Participation of people with haemophilia in clinical trials of new treatments: an investigation of patients' motivations and existing barriers	Quantitative descriptive	18 +	All	Adults with haemophilia
17	PUBMED	Lawrence, 2014, England	Patient and carer views on participating in clinical trials for prodromal Alzheimer's diseases and mild cognitive impairment	Qualitative	18+	All	Alzheimer's
18	PUBMED	Mancini, 2010, France	Participants' uptake of clinical trial results: a randomised experiment	Quantitative random-ized	18+	Women	HER2-positive non-metastatic breast cancer
19	PUBMED	Ssali, 2015, Africa	Volunteer experiences and perceptions of the informed consent process: Lessons from two HIV clinical trials in Uganda	Qualitative	18–40	All	HIV sero status

20	CINAHL	Dellson, 2016, Sweden	Patient representatives' views on patient information in clinical cancer trials	Qualitative	51–77	All	Colorectal cancer
21	CINAHL	DeWard, 2014 (USA, Canada)	Practical Aspects of Recruitment and Retention in Clinical Trials of Rare Genetic Diseases: The Phenylketonuria (PKU) Experience	Unknown	Unknown	Unknown	Rare diseases
22	CINAHL	Gaasterland, 2019, European	The patient's view on rare diseases trial design – a qualitative study	Qualitative	Unknown	Unknown	Rare diseases
23	CINAHL	Holman, 2010, USA	Patient-derived Determinants for Participation$_{SEP}$in Placebo-controlled Clinical Trials for Fibromyalgia	Quantative descriptive	18 +	All	Fibromyalgia
24	PUBMED	Dorcy, 2011, USA	I Had Already Made Up My Mind": Patients and Caregivers' Perspectives on Making the Decision to Participate in Research at a U.S. Cancer Referral Center	Qualitative	18 +	All	Cancer Hematopoietic cell transplants
25	Previous texts	Kinder, 2010, USA	Predictors for clinical trial participation in the rare lung diseases lymphangioleiomyomatosis	Quantitative descriptive	Age mean 53	Unknown	Rare lung diseases lymphangioleio-myomatosis
26	CINAHL	Cervo, 2013, Italy	An effective multisource informed consent procedure for research and clinical practice: an observational study of patient understanding and awareness of their roles as research stakeholders in a cancer biobank	Quantitative descriptive	18 +	All	Cancer
27	CINAHL	Fleming, 2015, Australia	Attitudes of the general public towards the disclosure of individual research results and incidental findings from biobank genomic research in Australia	Quantitative random-ized	18+	All	Patients with potential genetic risk
28	Previous texts	Eisenhauer, 2017, World	Participants' Understanding of Informed Consent for Biobanking: A Systematic Review	Systematic Review	Unknown	Unknown	Patients$_{SEP}$with many types of diseases
29	PUBMED	Toccaceli, 2014, Italy	Attitudes and Willingness to Donate Biological Samples for Research Among Potential Donors in the Italian Twin Register	Quantative descriptive	18 +	All	Twins with many types of diseases

(*Continued*)

Table 2
(*Continued*)

	Study			Population			
#	Sources	First author, years, Country	Title	Design study	Age	Sex	Diagnostic
30	Previous texts	Teare, 2015, England	Towards 'Engagement 2.0': Insights from a study of dynamic consent with biobank participants	Qualitative	Unknown	Unknown	Musculo-skeletal diseases Diabetes Cancer
31	Previous texts	Lemke, 2010, USA	Public and Biobank Participant Attitudes toward Genetic Research Participation and Data Sharing	Qualitative	18+	All	Patients$_{SEP}$ with many types of diseases
32	Previous texts	Allen, 2011, Australia	Reconsidering the value of consent in biobank research	Qualitative	54–80	All	Cancer
33	Previous texts	Johnsson, 2010, Sweden, Iceland, UK, Ireland, USA	Hypothetical and factual willingness to participate in biobank research	Unknown	Unknown	Unknown	Patients$_{SEP}$ with many types of diseases
34	Previous texts	Michie, 2011, USA	If I Could in a Small Way Help": Motivations for and Beliefs about Sample Donation for Genetic Research	Mixed methods	18+	All	Colorectal cancer
35	Previous texts	Mahnke, 2014, USA	A Rural Community's Involvement in the Design and Usability Testing of a Computer-Based Informed Consent Process for the Personalized Medicine Research Project	Mixed method	Unknown	Unknown	Rural community
36	Previous texts	Mancini, 2011, France	Consent for Biobanking: Assessing the Understanding and Views of Cancer Patients	Quantative descriptive	18 +	All	Colorectal cancer, breast cancer, hematological malignancy
37	Previous texts	Rahm, 2013, USA	Biobanking for research: a survey of patient population attitudes and understanding	Quantitative descriptive	18+	All	Patients$_{SEP}$ with many types of diseases

38	Previous texts	Toccaceli, 2009, Italy	Research understanding, attitude and awareness towards biobanking: a survey among Italian twin participants to a genetic epidemiological study	Quantitative descriptive	18+	All	Twins with many types of diseases
39	PUBMED	De Freitas, 2017, World	Public and patient involvement in needs assessment and social innovation: a people-centred approach to care and research for congenital disorders of glycosylation	Qualitative	Unknown	All	Congenital disorders of glycosylation
40	PUBMED	McGrath-Lone, 2015, England	Exploring research participation among cancer patients: analysis of a national survey and an in-depth interview study	Qualitative	18+	Women	Breast cancer patients
41	PUBMED	Mascalzoni, 2017, Europe	The Role of Solidarity(-ies) in Rare Diseases Research	Unknown	Unknown	Unknown	Rare diseases
42	PUBMED	Sacristán, 2016, Europe	Patient involvement in clinical research: why, when, and how	Unknown	Unknown	Unknown	Patients$_{SEP}$with many types of diseases
43	Embase	Coley, 2018, Finland, France, and the Netherlands.	Older Adults' Reasons for Participating in an eHealth Prevention Trial: A Cross-Country, Mixed-Methods Comparison	Mixed methods	+65 years old	All	Older adults
44	CINAHL	Chung, 2018, USA	Crohn's and Colitis Foundation of America Partners Patient-Powered Research Network$_{SEP}$Patient Perspectives on Facilitators and Barriers to Building an Impactful Patient-Powered Research Network	Qualitative	18 +	All	Chronic inflammatory diseases
45	CINAHL	Gysels, 2012, USA,$_{SEP}$UK$_{SEP}$, Australia$_{SEP}$	Patient, caregiver, health professional and researcher views and experiences of participating in research at the end of life: a critical interpretive synthesis of the literature$_{SEP}$	Synthesis of the literature$_{SEP}$	Unknown	Unknown	Patients end of life
46	CINAHL	Pollock, 2017, UK	Patient and researcher perspectives on facilitating patient and public involvement in rheumatology research	Unknown	Unknown	Unknown	Rheumatology diseases

(*Continued*)

Table 2
(*Continued*)

	Study			Population			
#	Sources	First author, years, Country	Title	Design study	Age	Sex	Diagnostic
47	Previous texts	Forsythe, 2014, USA	A Systematic Review of Approaches for Engaging Patients for Research on Rare Diseases	Systematic Review	Unknown	Unknown	Patients$_{SEP}$ with many types of diseases
48	PUBMED	Damman, 2016, Netherlands	Making comparative performance information more comprehensible: an experimental evaluation of the impact of formats on consumer understanding	Quantitative random-ized	18+	All	Patients$_{SEP}$ with many types of diseases
49	Embase	Ottman, 2018, USA, Argentina, Canada, Australia	Return of individual results in epilepsy genomic research : A view from the field.	Qualitative	18+	All	Epilepsy
50	Previous texts	Bendixen, 2016, USA	Engaging participants in rare diseases research: a qualitative study of duchenne muscular dystrophy	Qualitative	18+	All	Duchenne muscular dystrophy

3.6. Registries

The selected articles focused mainly on the importance of the active involvement of individuals with rare diseases in research (e.g., being more than a patient, becoming a partner by making their voices heard in decision-making) [12–14]. The main reasons for agreeing to participate in a registry study are to help family members, to help other individuals with rare diseases to improve their own health condition, or to improve care and services [12, 15]. Trust in the registries and healthcare professionals involved must be developed with individuals with rare diseases and their families, by actively engaging them in research and information sharing (e.g., ongoing studies and opportunities to participate, showing gratitude for their input) [14, 16, 17]. Their data in the registries should be kept confidential [16, 18]. These data should only be available to certain entities such as their healthcare team and researchers but not private insurance companies [16] (see Table 2).

3.7. Clinical trials

The topics covered in the selected articles on clinical trials focused mainly on individual benefits for trial participants [19–32]. For these people, their participation in research on their disease is the only option to improve their condition, sometimes even their last possible choice [19–21, 23, 26–29, 31]. Participating in a clinical trial allowed them to obtain additional healthcare services (especially for those without access to insurance) [19–28, 30, 32]. It also seemed very important to them that their participation in studies would benefit their loved ones (e.g., future generations in their families) [19–21, 23, 24, 27, 28, 30, 32]. Alternatively, the higher the risk of clinical trials (e.g., side effects), the more reluctant they were to participate [19, 21, 24, 25, 32]. They did not wish to be part of a control group [21, 25, 32]. According to them, therapeutic trials sometimes involved invasive procedures (e.g., biopsy) [19] which can cause pain [23] or can deteriorate their health for multiple reasons (e.g., discontinuation of current medication, drug interactions, decreased quality of care, increased testing) [21, 23, 30, 32–34]. This is the reason why their relationship with professionals and their trust in them, are very important [21, 24, 26, 28, 29, 32, 35]. To facilitate comprehension, they suggest structuring the information provided to them (e.g., graphics and images, underscoring, highlighting, bolding, and underlining text, using various colours, sections, and text sizes, as well as Braille, headers, first-impression appeal, meeting agendas, checklists) [27]. Finally, the preferred methods for providing information were through face-to-face meetings (allowing time for discussion and questions) [23, 25, 27, 28, 31, 34], websites and social media [28, 29, 33, 34], pamphlets and letters [25, 28, 33, 34], groups [34], conferences [29], videos [28], and phone calls [34] (see Table 2).

3.8. Biobanks

Reasons for participating in research centered mainly on helping others, including future generations [36–43], advancing research knowledge to improve services [36, 38, 39, 41–44] and helping oneself [36, 38, 40–42]. Individuals with rare diseases interviewed in these studies also emphasized the importance of establishing a trusting relationship with those involved in the creation and application of biobanks with knowledge of their credibility and sources [38–40, 44, 45]. According to them, biobanks can contribute to the common good by reducing health costs [37–40, 45]. However, they pointed out that their data must remain confidential [42, 44, 46]. Others highlighted the desire to obtain information on the risks associated with their participation in a biobank when collecting biological materials (e.g., injuries, pain) [38, 42, 46, 47], the benefits [36, 39, 46], and the results of their participation [42, 46, 47]. Some wanted to know if there are rules for sharing and accessing biological materials and other data [38, 46, 48], including what are the penalties if researchers do not respect confidentiality [38, 44,

46]. They would prefer being able to decide who can and cannot access their data [38, 44, 46]. In terms of how to provide information, the studies emphasized that their participants preferred that the content be made accessible to the general public according to their literacy and educational level [37, 38, 40, 46]. Repeating information and maintaining constant communication with research professionals are some of the proposed strategies [40, 44, 46]. Many emphasized the importance of having access to a person who is comfortable with providing information [44] and who is also able to answer their questions [37, 38, 40, 45, 46, 48] (see Table 2).

3.9. Academic research projects

The themes covered in the collected texts on academic research (e.g., qualitative with interviews, longitudinal) regarding research values important to individuals with rare diseases focused mainly on the trustworthiness of the information sources [49–54]. The willingness of researchers to give back to individuals with rare diseases [50], to express their gratitude to them [54, 55], to not being cold and distant [55], to promote solidarity (e.g., sharing information) in order to avoid competitiveness (e.g., retention of research results) [50], in developing a research culture accessible for all [55], in participants not feeling used for career advancement purposes by researchers [55] and confidentiality [50, 53, 56], are winning strategies, in their opinion, to help build trust among individuals with rare diseases. Participating in research is a way for individuals with rare diseases to obtain better treatment (e.g., more treatments, detection before deterioration, prevention, improvement of quality of life) and to develop new knowledge [51–53]. Regarding the information that individuals with rare diseases want to receive, the main topic discussed was benefits (e.g., reducing feelings of guilt, helping, socializing) [49–51, 57–58]. They want to receive the results of the study [50, 51, 58, 59]. They also want to receive information on risks before accepting to participate (e.g., anxiety about the results that may affect them regarding their disabilities) [49, 51], eligibility criteria [49, 54], goals and steps [51] and no impact quitting options [51]. Regarding how information is provided, studies emphasized that individuals with rare diseases preferred to receive information face-to-face [50, 51, 54, 55, 58, 59]. When discussing other means of disseminating information, several mentioned technology-based media (e.g., email, Skype, websites) [52–54, 56, 57, 59], while some mentioned more traditional media (e.g., flyers, poster) [51, 57, 59] (see Table 2).

4. Discussion

This study explored the decision-making needs (information and values) of individuals with neuromuscular disease when deciding to participate in research activities. Data revealed the following observations: first, they need to know more about research opportunities. Also, helping other generations with neuromuscular disease is an important part of the decision to participate in research. Finally, the creation of knowledge transfer tools to support the decision-making process on patient's engagement in a research project is also an important part of the decision to participate in research.

Regarding decision-making needs, several individuals with neuromuscular diseases highlighted the need to know more about opportunities to participate in ongoing projects and about scientific advances in the area of their disease. Individuals expressed a desire to know more about the importance of research participation to advance knowledge (e.g., scientific, clinical), and the potential benefits for themselves and other individuals with the same disease. As presented in the results, the development of research on DM1 is more advanced than on OPMD and ARSACS. Some patients have already participated in research projects. This is why it seems important to develop knowledge transfer tools, specific for each disease in relation to scientific advances. As Farha et al. (2020) emphasize, knowledge

can influence the willingness of patients to participate in research. Lavoie et al. (2020) add that DM1 patients can choose the knowledge transfer tool that matches their literacy level. It therefore seems important to focus on the vulgarization of all research documents in order to promote recruitment.

Regarding decisional outcomes, all patients highlighted the importance of helping other generations. Contributing to scientific progress and improved practices were themes promoted. Being part of the solution, not the problem, is an important element for individuals because many of them live with the guilt of being a carrier of an inherited disease. Helping others was a theme observed in recent literature [61, 62]. This aspect is important to highlight, because it can strongly influence a person's decision to participate in a research project. Given that there is no cure for DM1, OPMD or ARSACS and very few ongoing clinical trials, a person's participation in research may have more of an impact for the next generations than for themselves.

When creating knowledge transfer tools, it is important to take into account the specific needs of individuals with neuromuscular disease to support information retention using several methods, including repetition. They also suggested using several reminders (e.g., diary, calendar, phone call by a professional they know and trust). Recent studies highlighted the importance of involving healthcare professionals who are close to patients in knowledge transfer to build trust [61–63]. A trusting relationshipis very important and the strength of the Saguenay-Lac-Saint-Jean university-affiliated neuromuscular clinic is that the team (e.g., nurses, doctors, physiotherapists, social workers) ensures patient monitoring from birth to death. This contributes to the maturity of the relationship which promotes recruitment and participation in research projects (64). For example, some authors recommend highlighting items on documents that seem important to patients (65). To encourage dissemination, they proposed to involve individuals with neuromuscular disease in the creation of knowledge translation tools. Moreover, as Lavoie et al. (2020) underline, they observed that diversity of format makes it possible to respond to patient preference style of experiential learning. For the transmission of information in digital form, the study by Coathup et al. (2016) underlines that DM1 patients complain about not receiving enough medical information and they would be ready to receive more through digital tools. Especially videos on IPAD for patients with compromised health literacy to avoid abstraction [5].

5. Conclusions

One of this study's major strengths is the involvement of patient partners in documenting reviews, co-leading interviews and in data analysis to better target priority areas to be addressed. In addition, focus groups with professionals from the research community and the healthcare services allowed to expand the interdisciplinary expertise involved in the project. A limitation of the study is related to the sampling procedure that could lead to an ascertainment bias: 1) limited number of affected people recruited; 2) few rare disorders covered; 3) all followed at a specialty center. The literature review resulted in a significant amount of information collected regarding the needs of individuals with neuromuscular diseases related to various research activities (e.g., registries, clinical trials, biobanks). The literature review found that few articles take into account the concept of shared decision-making in research for individuals with neuromuscular diseases. Future studies could examine the impact of shared decision-making on research participation for individuals with neuromuscular diseases and their families. This paper provides useful recommendations to support researchers and clinicians in the development of material to inform individuals with neuromuscular diseases about research participation. Future studies could document the effectiveness of these recommendations to improve patients' understanding of research activities and support their decision-making process.

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Authors' contributions

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Conflict of interest

The authors declare that they have no conflict of interest.

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