



Understanding Rare Disease Experiences Through the Concept of Morally Problematic Situations

Ariane Quintal^{1,2,3} · Élissa Hotte^{1,3} · Caroline Hébert³ · Isabelle Carreau³ · Annie-Danielle Grenier³ · Yves Berthiaume^{3,4} · Eric Racine^{1,2,3,5}

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Abstract

Rare diseases, defined as having a prevalence inferior to 1/2000, are poorly understood scientifically and medically. Appropriate diagnoses and treatments are scarce, adding to the burden of living with chronic medical conditions. The moral significance of rare disease experiences is often overlooked in qualitative studies conducted with adults living with rare diseases. The concept of morally problematic situations arising from pragmatist ethics shows promise in understanding these experiences. The objectives of this study were to (1) acquire an in-depth understanding of morally problematic situations experienced by adults living with rare diseases in the province of Québec and (2) to develop an integrative model of the concept of morally problematic situations. To this end, an online survey targeting this population was developed through a participatory action research project. Respondents provided 90 long testimonies on the most important morally problematic situations they faced, often in healthcare settings. An integrative model was developed based on various qualitative analyses of these testimonies and relevant literature. The integrative model showcases that morally problematic situations have causes (i.e., contextual and relational factors, personal factors, jeopardized valuations), have affective repercussions (i.e., emotions and feelings, internal tensions), prompt action (i.e., through empowerment strategies leading to the evolution of situations), and elicit outcomes (i.e., factual consequences, residual emotions and feelings, positive or negative resolutions). In sum, this study advances understanding of the moral experiences of adults living with rare diseases while proposing a comprehensive conceptual tool to guide future empirical bioethics research on moral experiences.

Keywords Rare diseases · Ethics · Surveys and questionnaires · Community-based participatory research · Qualitative research · Pragmatist ethics

Caroline Hébert, Isabelle Carreau, Annie-Danielle Grenier and Yves Berthiaume have contributed equally to this work.

Extended author information available on the last page of the article

In the very process of conducting this research study, our group of authors was exposed to several shocking and inadmissible problematic situations. In one of the saddest cases, a young person died from not having received appropriate and timely care at the time this study was conducted. We stand in solidarity with individuals living with rare diseases alienated from mainstream healthcare. We sincerely hope that such situations cease without delay and that the human impact of these conditions is fully recognized.

Introduction

A disease is considered rare if its prevalence is below 1/2,000 (European Organisation for Rare Diseases, 2005). Rare diseases primarily have genetic causes, but may also arise due to infections, autoimmunity, exposure to toxins, or complications from drugs or surgery (Institute of Medicine (US), 2010). While not all rare diseases are life-threatening, they may result in significant and diverse functional limitations which may be exacerbated by inadequate care. Rare diseases are not viewed as a public health priority due to their individual low prevalence, but together may affect 8% of the population (Forman et al., 2012). As a result, limited funding and resources are directed towards rare disease research and treatment, preventing clinicians from having the knowledge and resources to adequately care for affected individuals (Forman et al., 2012).

Individuals living with rare diseases encounter a myriad of struggles linked to the rarity of their medical conditions and the social contexts in which they occur. They undergo diagnostic delays, are often misdiagnosed, and consult several doctors before being formally diagnosed (Bogart et al., 2022; Molster et al., 2016). They feel psychological distress due to their doctors' and their own lack of understanding of their rare diseases, adding to their felt uncertainty regarding illness progression and treatment (Garrino et al., 2015; Kesselheim et al., 2015; von der Lippe et al., 2017). When unable to establish a diagnosis, doctors sometimes refuse to further investigate these patients' cases and dismiss information brought by them (Grut & Kvam, 2013; Kesselheim et al., 2015). Individuals living with rare diseases must therefore bear the unsettling burden of being experts of their medical conditions (von der Lippe et al., 2017). These responsibilities add to the challenges of navigating social relationships, familial responsibilities, and (un)employment, often without proper financial support or services (Quintal et al., unpublished work 1 and 2).

Inherent to these concrete struggles, the rare disease experience is fundamentally a *moral* experience (Good, 1994). Morality corresponds to the group of dispositions and conduct that each individual judge to be acceptable or unacceptable. Morality is shaped by the things that individuals value (Hitlin, 2003; Racine, 2022). Moral experiences put at stake things that profoundly matter to people. Individuals value a variety of things based on their distinctive life histories, characteristics, and understandings, which can be jeopardized by illness experience (Kleinman & Benson, 2006):

Illness is everywhere a profoundly *moral* experience, since sufferers have things of great personal and collective value to gain or lose. Illness alters life plans and projects. And it provokes a response, however difficult and tenuous.

Illness matters because real things are on the line: self-identity, physical and mental health, life chances, social status, employment, finances, religious aspirations, or personal relationships. (Kleinman & Benson, 2006, 835)

Illness experiences may shatter self-conceptions and visions of the world due to the impairments and disability they bring about. Affected individuals may be forced to rethink their life paths while grappling with unknowns about their futures (Moreira, 2019). Yet, illness experiences may offer opportunities for imaginative reflection on purpose of life and personal growth (Moreira, 2019; Racine, 2022; Ryff & Singer, 2008).

Despite their existential importance, rare disease experiences are sometimes stripped of their moral significance in the literature (Kleinman & Benson, 2006). For example, these patients are said to experience a series of problems relating to healthcare, work, study, activities, or romantic relationships (Martinent et al., 2020; von der Lippe et al., 2017). Psychological hardships are often reported as a list of mental health issues such as anxiety and depression (Bogart & Irvin, 2017). Such abstract and simplified descriptions lose sight of the distress, suffering, uncertainty, and despair of patients that accompany illnesses (Kleinman & Benson, 2006; Racine, 2022).

Pragmatist Ethics and Morally Problematic Situations

Pragmatist ethics is premised on the interrelatedness of human experience and morality (Racine, 2022) hitherto described. It departs from traditional ethics theories claiming that moral conduct can be derived from a strict application of guidelines or rules (Aiguier & Loute, 2016; Dewey, 2003). It proposes that moral experiences are ultimately about *morally problematic situations*. In these situations, individuals are unable to enact their values due to the circumstances they face (Hunt & Carnevale, 2011). These situations undermine the ability of individuals to pursue activities or projects that are meaningful to them, jeopardizing their personal growth and flourishing (Pekarsky, 1990). Morally problematic situations hold existential weight. They give rise to internal tensions within individuals. Through such subjective malaise, individuals may feel abandoned, uncertain, misunderstood, or that their questions remain unanswered (Quintal et al., unpublished work 1). These situations may also constrain the agency of these individuals, leaving them unable to act according to their interests. As a result, they could experience powerlessness and disempowerment, disregard for their experience and knowledge by others, in addition to decisions taken on their behalf by others (Quintal et al., unpublished work 1).

To restore their sense of well-being and stability, individuals feel a pressing need to overcome morally problematic situations. Pragmatist ethics recognizes that individuals are endowed with *agency*, enabling them to actively shape their realities (Aiguier & Loute, 2016). Consequently, individuals, which are *agents*, may initiate an *inquiry* to address these situations. Through this process resembling ethical reasoning (Brendel & Miller, 2008), agents seek to fully understand their situations, imagine action scenarios that could resolve them, test the most promising scenarios, and improve them if needed (Fins et al., 1997). Solving these situations is a transactional,

intersubjective, and deliberative process. A successful scenario is compatible with the peculiarities of the environment and the values of the other agents facing the situation (Biesta, 2010; Racine, 2014). Morally problematic situations therefore offer an opportunity for biographical reconstruction. Agents can work towards new ideals of personal growth and flourishing if provided with favorable conditions for action (Quintal et al., unpublished work 2; Carricaburu & Pierret, 1995; Racine, 2022).

Objectives of this Study

Morally problematic situations experienced by adults living with rare diseases have been documented in previous studies, including a qualitative study (Quintal et al., unpublished work 1 and 2). These studies offer a first empirically grounded account of the morally problematic situations experienced by these individuals, while illuminating the internal tensions and the constraints to agency they elicit. Yet, these studies have two lacunae. First, both studies only name these situations rather than describing them in-depth. As a result, the primary objective of this current study was to acquire an in-depth understanding of the morally problematic situations experienced by adults living with rare diseases in the province of Québec.

Second, these studies do not provide a cohesive and extensive account of the components of morally problematic situations and how they interact in moral experiences. Hence, the secondary objective of this study was to develop an integrative model of the concept of morally problematic situations and their components. This conceptual framework would enable bioethics researchers to better capture and communicate distress, the existential anguish, and the uncertainty elicited by such situations and how agents can appropriate these situations to construct better futures for themselves.

Materials and Methods

The materials and methods reported below comply with the Standards for Reporting Qualitative Research of O'Brien et al. (2014).

Patient and Public Involvement Statement

This survey study is one of the steps in a more comprehensive participatory action research project exploring morally problematic situations experienced by adults living with rare diseases (Jagosh et al., 2012; Quintal et al., unpublished work 2). Pragmatist ethics recognizes the existence of plural conceptions of the good arising from varied backgrounds and experiences. It calls for the inclusion of diverse stakeholders in ethical deliberations to soundly understand the situations at hand (Lanre-Abass, 2010). Hence, the use of a large-scale survey, developed through participatory research and methods, is supported by pragmatist ethics.

The idea for this research project emerged through discussions with the founder of the *Regroupement québécois des maladies orphelines* (i.e., Québec Coalition of Orphan Diseases) and a clinician researcher specialized in cystic fibrosis. These discussions revealed the need to conduct a qualitative research project to document the difficult situations faced by adults living with rare diseases from a moral and existential perspective in partnership with patients. To this end, the *Ethics and Rare Diseases Working Group* was formed and consisted of patient partners living with rare diseases (authors 3, 4, and 5), representatives from the *Regroupement québécois des maladies orphelines*, and clinician researchers specializing in rare diseases (author 6). In addition, the first author is living with uncommon chronic conditions, while the last author has conducted previous research with individuals living with chronic conditions. Due to their respective backgrounds, the first and last authors were markedly interested by the experiences and knowledge of working group members, which fostered positive and enriching discussions during meetings with the working group.

Working group members were involved in the development of the survey and the interpretation of its results (Jagosh et al., 2012). More specifically, they refined the survey questions and the definition of the morally problematic situation provided therein based on their personal or professional experiences relating to rare diseases. Patient partners piloted the survey, which led to improvements in the formulation of questions. Working group members promoted the survey within their networks and suggested patient groups that could help promote it. They have provided comments on preliminary results, chose quotations to include in the article, provided input on the manuscript and approved it for submission.

Content of the Survey

The development of the survey has been described elsewhere (Quintal et al., unpublished work 2). Briefly, the survey was written in French, which is the official language of Québec. The survey was conducted in this Canadian province given that healthcare is mostly a provincial competency and shaped by the sociocultural characteristics of Quebec as a distinct nation. The survey was published on Eval&Go, an online survey design platform (<https://www.evalandgo.com/>). The consent section of the survey presented the objectives of the latter and specified that approximately 30 minutes would be required to complete the survey. Consistently with the inclusion criteria of the study, the eligibility section validated that survey respondents were 18 years old or older, lived in Québec, and had one or more rare disease(s) or self-identified with one or more undiagnosed rare disease(s).¹

The current manuscript reports the results of three relevant sections of the survey (see Supplementary file 1).² In the demographic and socioeconomic section, respondents were prompted to provide information about their gender, highest level

¹ Since individuals living with rare diseases are often confronted to diagnostic delays, those who self-identified with having one or more undiagnosed rare disease(s) were welcomed to participate to the study (Molster et al., 2016).

² The results of the other survey sections are reported elsewhere (Quintal et al., unpublished work 2).

of education achieved, occupation, annual family income, subjective socioeconomic positioning (Adler et al., 2000), first language, ethnic group (Government of Canada, 1998), and religiosity. In the subsequent section, respondents were asked to name the rare disease(s) they were diagnosed with or suspected having. They were also invited to specify the time elapsed until they were diagnosed or since the onset of their symptoms.

Later in the survey, respondents were asked to provide a long testimony of the most important morally problematic situation they had experienced in relation to their rare disease(s). To this end, respondents were given a plain language definition of the concept of morally problematic situations which had been improved with the help of working group members. Respondents could give a written testimony or an oral testimony, either by calling a designated phone number or by sending a voice recording by email. As first-person narratives, testimonies attest to the truth and suffering inherent to moral experiences (Moreira, 2019). At the end of the survey, respondents could participate to a draw of two \$50 gift cards and volunteer for follow-up interviews by leaving their contact information.

Participants and Recruitment

The survey was opened on March 12, 2021 and closed on May 9, 2021. The initial aim was to obtain a minimum of 50 completed questionnaires, and the survey was closed after far surpassing this aim. The *Regroupement québécois des maladies orphelines* advertised the survey twice on their social media accounts and in their newsletter. More than 80 patient associations and support groups were invited to promote the survey through their newsletters and Facebook accounts.

Data Analyses

Oral testimonies were transcribed by an external transcription service. Questionnaires were imported in an Excel spreadsheet, to which transcriptions of oral testimonies were subsequently added.

Exclusion of Questionnaires and Descriptive Statistics

Questionnaires were initially excluded from further analyses if they lacked a testimony or if respondents did not have a rare disease. Questionnaires were subsequently excluded if a testimony lacked enough information to be properly understood and analyzed, discussed a situation that was not morally problematic, or failed to address moral or ethical dimensions of moral experience as described above. One testimony was particularly long and alluded to several morally problematic situations. For simplicity reasons, the main situation described in the testimony was kept for the current analysis, while other parts of the testimony describing related situations were analyzed in another study (Quintal et al., unpublished work 2). Descriptive statistical analyses were conducted on demographic and socioeconomic data in addition to rare disease data.

Situational Analyses

Anonymized testimonies were subjected to in-depth novel situational analyses, closely informed by literature notably on pragmatist ethics and moral experience. These analyses sought to highlight the components of the morally problematic situations described in testimonies, consistently with an agential and situationalist understanding of morality (Racine, 2014). These components are understood to be closely interrelated, in constant interaction, and not independent from one another. To this end, situational analysis grids were prepared for each respondent in Word documents. Respondents' demographic and socioeconomic characteristics, rare disease(s), and testimonies were imported into each grid by the first author. Information on the components of the situation described in their testimony was extracted in designated spaces by the first and second authors based on the typology in Table 1 below.

Thematic Content Analysis

Grids generated in the previous steps were imported into the software MAXQDA 2022 (VERBI Software, 2021) for a thematic content analysis targeting specifically the information extracted from the testimonies. Thematic analysis consists in creating a coding guide, which is a hierarchical structure comprised of themes and subthemes present in a dataset. This structure was comprised of several codes, each of which can be linked to relevant text excerpts using the software (Braun & Clarke, 2006). The main themes of the coding guide were derived deductively from the components of morally problematic situations summarized in Table 1 while the subthemes were derived inductively (Nowell et al., 2017). The thematic analysis incorporated some aspects of content analysis by noting the frequency of certain codes among the testimonies (Vaismoradi et al., 2013). The analysis was conducted by the second author and verified by the first author to ensure correspondence of the coding guide with the content of testimonies. Ultimately, the thematic content analysis yielded a coding guide detailing how each component of the morally problematic situation materialized in respondents' testimonies, with supporting examples, in response to both objectives of the study.

Reporting of Results

The moral components of morally problematic situations characterized through the thematic content analysis are presented in distinct subsections in the Results section. Examples of these components are provided in accompanying texts and tables. To protect the confidentiality of respondents, not all their rare diseases were reported when they are quoted. In tables, results are generally ranked from the most to the least frequently mentioned in the testimonies.

Table 1 Components of morally problematic situations informed by previous literature

Progression of morally problematic situations*	Components of morally problematic situations	Insights based on previous literature
Causes of morally problematic situations	Contextual and relational factors	Morally problematic situations emerge in response to contextual and relational factors. Experiences are built intersubjectively through transactions with the environment, comprised of living and non-living entities (Biesta, 2010). Transactions between human beings have a distinctive feature, that by which “our cognitive ability will make us see our humanity in the ‘other,’ and the ‘other’ within ourselves” (Moreira, 2019, p. 3652). Hence, in medical settings, contextual factors could be comprised of conditions of the milieu, including scarce resources, institutional policies, legal and deontological obligations of clinicians, and clinical guidelines (Miller et al., 1997). Relational factors could namely include problematic discourses, actions, attitudes, and behaviors of clinicians or the family, or divergences between their interests (or valuations, defined below) and those of patients (Misak, 2013).
	Personal factors	Morally problematic situations also result from personal factors such as having a rare disease. Kleinman and Benson (2006, p. 835) write that “life thrusts people into vexing circumstances,” such as illness, which is part of “a broad category of personal [...] dangers and catastrophes.” They add that “illness is a catastrophe too, although a very intimate and bodily one.” Other personal factors may shape the way situations are experienced, such as agents’ unique profiles (e.g., sociodemographic status, life situations, lifestyle, language, culture and religiosity) and distinct prior life experiences (Miller et al., 1997).
	Jeopardized valuations	The above-mentioned factors prevent agents from actualizing their values in the morally problematic situations they face (Hunt & Carnevale, 2011). Things that are valued, or valuations, may encompass interests, priorities, needs, desires and preferences (Quintal et al., unpublished work 1) and are threatened by factors inherent to the situations (Kleinman & Benson, 2006). The jeopardization of values is what makes a situation morally problematic (Gallagher, 2014; Racine, 2022).

Table 1 (continued)

Progression of morally problematic situations*	Components of morally problematic situations	Insights based on previous literature
Affective repercussions of morally problematic situations	Emotions and feelings	<p>Agents experience a plethora of emotions and feelings in response to morally problematic situations. Moreira (2019, p. 3654) writes: “the condition of not choosing, and often the lack of family histories of rare illnesses, [evokes] feelings of having been invaded, surprised, and violently taken by something that does not belong to us.” Oppositely, a diagnosis can hold significance and be a source of relief especially for those who have undergone a long diagnostic delay (Jensen et al., 2019; Morgan et al., 2019). These emotions and feelings can occur initially in the situation, as it runs its course, or residually after the situation has subsided. In a paradigm of medicalization (Kleinman & Benson, 2006), morally problematic situation may sometimes be reduced to clinical cases (Jonsen, 1990; Jonsen et al., 2010; Racine, 2022), where psychological anguish may not be captured or at best, merely interpreted as manifestations of anxiety and mood disorders.</p>
Acting upon morally problematic situations	Internal tensions	<p>Morally problematic situations elicit internal tensions within agents. Internal tensions are experienced as subjective malaise regarding the ways in which agents’ valuations are opposed, undermined, or compromised in the situations (Quintal et al., unpublished work 1 and 2). These internal tensions attest to the complex, conflictual, and ambiguous essence of these situations (Dewey & Chipman, 1920; Racine, 2007). There may not be a clear-cut distinction between internal tensions and the emotions and feelings evoked by morally problematic situations.</p>
Acting upon morally problematic situations	Empowerment strategies	<p>The suffering inherent to moral experiences calls for an intervention (Moreira, 2019) through ethical inquiry. This process involves acquiring an in-depth understanding of situations through observation and experience and discussing and testing action scenarios susceptible to help resolve the situation at hand in collaboration with other stakeholders (Miller et al., 1997; Racine, 2016). To conduct this inquiry, agents may mobilize empowerment strategies to become active actors in the situations they face (Quintal et al., unpublished work 1). Through these strategies, they gather more information relevant to their situations, make their interests be heard, or nurture new life meanings amid health hardships (Quintal et al., unpublished work 1; Moreira, 2019).</p>

Table 1 (continued)

Progression of morally problematic situations*	Components of morally problematic situations	Insights based on previous literature
Evolution of situations	Evolution of situations	Morally problematic situations are susceptible to evolve following the use of empowerment strategies by agents in addition to the enactment of action scenarios (Racine et al., 2021). Newly emerging situations may not have reached a point where the enactment of action scenarios is possible due to the careful reflection necessary for proper ethical inquiry.
Outcomes of morally problematic situations	Consequences of situations	Consequences of morally problematic situations can be positive or negative depending on whether a resolution, if attained, is satisfactory or not. These consequences may be factual or emerge as residual emotions and feelings.
	Resolution of situations	Morally problematic situations may be resolved through the enactment of successful action scenarios and fruitful empowerment strategies. A resolved situation results in satisfaction, personal growth and flourishing of agents, since they learned to surmount a profound moral difficulty and made sure their valuations were considered (Pekarsky, 1990; Racine, 2016). Conversely, an unresolved situation elicits dissatisfaction attesting to a remaining moral problem (Epstein & Hamric, 2009; Racine, 2022).

*This progression of morally problematic situations is oriented by pragmatist ethics as a form of ethics which accompanies stakeholders in making sense of morally problematic situations, imagining responses to them, and enacting responses. It is not a description of any natural or mandatory progression of all morally problematic situations, but a process by which such situations can be tackled (Racine, 2016)

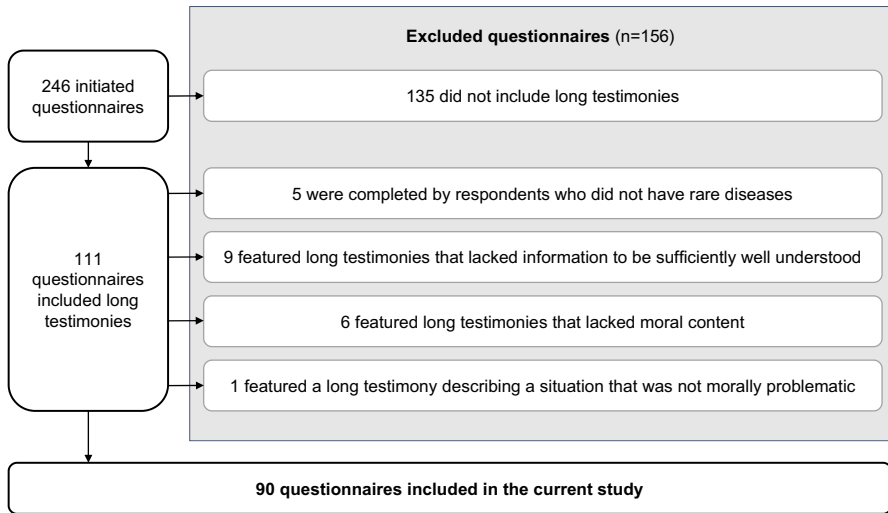


Fig 1 Flowchart for study sample

Ethics Approval

This study was approved by the human subject ethics committee of our institution (2021–1080). It complied with the Standards on Research Ethics and Scientific Integrity of the *Fonds de recherche du Québec—Santé* and the Tri-Council Policy Statement 2—Ethical Conduct for Research Involving Humans of the Panel on Research Ethics of the Canadian government. Survey respondents gave informed consent before participating to the study.

Results

Characteristics of the Respondents

Sociodemographic Characteristics of Respondents

Among the 246 initiated questionnaires, 90 were eligible to the current study, as shown in Fig 1. Respondents were aged between 19 and 75 years old, with a median age of 47.5 years, and most of them were women (84%). The sociodemographic characteristics of respondents are presented in Table 2.

Rare Diseases of Respondents

Together, respondents had 82 distinct rare diseases. Respondents who only had one rare disease constituted 72% of the sample. The highest number of rare diseases

Table 2 Sociodemographic characteristics of respondents

Characteristics of respondents	Number of respondents	% of sample respondents
<i>Gender</i>		
Women	76	84
Men	14	16
<i>Age</i>		
18–29	5	6
30–39	22	24
40–49	24	27
50–59	21	23
60–69	16	18
70–79	2	2
<i>Highest level of education completed</i>		
Elementary school	1	1
High school	9	10
Professional school	4	4
College ^a	22	24
Bachelor's degree	29	32
Graduate studies	24	26
Not specified	1	1
<i>Occupation</i>		
Student	3	3
Employed	30	33
Disability	41	46
Retired	9	10
Other	3	3
Prefer not to say	2	2
<i>Annual family income</i>		
Less than \$10,000	7	8
\$10,001–\$30,000	15	17
\$30,000–\$50,000	9	10
\$50,001–\$70,000	13	14
\$70,001–\$90,000	11	12
Over \$90,001	21	23
Prefer not to say	14	16
<i>Subjective socioeconomic positioning reported on a scale from 0 to 10</i>		
Low (scores of 0 to 4)	27	30
Medium (scores of 5 to 7)	45	50
High (scores of 8 to 10)	18	20
<i>First language</i>		
French	85	94
English	3	3
French and English	2	2

Table 2 (continued)

Characteristics of respondents	Number of respondents	% of sample
<i>Ethnic group</i>		
White	86	96
Other	2	2
Prefer not to say	2	2
<i>Religiosity</i>		
Yes	15	17
No	68	76
Don't know	1	1
Prefer not to say	6	7

^aIn the province of Québec, Canada, college education is offered in CÉGEPs (Collège d'enseignement général et professionnel), which are post-secondary education institutions offering 2-year pre-university training programs and 3-year training programs for trades

reported by a single respondent was 6. The most frequent rare diseases in the sample were Ehlers-Danlos syndromes, Sjögren syndrome, and scleroderma with respective frequencies of 16%, 6%, and 4% among respondents. The most frequent categories of rare diseases represented in the sample were: endocrine, nutritional, and metabolic diseases (16% of rare diseases); congenital malformations, deformations, and chromosomal abnormalities (15% of rare diseases); in addition to diseases of the musculoskeletal system and connective tissue (15% of rare diseases) (see Supplementary file 2 for detailed rare disease categories in the sample along with their frequencies).

Qualitative Results

Most respondents described the morally problematic situations they experienced by alluding to their causes, their affective repercussions, the intervention made upon them, and their outcomes.

Causes of Morally Problematic Situations

Most morally problematic situations described by respondents featured contextual and relational factors in addition to personal factors that clashed with their valuations, or things that fundamentally mattered to them (see Table 3).

Contextual and Relational Factors First, contextual and relational factors contributed to the genesis of morally problematic situations. These factors arose primarily in healthcare, followed by career and studies, social and family life, and personal and introspective life. These factors included problematic attitudes (e.g., contempt by clinicians or colleagues), inappropriate practices (e.g., physicians refusing to care for patients), limited resources (e.g., maladapted long-term care homes for non-elderly individuals), or demanding obligations or duties (e.g., difficulties caring for one's family) (see Table 3).

Table 3 Contextual and relational factors of the situations and associated valuations

Context	Sub-context	Contextual and relational factors of morally problematic situations	Valuations of the respondents at stake
Healthcare	Clinician attitudes	<ul style="list-style-type: none"> - Stigmatization and contempt - Skepticism and trivialization of illness experience - Lack of listening, of receptivity, and of empathy - Lack of transparency - Diagnostic delay - Physicians getting rid of patients - Negative implications of diagnostic delays - Difficulty in obtaining a diagnosis - Poorly announced diagnosis - No proper explanation of diagnosis 	<ul style="list-style-type: none"> - Recognition of the rare disease - Kindness and support - Respect - Being listened to - Honesty - Empathy
	Diagnostic odyssey	<ul style="list-style-type: none"> - Diagnostic delay - Physicians getting rid of patients - Negative implications of diagnostic delays 	<ul style="list-style-type: none"> - Proactivity and in-depth investigations - Not evoking having one's disease attributed to psychosomatisation
	Diagnosis	<ul style="list-style-type: none"> - Difficulty in obtaining a diagnosis - Poorly announced diagnosis - No proper explanation of diagnosis 	<ul style="list-style-type: none"> - Better knowledge of one's illness - Good support and care following the diagnosis
	Medical care and interventions	<ul style="list-style-type: none"> - Limited access to appropriate interventions - Being subjected to contraindicated or dangerous interventions or practices - Care delays 	<ul style="list-style-type: none"> - Access to care - Access to adapted, safe, and adequate interventions - Being able to make intervention decisions for oneself
	Medical follow-up	<ul style="list-style-type: none"> - Limited follow-up - Inadequate or absent follow-up 	<ul style="list-style-type: none"> - Respect of one's rights - More research on rare diseases - Prompt care - Being referred to appropriate specialists - Being offered adequate medical follow-up - Clinicians being knowledgeable about rare diseases
	Medical evaluations by insurers	<ul style="list-style-type: none"> - Limited eligibility to disability benefits - Complex process 	<ul style="list-style-type: none"> - Recognition of disability and invalidating symptoms by insurers - Respect from medical evaluators

Table 3 (continued)

Context	Sub-context	Contextual and relational factors of morally problematic situations	Valuations of the respondents at stake
Career and studies	Professional and academic objectives	<ul style="list-style-type: none"> - Work and study requirements incompatible with limitations - Work or study difficulties 	<ul style="list-style-type: none"> - Being able to work (or study) to favour one's flourishing - Pursuing professional objectives - Being able to prolong or complete studies
	Work and study conditions	<ul style="list-style-type: none"> - Absent, poor, or inadequate accommodations - Discrimination, harassment, and contempt - Having to work from home due to vulnerability to COVID-19 	<ul style="list-style-type: none"> - Adequate and adapted accommodations (e.g., flexible schedules in the workplace) - Employment benefits - Recognition for one's motivation despite hardships - Being harassment free
Social and family life	Close ones	<ul style="list-style-type: none"> - Inadequate support from close ones - Difficulties in caring for one's family - Denial of rare disease by close ones 	<ul style="list-style-type: none"> - Being a good parent, family member and friend - Having a family life - Being believed and supported by close ones - Being able to choose to have children or not
	Participation in society	<ul style="list-style-type: none"> - Poor societal interest towards rare diseases - Lack of patient associations for certain rare diseases 	<ul style="list-style-type: none"> - Being involved in one's rare disease community - Better awareness of the public towards rare diseases
	Social interactions	<ul style="list-style-type: none"> - Skepticism from others - Prejudice from others - Isolation - Having to justify and explain the rare disease 	<ul style="list-style-type: none"> - Being able to participate in social events

Table 3 (continued)

Context	Sub-context	Contextual and relational factors of morally problematic situations	Valuations of the respondents at stake
Personal and introspective life	Essential needs	<ul style="list-style-type: none"> - Maladapted long-term care homes for non-elderly individuals 	<ul style="list-style-type: none"> - Autonomy in daily tasks - Access to support resources - Adapted care and leisure in long-term care homes for non-elderly adults - Enjoying culinary pleasures - Being able to stay with one's family - Having the ability to go on an outing
	Socioeconomic condition	<ul style="list-style-type: none"> - Limited income - Significant medical expenses - Financial dependence - No access to disability benefits without a diagnosis 	<ul style="list-style-type: none"> - Financial security - Financial autonomy
	Personal growth	<ul style="list-style-type: none"> - N/A 	<ul style="list-style-type: none"> - Being able to do physical activity - Sense of accomplishment - Being able to participate in social activities
	Well-being	<ul style="list-style-type: none"> - N/A 	<ul style="list-style-type: none"> - Good health or quality of life - Not being afflicted by the rare disease as much

For example, a woman in her mid-forties living with hypermobile Ehlers–Danlos syndrome and primary immunodeficiency recounted that her internist had a patronizing attitude and failed to sufficiently investigate her case:

The internist had told me that she would look at all my folders, conduct tests, and conduct an analysis. Ultimately, she did absolutely nothing. Three months of waiting and examinations. When I met her, she had her arms crossed, her legs crossed, nothing on her desk, nothing on her [computer] screen, and told me “You have nothing” and it is fibromyalgia or myalgic encephalomyelitis. When I tried to talk to her about Ehlers-Danlos syndrome [...], she did not want to hear anything. She was full of herself, condescending, [and] close-minded.

A man in his early thirties living with empty nose syndrome, a rare distressing complication of nose surgery, was erroneously dismissed by the medical personnel as having a psychosomatic illness. He explained:

In the days following my septoplasty (nose surgery), [...] my symptoms were extremely severe, so I returned to the hospital to receive help and answers, but no doctor was able to explain what happened to me physically. All they did was to send me in the psychiatric department and give me drugs that did not work for me, because my problem is physical and not mental.

Personal Factors Second, respondents’ personal factors often played a complementary role in causing morally problematic situations. These factors were often immutable and usually related to their rare diseases. Examples of these factors include the fact that respondents’ rare diseases often required medical care and follow-up, had deleterious health impacts, were often accompanied by other rare diseases, and some were genetically transmittable, leading to hesitancy regarding having children. Respondents’ rare diseases involved functional limitations in everyday life, adaptations, significant food restrictions, and work and study-related limitations.

The testimony of a woman in her early sixties living with myasthenia gravis illustrates well how personal factors in addition to contextual and relational factors interacted in causing a morally problematic situation she experienced during a hospital stay. She was met with contempt when asking medical workers to ensure that the drugs they wanted to provide her were not contraindicated for her rare disease. She hypothesized that their negative attitude stemmed from prejudice linked to her socioeconomic status as a personal factor:

I was faced with an unpleasant attitude followed by intimidation. [...] I quickly realized after all these experiences that a person of sound mind and who exhibits sufficient strength and intelligence to defend themselves verbally, it is a situation that frustrates them even more. [...] Therefore, [they use] our social status (disability benefits) and our [lower levels of] education as means to crush us, by making us believe that we are completely lost and are completely unable to understand what is happening.

Jeopardized Valuations at Stake in the Situations Third, most respondents' valuations were jeopardized by contextual and relational factors in addition to personal factors specific to the respondents (see Table 3). The valuations alluded to by respondents encompassed values, ideals, expectations, needs, preferences, interests, and desires. The jeopardization of valuations made situations morally problematic. Valuations were primarily jeopardized in context of healthcare, followed by personal and introspective life, career and studies, and social and family life. Respondents sometimes alluded to their valuations more explicitly using expressions such as "I hope," "I would like /I would have liked," "I want to," "it is important that," "I would have needed," "being able to," "I prefer," and "I appreciate." Table 3 below contrasts contextual factors with respondents' valuations.

The testimony of a woman in her early thirties living with essential thrombocytosis, a rare disorder characterized by an elevated production of platelets, illustrates well how contextual and relational factors oppose the valuations anchored in her ideals and needs. She explained that upon her diagnosis, she was accompanied by her mother, who "was not reassuring at all, because she was scared for [her]" and the specialist she met was "especially intelligent" but "very cold." When he announced her diagnosis, he "did not understand why [she] cried, he attempted to reassure [her] by telling [her] that [her] life expectancy was normal." Instead of this inadequate support and lack of receptivity, the woman needed significantly more goodwill and support. She explained:

a meeting with a psychologist or a social worker could have made all the difference [...] I would have needed more reassurance and most importantly, to be linked to a community [of individuals] that live with this illness: having resources, tools, being told that things would go well and that I would be accompanied. It took me months to find precise resources that could help me, I was alone with my rare disease [...], and I was looking for a patient support group that I could not find.

More often, respondents alluded to their valuations implicitly. A woman in her thirties living with hypermobile Ehlers-Danlos syndrome explained that she expected to be listened to during clinical encounters; otherwise she would seek care elsewhere: "before taking appointments with a physiotherapist, a massotherapist, a dentist, an optometrist, I send an email to their office explaining my situation and I assess their answers based on how I sense their ability to listen. I go where I am listened to and where my condition is not minimized, and where [clinicians are ready] to learn about my situation." This strict expectation of being listened to emerged in reaction to contextual and relational factors related to her family doctor's problematic attitude, which "did not take [her] seriously" and who "was not interested to further investigate [her] case."

Affective Repercussions of Morally Problematic Situations

Morally problematic situations elicited internal tensions, in addition to emotions and feelings which materialized at the onset of the situation or residually.

Internal Tensions Raised by the Situations Internal tensions were experienced by several respondents when initially faced with their situations. They consisted of subjective uneasiness due to valuations being undermined in the situation. Internal tensions appeared implicitly in several respondents' testimonies and were overwhelmingly experienced as powerlessness, followed by dilemma, misunderstanding, internal conflict, uncertainty, hesitation, questioning, doubt, and ambiguity (see Supplementary file 3).

For example, a profound internal tension of powerlessness was reported by a woman in her mid-thirties living with Ehlers-Danlos syndrome and several other rare diseases. She explained that in her late twenties, she was not able to study and work anymore due to her undiagnosed rare diseases. Since she was not eligible to disability benefits, she then experienced the four most difficult years of her life: "I had to live with seven thousand dollars per year, alone and sick [...]. I underwent a severe depression and my physical condition significantly worsened." She explained that her situation changed today "only because [she] accepted to depend on a man, but [she] would end up in the same situation if he decides to leave [her]."

Initial Emotions and Feelings When initially faced with morally problematic situations, most respondents reported experiencing a plethora of emotions and feelings. While internal tensions were experienced in reaction to undermined valuations, emotions and feelings were understood as relating to the psychological state of mind of respondents. In order of frequency, most respondents alluded to emotions and feelings of distress, worry, sadness, isolation, anger, helplessness, disrespect, guilt, exhaustion, shock, and poor self-esteem. Very few respondents reported positive emotions and feelings. The respondents mostly named their emotions and feelings explicitly and using terms related to those (see Table 4).

A woman in her early fifties living with Stickler syndrome lamented having to "fight a rare, invisible, progressive, and invalidating illness" and having to "fight [her] family doctor and the healthcare system" to receive proper recognition and care. She added that over the years, her "mental health was seriously affected to the point that [she] became suicidal and attempted suicide." Similarly, a woman in her early fifties living with Ehlers-Danlos syndrome recounted the turmoil she experienced in relation to the evaluation of her case by her insurance company. She explained what happened with the physician:

He absolutely did not take into account my health condition, he has distorted my remarks, he insinuated that I sought to deceive him, and he recommended that I return to work full time starting the following week. But the most difficult [event] was when my family doctor [...] told me that he had been won over the expert physician's assessment. I felt betrayed and abandoned by my family doctor who was aware of [...] my numerous

Table 4 Diversity of emotions and feelings reported by respondents in response to morally problematic situations

Types of emotions and feelings	Emotions and feelings only present initially	Emotions and feelings only present residually	Emotions and feelings present both initially and residually
Distress	Finding the situation difficult	Persistent existential questions	Feelings of injustice
	Wanting to die	Having a sword of Damocles over one's head	Feeling like living a nightmare
	Psychological suffering	Trauma	
	Falling into a void	Social phobia with panic attacks	
	Distress		
	Falling apart		
	Not being able to hold on to anything anymore		
	Desolation		
	Turmoil		
	Neurosis		
Worry	Not being able to think about other things	Nervousness	Stress
	Doubt	Not being reassured	Anxiety
	Insecurity		Worry
	Uncertainty		Anguish
			Fear
Sadness	Discouragement		Sadness
	Feeling hurt		Grief
	Despair		Depression
	Disappointment		
	Isolation		
Isolation	Feeling mistreated and left aside		Loneliness
			Feeling misunderstood
Powerlessness			Feeling abandoned
			N/A

Table 4 (continued)

Types of emotions and feelings	Emotions and feelings only present initially	Emotions and feelings only present residually	Emotions and feelings present both initially and residually
Anger	Frustration Rage Exasperation	Repressed emotions	Anger
Feeling disrespected	Betrayal Humiliation Not feeling being taken seriously Feeling judged Being annoyed by a lack of recognition (at work) Being surprised regarding prejudices Not feeling welcomed Feeling dehumanized N/A	Feeling that one's rights have been flouted	N/A
Loss of trust	N/A	Loss of trust Distrust	N/A
Poor self-esteem	Insecurity Feeling unable to fit in	Poor self-esteem Self-doubt Feeling of failure Feeling of having missed out	N/A
Guilt	Feeling like a nuisance to others Shame	Temptation not to follow dietary restrictions	Guilt
Exhaustion	Fatigue Feeling overtaxed	N/A	Exhaustion
Shock	Shocked Being distraught	N/A	N/A

Table 4 (continued)

Types of emotions and feelings	Emotions and feelings only present initially	Emotions and feelings only present residually	Emotions and feelings present both initially and residually
Positive emotions and feelings ^a	Hope Feeling at ease regarding the current situation	Gratitude Less guilt Feeling calmer Being satisfied with the approach taken Improved mental health Feeling liberated	Feeling understood and accompanied by clinicians

^aAs indicated in the Methodology section, results are ranked from the most to the least frequently mentioned in the long testimonials. However, positive emotions and feelings are reported at the end of this table due to their fundamental difference with the remaining negative emotions and feelings

[...] health issues for the last 20 years [...]. I felt very lonely and without resources during these dark days. I lost trust in my family doctor due to lack of support from him.

A woman in her late thirties living with cystic fibrosis had to stop working to better care for her health. This decision led her to ask for disability benefits, lower-income housing, and moving back with her parents in the meantime. Through these challenging life changes, she experienced strong emotions “ranging from sadness to anger, grieving an important value for [her], a feeling of intense solitude by feeling ‘abandoned by the system’ and being entirely left on [her] own.” This decision may have been “one of the most difficult” she took because it opposed two of her valuations: work as a representation of her autonomy and as integral to her identity and being able to take better care of her health.

Residual Emotions and Feelings Along the factual consequences of morally problematic situations, several respondents experienced durable emotions and feelings in relation to what remained of the situation. In order of occurrence, several respondents alluded to emotions and feelings of worry, distress, loss of trust, sadness, poor self-esteem, isolation, anger, guilt, helplessness, feeling disrespected, and exhaustion. Some respondents also alluded to positive feelings. Respondents primarily named their emotions and feelings explicitly and using related terms (see Table 4). Distress, worry, and sadness were prevalent and constant as the situations evolved.

A young man living with Ehlers-Danlos syndrome gave a poignant testimony on the lack of recognition he endured in the medical system, and the severe psychological impacts it had on him:

I started having difficulties eating [about half a year ago]. I have met with several doctors who did not take me seriously or who minimized my symptoms. [...] I have been accused of anorexia by [emergency doctors] which [implied] that I was inflicting this to myself. I have cried dozens of times; I have nightmares every night or almost where I look for help but do not receive any. I suffer every minute. I do not have enough energy anymore to fight against the medical system. [...] I feel abandoned, misunderstood, like a nuisance for physicians and emergency doctors. The disease is already an ordeal, having good timely care wouldn't be too much to ask for! I avoid medical consultations or going to the emergency room because I lost all my trust in the medical system. I don't feel taken seriously.

A woman in her late sixties living with achondroplasia endured prolonged harassment and intimidation at the hospital where she worked, in relation to her physical difference. Despite having retired, she confided: “I [...] developed social phobia and panic attacks. I distrust everyone due to how I am afraid of being ridiculed, humiliated, and denigrated. I have lost my self-esteem and confidence.”

In contrast, some respondents communicated positive emotions and feelings when reflecting on their journeys. A few expressed gratitude, notably regarding one's privileged access to a liver graft and specialized care in Canada, while another celebrated her chance of having been able to work, unlike many others living with rare diseases.

Others discussed the relief they experienced when being accurately diagnosed, which held an explanatory and validating value for them.

Acting Upon Morally Problematic Situations

Several respondents described empowerment strategies they mobilized to face their morally problematic situations. Some of these strategies led to changes in the situations, enabling their evolution.

Empowerment Strategies Prompted by internal tensions and emotions and feelings, several respondents described a variety of empowerment strategies that they used to attempt to overcome morally problematic situations. Through the adoption of these strategies (see Table 5), these respondents positioned themselves as active actors in the situations they faced. They acted proactively and empowered themselves in the face of adversity notably by adopting a positive mindset or surrounding themselves with allies. Through the adoption of these strategies, the respondents were sometimes able to initiate changes in the contextual and relational factors of their situations. They worked towards cultivating the best conditions for attaining their objectives and to nurture their well-being and growth amid hardships.

Evolution of the Situations The adoption of the above-mentioned strategies generally resulted in situations evolving, which was addressed in most testimonies (66%). More specifically, 11% of situations were ongoing and 44% situations had ended. In contrast, the evolution of 44% situations had not been clearly described by respondents, sometimes because the situations did not change beyond their initial state.

A woman in her late forties living with confirmed severe pelvic congestion, highly suspected Ehlers-Danlos syndrome and Nutcracker syndrome in addition to other undiagnosed rare diseases detailed the evolution of her situation through the lens of the various strategies she mobilized:

Since the beginning of my important symptoms two years ago, I have repeatedly seen my family doctor and 6 specialists, 2 visits to the emergency room with various kinds of exams, including 4 scans. Aside from pelvic congestion, they did not find anything, and they all agreed that [this diagnosis] did not explain my most important symptoms. I had to become informed, do my research, find people with similar symptoms and suggest avenues to my physicians. [...] When physicians here said that they do not know what to do anymore to diagnose me as my symptoms worsened, I decided to travel to see a specialist in Germany known to have diagnosed several [...] Canadians [...]. It was a very difficult decision due to [COVID] and the important travel and consultation fees.

Outcomes of the Situations and Their Consequences

Following interventions in the situations and their evolution, some situations had positive consequences on respondents while others had negative consequences. These situations were thus resolved satisfactorily or unsatisfactorily respectively.

Table 5 Empowerment strategies

Context	General strategies mobilized	Specific strategies mobilized
Healthcare	<p>Being a proactive patient</p> <p>Caring for one's health more independently</p> <p>Optimizing clinical encounters</p> <p>Working towards obtaining income</p> <p>Ensuring one's safety and security during hospital stays</p>	<p>Consulting several clinicians and specialists</p> <p>Returning to the hospital to seek answers</p> <p>Asking for more in-depth medical consultations despite waiting times</p> <p>Turning to local health resources</p> <p>Launching a GoFundMe campaign to fund one's treatments</p> <p>Searching for information online</p> <p>Managing one's rare disease and symptoms carefully</p> <p>Turning to private healthcare</p> <p>Identifying international experts</p> <p>Avoiding consultations with physicians</p> <p>Soliciting the help of patient associations</p> <p>Bringing written personal information to physicians, often in a list format (medical history, symptoms, possible diagnoses, recommended and contraindicated drugs)</p> <p>Bringing scientific literature and documentation to physicians</p> <p>Being insistent</p> <p>Carefully selecting the information to be shared with physicians (i.e., to avoid prejudice)</p> <p>Being accompanied by a close one or a representative of the hospital's users' committee</p> <p>Turning to governmental bodies for disability benefits</p> <p>Soliciting the employer's help</p> <p>Asking a relative to be present at one's side at the hospital</p> <p>Providing healthcare professionals with a list of contraindicated drugs for one's rare disease</p> <p>Being vigilant</p>

Table 5 (continued)

Context	General strategies mobilized	Specific strategies mobilized
Personal and introspective life	Positive and constructive attitudes to keep morale	<ul style="list-style-type: none"> Cultivating resilience Undertaking personal reflections Staying positive Managing on one's own Putting things in perspective Putting one's interests first
	Striving for fulfilment	<ul style="list-style-type: none"> Seeking mental health support Having hobbies Participating in research
	Asking for reparation and justice	<ul style="list-style-type: none"> Lodging a complaint towards a physician Consulting one's union Consulting a lawyer Denouncing an abusive boss Participating in a commission of inquiry
Social and family life	Soliciting the support of close ones and of the community	<ul style="list-style-type: none"> Soliciting or benefiting from the support of close ones Looking for support groups or individuals with similar rare diseases Creating a family routine to better accommodate the rare disease
Career and studies	Equip oneself to work and study more easily	<ul style="list-style-type: none"> Asking for accommodations (e.g., part-time work) Benefiting from the support of one's boss and colleagues Reorientation of one's career Educating oneself through online material and events for lack of access to the education system

Consequences of the Situations The morally problematic situations encountered by respondents had widespread and diverse factual consequences on their lives. These consequences were mostly negative, although some patients saw positive outcomes. Since the consequences of the situation were often inextricable from the reality of living with rare diseases and their associated constraints, they are not reported extensively here. In short, these consequences arose primarily in relation to some respondents' personal and introspective life, followed by healthcare, career and studies, and social and family life. The man living with empty nose syndrome cited above summarized the consequences of his situation as such: "what I cannot conceive is the lack of follow-up, the lack of recognition and the denial of an illness so invalidating that had important repercussions on my general health, my academic and professional life, my finances, and my relationship with my family." Moreover, morally problematic situations ultimately resulted in significant residual emotions and feelings, as described above.

Resolution of the Situations Some of the morally problematic situations introduced above had ended. They were characterized as having an unsatisfactory resolution in 65% of cases and as having a satisfactory resolution in 35% of cases. The resolution of situations was contingent on their negative or positive consequences.

In situations leading to unsatisfactory resolutions, some respondents reluctantly settled for undesired life choices or decisions that were imposed on them. In some cases, despite acting in the situation, the moral problem inherent to the situation persisted and the respondents felt confronted with the same battle.

This is the experience of a woman in her early thirties living with interstitial cystitis. Newly published studies suggested that her pain-relieving drug could result in long-term visual issues. She pondered: "do I stop taking this drug or not, given these new studies?" She attempted to stop the drug, but due to her "atrocious pain," she ultimately reduced her daily dosage. Since there are no accessible alternative drugs, she felt distraught and disempowered: "the situation did not necessarily improve, but by discussing with my doctors, by undergoing regular ocular testing, and by reducing my dosage, [...] I can better manage the stress relating to a [related] vision impairment in the future."

In another situation, a man in his late fifties living with stage IV pulmonary hypertension had to wait too long for a proper diagnosis and care. He lamented that "a timely diagnosis would have allowed me not to reach the last stage of the illness." He added that his situation was far from ideal: "Disability benefits do not enable me to live because my revenue is below the poverty threshold and I cannot afford the drug that I need."

In contrast, a few morally problematic situations led to satisfactory resolutions. Key situations with satisfactory resolutions featured the enactment of the best decision for oneself (e.g., by leaving a job to better care for one's rare disease), the acknowledgement of one's experience (e.g., through a formal rare disease diagnosis with explanatory value), overcoming hardships through resilience (e.g., dissipation of suicidal thoughts), taking risks which paid off (e.g., a successful lung transplant for cystic fibrosis), or giving back to others (e.g., creating

a patient support group). All these situations had the common characteristic of making respondents' lives feel more bearable over time.

Discussion

This study offers a first in-depth understanding of morally problematic situations experienced by adults living with rare diseases. It uncovers their various components and how they relate with one another, sometimes with excerpts of testimonials from survey respondents. Based on these results, an integrative model of the components of morally problematic situations is proposed in Fig. 2. The model integrates study results (numbers 1 to 10) and related concepts from the literature on pragmatist ethics and moral experience (letters a to e).

Features of the Integrative Model

The integrative model of the morally problematic situation illustrates how causes, affective repercussions, actions, and outcomes are constitutive of morally problematic situations. Situations are caused by contextual and relational factors in addition to personal factors (components 1 and 2).³ As these factors interact, the valuations of agents are jeopardized and situations become morally problematic (component 3). These conflicts have affective repercussions which take the form of internal tensions and emotions and feelings (components 4 and 5). This felt uneasiness can motivate agents to act upon their situations using empowerment strategies (component 6) to prompt the evolution of their situations (component 7). Ultimately, these strategies elicit factual consequences (component 8) which define a new version of the situation. Residual emotions and feelings may result (component 9) and if applicable, agents may perceive this resolution as either satisfactory or unsatisfactory (component 10).

The integrative model is enriched from concepts discussed in the literature. In short, contextual and relational factors may be experienced as constraints to agency (concept a), reinforcing how agents' valuations are jeopardized. Agents may use empowerment strategies as they deploy an action scenario to move towards a resolution (concept b). If they manage to satisfactorily resolve their situations, agents undergo personal growth and flourishing (concept c). Alternatively, they experience moral residue resulting from residual negative emotions and feelings and the unsatisfactory resolution of their situations (concept d). Moral residue prompts agents to conduct further inquiry (concept e) by enacting alternative solutions (concept b) that could promise to resolve their situations more satisfactorily. In the integrative model, we also propose that morally problematic situations acquire moral and existential significance, for example as they jeopardize valuations (component 3), elicit

³ In the Discussion, the numbers and letters in parentheses refer to their equivalents in Fig. 2.

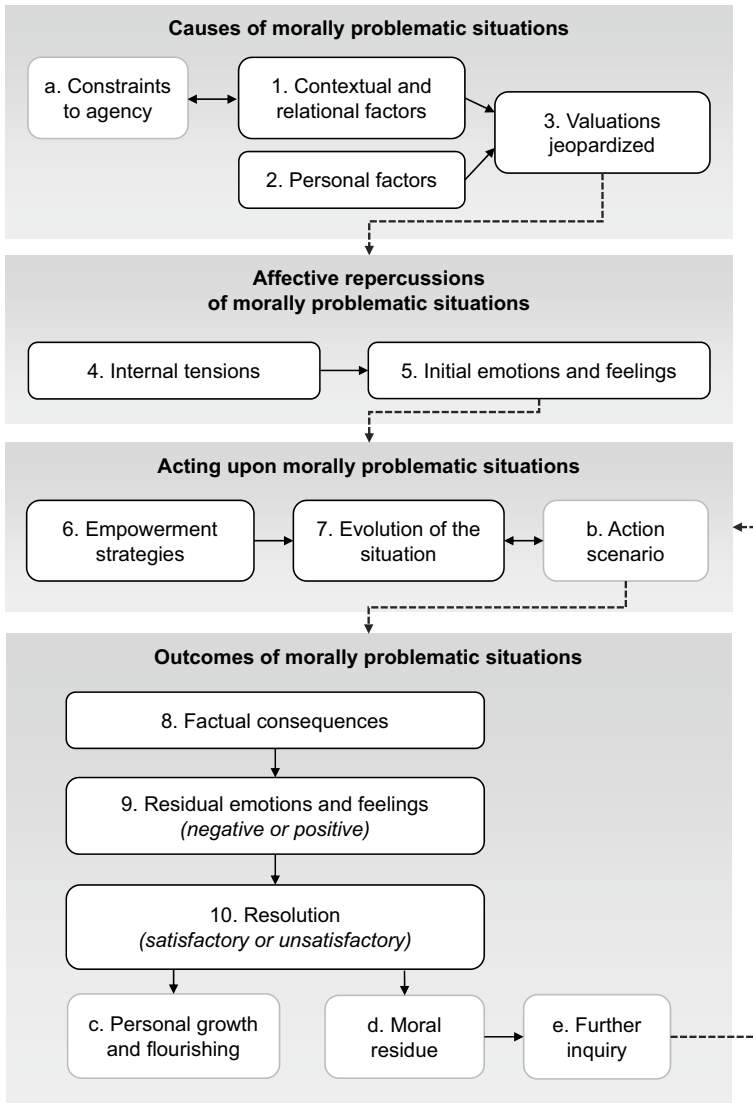


Fig. 2 An integrative model of morally problematic situations. Numbers 1-10: Components of morally problematic situations based on study results; letters a-e: relevant concepts from the literature on pragmatist ethics and moral experience. We acknowledge that each situation is experienced differently. To account for this diversity, the aim of this model is to synthesize insights on morally problematic situations derived from patient experiences and pragmatist ethics. This model is not meant to present how agents progress through morally problematic situations following an algorithmic logic. It also is not meant to represent all interactions occurring between its components extensively. Rather as indicated in the note for Table 1, it is a process suggested and supported by pragmatist ethics by which such situations can be tackled and learned from.

internal tensions (component 4), evoke emotions and feelings (components 5 and 9), and involve satisfactory or unsatisfactory resolutions (component 10).

The next sections further examine the components and interactions constitutive of morally problematic situations faced by adults living with rare diseases by building on literature on pragmatist ethics and moral experience. Some of these challenges may also be experienced in the context of other non-rare chronic illnesses (e.g., Asbring & Närvänen, 2003; Baarsma et al., 2022; Blease et al., 2017), but our focus here is on exploring the value of the integrative model to explain the difficult experiences of patients with rare diseases.

Moral and Existential Significance of Morally Problematic Situations

Morally problematic situations hold moral and existential significance for agents. This significance first arises from jeopardized valuations, which elicit internal tensions in addition to initial emotions and feelings (components 3–5). This significance also arises from personal growth and flourishing or moral residue (concepts c and d) being intrinsically linked to satisfactory or unsatisfactory resolutions of situations (component 10).

This two-fold account of the moral and existential significance of situations is compatible with understandings of morality and valuations. Morality designates the behaviors and inclinations judged to be acceptable or unacceptable by agents. Each agent has a personal morality that emerges through life experiences (Hitlin, 2003; Racine, 2022). Valuations are defined through life experiences and can take the shape of preferences, desires, interests, needs, and priorities (Miller et al., 1997; Quintal et al., unpublished work 1). Morality and valuations are thus closely intertwined. They shape agents' actions and more broadly, their understanding of their existential purpose, or what they want to do with their lives (Hartman et al., 2019; Hitlin, 2003; Racine, 2022). The powerlessness, distress, worry, and sadness (components 4 and 5) experienced by respondents are indicative of their inability to enact their existential purpose and important valuations (component 3) within the situations they face.

Due to its moral and existential weight, this moral suffering calls for ethical interventions (Moreira, 2019). Morality is experienced intersubjectively, as agents interact with others and their physical environment through transactions (Biesta, 2010). As a result, these ethical interventions could be deployed in intersubjective settings. Healthcare settings offer opportunities for these interventions (Moreira, 2019). Clinicians could seek to acknowledge the moral suffering of patients by expressing empathy and attentiveness to their patients' valuations jeopardized by rare disease experiences. They could validate patients' experiences, thus alleviating the moral suffering of patients (Kleinman & Benson, 2006; Levinas, 2002). Such interventions would support patient well-being, which is often at stake among individuals living with rare diseases given the scarcity of potential therapeutic interventions. Conversely, negative clinician attitudes, which often amounted to a lack of empathy, were profoundly devastating for respondents due to the intersubjective nature of moral experience (see Table 3).

More research would be needed to thoroughly investigate the causes for these negative clinician attitudes frequently reported by participants. In the meantime, conditions should be improved to allow for more ethical interventions in rare disease care characterized by empathetic and welcoming attitudes. Negative clinicians' attitudes, which constitute relational factors (see Table 3), could be mitigated through training opportunities such as workshops offered as continued medical training with an evaluation component from patients, discussions with specific care teams, such as emergency personnel, and awareness campaigns conducted in partnership with clinicians who have first-hand experiences of living with a rare disease. Gearing medical school admission criteria towards more empathetic personality traits could be another course of action to consider. In addition, organizational changes, such as the creation of multidisciplinary rare disease clinics and ensuring that physicians can dedicate enough time to their patients, could also foster these ethical interventions. Such interventions could be an impactful first step to humanize medical care in the realm of rare disease while supporting patients in renewing their existential purpose. Yet, we recognize that such courses of action are complex and warrant further exploration before being put into practice.

Empowerment Strategies and Constraints to Agency

The integrative model illustrates how affective repercussions of morally problematic situations (i.e., emotions and feelings, in addition to internal tensions) motivate agents to adopt empowerment strategies to make their situations evolve (components 6 and 7). Agents therefore take on an active posture to tackle the situation, as opposed to passively witnessing the course of events (Racine, 2022). Moreira (2019) describes the desire for empowerment of individuals living with rare diseases in a context of medicalization that obscures moral experience as follows:

struggles for recognition and power, here interpreted as power, interpreted here as retaking the place of speech [...], commonly called empowerment are at stake [...]. The central struggle here is between the first [...] person authorial discourse in the face of the moral experience of being ill, and the third person biomedical discourse, which may justify an artificial isolation of the disease from the life context in which it is anchored. (Moreira, 2019, 3657)

The empowerment strategies used by respondents included taking charge of their health, cultivating positive attitudes to cope with their challenging situations, and soliciting the support of others (see Table 5). These empowerment strategies are directed towards contextual and relational factors that define their situations (components 1 and 6). They highlight respondents' receptivity towards their surroundings, their nimbleness, their resilience, and their commitment to their personal well-being and growth amid health challenges and existential hardships.

However, empowerment strategies may not always be fruitful in tackling the contextual and relational factors responsible for situations. On one hand, it may be practically impossible for agents to intervene upon factors arising from organizational constraints. Diagnostic and care delays, in addition to inadequate follow-ups

(see Table 3) likely arise due to limited resources directed towards rare disease care and research. These conditions oppose some valuations held by respondents, such as benefiting from prompt care, in-depth investigations, and good support following a diagnosis (see Table 3). Moreover, agents may feel that some contextual and relational factors, such as prejudices directed by clinicians against individuals living with rare diseases, are out of their reach given how normalized and widespread they are (Racine, 2022; Quintal et al., unpublished work 2).

In both cases, these contextual and relational factors may be experienced as *constraints to agency*, where agents feel unable to act upon the circumstances they are stricken with (Quintal et al., unpublished work 1). Constraints to agency hold existential importance because the ability to act upon life circumstances is key to human flourishing and fulfilment (Ryff, 1989; Ryff & Singer, 2008). More specifically, one dimension of the synthetic and multidimensional influential model of psychological well-being of Ryff and Singer is environmental mastery, or the ability to “manipulate and control complex environments” (2008, p. 22). Hence, given the importance of enacting valuations for moral and existential fulfilment, constraints to agency call for establishing conditions favorable to agents’ empowerment.

First, organizational constraints responsible for inadequate rare disease care should be addressed through a political inquiry featuring deliberations with key stakeholders (Shook, 2010). To this end, the Québec government has created a working group tasked to reflect on a national rare disease strategy, composed of medical specialists, patient associations, and patient partners. Following a first round of deliberations, Québec’s Policy for Rare Diseases was published in 2022. The policy suggests improving training of healthcare professionals, access to diagnosis and care for patients, in addition to research (Ministère de la Santé et des Services sociaux, 2022). Translating this policy into organizational changes could yield improved conditions for empowerment in living with rare diseases.

Second, contextual and relational factors appearing insurmountable to agents due to their prevalence, such as negative clinician attitudes, could be alleviated through a diversity of interventions. In combination with improved training opportunities for clinicians and organizational changes as suggested above, complementary interventions targeting patients could be implemented. These interventions would expose them to a diverse range of empowerment strategies such as those presented in Table 5. These interventions could be offered through webinars, workshops, or information videos. These interventions could be developed in collaboration with patient partners and clinician researchers in the spirit of the pluralist and participatory orientation of pragmatist ethics (Racine, 2014).

Growth and Flourishing Amid Moral Residue

In the integrative model, the use of empowerment strategies (component 6) in the application of action scenarios (concept b) may result in the evolution of situations (component 7). The resulting factual consequences (component 8) may modify the contextual and relational factors inherent to the situation, provided that they are amenable to change (components 1 and 2). Residual emotions and feelings, either

positive or negative (component 9) may occur, depending on the extent to which agents' valuations are still jeopardized. Agents may ultimately consider that the situation is solved, either satisfactorily or unsatisfactorily (component 10).

The enactment of action scenarios is central to ethical inquiry, a process extensively described in the pragmatist ethics literature. Agents use action scenarios as strategies to tackle the moral and existential struggles inherent to situations (Miller et al., 1996). They imagine and refine these action scenarios as they gather empirical data on the components of their situations and through deliberation with others (Hartman et al., 2019).

As agents deploy successful action scenarios, they can experience personal growth and flourishing (component 10). Agents become stronger and clearer advocates for their own valuations. They reinforce their agency by appropriating fruitful empowerment strategies and critically reflecting upon their actions (Aiguier & Loute, 2016; Racine, 2022). They refine their personal morality as they enact and reinforce valuations that align with their existential purpose (Racine, 2022):

knowledge of moral experience is considered instrumental in assessing the kinds of moral beliefs and behaviours that are most conducive to human flourishing. The real test of truth of moral judgments and beliefs lies in their ability to promote enriched experiences that align with human flourishing. (Racine & Cascio, 2020, 6)

Human flourishing is a state and a process where agents pursue objectives they judge to be meaningful and exercise their potential towards them (Lanteigne et al., 2021; Witten et al., 2019). Working towards positive relations with others, purpose in life, greater autonomy, and self-acceptance is conducive to human flourishing, along the environmental mastery and personal growth previously discussed (Lanteigne et al., 2021; Ryff, 1989; Ryff & Singer, 2008).

Alternatively, action scenarios may not always successfully enable agents to enact their valuations. Agents may experience moral residue (concept d) in response to negative emotions and feelings (component 9), such as malaise, distress, and frustration, and the unsatisfactory resolution of their situations (component 10). Moral residue prompts further inquiry (concept e), where agents imagine and enact alternative action scenarios (concept b) (Racine, 2016). The need for such scenarios in several cases is indicative of the iterative and open-ended nature of ethical inquiry (Miller et al., 1996; Racine, 2022). Moral residue may also arise along situations that are overall satisfactorily resolved.

Nonetheless, situations often constitute opportunities for growth and flourishing even if they do not evolve towards a satisfactory resolution. This reality mirrors how illness experiences are conceptualized by Moreira (2019, p. 3654): they “imply continuous and intimate care, and carry with them the images of terminality and suffering, and may also imply hope, happiness, as a possibility of reinvention and creativity exercise.” In sum, the existential importance of moral experiences and inquiry for personal growth and flourishing reiterates the need for interventions supporting the empowerment of individuals living with rare diseases.

Limitations of the Study

A first limitation of the current study is that respondents may have needed elevated levels of literacy to provide a long testimony. Thus, individuals with lower literacy may inadvertently be underrepresented in the study sample, as evidenced by the exclusion of a high number of incomplete or ineligible questionnaires. A second limitation of the study is that men and visible minority groups are also underrepresented in the sample, which could have limited the diversity of morally problematic situations identified. Men and visible minority groups make up 49% and 13% of the Québec population respectively according to Statistics Canada but represent 16% and 2% of study respondents. A third limitation of the study is that respondents' testimonies provide key information allowing to understand their experiences but may inevitably leave out other components that morally and existentially matter to them. This could be due to time limitations but also due to the sometimes narrow understanding that individuals have regarding their moral lives despite providing them with a simple definition of the morally problematic situation. More fundamentally, a fourth limitation of our study is that it does not identify which morally problematic situations are uniquely experienced or exacerbated in the context of rare diseases given the non-comparative study design.

Conclusion

This study, developed through participatory research and methods, presented the moral experiences of adults living with rare diseases through the lens of the morally problematic situation and its components. Ultimately, the study puts forward an integrative model which presents the components of morally problematic situations and explains their main interactions. The situations experienced by adults living with rare diseases, of great moral and existential importance, call for changes in clinical practices and empowerment of patients. In parallel, this model, derived from the results of this study and literature on pragmatist ethics and moral experience, could constitute a useful tool to guide future empirical bioethics research on moral experiences. Conversely, the components of the model and their interactions could be refined and validated through empirical bioethics research conducted with various patient populations and diverse stakeholders, such as clinicians and families. A comparative study between individuals living with rare diseases and those living with common chronic conditions could also be undertaken to discern which situations are unique or salient for the former population. It is possible—although to be verified—that the distress occasioned by the lack of diagnosis, the unknowns surrounding treatments and prognosis associated with many rare diseases and reported in the current study, all compound how illness impacts moral life and attitudes toward what is valued. This would increase the importance of understanding and addressing the connection between the experiences associated with rare diseases and their impact on moral life.

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Author Contributions AQ and ER designed most of the study and secured funding. AQ, ÉH, and ER analyzed the survey data. AQ was responsible for writing the manuscript. CH, IC, ADG, and YB significantly contributed to improving the study design, interpreting its results, and enriched the manuscript. All authors have approved the final manuscript. AQ and ER accept full responsibility for the finished work, had access to the data and controlled the decision to publish.

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Data Availability The datasets generated and analysed during the current study are not publicly available in order to protect respondents' confidentiality.

Declarations

Competing interests The authors have no relevant financial or non-financial interests to disclose.

Ethical Approval This study was approved by the human subject ethics committee of our institution (2021-1080). It complied with the Standards on Research Ethics and Scientific Integrity of the Fonds de recherche du Québec—Santé and the Tri-Council Policy Statement 2—Ethical Conduct for Research Involving Humans of the Panel on Research Ethics of the Canadian government. Survey respondents gave informed consent before participating to the study.

Consent to Participate Free and informed written consent was obtained from all survey respondents. The respondents have given written consent to the inclusion of material pertaining to themselves in the manuscript, acknowledging that they cannot be identified in the manuscript and that their statements are fully anonymized.

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References

- Adler, N. E., Epel, E. S., Castellazzo, G., & Ickovics, J. R. (2000). Relationship of subjective and objective social status with psychological and physiological functioning: Preliminary data in healthy, white women. *Health Psychology, 19*(6), 586–592. <https://doi.org/10.1037/0278-6133.19.6.586>
- Aiguier, G., & Loute, A. (2016). L'intervention éthique en santé: Un apprentissage collectif. *Nouvelles Pratiques Sociales, 28*(2), 158–172. <https://doi.org/10.7202/1041185ar>
- Åsbring, P., & Närvänen, A. L. (2003). Ideal versus reality: Physicians perspectives on patients with chronic fatigue syndrome (CFS) and fibromyalgia. *Social Science & Medicine, 57*(4), 711–720. [https://doi.org/10.1016/S0277-9536\(02\)00420-3](https://doi.org/10.1016/S0277-9536(02)00420-3)
- Baarsma, M. E., Claassen, S. A., van der Horst, H. E., Hovius, J. W., & Sanders, J. M. (2022). Knowing the entire story—A focus group study on patient experiences with chronic Lyme-associated

- symptoms (chronic Lyme disease). *BMC Primary Care*, 23(139), 1–12. <https://doi.org/10.1186/s12875-022-01736-5>
- Biesta, G. (2010). Pragmatism and the philosophical foundations of mixed methods research. In A. Tashakkori & C. Teddlie (Eds.), *Sage handbook of mixed methods in social and behavioral research* (pp. 95–118). Sage Publications.
- Blease, C., Carel, H., & Geraghty, K. (2017). Epistemic injustice in healthcare encounters: Evidence from chronic fatigue syndrome. *Journal of Medical Ethics*, 43(8), 549–557. <https://doi.org/10.1136/medethics-2016-103691>
- Bogart, K., Hemmesch, A., Barnes, E., Blissenbach, T., Beisang, A., & Engel, P. (2022). Healthcare access, satisfaction, and health-related quality of life among children and adults with rare diseases. *Orphanet Journal of Rare Diseases*, 17(1), 1–18. <https://doi.org/10.1186/s13023-022-02343-4>
- Bogart, K. R., & Irvin, V. L. (2017). Health-related quality of life among adults with diverse rare disorders. *Orphanet Journal of Rare Diseases*, 12(1), 1–9.
- Braun, V., & Clarke, V. (2006). Using thematic analysis in psychology. *Qualitative Research in Psychology*, 3(2), 77–101. <https://doi.org/10.1191/1478088706qp0630a>
- Brendel, D. H., & Miller, F. G. (2008). A plea for pragmatism in clinical research ethics. *The American Journal of Bioethics*, 8(4), 24–31.
- Carricaburu, D., & Pierret, J. (1995). From biographical disruption to biographical reinforcement: The case of HIV-positive men. *Sociology of Health & Illness*, 17(1), 65–88. <https://doi.org/10.1111/1467-9566.ep10934486>
- Dewey, J. (2003). *Oeuvres complètes: Tome 1, Reconstruction en philosophie*. Editions Farrago.
- Dewey, J., & Chipman, A. H. (1920). *Letters from China and Japan*. EP Dutton Company.
- Epstein, E. G., & Hamric, A. B. (2009). Moral distress, moral residue, and the crescendo effect. *Journal of Clinical Ethics*, 20(4), 330–342.
- European Organisation for Rare Diseases. (2005). *Rare diseases: Understanding this public health priority*. Eurordis. Retrieved November 18, 2022, from https://www.eurordis.org/wp-content/uploads/2009/12/princeps_document-EN.pdf
- Fins, J. J., Bacchetta, M. D., & Miller, F. G. (1997). Clinical pragmatism: A method of moral problem solving. *Kennedy Institute of Ethics Journal*, 7(2), 129–145. <https://doi.org/10.1353/ken.1997.0013>
- Forman, J., Taruscio, D., Llera, V. A., Barrera, L. A., Coté, T. R., Edjäll, C., Gavhed, D., Haffner, M. E., Nishimura, Y., Posada, M., Tambuyzer, E., Groft, S. C., & Henter, J.-I. (2012). The need for worldwide policy and action plans for rare diseases. *Acta Paediatrica*, 101(8), 805–807.
- Gallagher, S. (2014). Pragmatic interventions into enactive and extended conceptions of cognition. *Philosophical Issues*, 24(1), 110–126. <https://doi.org/10.1111/phis.12027>
- Garrino, L., Picco, E., Finiguerra, I., Rossi, D., Simone, P., & Roccatallo, D. (2015). Living with and treating rare diseases: Experiences of patients and professional health care providers. *Qualitative Health Research*, 25(5), 636–651. <https://doi.org/10.1177/1049732315570116>
- Good, B. (1994). *Medicine, rationality and experience: An anthropological perspective*. Cambridge University Press.
- Government of Canada. (1998, July 15), last modified May 24, 2017. *Previous standard—ethnicity*. Statistics Canada. Retrieved November 18, 2022, from <https://www.statcan.gc.ca/en/concepts/definitions/previous/ethnicity2>
- Grut, L., & Kvam, M. H. (2013). Facing ignorance: People with rare disorders and their experiences with public health and welfare services. *Scandinavian Journal of Disability Research*, 15(1), 20–32. <https://doi.org/10.1080/15017419.2011.645870>
- Hartman, L., Metselaar, S., Widdershoven, G., & Molewijk, B. (2019). Developing a ‘moral compass tool’ based on moral case deliberations: A pragmatic hermeneutic approach to clinical ethics. *Bioethics*, 33(9), 1012–1021. <https://doi.org/10.1111/bioe.12617>
- Hitlin, S. (2003). Values as the core of personal identity: Drawing links between two theories of self. *Social Psychology Quarterly*, 66(2), 118–137. <https://doi.org/10.2307/1519843>
- Hunt, M. R., & Carnevale, F. A. (2011). Moral experience: A framework for bioethics research. *Journal of Medical Ethics*, 37(11), 658–662. <https://doi.org/10.1136/jme.2010.039008>
- Institute of Medicine (US) Committee on Accelerating Rare Diseases Research and Orphan Product Development. (2010). Profile of rare diseases. In M. J. Field & T. F. Boat (Eds.), *Rare diseases and orphan products: Accelerating research and development* (pp. 41–72). National Academies Press. <https://www.ncbi.nlm.nih.gov/books/NBK56184/>
- Jagosh, J., Macaulay, A. C., Pluye, P., Salsberg, J., Bush, P. L., Henderson, J., Sirett, E., Wong, G., Cargo, M., Herbert, C. P., Seifer, S. D., Green, L. W., & Greenhalgh, T. (2012). Uncovering the benefits

- of participatory research: Implications of a realist review for health research and practice. *Milbank Quarterly*, 90(2), 311–346. <https://doi.org/10.1111/j.1468-0009.2012.00665.x>
- Jensen, B., Broesby-Olsen, S., Bindslev-Jensen, C., & Nielsen, D. S. (2019). Everyday life and mastocytosis from a patient perspective—a qualitative study. *Journal of Clinical Nursing*, 28(7–8), 1114–1124. <https://doi.org/10.1111/jocn.14676>
- Jonsen, A. R., Siegler, M., & Winslade, W. (2010). *Clinical ethics: A practical approach to ethical decisions in clinical medicine* (7th ed.). McGraw Hill Professional.
- Jonsen, A. R. (1990). Case analysis in clinical ethics. *Journal of Clinical Ethics*, 1(1), 63–66.
- Kesselheim, A. S., McGraw, S., Thompson, L., O’Keefe, K., & Gagne, J. J. (2015). Development and use of new therapeutics for rare diseases: Views from patients, caregivers, and advocates. *Patient*, 8(1), 75–84.
- Kleinman, A., & Benson, P. (2006). Culture, moral experience and medicine. *The Mount Sinai Journal of Medicine*, 73(6), 834–839.
- Lanre-Abass, B. (2010). Racism and its presuppositions: Towards a pragmatic ethics of social change. *Human Affairs*, 20(4), 364–375. <https://doi.org/10.2478/v10023-010-0037-5>
- Lanteigne, A., Genest, M., & Racine, E. (2021). The evaluation of pediatric-adult transition programs: What place for human flourishing? *SSM: Mental Health*, 1(100007), 1–8. <https://doi.org/10.1016/j.ssmmh.2021.100007>
- Levinas, E. (2002). Useless suffering. In R. Bernasconi & D. Wood (Eds.), *The provocation of Levinas* (pp. 168–179). Routledge.
- Martinet, G., Carrot, M., Chirac, A., Dupuis-Girod, S., Fargeton, A. E., Da Conceição, S. B., & Fourdrinoy, S. (2020). Hereditary hemorrhagic telangiectasia and health-related quality of life: A qualitative investigation. *Quality of Life Research*, 29(5), 1291–1299.
- Matta, R. da. (1981). *Relativizando: Uma introdução à antropologia social*. Editora Vozes.
- Miller, F. G., Fins, J. J., & Bacchetta, M. D. (1996). Clinical pragmatism: John Dewey and clinical ethics. *Journal of Contemporary Health, Law & Policy*, 13(1), 27–51.
- Miller, F. G., Fletcher, J. C., & Fins, J. J., et al. (1997). Clinical pragmatism: A case method of moral problem solving. In J. C. Fletcher (Ed.), *Introduction to clinical ethics*, second edition (pp. 21–34). University Publishing Group.
- Ministère de la Santé et des Services sociaux. (2022). *Politique québécoise pour les maladies rares*. Bibliothèque et Archives nationales du Québec. Retrieved July 4, 2022, from <https://publications.msss.gouv.qc.ca/msss/document-003397/>
- Misak, C. (2013). *The American pragmatists*. Oxford University Press.
- Molster, C., Urwin, D., Di Pietro, L., Fookes, M., Petrie, D., van der Laan, S., & Dawkins, H. (2016). Survey of healthcare experiences of Australian adults living with rare diseases. *Orphanet Journal of Rare Diseases*, 11(1), 1–12.
- Moreira, M. C. N. (2019). Trajectories and moral experiences of rare and chronic illness in biographies: A theoretical essay. *Ciencia & Saude Coletiva*, 24(10), 3651–3661. <https://doi.org/10.1590/1413-812320182410.33532018>
- Morgan, A., Eccles, F. J. R., & Greasley, P. (2019). Experiences of living with dystonia. *Disability and Rehabilitation*, 43(7), 944–952.
- Nowell, L. S., Norris, J. M., White, D. E., & Moules, N. J. (2017). Thematic analysis: Striving to meet the trustworthiness criteria. *International Journal of Qualitative Methods*, 16(1), 1–13. <https://doi.org/10.1177/1609406917733847>
- O’Brien, B. C., Harris, I. B., Beckman, T. J., Reed, D. A., & Cook, D. A. (2014). Standards for reporting qualitative research: A synthesis of recommendations. *Academic Medicine*, 89(9), 1245–1251. <https://doi.org/10.1097/ACM.0000000000000388>
- Pekarsky, D. (1990). Dewey’s conception of growth reconsidered. *Educational Theory*, 40(3), 283–294. <https://doi.org/10.1111/j.1741-5446.1990.00283.x>
- Racine, E. (2007). HEC member perspectives on the case analysis process: A qualitative multi-site study. *HEC Forum*, 19(3), 185–206. <https://doi.org/10.1007/s10730-007-9046-1>
- Racine, E. (2014). Pragmatism and the contribution of neuroscience to ethics. In T. Solymosi & J. R. Shook (Eds.), *Neuroscience, neurophilosophy and pragmatism: Brains at work with the world* (pp. 243–263). Palgrave Macmillan.
- Racine, E. (2016). Can moral problems of everyday clinical practice ever be resolved? A proposal for integrative pragmatist approaches. In P. L. Rosenbaum, G. M. Ronen, E. Racine, J. Johannesen, & D. Bernard (Eds.), *Ethics in child health: Principles and cases in neurodisability* (pp. 33–48). Mac Keith Press.

- Racine, E. (2022). How ethics liberates experience: Insights from pragmatist theory and contemporary research. *Journal of Speculative Philosophy*, 36(4), 517–536.
- Racine, E., & Cascio, M. A. (2020). The false dichotomy between empirical and normative bioethics. *AJOB Empirical Bioethics*, 11(1), 5–7.
- Racine, E., Kusch, S., Cascio, M. A., & Bogossian, A. (2021). Making autonomy an instrument: A pragmatist account of contextualized autonomy. *Humanities and Social Sciences Communications*, 8(1), 1–15.
- Ryff, C. D. (1989). Happiness is everything, or is it? Explorations on the meaning of psychological well-being. *Journal of Personality and Social Psychology*, 57(6), 1069–1081.
- Ryff, C. D., & Singer, B. H. (2008). Know thyself and become what you are: A eudaimonic approach to psychological well-being. *Journal of Happiness Studies*, 9(1), 13–39.
- Shook, J. R. (2010). Pragmatism, pluralism, and public democracy. *Revue française d'études américaines*, 124(2), 11–28. <https://doi.org/10.3917/rfea.124.0011>
- Vaismoradi, M., Turunen, H., & Bondas, T. (2013). Content analysis and thematic analysis: Implications for conducting a qualitative descriptive study. *Nursing & Health Science*, 15(3), 398–405. <https://doi.org/10.1111/nhs.12048>
- von der Lippe, C., Diesen, P. S., & Feragen, K. B. (2017). Living with a rare disorder: A systematic review of the qualitative literature. *Molecular Genetics & Genomic Medicine*, 5(6), 758–773.
- Witten, H., Savahl, S., & Adams, S. (2019). Adolescent flourishing: A systematic review. *Cogent Psychology*, 6(11640341), 1–16. <https://doi.org/10.1080/23311908.2019.1640341>

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Authors and Affiliations

Ariane Quintal^{1,2,3} · Éliissa Hotte^{1,3} · Caroline Hébert³ · Isabelle Carreau³ · Annie-Danielle Grenier³ · Yves Berthiaume^{3,4} · Eric Racine^{1,2,3,5}

✉ Eric Racine
eric.racine@ircm.qc.ca

¹ Pragmatic Health Ethics Research Unit, Institut de recherches cliniques de Montréal, 110 Pine Avenue West, Montréal, QC H2W 1R7, Canada

² Département de médecine sociale et préventive, Université de Montréal, Montréal, QC, Canada

³ Ethics and Rare Diseases Working Group, Institut de recherches cliniques de Montréal, Montréal, QC, Canada

⁴ Département de médecine, Université de Montréal, Montréal, QC, Canada

⁵ Division of Experimental Medicine, and Department of Neurology and Neurosurgery, Department of Medicine, McGill University, Montréal, QC, Canada