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To cite this article: Ariane Quintal, Élissa Hotte, Annie-Danielle Grenier, Caroline Hébert, Isabelle Carreau, Yves Berthiaume & Eric Racine (14 Dec 2023): Morally Problematic Situations Encountered by Adults Living With Rare Diseases, AJOB Empirical Bioethics, DOI: [10.1080/23294515.2023.2274587](https://doi.org/10.1080/23294515.2023.2274587)

To link to this article: <https://doi.org/10.1080/23294515.2023.2274587>




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Morally Problematic Situations Encountered by Adults Living With Rare Diseases

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ABSTRACT

Background: Rare diseases are generally poorly understood from scientific and medical standpoints due, to their complexity and low prevalence. As a result, individuals living with rare diseases struggle to obtain timely diagnoses and suitable care. These clinical difficulties add to the physical and psychological impacts of living with chronic and often severe medical conditions. From the standpoint of pragmatist ethics, the morally problematic situations that adults living with rare diseases experience matter crucially. However, there is little known about these experiences.

Methods: A survey study was conducted with 121 adults living with rare diseases in Québec, Canada, to identify morally problematic situations encountered in the healthcare system and everyday life as part of a participatory action research project. Morally problematic situations elicited internal tensions and constraints to agency.

Results: Adults living with rare diseases experienced morally problematic situations of stigmatization, disbelief, and sometimes abuse in the healthcare system. These situations were compounded by diagnostic delays, inadequate care, and suboptimal follow-up, and led some individuals to opt-out of medical care. In their personal lives, these individuals sometimes found themselves in situations of physical and financial dependency. They often also had to give up professional occupations, academic training, or life projects.

Conclusions: Adults living with rare diseases experience important morally problematic situations navigating the healthcare system and their everyday lives, some of which could be alleviated through interventions developed through future participatory action research.

KEYWORDS

Rare diseases; ethics; surveys and questionnaires; community-based participatory research; pragmatist ethics; pragmatism

Introduction¹

The common understanding and definition of rare diseases is that their prevalence is under 1/2,000 (European Organisation for Rare Diseases 2005). Due to the low prevalence of rare diseases, scientific advances, physician training opportunities, and clinical practice guidelines relating to rare diseases are scarce. Medical students are taught to look out for common rather than rare conditions through the adage, “when you hear hoofbeats, think horses not zebras” (Cormier and Buikstra 2021, 196). As a result, rare diseases might not be properly investigated, promptly diagnosed, or adequately treated (Huyard 2009;


Ramalle-Gómara et al. 2020). These medical hardships add to the physical and social limitations, stigmatization, and resulting psychological distress that individuals living with rare diseases endure (von der Lippe, Diesen, and Feragen 2017).

Pragmatist ethics and the need to understand morally problematic situations

The moral implications of the situations that individuals living with rare diseases face have not been previously tackled or made explicit, despite some studies addressing the moral issues raised by rare diseases at a societal level, notably regarding clinical trials and

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 Supplemental data for this article can be accessed online at <https://doi.org/10.1080/23294515.2023.2274587>

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resource allocation (Kesselheim et al. 2015; Neil and Craigie 2004; Picavet et al. 2013).

Pragmatist theory proposes that ethics adopts a situational, embodied, and contextualized outlook on moral situations, to show how ordinary experiences are actually morally salient (Fesmire 2003; Gross 2009; Pappas 1997). Further, it advances that moral problems are experienced in social situations by individuals who are *agents*. That is to say, individuals, as active agents, construct an interpretation of these situations and ideally act upon them to bring about their resolution (Aiguier and Loute 2017; Racine et al. 2021). Dewey pioneered the concept of *morally problematic situations* to describe such episodes of life that are “disturbed, troubled, ambiguous, confused, full of conflicting tendencies, obscure, etc.” (Dewey 1938, 105; Fesmire 2003). Morally problematic situations are experienced as such because they elicit an internal tension within the agent or constrain their agency such that they generate conflicting and vexing feelings as described by Dewey (Quintal et al. [under review](#); Racine 2022). In these situations, agents experience distress because they are unable to fully enact their values because of unsatisfactory courses of action (Racine 2007).

Morally problematic situations can be understood through inquiry to foster their eventual resolution (Foucart 2014). More specifically, inquiry is a deliberative and reflective process involving imagining, testing, and refining action scenarios to conciliate agents’ values with contextual factors inherent to the situation (Inguaggiato et al. 2019). Resolving a morally problematic situation can elicit growth and flourishing within agents who overcome a problem and actualize themselves as ethical agents (Racine et al. 2019).

A previous interpretive qualitative literature review revealed that adults living with rare diseases experience morally problematic situations throughout their medical journeys (Quintal et al. [under review](#)). Some situations evoked internal tensions within these individuals, such as unfulfilled expectations and the feeling of being misunderstood. In contrast, others featured constraints to agency by disempowering these individuals or involving decisions taken on their behalf (Quintal et al. [under review](#)). However, the qualitative studies included in this literature review were not designed with the concept of the morally problematic situation in mind. As a result, the literature review only provides a limited account of these situations while being restricted to situations arising in healthcare settings. Hence, more in-depth empirical work closely informed by the concept of the morally problematic situation is needed. This would enable a

deeper understanding of the moral salience of these situations within healthcare settings and beyond, especially since the ordinary experiences of people with rare diseases appears to be trivialized as being similar to those of other people, thus further fueling stigma against them.

Objectives of the study

The two objectives of this study were to: (1) highlight the scope of morally problematic situations encountered by adults living with rare diseases² in the province of Québec; and (2) acquire an in-depth understanding of the moral dimensions of these morally problematic situations. This study focused on adults given that they face poorer recognition by healthcare professionals than children living with rare diseases (Esquivel-Sada and Nguyen 2018). This study uses a pragmatist ethics lens to highlight the moral significance of rare disease experience which can otherwise be mistakenly assumed to non-morally problematic experience of healthcare as in most other health conditions.

Materials and methods

The methodology used to conduct this survey is described briefly here and more extensively in Appendix 1.

Survey design and launch

The survey was developed and interpreted through a participatory action research approach (Cordeiro and Soares 2018), with the involvement of a purposely created *Ethics and Rare Diseases Working Group*. The working group was comprised of two bioethics researchers (authors 1 and 7), three patient partners living with rare diseases (authors 3–5), representatives from the Québec Coalition of Orphan Diseases, two clinician scientists (including author 6), and a research assistant (author 2).

The survey was written in French and hosted on the Eval&Go survey design platform (<https://www.evalandgo.com/>). The survey targeted adults living with rare disease(s) residing in Québec, officially diagnosed or not. Recruitment was focused to this socio-political context consistent with a participatory action research approach. The survey included a consent form and questions on demographic and socioeconomic profiles, rare disease profiles, and multifaceted impacts of rare diseases (see Appendix 2). Respondents

were provided with a simple definition of the morally problematic situation compatible with the literature reviewed in the Introduction, that is:

A morally problematic situation is a difficult life experience evoking uncertainty, distress, or a dilemma regarding the best way to act. We all come across such situations that are morally arduous. These situations collide with our values, our preferences, and our priorities.

These situations can emerge in everyday life. They can relate to family, routine, leisure, moving around, interactions with others, or personal finances. These situations can also arise in relation to important life decisions, like choosing a medical treatment, choosing to have a child or not, or choosing a job. Adults living with rare diseases are very susceptible to experience morally problematic situations. They experience various constraints in relation to their health, forcing them to make difficult and heartbreaking choices based on what matters most to them in life. Despite the importance of these situations, they are poorly understood by researchers and healthcare professionals. This lack of knowledge makes it even more difficult to support individuals facing these hardships.

Respondents ranked three to eight morally problematic situations they have faced in order of importance for them. They then described more extensively the most important situation they faced. At the end of the survey, respondents could provide their contact

information to enroll in a draw and to volunteer for subsequent phases of the project. The survey was launched on March 12, 2021, and ended on May 9, 2021. The Québec Coalition of Orphan Diseases, along with more than 80 patient associations and online support groups, were invited to advertise the survey (see Appendix 3).

Data analyses

Responses to all survey sections were analyzed except for the extensive descriptions of situations, which will be treated elsewhere using a fitting methodology.

Quantitative analyses

The survey data was imported into a Microsoft Excel (2019) spreadsheet. Questionnaires were excluded if they were duplicates, if they lacked information on respondents' profiles, or if respondents did not have a rare disease. A second exclusion phase involved discarding questionnaires which did not discuss situations that were morally problematic (see Figure 1). Descriptive statistics were conducted on respondents' profiles and the multifaceted impacts of their rare diseases. Chi-squared analyses were conducted on questionnaires discarded during the second exclusion phase to establish whether the included and excluded

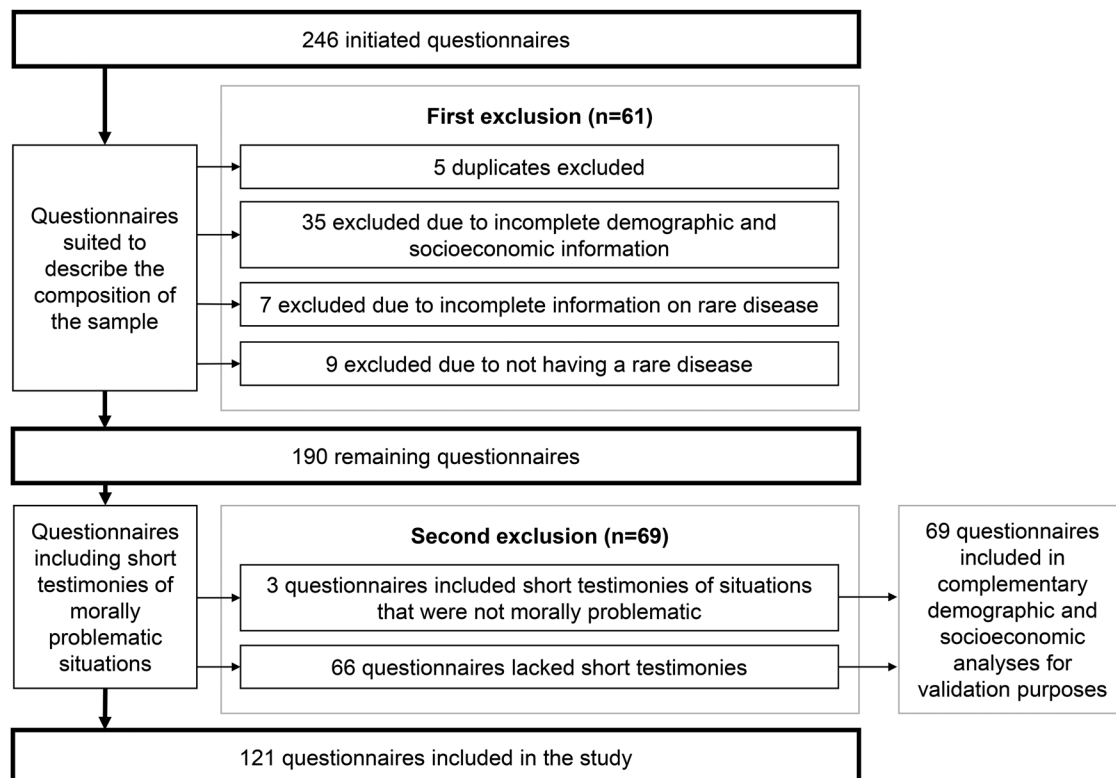


Figure 1. Exclusion process leading to the inclusion of 121 eligible questionnaires.

respondents were statistically different. A p -value inferior to 0.05 was considered statistically significant.

Qualitative analyses

The morally problematic situations described in the short testimonies were imported into Microsoft Word (2019) documents. They were summarized in concise sentences ordered by the first and second authors while preserving the ranking attributed to them by respondents. All summaries were reviewed by both authors. Summaries were excluded from further analyses if they lacked moral content (see Appendix 1 for exclusion criteria). Included summaries were compatible with the given definition of the morally problematic situation and with the literature reviewed in the Introduction. The included summaries were imported into MaxQDA (2020) and subjected to an inductive thematic analysis (Braun and Clarke 2006; Nowell et al. 2017). The initial coding guide was initiated by the first author and completed by the second author. For practical reasons, intercoder reliability testing was not conducted. Nonetheless, to favor the reliability of the coding process, the first author instructed the second author on how to conduct the subsequent coding phases, and to ensure consistency and discuss disagreements. The last author also reviewed the completed coding.

The resulting primary themes denoted life domains where the situations emerged (e.g., personal life). Secondary themes designated spheres of activity relating to these domains (e.g., “financial hardships” was a secondary theme classified under “personal life”). Tertiary themes denoted types of situations (e.g., “expensive treatments and care” was a tertiary theme classified under “financial hardships”). Quaternary themes referred to concrete situations encountered by respondents.

Patient partners each prioritized the 6–12 most important situations for them among those ranked as the most or second to most important situations by respondents. A deductive thematic analysis was conducted on these situations as well as on a few complementary situations selected by our research team (Nowell et al. 2017). This analysis supported the identification of these situations’ moral dimensions. This analysis was informed by preexisting themes, namely internal tensions and constraints to agency, which are two moral dimensions of morally problematic situations (Quintal et al. [under review](#)). An internal tension corresponded to a subjective malaise felt by an agent overwhelmed by a situation that provokes questioning about important human values and dimensions of human existence. Constraints to agency were experienced as

limitations to one’s ability to act on a morally important aspect of the situation one is faced with. Types of internal tensions and constraints to agency have been described elsewhere (Quintal et al. [under review](#)).

Reporting of results

Summaries of morally problematic situations are synthesized in the tables following the inductive thematic analysis. Examples of important morally problematic situations, along with their moral dimensions, are provided in the accompanying text. These results, elucidated through the deductive thematic analysis, constitute an explicit effort to interpret the respondents’ statements using a pragmatist ethics lens which helps show and explain (e.g., *via* the concepts of internal tensions and constraints to agency) how some seemingly ordinary and non-moral experiences are actually morally problematic for those living them. Quotations were translated from French to English by the first author, and an anglophone ensured that the translations accurately conveyed the words used by respondents.

Ethics approval

This study has been approved by the human subject ethics committee of the Montreal Clinical Research Institute (IRCM). It was conducted in compliance 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.

Results

Sample characteristics

Demographic and socioeconomic characteristics of respondents

Demographic and socioeconomic analyses were conducted on 121 eligible questionnaires (see [Figure 1](#)). Most respondents self-identified as women (79%). Respondents were aged between 18 and 79 years old, had a mean (SD) age of 48.5 (12.4) years old, and 61% were aged between 40 and 59 years old. [Table 1](#) presents the demographic and socioeconomic characteristics of the 121 respondents.

The demographic and socioeconomic characteristics of the included respondents ($n=121$) and respondents excluded during the second exclusion phase ($n=69$) were mostly not statistically different. Notably, the included sample was proportionally comprised of

Table 1. Demographic and socioeconomic characteristics of respondents.

Characteristics of respondents	Number of respondents	% of sample
With what gender do you self-identify?		
Men	25	21
Women	96	79
What is your age?		
18–29	6	5
30–39	26	22
40–49	33	27
50–59	28	23
60–69	24	20
70–79	4	3
What is the highest-level education that you have achieved?		
Elementary school	1	1
High school	14	12
Professional school	6	5
College*	33	27
Bachelor's degree	37	31
Graduate studies	29	24
Not specified	1	1
What is your main occupation?		
Student	2	3
Employed	41	34
Disability	53	44
Retired	19	16
Other	3	3
Prefer not to say	2	2
What is your annual familial income?		
Less than \$10,000	9	7
\$10,001–30,000	20	17
\$30,000–50,000	18	15
\$50,001–70,000	17	14
\$70,001–90,000	15	12
\$Over 90,001	27	22
Prefer not to say	15	12
Please indicate how you would position yourself in terms of socioeconomic status on a scale from 0 to 10.		
Low (scores of 0–4)	37	31
Medium (scores of 5–7)	62	51
High (scores of 8–10)	22	18
What is your first language?		
French	113	93
English	4	3
French and English	2	2
Other	2	2
Which ethnic group do you self-identify as belonging to?		
White	117	97
Other/prefer not to say	4	4
Do you practice a religion?		
Yes	22	18
No	90	74
Don't know	1	1
Prefer not to say	8	7

*College education in the province of Québec in Canada is typically offered by CEGEPs (Collège d'enseignement général et professionnel). CEGEPs are post-secondary education institutions exclusive to Québec which offer 3-year training programs for trades and 2-year preparatory training programs for university studies.

more individuals aged between 30 and 39 years old, more individuals unable to work due to disability, and fewer retired individuals (see Appendix 4).

Categories of rare diseases represented in the sample and their prevalence across respondents

In total, 95 rare diseases were represented among respondents. The four most common rare diseases among respondents were Ehlers-Danlos syndromes

Table 2. Categories of rare diseases represented in the study and their prevalence across respondents.

Category	Number of rare diseases	% of all rare diseases in the study
Endocrine, nutritional and metabolic diseases	16	17
Congenital malformations, deformations, and chromosomal abnormalities	15	16
Diseases of the musculoskeletal system and connective tissue	13	14
Diseases of the nervous system	11	12
Diseases of the circulatory system	9	9
Neoplasms	5	5
Diseases of the skin and subcutaneous tissue	5	5
Diseases of the blood and blood-forming organs and certain disorders involving the immune system	4	4
Diseases of the respiratory system	4	4
Diseases of the genitourinary system	3	3
Certain infectious and parasitic diseases	3	3
Diseases of the digestive system	2	2
Diseases of the eye and adnexa	2	2
External causes of morbidity and mortality	1	1
N/A	2	2
Total	95	100

(14%), Sjogren's syndrome (7%), scleroderma (5%), and cystic fibrosis (4%). From the sample, 92% of respondents had a diagnosis for their rare disease, 6% suspected having a specific rare disease without having a formal diagnosis, and 2% suspected having an unknown rare disease. Table 2 classifies the rare diseases affecting respondents according to the 10th revision of the International Statistical Classification of Diseases and Related Health Problems (<https://icd.who.int/browse10/2016/en>).

Diagnostic delays experienced by respondents

Diagnostic delays varied between less than a year (23%), 1–5 years (23%), 6–10 years (14%), 11–30 years (17%), and more than 30 years (14%), or was unknown (3%) or unclearly specified (6%).

Multifaceted impacts of rare diseases

Rare diseases had multifaceted and sometimes severe impacts on respondents (see Table 3), causing prevalent limitations to their everyday lives. Among notable results (i.e., where the sum of the scores for the “strongly agree” and “agree” categories exceeded 80%), having a rare disease affected relationships with close

Table 3. Multifaceted impacts of rare diseases.

Statement	% of agreement (n)			
	Agreement	Neutral	Disagreement	Unknown or not applicable
Your rare disease(s) has/have an impact on your relationships with your close ones (partner, kids, family, friends).	83% (101)	6% (7)	10% (12)	1% (1)
Your rare disease(s) has/have impacted your career.	82% (100)	7% (9)	6% (7)	4% (5)
Your rare disease(s) lead(s) to the degradation of your physical health or mental capacities.	82% (99)	9% (11)	7% (8)	2% (3)
Your rare disease(s) limit(s) your ability to participate to leisure activities that you enjoy.	80% (97)	5% (6)	15% (18)	0% (0)
Your rare disease(s) limit(s) your capacity to participate in social activities.	77% (94)	8% (10)	13% (16)	1% (1)
Your rare disease(s) limit(s) your mobility.	74% (89)	11% (13)	14% (18)	1% (1)
You rely on medication to maintain a good quality of life.	73% (88)	5% (6)	13% (16)	9% (11)
Your rare disease(s) impose(s) diet constraints.	67% (81)	14% (17)	16% (20)	2% (3)
You rely on medication to survive.	50% (60)	10% (12)	23% (28)	17% (21)
Your rare disease(s) reduce(s) your life expectancy.	45% (54)	18% (22)	11% (14)	26% (31)
Your rare disease(s) impact(s) your decision to have or not to have kids.	41% (50)	7% (8)	8% (9)	45% (54)
Your rare disease(s) has/have impacted your studies.	34% (41)	14% (17)	24% (29)	28% (34)
Living with your rare disease(s) also implies living with a malformation.	30% (36)	8% (10)	30% (37)	32% (38)

ones for 83% of respondents, impacted the careers of 82% of respondents, decreased the mental or physical state of 82% of respondents, and restricted leisure activities for 80% of respondents.

Qualitative results

The 121 analyzed questionnaires included 615 short testimonies, from which were generated 665 summaries of morally problematic situations. Based on an existing framework (Quintal et al. [under review](#)), internal tensions could materialize as abandonment, uncertainty, loss of trust, unfulfilled expectations, injustice, being lied to, not being listened to, ambivalence, being misunderstood, or unanswered questions. In contrast, constraints to agency were experienced as internalized self-doubt, powerlessness, having one's experience and knowledge disregarded, not being provided with adequate information, having decisions taken on one's behalf, dehumanization, compartmentalized understanding of the self by others, and moralization (Quintal et al. [under review](#)).

Morally problematic situations encountered in healthcare

Respondents experienced morally problematic situations in healthcare. These distressing situations arose when seeking a diagnosis, during interactions with healthcare workers, in relation to medical interventions, and during medical follow-ups, and had long-lasting consequences (see [Table 4](#)).

Stigmatization and disbelief. Several respondents were afflicted by situations of stigmatization and disbelief

by clinicians. They were belittled by clinicians in addition to being met with a lack of compassion, and a lack of recognition that their illnesses had physiological causes (see [Table 4](#)). Disturbingly, a woman explained:

When I experienced a carotid artery dissection related to my illness, doctors initially suspected that I was simulating pain to obtain drugs. I was first evaluated by a psychiatrist and placed in isolation because my cries of pain were disturbing. Once the pain subsided, I was promptly discharged. The following day, the same excruciating pain resurfaced, prompting me to seek care at a different hospital. Finally, a proper diagnosis of the dissection and [my fibromuscular dysplasia] was made.

Another notable example: a former professional engineer in his late thirties, living with Ehlers-Danlos syndrome, pudendal neuralgia, and rare complications from a commonly prescribed drug, was unable to be diagnosed in a timely fashion and with respect:

I have lived a nightmare in the Québec healthcare system due to a lack of recognition [of my illness] and a diagnostic odyssey [...]. I have literally been abandoned to my fate. My systemic conditions have, for a long time, erroneously been associated with deconditioning and psychiatric/psychological issues by doctors from diverse specialties [...] The majority of doctors have failed to recognize my loss of functional capacity [despite] the fact that I am confined to my house or to my bed most of the time.

These situations were morally problematic, as evidenced by the internal tensions experienced by respondents whom clinicians markedly misunderstood.

Table 4. Morally problematic situations encountered in healthcare.

Sphere	Sub-sphere	Types of morally problematic situations described by respondents
Negative interactions with healthcare workers	Stigmatization	Experiencing condescension, intolerance, or lack of compassion from clinicians. Not being listened to, being ridiculed, or accused of lying.
	Disbelief	Experiencing indifference, not being listened to, or being ignored. Being repetitively and erroneously told by clinicians that the rare disease has psychosomatic roots. Inappropriate referrals to psychotherapists or psychiatrists.
	Psychological and physical abuse	Being insulted, intimidated, mocked, gaslit, or subjected to physical violence by some healthcare workers during hospital stays. Being denied proper hygiene or feeding assistance due to incorrect suspicions of psychosomatic illness.
Diagnosis	Diagnostic delays	Lack of prompt recognition of rare diseases by clinicians. Undergoing taxing medical tests and consultations of limited utility while waiting for a diagnosis, while health deteriorates.
Clinical care and treatment	Non-definitive diagnoses	Rare disease diagnoses being questioned or reversed by other clinicians.
	Medical risks, errors, and complications	Worsening health outcomes resulting from medical interventions inadequately tailored to respondents' unique situations (e.g., surgeries, prescribed orthopedics, contraindicated drugs). Hesitating to undergo risky surgical procedures or being refused these procedures due to clinicians' limited expertise. Difficulties with providing informed consent to surgical procedures due to uncertainty and lack of adequate information.
Medical follow-up	Limited availability of drugs and adequate care	Drugs, interventions, or specialists not being available remotely. Having to travel hundreds of kilometers to receive adequate care. Medical material adapted to one's physical difference (e.g., short stature) being unavailable in hospitals.
	Lack of proactivity	Unwillingness of clinicians to further investigate one's rare disease. Lack of receptivity to the information brought by respondents. Refusal to collaborate with international experts on the rare disease.
Long-lasting consequences	Inadequate follow-up	Not being provided with a treatment plan, support, or follow-up after being diagnosed. Undergoing "medical ping-pong" i.e., successive referrals from one specialist to the next. Having to fiercely advocate for oneself to obtain adequate care.
	Psychological distress	Doubting one's illness experience following unsatisfactory care. Consulting psychotherapists to primarily address distressing healthcare experiences instead of the impacts of the rare disease.
	Opting out from the healthcare system	Losing trust in clinicians, thereby avoiding future clinical consultations. Opting out from follow-up appointments due to the absence of therapeutic avenues.

Respondents considered that these clinicians disregarded their experience and knowledge, did not listen to them, and abandoned them.

Medical risks, errors, and complications. Many respondents were exposed to risks, errors, and complications related to medical interventions. A woman living with Ehlers-Danlos syndrome experienced "symptom denial by a healthcare professional who prescribed me with orthotics that made my condition worse. I foolishly [...] wore the orthosis for a few hours, then I could not walk normally for months." Others hesitated to undergo risky surgical procedures due to the clinicians' limited expertise (see Table 4). For example, a man living with Klippel-Feil syndrome and lipomatosis needed to undergo a surgical procedure. He experienced a morally problematic situation related to the "decision to have surgery or not, knowing that there is no Canadian expert [on my condition] and that the scientific literature recommends real experience in this type of surgery." This situation was morally problematic because it required this man to make a life-changing decision while eliciting internal tensions such as ambivalence and unanswered questions, compounded by several layers of medical uncertainty (Quintal et al. [under review](#)).

Inadequate follow-up. Several respondents were left with inadequate medical follow-ups. For some respondents, this meant the lack of a treatment plan and periodic appointments with specialists following a diagnosis (see Table 4). A man in his sixties explained that when he was diagnosed with cerebellar ataxia of genetic origin, "I was not provided with any treatment. I found it difficult to have no medical follow-up." The situation left him feeling abandoned, powerless and unable to act proactively upon his health, unlike what he had hoped.

A woman living with scleroderma and Sjogren syndrome waited 8 years to be diagnosed. She explained "the most traumatizing situation relating to my illness was being left alone and having to fight against the healthcare system to receive proper care." In addition to the powerlessness she endured, this woman felt a distressing internal tension of abandonment, wherein the lack of medical support condemned her to suffer alone without any help. Moreover, other respondents deplored taxing experiences of "medical ping-pong"—being referred from one unknowledgable specialist to another.

Opting out from the healthcare system. Some respondents had opted out of the medical system due to their past negative experiences or the lack of

treatment for their conditions (see Table 4). This was the case of a young man living with Ehlers-Danlos syndrome who chose to avoid medical consultations:

Doctors in the emergency room lack understanding of my illness. They [doubt] my illness, because it is not very visible and because of my young age. Therefore, I avoid getting medical consultations, including in the emergency room. This puts me in danger, while also causing me constant stress. It is very difficult to feel misunderstood, alone, and not cared for.

Through his tribulations, this young man experienced internal tensions of feeling abandoned, misunderstood, and losing trust. His decision forced him to choose between two paradoxical values, and the desire to be respected as a worthy human being ultimately superseded his need for better care and support.

Morally problematic situations encountered in personal life

Morally problematic situations were encountered in the respondents' personal lives. They related to struggles with illness acceptance or disclosure, practical difficulties in fulfilling one's basic needs and daily tasks, financial hardships, and hindered personal growth (see Table 5).

Dependency in living spaces and personal finances. Some respondents reluctantly transitioned into various situations of dependency due to their rare diseases. Some have become dependent on caregivers to fulfill their basic needs. Others have become dependent on financial support from a partner or disability benefits (see Table 5). A woman living with idiopathic ataxia was devastated as the sudden onset of her illness in her early fifties, which left her with no other choice but to "leave her house and her memories to move to a CHSLD³" where she was no longer visited by her family. Dishearteningly, another woman, in her mid-thirties and living with several rare diseases, deplored that her financial dependence upon her partner weighed significantly on her. She explained:

I am lucky to have an exceptional spouse. However, my physical and financial dependence on him weighs on me and transforms any little quarrel into an excessive fear of ending up in a situation of homelessness and destitution. I have [significant] student debts that will never be paid. I live under [his] financial guardianship [...] despite having intact intellectual faculties. I am not eligible for credit and cannot have possessions, or buy bubble gum, without asking for permission. And only because I am sick. I have no control over my life, except at the psychological level.

Table 5. Morally problematic situations encountered in personal life.

Sphere relating to personal life	Sub-sphere	Types of morally problematic situations described by respondents
Acceptance	Difficulties with acceptance	Difficulties in accepting a diagnosis of a rare disease, especially if incurable, and its associated physical changes. Mourning loss of capacities. Feelings of guilt for incurring costs to society or having likely genetically transmitted a rare disease to children prior to being diagnosed.
Disclosure	Invisible rare diseases	Preference to silently endure symptoms rather than disclosing the rare disease. Frequent need to answer sensitive questions about the rare disease. Distress when announcing a diagnosis of a deadly and rare degenerative disease to close ones.
	Visible rare diseases	Attempting to conceal the physical manifestations of the rare disease. Avoiding social activities to avoid the stares of others.
Basic needs	Undesired or maladapted living spaces	Being forced to change living environments (e.g., moving into a long-term care facility or low-income housing). Difficulty to find adapted living spaces.
	Food restrictions	Having to follow complex, frustrating, and sometimes unappetizing diets or food restrictions that are often misunderstood by others.
	Mobility issues	Incapability of moving freely around (e.g., inability to drive, unsuitable public transportation).
Daily tasks	Strict schedules	Having to carefully plan one's schedule, routines, and limited outings.
	Household chores	Struggling to complete household chores. Having to choose between chores and other occupations (e.g., professional duties, social events).
Personal finances	Limited access to resources and support	Facing delays and various constraints when attempting to access publicly funded psychological support, disability benefits, and domestic assistance.
	Limited income	Inability to work. Frequent ineligibility to governmental disability benefits or to compensation by insurers, notably due to a lack of recognition of rare diseases.
	Medical expenses	Expensive treatments, medical supplies, and care from private specialists (e.g., osteopaths) whose coverage is limited by medical insurers.
Personal growth	Financial hardships	Unaffordable basic expenses due to limited income and medical expenses. Financial dependence upon a partner or family.
	Obstacles in pursuing leisure, activities, and hobbies	Inability to fully engage in leisure, physical activities, outings, and social activities.
	Changing life projects	Being forced to abandon cherished lifelong dreams due to rare diseases (e.g., owning a property, traveling, pursuing a particular career). Frequently needing to reassess life priorities.

Table 6. Morally problematic situations encountered in career and studies.

Sphere relating to career and studies	Sub-sphere	Types of morally problematic situations described by respondents
Career	Changing or abandoning career objectives	Settling for jobs compatible with one's limitations, with minimal demands or favorable employee benefits, but which may be less stimulating.
	Challenging work performance	Being unable to work full time. Facing constraints at work. Jeopardizing one's health to continue working.
	Discrimination and prejudice	Avoiding rare disease disclosure in work environments to avoid discrimination, at the costs of being criticized for poor work performance or being misunderstood. Being ridiculed, discriminated, and humiliated in relation to the rare disease.
	Disability	Losing fulfilling jobs due to repeated absences (e.g., medical appointments, flare-ups). Having to go on prolonged medical leave. Being unable to work and needing to request disability benefits.
Studies	Prolonging or abandoning studies	Prolonging studies, sometimes with mitigated success and health-related costs, or abandoning studies reluctantly.
	Discrimination	Encountering unaccommodating teachers or internship supervisors who jeopardize one's ability to complete a study program. Being refused admission in a study program.

These situations were experienced as morally problematic due to the powerlessness they elicited among respondents, which limited in their ability to act upon the difficult life circumstances imposed on them.

Financial struggles due to poor governmental support. Some respondents reported facing financial hardships and vulnerability. They were often ineligible for financial support or compensation due to poor recognition of their rare diseases (see Table 5). Additionally, a woman living with Behcet's disease lamented that "I have no insurance and there is no social safety net for me. [...] I feel that one day I will have to refuse treatment if my health does not improve because without income, I will no longer have the means to continue living". Such financial struggles are evocative of internal tensions of injustice and abandonment in respondents.

Leisure, activities, and hobbies. A rare disease can significantly hinder personal growth, which limits one's capacity to engage in leisure, activities, and hobbies fully. Several respondents were forced to abandon physical activities (i.e., from short walks to dancing), to limit social activities (e.g., meeting with friends, visiting family), and to renounce to some leisure (e.g., gardening) due to rare-disease-associated symptoms, needs, and limitations (see Table 5). A woman living with hereditary angioedema explained that "learning to say no and to never promise anything to someone is often difficult. I cannot predict whether I will feel good enough for dinner with friends [...]. It is only on the same morning that I can tell," attesting to internal tensions of ambivalence and uncertainty.

Morally problematic situations encountered in career and studies

Respondents experienced morally problematic situations in their careers and studies, which often materialized as challenges in attaining one's objectives or as discrimination (Table 6).

Abandoning career or study objectives due to rare diseases. Some respondents were forced to quit their job or abandon their studies due to their rare diseases, were unable to fulfill professional or academic demands, or had to relinquish their professional or academic objectives (see Table 6). This was the case of a woman living with oculopharyngeal muscular dystrophy. She dreamt of completing a master's degree in rehabilitation and a certificate in gerontology to teach after retiring from her current profession as a nurse manager, but sadly had to give up both aspirations. A woman living with juvenile dermatomyositis explained: "I chose to end my studies because they exhausted me, to the point where I was hospitalized for months at once due to my illness flaring up". She added: "I had to grieve because I always enjoyed going to school and learning." Grieving the loss of such life projects is experienced as morally problematic for respondents who were left with the impossible choice of giving up their dreams. As a result, they had internal tensions of unfulfilled expectations regarding their futures.

Discrimination and prejudice. Some respondents were exposed to alarming situations of discrimination and prejudice in the workplace or their academic pursuits. Avoiding the disclosure of their rare disease to colleagues or superiors was a reported coping strategy.

Conversely, others recounted instances of discrimination and prejudice they had faced, eliciting a variety of internal tensions (see Table 6). A woman living with Dandy-Walker syndrome explained that she faced disbelief and was humiliated by her superior and colleagues regarding her medical absence:

One day, my superior told me it was impossible that I was so sick that a simple headache would prevent me from working and require me to be hospitalized. [On the same day], given that my intracranial pressure rose, I had to undergo emergency surgery. A week later, I was discharged from the hospital and was handing in my medical note. The administrative assistant [...] asked me to come to my superior's office to show him my scars. I never dared to go back to work at that place.

In this situation, the woman was dehumanized, and her experience and knowledge were disregarded, limiting her ability to take ownership of the situation. She was profoundly misunderstood by both her superior and her colleague.

In another situation, a woman in her late thirties living with steatocystoma multiplex was a victim of discrimination when applying to a specialized master's program, as she was waiting for a diagnosis. She explained that:

I asked for part-time studies at my university due to my illness. The administration told me that part-time studies were not offered in my program [...]. They believed that if I received an official diagnosis during my enrollment, I would no longer have the capacity to complete my study program anyway.

In this context, her ability to exert agency on her professional future was compromised by the administration's decision-making on her behalf, which was tainted by paternalism and rigidity.

Morally problematic situations encountered in social interactions and relationships

Morally problematic situations were encountered in social interactions and relationships, notably in relation to family, conjugal life, and friends. Several situations were experienced, such as marginalization and isolation (Table 7).

Difficulties arising in family and conjugal life. Choosing to have children or not is at the heart of morally problematic situations for respondents. Some respondents were infertile, and others hesitated or decided not to have children due to practical constraints, including genetic transmissibility of the illness, poor or uncertain prognosis, limited capacity to take care of children, or fears of pregnancy-associated complications (see Table 7). Nevertheless, respondents exerted their agency to make decisions that best served their interests. For example, a man living with severe hemophilia explained: "Several years ago, I made the decision not to have children in order to protect them from my hereditary condition, severe hemophilia, and to spare them from the pain and numerous hospitalizations that I have experienced."

Respondents also reported challenges in fulfilling their roles as parents and spouses. Parents living with

Table 7. Morally problematic situations encountered in social interactions and relationships.

Sphere relating to social interactions and relationships	Sub-sphere	Types of morally problematic situations described by respondents
Family	Choosing to have children or not	Hesitation or choice not to have children notably due to potential genetic transmission of the rare disease, poor or uncertain prognosis, or limited capacity to take care of children. Desire to have children but low fertility.
	Difficulties raising children	Inability to satisfactorily care for children, play with them, accompany them to physical or recreational activities, or travel with them.
	Difficulties visiting family members	Inability to travel to see family members that live further away. Inability to attend family reunions or important events (e.g., weddings) due to symptoms, immunodeficiency, or worsening of health following the events.
Conjugal life	Difficulties finding a partner	Difficulties finding a partner due to poor financial situation, inability to travel, fear of dependency, or the other's fear of the rare disease and its complications.
	Relationship challenges	Partners composing with the limitations posed by the respondents' rare diseases, notably regarding physical activities, leisure, life projects, and sexuality.
	Separation or divorce	Partners may initiate a separation or a divorce due to the weight of the rare disease limitations or the fear associated with the progression of the illness.
Friends	Losing friends	Loss of friends due to not being able to participate to activities they used to enjoy together. Lacking the energy to maintain friendships.
Marginalization and isolation	Limited social activities	Inability to go to the restaurant with close ones or share meals with them due to dietary restrictions (e.g., phenylketonuria) or physical limitations (e.g., oropharyngeal muscular dystrophy). Difficulties in participating to various social and community activities.
	Isolation	Inability to meet new people due to mobility issues, limited ability to participate in activities, and immunodeficiency.
	Misunderstanding, disbelief, and prejudice	Misunderstanding of the rare disease and its impacts. Being perceived as lazy, disinterested, weird, or dramatizing a minor illness. Having to convince others that the rare disease is real.

rare diseases may struggle to care for their children, play with them, accompany them to physical or recreational activities, or travel with them. As their spouse, they may have difficulties engaging in their significant other's leisure and physical activities, and may halt important lifelong dreams (see Table 7). For instance, a woman in her late thirties living with narcolepsy admitted: "I would like to be more present for my spouse and my children but [they] must deal with a mother or a spouse that sleeps [often]."

Additionally, a man living with primitive lateral sclerosis tragically explained that: "My partner and son left me because of my illness. They couldn't handle seeing my physical abilities decline. Recently, I received the official request for divorce and sole custody of my son from my partner of the past 26 years." These difficult family situations evoked powerlessness within respondents concerning the burden posed by their health on their close ones.

Marginalization and isolation. Respondents experienced various instances of marginalization. They were perceived by others as lazy, disinterested, weird, or exaggerating their illness. Being significantly misunderstood, they have been excluded from social events or chose to avoid them. For example, a woman living with hemifacial spasms and suspected Ehlers-Danlos syndrome confided that: "I refused to attend social gatherings because speaking, smiling, or laughing sharply intensifies the pain in my face. When I did participate, I limited my involvement to minimal interactions."

Conversely, a woman in her late forties living with Ehlers-Danlos syndrome recounted: "I feel excluded on the basis that I cannot participate in an activity. For example, when my sister-in-law organized a fundraising campaign for a school trip for one of my nieces, she did not invite me under the pretext that I would not be interested in attending since I cannot dance [due to my illness]." Through this rejection paved with misunderstanding, the woman was hurt by others taking a decision on her behalf. In both situations, the marginalization the respondents experienced limited their ability to participate as active members of their family, community, or society.

Discussion

This survey study described the morally problematic situations experienced by adults living with rare diseases in healthcare, personal life, career, and studies, as well as relationships and social interactions. We

discuss our findings in light of pragmatist theory calling for the recognition of the experiential nature and existential impacts of morally problematic situations and the recognition of the moral relevance of experiences which, from the point of view of others, such as ethics experts and healthcare professionals, may not appear to be morally salient.

Healthcare struggles: stigmatization to inadequate care

The survey showed that, in a given healthcare system aiming to provide care to all (Maioni 2018), adults living with rare diseases face significant and repeated stigmatization, disbelief, and sometimes psychological and physical abuse from healthcare workers. Such attitudes may prevent individuals from receiving crucial or life-saving medical interventions.

Despite their existential nature and moral salience, such situations are mostly overlooked in the ethics literature on rare diseases, which is mostly centered on research ethics and resource allocation (e.g., Barrera and Galindo 2010; Borski 2015; Schieppati et al. 2008). Stigmatization in healthcare may be the hallmark of living with a rare disease, as opposed to common diseases which are more widely recognized. These challenges are evidenced in qualitative studies, not specifically in morally problematic situations. For example, adults living with rare diseases in the United States were subjected to professionals' dismissive attitudes and limited knowledge, erroneous accusations of lying, misplaced diagnoses of mental health issues, and disinterest in treating rare and complex conditions (Munro, Cook, and Bogart 2022).

Yet, such qualitative studies do not bring to light the moral importance of those situations with respect to human flourishing and how such diseases profoundly challenge their values and life trajectories. Our study shows that these unsettling experiences of stigmatization are morally salient because they elicit internal tensions of being profoundly misunderstood, not being listened to, and having one's knowledge and experience disregarded by clinicians. They undermine the sense of self-worth, the capacity to act upon their situation, and, more globally, the respect of these adults living with rare diseases as individuals.

The impact of physical and financial dependency on authenticity and self-realization

Physical and financial dependency were inherent to some morally problematic situations, such as having to rely on caregiver support or on a partner for

financial stability. Such dependency situations may elicit powerlessness within some individuals and may limit their ability to act authentically (Racine et al. 2021).

Authenticity is a critical subcomponent of autonomy (Racine et al. 2021). It is achieved when decisions closely align with oneself, including personal integrity, individuality, and character (Ganzini and Lee 1993; Racine et al. 2021). In these situations of dependency, respondents' scope of action is constrained by physical limitations or financial precariousness. In response, their authenticity could be promoted by offering them opportunities to voice their needs and desires, provided that individuals in their support system listen to them and are open to enacting their values. Otherwise, considerable harm can be done to them as people.

Overcoming biographical disruption through self-awareness and renewed purpose in life

Respondents were confronted with morally problematic situations in which they were unable to reach cherished personal milestones, relating to their careers and studies. Many of these situations were reminiscent of *biographical disruptions*, a lens to interpret life trajectories that are abruptly questioned or halted by illness (Williams 2023). The impact of biographical disruptions can be understood through the inseparability of context and meaning expounded by pragmatist ethics (Brinkmann 2011; Bury 1991; Fesmire 2003; Gallagher 2014). Existential meaning, or purpose in life, is inseparable from life context because it is the way through which individuals transact with their environments and make sense of their situations and of their actions (Brinkmann 2011; Kestenbaum 1992).

Individuals facing a biographical disruption may mobilize various social, cultural, medical, and physical resources to overcome this existence-shaking adversity (Williams 2023). Through biographical reconstruction (Carricaburu and Pierret 1995), the meaning of the situation changes because agents find ways of overcoming their difficulties. Previous literature has shown that patients use empowerment strategies to surmount morally problematic situations, reinforcing the possibility for biographical reconstruction in light of these situations (Quintal et al. 2023). Individuals living with rare diseases often learn to work around their limitations and aim for alternative, albeit fulfilling, life objectives that can depart from societal expectations. Such reimaginings may be a key step in overcoming biographical disruptions.

Reflecting on this idea, William James wrote that existential anguish is the price to pay for moments of growth, when in response to perplexities, following

periods of what James called “incubation” and “fermentation,” “the results hatch out, or burst into flower” (Fesmire 2003, James cited p. 36). Nonetheless, living with a rare disease, managing it, and fulfilling one's basic needs may be exhausting and demanding (Bathen et al. 2022). This can pose significant challenges in overcoming biographical disruptions. Here, peer and community support could be key in helping make sense of rare disease experiences and developing strategies to overcome what can sometimes be overwhelming for an individual.

Limitations of the study

The first limitation of the current study is an over-representation of individuals with high socioeconomic status. A second limitation is that men and visible minority groups were underrepresented in the sample with respect to the Québec population. Less than 5% of the study respondents were part of visible minority groups, which constitute 13% of the Québec population. A third limitation of the study is that some rare disease patients may presumably not be familiar with the Québec Coalition of Orphan Diseases or other associations which played a key role in advertising the survey. Consequently, the morally problematic situations listed herein may not represent the diversity and scope of situations encountered by all adults living with rare diseases in the province of Québec. Nonetheless, the composition of the sample featured in this study resembles that of other survey studies with similar representation of conditions (Molster et al. 2016), official diagnoses (Molster et al. 2016), multifaceted impacts (Heuyer, Pavan, and Vicard 2017), and income (Bogart and Irvin 2017). One pending issue posed by the current study's design is the lack of comparison of rare disease patient experiences with the experience of people with common chronic disorders. It can be suspected that, for example, the systematic and substantive discrimination, stigmatization, gaslighting, belittlement, as well as the tolling diagnostic odysseys (often amounting to 5, 10, 15 years) and therapeutic nihilism encountered by people with rare diseases is not as frequently encountered or encountered in the same manner by people with common chronic conditions, but this remains something to be established *via* comparative research.

Conclusion

This survey study highlighted morally problematic situations faced by adults living with rare diseases using

a participatory approach inspired by pragmatist ethics. These situations are morally problematic due to the salient internal tensions and constraints to agency they elicit. From a theoretical and methodological standpoint, this study demonstrates that the operationalization of the concept of the *morally problematic situation* in empirical bioethics research is useful for capturing meaningful aspects of moral experience. This study also advances the added value of participatory methods for conducting ethics research on moral experiences. Future participatory action research could help further the integration of ethics in rare disease care and find constructive strategies to overcome the significant and tolling morally problematic experiences of individuals living with rare diseases.

Notes

1. 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. We stand in solidarity with individuals living with rare diseases alienated from mainstream healthcare.
2. In this article, adults living with rare diseases are occasionally designated as *individuals* for concision purposes. They are referred to as *agents* in theoretical discussions inspired by pragmatist ethics, and those who answered the survey are referred to as *respondents*.
3. A CHSLD is a publicly funded long-term care centre in the province of Québec, Canada.

Disclosure statement

No potential conflict of interest was reported by the author(s).

Funding

This work was supported by the Canadian Institutes of Health Research (Doctoral research scholarship), Fonds de Recherche du Québec - Santé (Career award) and the Université de Montréal (Graduate student awards).

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