

Morally problematic situations experienced by adults living with rare diseases, including rare neurological diseases

Authors

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* Equivalent contributions

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Comments or questions?



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1. Background

Rare diseases

- A **rare disease** has a prevalence inferior to 1/2000 individuals.
- Rare diseases are **poorly understood** due to their rarity. Hence, **diagnostic delays and lack of proper treatment** are frequent.
- Bioethics discourse focuses on resource allocation and research participation [1,2].
- Moral experiences** of **adults** living with rare diseases have not been extensively studied.

Morally problematic situations

- A concept from **pragmatist ethics**
- Morality is experienced through **lived situations** [3]
- These situations hold **existential** importance by destabilizing cherished **values** [4]
- Yet, this concept has **never been operationalized** in empirical bioethics research.

2. Aims

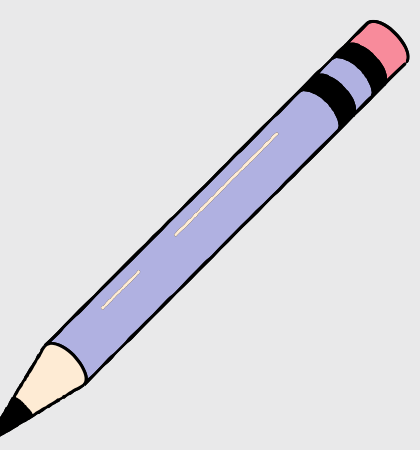
Aim 1

- Identify **morally problematic situations** experienced by **adults** living with rare diseases



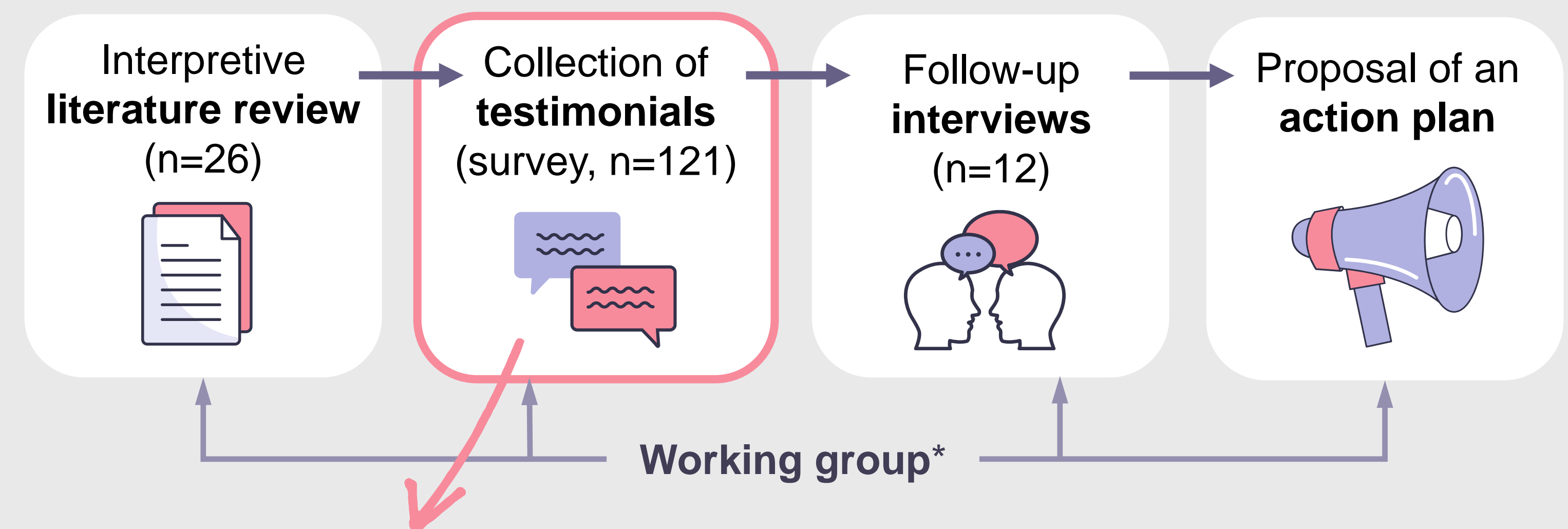
Aim 2

- Develop the concept** of the morally problematic situation, notably by identifying its components.



3. Methods

A **participatory action research** project [5] guided by **pragmatist ethics** [6], and comprised of several steps



* Working group:

Composition:

- 3 patient partners
- 2 clinician researchers
- Representatives from Québec's rare disease association

Contributions:

- Refining protocols and data collection tools
- Interpreting results
- Enriching manuscripts

4. Featured results

Aim 1 – Situations

Examples below were experienced by adults living with **rare neurological disorders**.

Healthcare

Disbelief by clinical staff during hospitalization:

“My illness was believed to be psychiatric in nature, even if I was diagnosed 17 years ago. I was left in my bed for 10 days, deprived of hygiene [...] and despite the spasticity, I had to sit upright in my bed to have breakfast without any help”.

— 58-year-old woman living with stiff person syndrome

Daily life and relationships

Being **abandoned** and **isolated** in a long-term care home:

“I was placed in a long-term care home. I had to say goodbye to my workplace, [...] my memories, and my house to end up among the undead. The care home is not adapted with care and leisure for younger patients. My family has abandoned me because they find it difficult to see my health declining”.

— 50-year-old woman living with rapidly evolving idiopathic ataxia

Career and studies

Uncertainty amid new life projects:

“I just changed jobs. I debated doing so for a while because changes in my routine lead me to experience health crises for weeks until I adjust to a new routine”.

— 37-year-old woman living with narcolepsy with cataplexy

Aim 2 – Conceptual development

Key components of morally problematic situations:

- Internal tensions**
- Jeopardized **values**
- Emotions** and **feelings**

Additional components may reveal themselves through **agency** in the situations:

- Empowerment** strategies
- Personal **growth** and **flourishing** or alternatively, moral **residue**