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# Patient-Centric Approach to Improve the Quality of Life and Healing Efficacy for Patients With Fabry Disease: A Grounded Theory-Based Analysis

**Seo-Yeon Gwak**,[1] **Ha Jeong Lim**,[2] **Iksung Cho**,[1] **Chi Young Shim**,[1] **Kyu Kim**,[1] **Hyun-Jung Lee**,[1] **Jang-Won Son**,[3] and **Geu-Ru Hong**[1]

[1]Division of Cardiology, Severance Cardiovascular Hospital, Yonsei University College of Medicine, Seoul, Korea
[2]Division of Cardiology, CHA Bundang Medical Center, CHA University, Seongnam, Korea
[3]Division of Cardiology, Department of Internal Medicine, Yeungnam University Medical Center, Daegu, Korea

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**Address for Correspondence:**
**Jang-Won Son, MD, PhD**
Division of Cardiology, Department of Internal Medicine, Yeungnam University Medical Center, 170 Hyeonchung-ro, Nam-gu, Daegu 42415, Republic of Korea.
Email: gubjae@yu.ac.kr

**Geu-Ru Hong, MD, PhD**
Division of Cardiology, Severance Cardiovascular Hospital, Yonsei University College of Medicine, 50-1 Yonsei-ro, Seodaemun-gu, Seoul 03722, Republic of Korea.
Email: grhong@yuhs.ac

**ORCID iDs**
Seo-Yeon Gwak
https://orcid.org/0000-0002-5550-4156
Ha Jeong Lim
https://orcid.org/0000-0002-3567-2347
Iksung Cho
https://orcid.org/0000-0001-5927-5410

## ABSTRACT

**Background:** Fabry disease (FD) poses multifaceted challenges for patients in Republic of Korea (South Korea), but an in-depth qualitative understanding of their experiences and support needs is still lacking. This study aimed to qualitatively explore the experiences and coping strategies of Korean patients with FD who reported dissatisfaction with their well-being and faced social, occupational, or personal difficulties, to inform patient-centered care and healthcare policy.
**Methods:** We conducted a qualitative study using a grounded theory approach to explore the experiences of individuals with FD in Korea. Eligible participants were aged 19–70 years, had been diagnosed for more than three years or treated for at least one year, and reported dissatisfaction with their well-being alongside social or personal disadvantages. Each participant completed three in-depth interviews. Data were collected iteratively and analyzed through open and axial coding until theoretical saturation was reached.
**Results:** Theoretical saturation was achieved after conducting three in-depth interviews with each of the 10 participants (5 males, 5 females; age range: 34–66 years). A total of 273 initial concepts were generated and refined into 21 categories across six conceptual domains: causal conditions, contextual conditions, central phenomenon, intervening conditions, actions/interactions, and consequences. Patients reported persistent physical and emotional burdens, financial strain, limited systemic support, and stigma. However, they developed adaptive coping strategies, including lifestyle modifications, medical engagement, and meaning-making. The coping process followed five progressive stages: diagnostic odyssey, treatment entry, life reconstruction, symptom management, and hope, highlighting dynamic psychological adaptation. Three distinct coping styles emerged: religious will, optimistic acceptance, and altruistic solidarity.
**Conclusion:** Fabry patients experience physical pain, emotional and socioeconomic burdens, and limited systemic and social support, yet they develop adaptive coping strategies and resilience. These findings highlight the importance of developing patient-centered, context-sensitive care approaches in Korea.

**Keywords:** Fabry Disease; Grounded Theory; Patient-Centric Approach; Quality of Life

# INTRODUCTION

Fabry disease (FD) imposes a wide range of physical, systemic, and psychosocial challenges on patients. These include chronic pain, treatment fatigue, delayed diagnosis, limited insurance coverage, low awareness of the disease's genetic nature, social stigma, poor communication with healthcare providers, and inadequate support systems. Previous studies have also identified financial strain, stigma, diminished self-confidence, and policy-related barriers as major factors negatively affecting patients' quality of life (QoL).[1]

The EuroQol 5-dimensional (EQ-5D) and 5-level EQ-5D have been used to assess QoL in FD, covering mobility, self-care, daily tasks, pain, and mental health. Studies showed that FD patients have significantly impaired health-related quality of life (HRQOL) across these areas.[2-4] However, these may not capture the full depth of patient experiences. To address this, the French National Authority for Health (HAS) proposed a patient-centered framework emphasizing shared decision-making and developed a self-reported questionnaire reflecting lived experiences.[5]

In-depth qualitative research on FD remains scarce in Republic of Korea (South Korea), where psychosocial challenges receive limited attention. To address this gap, we aimed to explore the experiences qualitatively, coping strategies, and unmet needs of Korean FD patients with reduced well-being, and to propose a patient-centered care framework that informs policy and improves QoL.

# METHODS

## Study design

This study employed a qualitative approach to explore the in-depth experiences and perspectives of patients with FD.[6] The interview guide was developed based on the patient-centric framework proposed by the French HAS, which emphasizes lived experiences and contextual challenges affecting QoL.[5] Grounded theory methodology by Corbin and Strauss guided the research design, as it is well-suited for exploring complex psychosocial phenomena and generating theoretical insights from participants' narratives.[7] The study followed core principles of grounded theory, including theoretical sampling, iterative data collection and analysis, and the pursuit of theoretical saturation rather than statistical generalizability.[8]

## Participants

Eligible participants met the following inclusion criteria: individuals aged 19 to 70 years who had been diagnosed with FD for more than three years or had initiated treatment at least one year prior; those who had experienced delays in diagnosis or had previously refused treatment; those capable of verbally articulating their experiences and providing informed consent (excluding individuals under adult guardianship); and those who reported dissatisfaction with their HRQOL and faced disadvantages in areas such as education, employment, family, social, or romantic relationships. Written informed consent was obtained after participants received a full explanation of the study's purpose and methodology.

## Data collection

Data were collected through three in-depth interviews per participant, conducted in Korea between January and February 2023. Following Creswell's guidelines for qualitative

Chi Young Shim
https://orcid.org/0000-0002-6136-0136
Kyu Kim
https://orcid.org/0000-0002-3632-0183
Hyun-Jung Lee
https://orcid.org/0000-0002-7498-0705
Jang-Won Son
https://orcid.org/0000-0002-8109-5018
Geu-Ru Hong
https://orcid.org/0000-0003-4981-3304

**Disclosure**

The authors have no potential conflicts of interest to disclose.

**Author Contributions**

Conceptualization: Gwak SY, Son JW, Hong GR. Data curation: Gwak SY, Lim HJ, Kim K, Lee HJ, Shim CY, Son JW. Investigation: Gwak SY, Lee HJ. Methodology: Gwak SY, Lim HJ, Kim K, Cho I. Supervision: Hong GR. Writing - original draft: Gwak SY. Writing - review & editing: Son JW, Hong GR.

research, the process adhered to a grounded theory approach, with no predefined sample size.[6] Instead, interviews were conducted iteratively, beginning with one participant, and subsequent interviews were guided by concepts that emerged from earlier sessions. This cyclical process continued until theoretical saturation was achieved, with no new concepts or categories emerging in the final interviews.[8] Each interview lasted approximately 50–60 minutes. All interviews were audio-recorded with consent and fully transcribed for analysis.

### Data analysis and coding

Data were analyzed using the grounded theory methodology proposed by Corbin and Strauss,[7] incorporating both open and axial coding. In the open coding phase, a line-by-line analysis of the transcripts was performed to derive initial concepts. These concepts were grouped based on thematic similarities into subcategories and broader categories. During the axial coding phase, the categories were systematically reassembled to construct a paradigm model, which mapped the structure of the patient experience across six major domains: causal conditions, contextual conditions, central phenomenon, actions/interactions, intervening conditions, and consequences.

### Ethics statement

The study protocol was reviewed and approved by the Yonsei University Institutional Review Board (IRB) in accordance with the Bioethics and Safety Act (IRB No. 4-2022-1347). All participants were fully informed about the study's purpose and methodology, and written informed consent was obtained before participating. **Supplementary Data 1** provides a detailed description of the ethical considerations, including participant consent procedures, data protection measures, and adherence to research ethics guidelines.

## RESULTS

### Sociodemographic and clinical profile of participants

Interviews were conducted from January to February 2023. Theoretical saturation was reached after 10 interviews, with no new concepts emerging. Each participant completed three in-depth interviews, allowing for comprehensive exploration and validation of emerging themes over time. Ten individuals (5 males, 5 females) aged 34–66 years were included; nine had classic FD. Age at diagnosis ranged from 21 to 53 years. Most had received treatment for over a year (mean 7.3 ± 4.5 years). Organ involvement and occupations varied. Family history was diverse, from no known cases to multiple affected relatives. Detailed baseline characteristics are provided in **Table 1**.

### Structure of disease-related QoL, coping strategies, and healing efficacy in patients with FD

The finalized questionnaire included 45 items (**Supplementary Data 2**). New concepts emerged up to the 8th interview, while the 9th and 10th interviews reinforced existing categories. By the third interview with the tenth participant, no new themes appeared, confirming saturation. Line-by-line analysis of interview transcripts produced 273 initial concepts, which were grouped into 77 subcategories based on shared experiences (**Supplementary Fig. 1**). These were further refined into 21 categories. As shown in **Fig. 1**, the categories are grouped into six domains: causal conditions, contextual conditions, central phenomenon, intervening conditions, actions and interactions, and consequences, reflecting the paradigm model used in grounded theory.

**Table 1.** Sociodemographic characteristics and FD-related characteristics of the participants

| Patient No. | Sex | Age, yr | Classic type | Organ involvement | Occupation | Diagnosis (start of treatment) | Status of family members diagnosed with FD | Comorbidities |
|---|---|---|---|---|---|---|---|---|
| 1 | M | 35 | Yes | Heart, kidney, nervous system, skin | Office worker | 2011 (22 yr old) | The older brother was healthy with no FD, while the younger brother was diagnosed with FD. | None |
| 2 | F | 59 | No | Heart, kidney, skin, gastrointestinal system | Housewife | 2010 (53 yr old) | No manifestation of FD among children. | Visual impairment |
| 3 | M | 35 | Yes | Heart, nervous system, ears, skin, gastrointestinal system | Self-employed | 2008 (21 yr old) | Elder sister did not present with any symptoms of FD. | None |
| 4 | F | 42 | Yes | Kidney, nervous system, skin, eyes, ears, gastrointestinal system | Housewife | 2015 (35 yr old) | Two sons (no symptoms) | None |
| 5 | F | 50 | Yes | Heart, nervous system, eyes, ears, gastrointestinal system | Office worker | 2015 (45 yr old) | One of the two daughters was diagnosed with FD; two siblings were also patients with FD. | Cancer |
| 6 | F | 50 | Yes | Heart, kidney, nervous system, ears, skin, gastrointestinal system | Office worker | 2019 (47 yr old) | Son presented with the onset of FD, while daughter did not present with any symptoms. | None |
| 7 | M | 49 | Yes | Heart, kidney, nervous system, skin, eyes, gastrointestinal system | University lecturer | 2021 (48 yr old) | No children | None |
| 8 | M | 51 | Yes | Heart, kidney, nervous system, skin, eyes, ears, gastrointestinal system | Self-employed | 2020 (50 yr old) | Son did not manifest any symptoms; older brother was diagnosed with FD; the son of the older sister was diagnosed with FD. | Sequelae of cerebral infarction |
| 9 | M | 55 | Yes | Heart, kidney, nervous system, eyes, ears, gastrointestinal system | Self-employed | 2017 (50 yr old) | Two sons and one daughter were diagnosed with FD. | None |
| 10 | F | 50 | Yes | Heart, kidney, nervous system, skin, eyes, ears, gastrointestinal system | Self-employed | 2021 (48 yr old) | All three children were diagnosed with FD and were undergoing treatment. | None |

FD = Fabry disease, M = male, F = female.

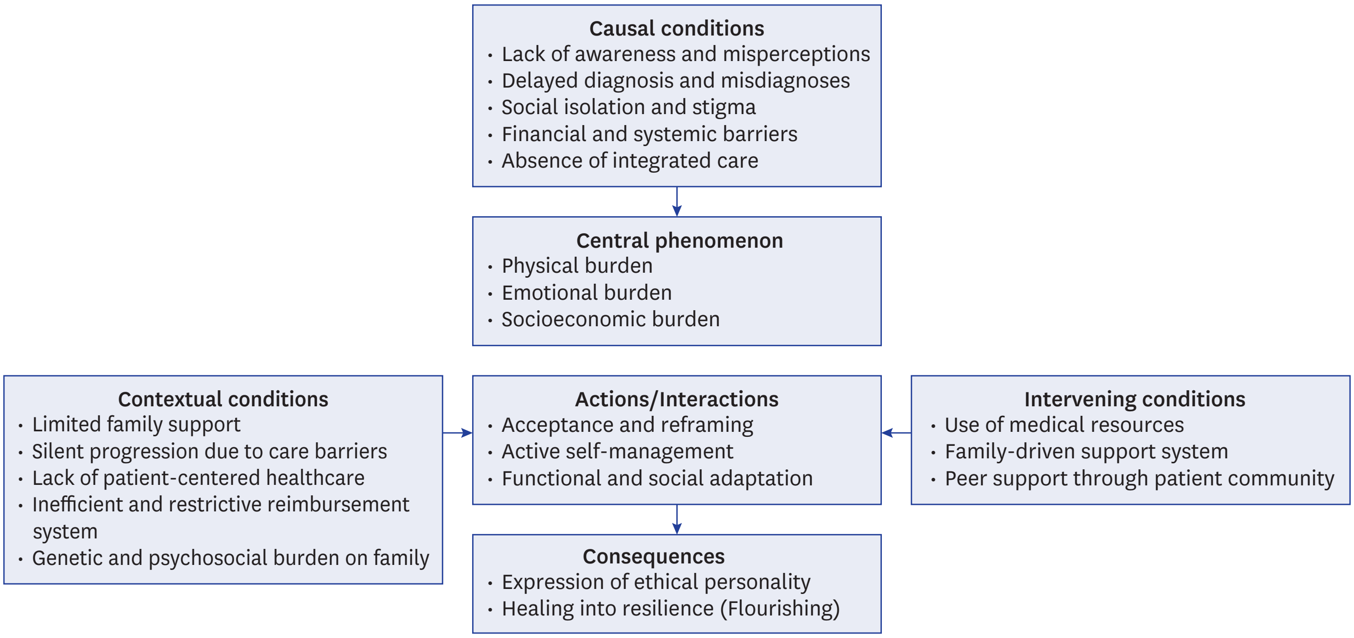

**Fig. 1.** Structural framework of patient experiences with FD based on grounded theory analysis. Conceptual framework derived from grounded theory analysis, outlining the lived experiences of patients with FD. The diagram presents six core components: causal conditions (e.g., lack of awareness, delayed diagnosis, stigma), contextual conditions (e.g., limited family support, systemic and reimbursement barriers), central phenomenon (physical, emotional, and socioeconomic burden), intervening conditions (e.g., medical, family, and peer support), actions/interactions (e.g., acceptance, self-management, and adaptation), and consequences (e.g., ethical awareness and resilience). Each component reflects the dynamic process through which patients understand, respond to, and live with FD.
FD = Fabry disease.

## Central phenomenon

The central phenomenon refers to the core experience of patients with FD, encompassing physical, emotional, and socioeconomic burdens. Physically, both men and women reported fatigue, immune weakness, and difficulties managing fever; men frequently recalled severe childhood pain. Emotional burden involved daily anxiety from pain, fear of flare-ups despite treatment, and uncertainty about policy or financial support. These feelings were intensified by the rarity and incurability of FD. Socioeconomic burden affected those unable to work or forced to change jobs due to disease-related limitations.

## Causal conditions

We identified several causal conditions contributing to the central phenomenon. Limited awareness and misconceptions about FD led to a lack of understanding and support from others, creating communication barriers. Delayed and incorrect diagnoses prolonged patients' suffering through repeated hospital visits, misdiagnoses, and pain that restricted social participation. These challenges often resulted in social isolation, stigma, and strained personal and professional relationships. Even after diagnosis, financial and systemic burdens remained, as strict criteria limited access to treatment support. The absence of integrated care and restrictive insurance policies further deepened patients' sense of helplessness.

## Contextual conditions

Contextual conditions affecting the identified phenomena included family support, access to care, patient-centric approaches, healthcare policies, and the genetic nature of FD. Few patients benefited from family support, while most faced FD-related challenges in isolation. Inadequate care and diagnostic and treatment barriers further worsened FD symptoms. Some patients experienced non-patient-centric approaches even after being diagnosed and treated for FD. The co-payment reduction system required extensive documentation and often lacked empathy or compassion from administrative staff. The reimbursement process was burdensome, involving substantial upfront payments (KRW 400 million monthly for a year before receiving support) and disclosure of family details, negatively impacting patients' QoL. Additionally, due to a systemic loophole, patients became ineligible for health insurance benefits if their income increased, causing them to avoid work or lose motivation to progress professionally. Some patients chose not to have children out of fear of passing FD to future generations.

## Intervening conditions

Intervening conditions positively shaped patients' coping strategies. After diagnosis, patients actively used medical resources and adopted a positive mindset supported by accurate information and empathetic communication with healthcare professionals. Hospital visits were seen not only as treatment but also as opportunities for emotional healing. Family-driven support systems, especially spousal and parental support, helped patients overcome difficulties. Communication and understanding from friends also enhanced their QoL and motivation. Following diagnosis, participants managed FD at the family level. Peer support from FabryKorea fostered a sense of community through shared experiences and information, helping patients cope through altruism and companionship.

## Actions/interactions

Participants responded to the challenges of FD through acceptance and cognitive reframing. Many embraced the diagnosis early and approached it with optimism, gratitude, and trust in treatment, which helped reduce uncertainty and make sense of their symptoms. Active self-management included establishing routines, adopting healthy behaviors, avoiding harmful habits, and engaging in regular exercise to promote well-being and hope. Functional and social adaptation was evident as participants integrated treatment into their daily lives and continued to fulfill family and work roles, striving to maintain a sense of normalcy despite ongoing symptoms.

### Consequences

The actions and interactions of patients led to various outcomes. Although some continued to face long-term disease burden, emotional distress, and treatment fatigue, these challenges often coexisted with psychological growth and renewed purpose. Many adapted and found meaning in life with FD. Some developed a stronger sense of ethical responsibility, appreciating public support and choosing to use welfare resources responsibly. In some cases, participants declined aid out of respect for taxpayers. Others gained empathy and committed to volunteer work. For some, religious faith provided moral grounding. Despite the rarity and severity of FD, most patients showed resilience and a sense of flourishing beyond survival.

### Coping trajectory and stages of disease acceptance in FD

Based on the analysis of patient narratives, we identified a five-stage trajectory that characterizes the process of coping with and accepting FD. These stages include diagnostic odyssey, entry into FD treatment environment, life reconstruction, progress in symptom management, and grasping hope, and reflect patients' evolving psychological and behavioral responses to their diagnosis and treatment. This coping trajectory is illustrated in **Fig. 2** and reflects the evolving interplay between medical, emotional, and social dimensions of the patient experience.

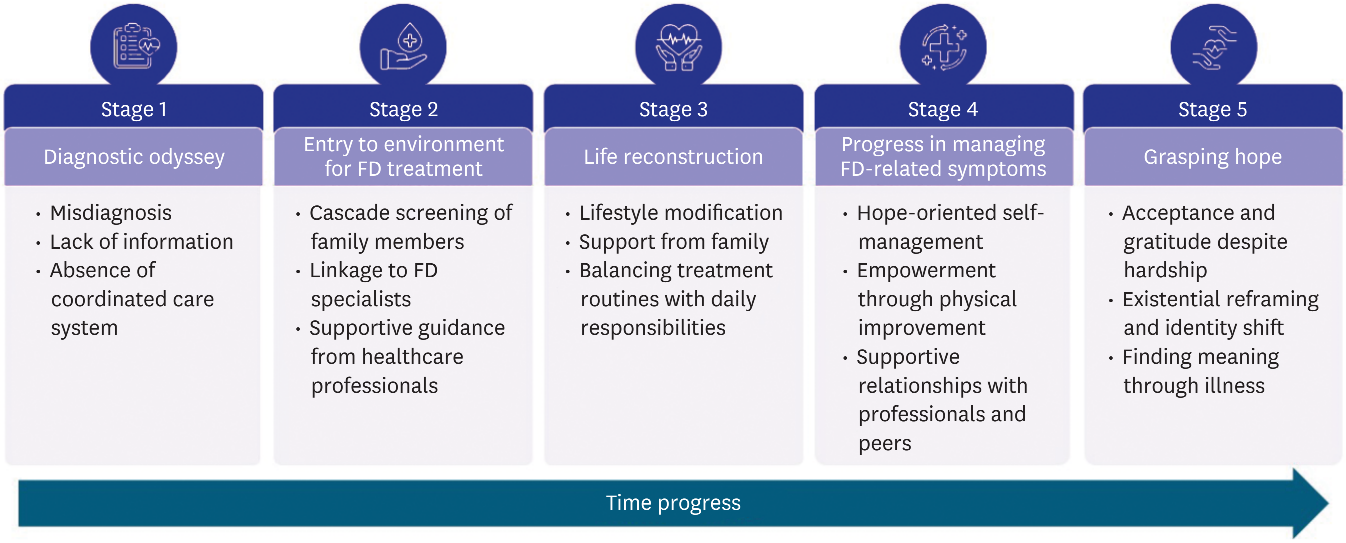

**Fig. 2.** Coping process stages among patients with FD. Sequential stages of coping with FD, derived from grounded theory analysis. The process comprises five progressive stages: Stage 1 (diagnostic odyssey), Stage 2 (entry to the environment for treatment of FD), Stage 3 (life reconstruction), Stage 4 (progress in managing FD-related symptoms), and Stage 5 (grasping hope). Each stage is shaped by specific contextual factors, such as misdiagnosis, family support, trust in healthcare professionals, and existential awakening. The horizontal arrow indicates the temporal progression of patients' psychological and practical adaptation to FD.
FD = Fabry disease.

### Diagnostic odyssey

At this stage, the participants had endured FD for a prolonged period. However, due to circumstances such as physician misdiagnosis and lack of awareness about FD, they felt isolated and confused about their condition. The absence of a collaborative medical care system in South Korea subjected patients to a "diagnostic odyssey." This caused missed opportunities for timely treatment, resulting in undetected disease progression.

### Entry into the FD treatment environment

At this stage, not only were the participants themselves diagnosed with FD, but the diagnosis often extended to other family members through cascade screening. Although their diagnostic odyssey had concluded and they had received accurate diagnoses and treatment, they often lacked information about FD or the eligibility criteria for the co-payment reduction system. As a result, many assumed that treatment would involve high medical costs. Support from healthcare professionals was therefore essential, as they provided critical information and resources regarding various aspects of treatment.

### Life reconstruction

At this stage, the participants actively sought to adjust their career paths and modify their lifestyles. They abstained from alcohol and smoking while striving to balance their livelihoods with treatment schedules. Despite the biweekly hospital visits necessitated by FD treatment, the participants adapted to this routine, with some perceiving these visits as opportunities for emotional and physical healing.

### Progress in managing FD-related symptoms

At this stage, the participants prioritized hope over despair and actively sought forms of exercise or work that aligned with their individual circumstances. All participants reported engaging in regular exercise, often to the point of sweating, which alleviated lethargy. Notable improvements in their laboratory values brought a sense of happiness and fostered a feeling of control in managing their FD-related symptoms. This process was also supported by trust in healthcare professionals and solidarity with others living with FD.

### Grasping hope

At this stage, the participants no longer perceived their lives as unfortunate or unhappy. Despite enduring hardships, they actively sought experiences that inspired gratitude, embracing the understanding that while FD cannot be cured, symptom aggravation can be prevented, enabling them to lead a normal life. This stage marked an existential awakening, as the participants ceased identifying solely as patients and began viewing themselves as individuals undergoing healing and recovery. They also recognized that the gains attributed to FD, such as humility and familial love, outweighed their losses.

### Coping styles among patients with FD

To better understand individual variations in how patients manage FD, we categorized their coping styles into three distinct types: religious will, optimistic acceptance, and altruistic solidarity. These typologies reflect the dominant attitudes and behaviors that shaped each participant's approach to living with FD. While some drew strength from faith and spiritual acceptance, others relied on family support and a positive outlook, or found meaning through helping others in the patient community. This classification, summarized in **Table 2**, highlights the diverse yet resilient ways patients adapt to the challenges of FD.

**Table 2.** Coping styles and corresponding participant characteristics in patients with FD

| Types | Participants | Characteristics |
|---|---|---|
| Religious will | 1, 3, 5 | Accepted FD through prayer and religious faith<br>Content with the current level of support<br>Grateful for FD instead of considering it an unfortunate event |
| Optimistic acceptance | 2, 7, 8, 10 | Strong support from spouse and other family members<br>Appreciate small achievements in daily life<br>Self-compassion and encouragement |
| Altruistic solidarity | 4, 6, 9 | Attended/conducted activities in patient organizations, like FabryKorea<br>Provided support for the diagnosis and medical care of patients with FD<br>In pursuit of altruism and meaning/purpose in life |

FD = Fabry disease.

### Religious will

Participants 1, 3, and 5 were categorized under the religious will group. All three participants identified as religious and relied on prayer and faith as central coping mechanisms in their battle against FD, both during their youth and continuing into the present. Additionally, they expressed satisfaction with the financial support and social welfare services available to them.

### Optimistic acceptance

Participants 2, 7, 8, and 10 were categorized under the optimistic acceptance group. They reported receiving strong support from their spouses and family members. For instance, Participant 2's husband actively assisted with homemaking, accompanied her to hospital visits, and provided care at home. During hospital visits, Participant 2 and her husband consistently displayed a cheerful demeanor. They prioritized small, everyday achievements over larger milestones, valuing these moments of happiness. Additionally, these participants exhibited a strong inclination toward self-compassion and self-encouragement.

### Altruistic solidarity

Participants 4, 6, and 9 were categorized under the altruistic solidarity group. They actively engaged in activities organized by patient organizations, sharing hope and valuable information with other patients with FD. These participants demonstrated selflessness and a commitment to leading meaningful lives. Participant 9, despite believing he had less than five years to live, was resolute in his pursuit of purpose and fulfillment. He chose to contribute to society until his final moments, finding that his dedication to altruism became the greatest source of support in his life and journey as a patient.

## DISCUSSION

This qualitative study explored the lived experiences of FD patients in Korea who reported psychosocial challenges and dissatisfaction with their QoL. Grounded theory analysis of in-depth interviews revealed persistent physical and emotional burdens, systemic barriers, and diverse coping strategies. Despite diagnostic delays, financial strain, and limited support, many participants adapted through medical engagement, lifestyle adjustments, and emotional resilience built on family, faith, and community support. These findings emphasize the need for context-sensitive, patient-centered strategies in FD care.

The sample size was considered sufficient to achieve data saturation and conceptual depth, given the qualitative design, the rarity of FD (1 in 40,000–170,000 worldwide; approximately 200 cases in Korea), and prior evidence that thematic saturation is typically reached with

9–17 interviews in similar samples.[9] Although all participants were from one tertiary center, the grounded theory approach minimizes selection bias by focusing on theoretical relevance. Severance Hospital has received referrals for FD diagnosis and management from 15 hospitals nationwide, as well as from patients who independently sought care. Patient distribution was Seoul (20.0%), Gyeonggi (29.4%), Incheon (9.4%), Chungcheong (15.3%), Gangwon (3.5%), Yeongnam (11.8%), and Honam (10.6%). Given the rarity of FD and centralized care at tertiary centers in Korea, this single-center sample may still represent a diverse range of patient experiences nationwide.

Some patients adopted strong self-management habits, supported by better disease understanding and provider encouragement. Positive feedback from medical staff fostered hope and treatment adherence. Acceptance of their genetic condition eased family tensions and enabled future planning, highlighting the role of psychological support in rare disease care. In this context, the patient organization FabryKorea played a pivotal role in emotional support and education. Activities such as lectures for family members encouraged diagnostic testing and promoted care engagement, highlighting the vital contribution of patient-led initiatives.

Despite recent progress, our study revealed persistent barriers in diagnosing and managing FD. Patients reported financial burdens, limited eligibility for support, and abrupt loss of coverage, underscoring the need for increased funding and staffing for specialized clinics and research (**Supplementary Data 3**). One participant noted, “Paperwork for continued treatment is complicated and stressful every year,” illustrating the need to simplify requalification procedures. Access to enzyme replacement therapy remains restricted under Korea’s stringent reimbursement criteria, with additional barriers for women, highlighting the need for policy reform to ensure equitable care. Family-based cascade testing, though standard globally, was declined by relatives in 9 of 10 cases, mainly due to stigma, cost, or fatalism. As one explained, “They worry about being labeled and about the financial burden,” this “hidden” population complicates early detection. While early diagnosis may raise concerns such as labeling or overmedicalization, participants acknowledged its benefits, aligning with prior studies.[10,11] Many also supported newborn screening, recognizing its role in early intervention and preventing progression.[12,13] Improving outcomes in Korea requires systems for early diagnosis, broader multidisciplinary access, revised therapeutic eligibility, and gender equity in reimbursement.

Though participants didn’t mention national policies directly, many preferred care at the FD Clinic at Severance Hospital due to trust, expertise, and consistent communication. These views highlight the value of centralized and specialized care. The Rare Disease Management Act supports this by recommending designated centers through the Korea Disease Control and Prevention Agency.[14]

This study has several limitations. First, the small sample size, despite including varied genders, ages, and organ involvement, limited broader demographic diversity. Second, as the study focused on patients with psychosocial challenges, it may not reflect all FD experiences, though it offers valuable insights into those most affected. Third, findings from a single-country, qualitative study may not be generalized internationally. Further research involving a wider range of stakeholders and longitudinal designs is needed to validate and expand these insights. Fourth, since all participants were from the FD Clinic at Severance Hospital, their preferences may reflect familiarity rather than objective care evaluation. Broader, multi-center, and longitudinal studies are needed to validate and expand these findings.

In conclusion, Fabry patients experience physical pain, emotional and socioeconomic burdens, and limited systemic and social support, yet they develop adaptive coping strategies and resilience. These findings highlight the importance of developing patient-centered, context-sensitive care approaches in Korea.

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## SUPPLEMENTARY MATERIALS

### Supplementary Data 1

Ethical consideration

### Supplementary Data 2

Interview questionnaire

### Supplementary Data 3

Verbatim Patient Quotes Supporting Key Findings

### Supplementary Fig. 1

Thematic Saturation Curve

## REFERENCES

1. Gold KF, Pastores GM, Botteman MF, Yeh JM, Sweeney S, Aliski W, et al. Quality of life of patients with Fabry disease. *Qual Life Res* 2002;11(4):317-27. PUBMED | CROSSREF
2. Jovanovic A, Miller-Hodges E, Castriota F, Takyar S, Howitt H, Ayodele O. A systematic literature review on the health-related quality of life and economic burden of Fabry disease. *Orphanet J Rare Dis* 2024;19(1):181. PUBMED | CROSSREF
3. Andonian C, Beckmann J, Mayer O, Ewert P, Freiberger A, Huber M, et al. Quality of life in patients with Fabry's disease: a cross-sectional study of 86 adults. *Cardiovasc Diagn Ther* 2022;12(4):426-35. PUBMED | CROSSREF
4. Polistena B, Rigante D, Sicignano LL, Verrecchia E, Manna R, d'Angela D, et al. Survey about the quality of life of Italian patients with Fabry disease. *Diseases* 2021;9(4):72. PUBMED | CROSSREF
5. Noël E, Dussol B, Lacombe D, Bedreddine N, Fouilhoux A, Ronco P, et al. Treatment needs and expectations for Fabry disease in France: development of a new Patient Needs Questionnaire. *Orphanet J Rare Dis* 2019;14(1):284. PUBMED | CROSSREF
6. Creswell JW, Poth CN. *Qualitative Inquiry and Research Design*. Thousand Oaks, CA, USA: Sage Publications; 2007.
7. Corbin J, Strauss A. *Basics of Qualitative Research: Techniques and Procedures for Developing Grounded Theory*. 4th ed. Thousand Oaks, CA, USA: Sage Publications; 2014.
8. Kuzel AJ. Sampling in qualitative inquiry. In: Crabtree BF, Miller WL, editors. *Doing Qualitative Research*. Thousand Oaks, CA, USA: Sage Publications; 1992, 31-44.
9. Hennink M, Kaiser BN. Sample sizes for saturation in qualitative research: a systematic review of empirical tests. *Soc Sci Med* 2022;292:114523. PUBMED | CROSSREF

10. Bouwman MG, de Ru MH, Linthorst GE, Hollak CE, Wijburg FA, van Zwieten MC. Fabry patients' experiences with the timing of diagnosis relevant for the discussion on newborn screening. *Mol Genet Metab* 2013;109(2):201-7. **PUBMED** | **CROSSREF**

11. Lisi EC, McCandless SE. Newborn screening for lysosomal storage disorders: views of genetic healthcare providers. *J Genet Couns* 2016;25(2):373-84. **PUBMED** | **CROSSREF**

12. Wasserstein MP, Caggana M, Bailey SM, Desnick RJ, Edelmann L, Estrella L, et al. The New York pilot newborn screening program for lysosomal storage diseases: report of the first 65,000 infants. *Genet Med* 2019;21(3):631-40. **PUBMED** | **CROSSREF**

13. von der Lippe C, Frich JC, Harris A, Solbrække KN. Experiences of being heterozygous for Fabry disease: a qualitative study. *J Genet Couns* 2016;25(5):1085-92. **PUBMED** | **CROSSREF**

14. Ministry of Health and Welfare (KR). Article 14 (Designation of Rare Disease Specialized Institutions, etc.). Rare Disease Management Act. https://www.law.go.kr/%EB%B2%95%EB%A0%B9/%ED%9D%AC%EA%B7%80%EC%A7%88%ED%99%98%EA%B4%80%EB%A6%AC%EB%B2%95/%EC%A0%9C14%EC%A1%B0. Updated 2023. Accessed July 22, 2024.