Brief Report

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Emotional journey of patients with specified intractable diseases in Japan

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SUMMARY This study aimed to depict the emotional journey of Japanese patients with specific intractable diseases facing challenges associated with a delayed diagnosis. Specifically, our focus was on elucidating the emotional journey of patients and identifying the unmet needs caused by a delayed diagnosis. We conducted a web-based survey targeting 179 patients with 11 specified intractable diseases. They reported their emotional states during each journey phase using a 10-point scale. The results revealed that the period from noticing bodily changes to clinic visits was characterized by the most negative emotional states. Furthermore, the patients experienced a gradual shift towards positive emotional states as they decided to complete a consultation at a specialized hospital. They reached their most positive emotional states when they received a definitive diagnosis, subsequent treatment, and care. The thematic classification of emotional changes at the time of definitive diagnosis showed that "relief" was the most prevalent emotion (41.9%), followed by "no change" (19.9%), "anxiety" (14.0%), "shock" (13.4%), and "resignation" (6.5%). Additionally, when classifying the thematic changes in emotions during the period of bodily changes and clinic visits, "frustration" was the most common (51.3%), followed by "fear and anxiety" (43.6%). Patients tended to be most psychologically distressed during the period leading up to the definitive diagnosis. These results reveal that patients with intractable diseases are seeking a fast and accurate diagnosis, and that achieving these is a key unmet need for the patients.

Keywords patient journey, emotional journey, definitive diagnosis, diagnostic delay

1. Introduction

Although rare diseases individually affect a small number of patients, they collectively comprise over 7,000 conditions worldwide, affecting an estimated 350 million individuals. Patients with rare diseases face the challenges of difficult diagnoses and limited access to specialized medical expertise; together, these issues result in misdiagnoses or delayed diagnoses for many patients (*1,2*). Consequently, patients with rare diseases often face significant difficulties in their daily lives, highlighting the need to address this public health challenge (*3*).

In Japan, rare diseases are generally defined as medical conditions involving less than 50,000 patients. Rare diseases that do not meet this Ministry of Health, Labour and Welfare criterion and that have established objective diagnostic criteria are designated as "specified intractable diseases". The government recognizes these rare diseases and includes them in its medical expense subsidy programs (*4*). Despite these efforts to establish a medical support network for rare diseases and reduce the amount of time for definitive diagnosis in Japan, many patients still wait a long time before receiving a confirmed diagnosis (*5*). This matters because early diagnosis is crucial for patients with rare diseases. Even in cases in which effective treatments are lacking, early diagnosis enables the implementation of strategies to manage or slow disease progression, contributing to potential improvements in a patient's quality of life (QOL) (*6,7*).

Therefore, this study aimed to capture the emotional states of patients throughout the entire medical process – from the onset of symptoms through to diagnosis, treatment, and care. To capture the experiences and emotional changes of patients at each stage, we employed the widely used method of depicting the "patient journey". Several studies targeting patients with rare diseases have used this approach (*8,9*). However, most of these studies illustrated the experiences of a few representative patients for each disease based on non-structured interviews. Although this approach is effective for exploring diverse patient journeys and gaining deep

insights into patient experiences, it can only survey a limited number of patients. Furthermore, interviewing individual patients may also reveal strong personal emotions related to each patient's specific situation, which can make it challenging to capture the overall emotional landscape of patients with a particular disease. Few studies on rare diseases have targeted a large number of patients to capture their emotional changes and identify their unmet needs. Therefore, this study aimed not to focus on a very limited number of patients through interviews but rather to comprehensively capture emotional changes from a multitude of patients using a web-based survey method, with the goal of identifying the unmet needs of patients. We selected rare diseases based on the results of our previous research, which identified rare diseases associated with a high likelihood that patients would delay seeking medical care despite feeling abnormal symptoms and wait a long time before receiving a definitive diagnosis (*10*).

2. Materials and Methods

2.1. Study population

As noted above, our previous research in Japan targeting patients with specific intractable diseases revealed that they tended to wait a long time after the onset of abnormal symptoms to seek medical care; further, we also found that these patients tended to experience long delays (*e.g.*, over a year) in receiving a definitive diagnoses (*10*). Building on these findings, this study focused on diseases in which these observed outcomes occurred frequently.

We conducted a web-based questionnaire in November 2023 using patient panels owned by Rakuten Insight, Inc. Eleven designated intractable diseases (Crohn's disease, Sjögren's syndrome, polycystic kidney disease, IgA nephropathy, systemic lupus erythematosus, Parkinson's disease, idiopathic dilated cardiomyopathy, multiple sclerosis/neuromyelitis optica, spinocerebellar degeneration (excluding multiple system atrophy), idiopathic interstitial pneumonia, and eosinophilic sinusitis) were targeted, and participants were recruited from patient panels maintained by the company. The survey methodology involved an Internet-based questionnaire. The initial sample comprised 212 respondents. After excluding cases deemed analytically unfeasible, the final sample for the analysis included 179 respondents (this was a valid final sample size).

2.2. Questionnaires

In this study, a web-based questionnaire approach was utilized instead of interviews to survey participants. This approach allows for broad participation by reaching out to a wide range of participants. Additionally, the use of a questionnaire ensures that uniformity is maintained in the wording of the questions and the scale, providing the advantage of achieving consistency in patient responses. The participants were informed about the purpose of the research, and their participation implied consent. The questionnaire asked them to state their demographic information, such as the name of the disease with which they had been diagnosed and their age, sex, marital status, and educational background. The participants selected their responses from predefined options.

The survey focused on emotional states at various stages of the medical journey, including the onset of awareness of bodily changes, emotions felt upon learning about bodily changes from health check-up results, emotions experienced from the onset of feeling bodily changes to visiting a clinic, emotional states experienced during the transition from a clinic to a specialized hospital, emotions felt during the initiation of examinations at a specialized hospital, emotional states at the time of definitive diagnosis at a specialized hospital, and emotions experienced at the commencement of treatment and care. Respondents were asked to rate these emotional states on a scale of 1–10, with 5 representing a neutral or usual emotional state. Additionally, the participants were encouraged to provide free-text descriptions of their emotions at each stage and elaborate on any changes in their emotions upon receiving a confirmed diagnosis (Supplemental Table S1, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=181*).

2.3. Analysis

For the obtained 10-point emotional states, the average value was calculated and utilized as the "emotional status". Given the subjective nature of individual responses at each stage, we chose not to account for variability in the numerical values when calculating the average.

Qualitative data collected from the web survey were subjected to thematic analysis (*11*) using MAXQDA 2022, software by VERBI GmbH. This approach involves a thorough examination of the dataset to deconstruct qualitative data, reveal patterns of underlying meaning, and thereby identify comprehensive themes. Codes were assigned to themes, which were further clustered. Themes related to emotional changes upon receiving a confirmed diagnosis were categorized into "relief", "anxiety", "shock", "no change", and "other factors". Themes associated with the stage where participants reported the most negative emotions before visiting a specialized hospital were classified into "frustration", "fear and anxiety", "aversion to visiting the hospital", and "other factors". The quoted passages in the emotional journey were included to facilitate the understanding of the phenomenon.

2.4. Ethical approval

This study, conducted using a fully anonymized questionnaire survey, underwent an ethical review and was approved by the Research Ethics Review Committee of the School of Health Innovation at Kanagawa University of Human Services. The application was submitted according to prescribed procedures before the commencement of the study, and ethical approval was granted (registration number: SHI No. 35). In addition, informed consent was obtained from all of participants on the website.

3. Results and Discussion

Table 1 presents the characteristics of the participants, with the number of patients in each disease group ranging from 9 to 25. While the overall sex distribution was 62.6% male, diseases such as Sjögren's syndrome, systemic lupus erythematosus, and multiple sclerosis/optic neuromyelitis spectrum disorder were more prevalent among females.

Next, we depicted the emotional journey of patients from the onset of recognizing bodily anomalies to receiving treatment and care (Figure 1). The emotional status was 2.50 at the stage of recognizing bodily anomalies, 2.45 when visiting clinics, and 2.95 upon arrival at the specialized hospitals. Thus, the patients' emotional states during the period from recognizing bodily anomalies to visiting specialized hospitals were predominantly negative. Moreover, their emotional status gradually increased as they entered the diagnostic phase (3.23), reached a confirmed diagnosis (3.24), and began treatment and care (3.84). In sum, these results suggest that patients experience a positive shift in their emotional status upon entering the diagnostic phase at a specialized hospital, which intensifies during the definitive diagnosis and treatment phases. Notably, patients who visited specialized hospitals directly after receiving their health check results maintained an emotional status between 4.14 and 4.35, with minimal fluctuations.

Furthermore, we classified the participants' emotional responses to a definitive diagnosis into six themes: relief, anxiety, shock, no change, and other factors. The results in Figure 2 show that relief was the most prevalent theme (41.9%), followed by no change (19.9%), anxiety (14.0%), shock (13.4%), and resignation (6.5%). After a definitive diagnosis, participants experienced a mix of shock and relief, with many expressing relief at the end of their understanding of the cause of their symptoms. Interestingly, 19.9% of the participants reported no significant emotional change, positively accepted their diagnosis, and approached treatment and care from a forward-looking perspective.

Moreover, we explored specific emotions during the period when patients felt most negatively about their journeys, namely, leading up to specialized hospital visits. Responses were categorized into four themes: frustration, fear, anxiety, aversion to hospital visits, and other factors (Figure 3). Frustration was the predominant theme (51.3%), followed by fear and anxiety (43.6%). The results indicated that patients experiencing frustration were often unable to identify the cause of their symptoms even after multiple clinic visits and tests, which contributed to their negative emotional state. Additionally, the fear of deteriorating health during this process proved to be a significant contributor to negative emotions. One of the causes for the decline in the emotional state of these patients is the delay in referrals from general practitioners (GPs), who often have the opportunity to initially diagnose patients with rare diseases, to specialized hospitals. Many comments from patients indicated that GPs did not refer them to specialized hospitals without suspecting a rare disease. It became evident that addressing how to promptly refer patients from GPs to specialized hospitals is a crucial issue that needs to be resolved. These findings provide crucial insights into the unmet needs of patients with rare diseases; notably, they emphasize the importance of an early confirmed diagnosis for QOL.

In our previous study, we found that a prolonged duration before a definitive diagnosis leads to a decline in physicians' trust (*12*). A weak trust relationship may result in patients being less inclined to communicate detailed symptoms to physicians, potentially hindering a thorough understanding of their conditions. In the case of rare diseases, expediting the steps to visiting a specialized hospital before reaching a definitive diagnosis is crucial. GPs play a significant role in this work; however, GPs should build strong relationships with specialized hospitals to ensure they can efficiently diagnose rare diseases while treating multiple patients.

Further, it is essential for patients to contemplate what health information they should communicate to physicians. Notably, social media initiatives have recently emerged that enable patients with similar health issues to actively exchange opinions and find clinicians with knowledge of rare diseases (*13*). Along these lines, Yamaguchi *et al.* (*14*) explored whether social media postings of the medical history of rare disease patients may shorten the timeline for diagnosis and treatment. Such endeavors not only benefit researchers but may also motivate undiagnosed patients worldwide to seek medical attention.

This study has two strengths. First, it demonstrates the ability to depict the journey of a patient with a rare disease and capture changes in their emotional status at each stage; this approach can notably be used to identify the unmet needs of patients with rare diseases. Second, the study evidences that a web survey, which allowed for an analysis with a large sample size, is a valuable approach for capturing changes in patients' emotional statuses.

However, this study has several limitations. If it is

Table 1. Characteristics of the study sample

Characteristic	Study sample		Crohn's disease		Idiopathic interstitial pneumonia		Eosinophilic sinusitis		Sjögren's syndrome		Polycystic kidney disease		IgA nephropathy		Systemic lupus erythematosus		Parkinson's disease		Idiopathic dilated cardiomyopathy		Multiple sclerosis/ neuromyelitis optica		Spinocerebellar degeneration	
	n	(%)	*n*	(%)	*n*	(%)	*n*	(%)	*n*	(%)	*n*	(%)	*n*	(%)	*n*	(%)	*n*	(%)	*n*	(%)	*n*	(%)	*n*	(%)
Total	179	(100.0)	22	(100.0)	9	(100.0)	13	(100.0)	16	(100.0)	20	(100.0)	24	(100.0)	25	(100.0)	14	(100.0)	14	(100.0)	13	(100.0)	9	(100.0)
Sex																								
Male	112	(62.6)	18	(81.8)	9	(100.0)	12	(92.3)	2	(12.5)	15	(75.0)	15	(62.5)	6	(24.0)	9	(64.3)	13	(92.9)	6	(46.2)	7	(77.8)
Female	67	(37.4)	4	(18.2)	0	(0.0)	1	(7.7)	14	(87.5)	5	(25.0)	9	(37.5)	19	(76.0)	5	(35.7)	1	(7.1)	7	(53.8)	2	(22.2)
Age																								
< 29 years	3	(1.7)	0	(0.0)	0	(0.0)	0	(0.0)	2	(12.5)	0	(0.0)	0	(0.0)	0	(0.0)	0	(0.0)	0	(0.0)	1	(7.7)	0	(0.0)
29–49 years	40	(22.3)	10	(45.5)	0	(0.0)	2	(15.4)	4	(25.0)	5	(25.0)	5	(20.8)	6	(24.0)	1	(7.1)	2	(14.3)	3	(23.1)	2	(22.2)
50–59 years	66	(36.9)	9	(40.9)	1	(11.1)	3	(23.1)	6	(37.5)	8	(40.0)	10	(41.7)	12	(48.0)	2	(14.3)	3	(21.4)	7	(53.8)	5	(55.6)
60–69 years	50	(27.9)	2	(9.1)	5	(55.6)	8	(61.5)	2	(12.5)	2	(10.0)	9	(37.5)	4	(16.0)	7	(50.0)	7	(50.0)	2	(15.4)	2	(22.2)
> 69 years	20	(11.2)	1	(4.5)	3	(33.3)	0	(0.0)	2	(12.5)	5	(25.0)	0	(0.0)	3	(12.0)	4	(28.6)	2	(14.3)	0	(0.0)	0	(0.0)
Marriage																								
Married	118	(65.9)	12	(54.5)	8	(88.9)	9	(69.2)	7	(43.8)	15	(75.0)	20	(83.3)	15	(60.0)	9	(64.3)	10	(71.4)	8	(61.5)	5	(55.6)
Other	61	(34.1)	10	(45.5)	1	(11.1)	4	(30.8)	9	(56.3)	5	(25.0)	4	(16.7)	10	(40.0)	5	(35.7)	4	(28.6)	5	(38.5)	4	(44.4)
Education Background																								
Junior school	4	(2.2)	0	(0.0)	0	(0.0)	1	(7.7)	0	(0.0)	1	(5.0)	1	(4.2)	0	(0.0)	1	(7.1)	0	(0.0)	0	(0.0)	0	(0.0)
High school	46	(25.7)	5	(22.7)	0	(0.0)	6	(46.2)	5	(31.3)	4	(20.0)	4	(16.7)	10	(40.0)	2	(14.3)	6	(42.9)	4	(30.8)	0	(0.0)
Professional school	28	(15.6)	4	(18.2)	1	(11.1)	1	(7.7)	0	(0.0)	4	(20.0)	3	(12.5)	4	(16.0)	2	(14.3)	1	(7.1)	5	(38.5)	3	(33.3)
Junior college	20	(11.2)	3	(13.6)	0	(0.0)	0	(0.0)	3	(18.8)	1	(5.0)	4	(16.7)	3	(12.0)	1	(7.1)	2	(14.3)	2	(15.4)	1	(11.1)
Bachelor's degree	68	(38.0)	7	(31.8)	7	(77.8)	5	(38.5)	6	(37.5)	9	(45.0)	7	(29.2)	8	(32.0)	8	(57.1)	4	(28.6)	2	(15.4)	5	(55.6)
Master's degree and above	12	(6.7)	3	(13.6)	1	(11.1)	0	(0.0)	1	(6.3)	1	(5.0)	5	(20.8)	0	(0.0)	0	(0.0)	1	(7.1)	0	(0.0)	0	(0.0)
Other	1	(0.6)	0	(0.0)	0	(0.0)	0	(0.0)	1	(6.3)	0	(0.0)	0	(0.0)	0	(0.0)	0	(0.0)	0	(0.0)	0	(0.0)	0	(0.0)
Medical visit pattern																								
Initially visited the clinic	115	(64.2)	17	(77.3)	3	(33.3)	10	(76.9)	11	(68.8)	6	(30.0)	15	(62.5)	18	(72.0)	11	(78.6)	13	(92.9)	7	(53.8)	4	(44.4)
Visited specialized hospitals from the beginning	43	(24.0)	3	(13.6)	3	(33.3)	2	(15.4)	3	(18.8)	9	(45.0)	8	(33.3)	4	(16.0)	2	(14.3)	1	(7.1)	4	(30.8)	4	(44.4)
Others	21	(11.7)	2	(9.1)	3	(33.3)	1	(7.7)	2	(12.5)	5	(25.0)	1	(4.2)	3	(12.0)	1	(7.1)	0	(0.0)	2	(15.4)	1	(11.1)

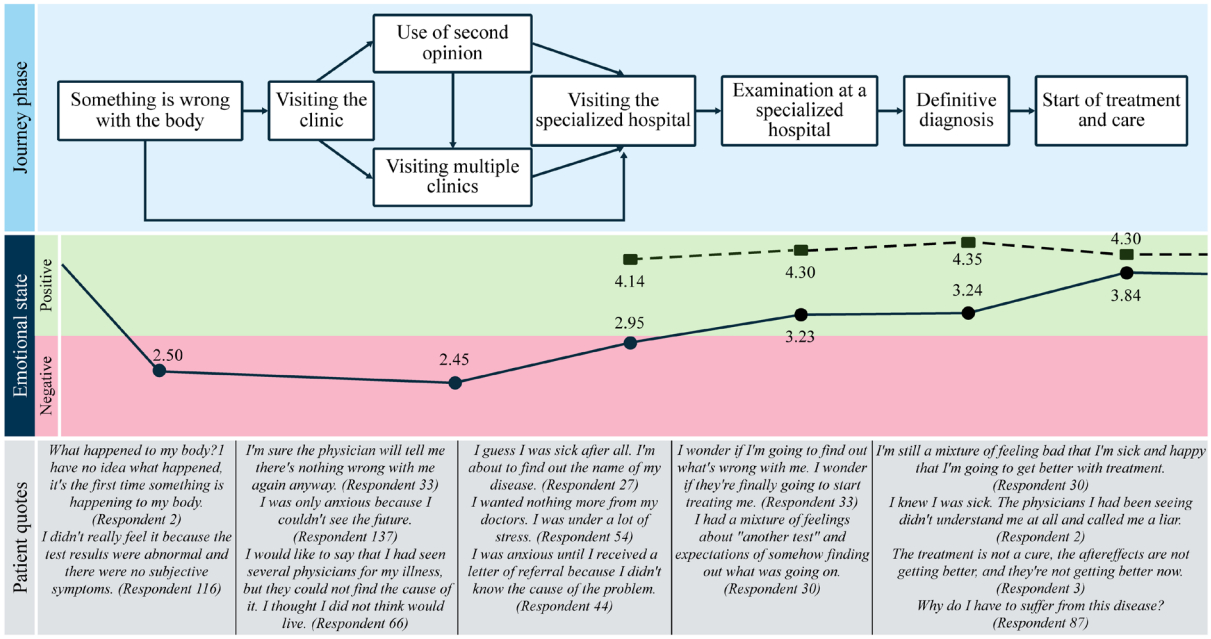

Figure 1. Emotional journey of patient with intractable disease.

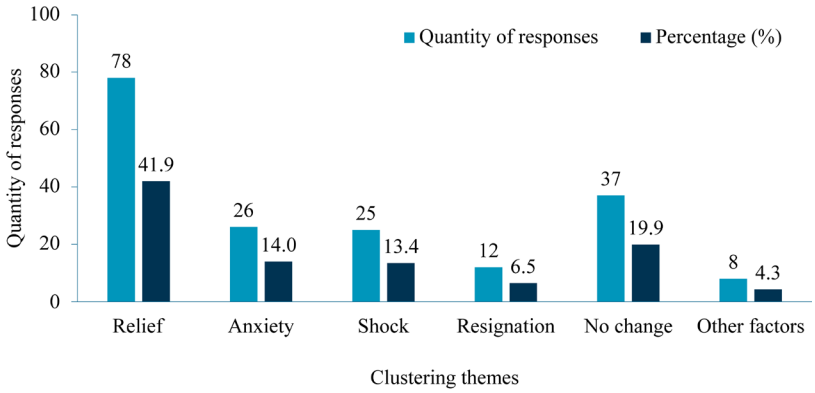

Figure 2. The emotional shift at the time of the definitive diagnosis.

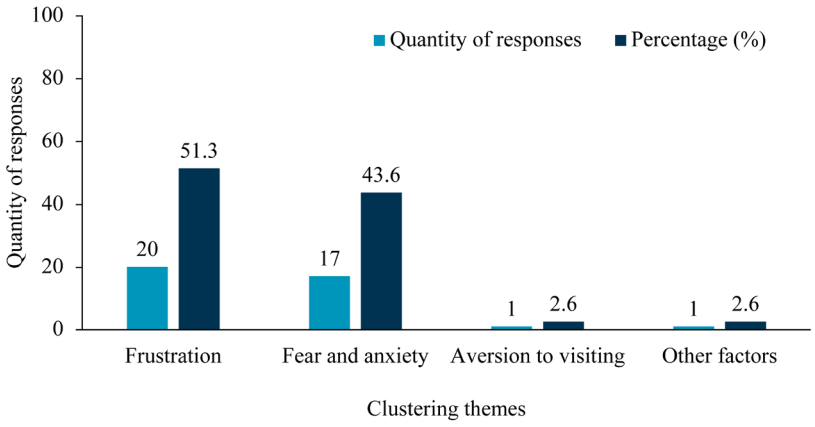

Figure 3. Clustering themes of specific emotions during the period when patients felt most negatively until they visited a specialized hospital.

anticipated that there are numerous patient journeys for each persona, thorough pre-investigation is necessary. The method of understanding the emotional states of patients through web surveys, as done in this study, may be more suitable for diseases that follow a somewhat typical medical course. In other words, this study focused on the challenges in the definitive diagnosis of specific rare diseases in Japan, and the selected disease group

allowed for some degree of prior prediction of the patient journey. In future research, it is necessary to verify the effectiveness of this survey method using a broader range of disease groups and sample sizes.

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