

**Supplemental Table S1. Questionnaire survey conducted to assess the emotional status**

Generally, in the case of rare diseases, patients go through the following steps:		
<ol style="list-style-type: none"> <li>1. When they first notice abnormalities in their bodies</li> <li>2. Initial visits to a clinic, utilization of second opinions, and visits to multiple hospitals</li> <li>3. When they visit a specialized hospital</li> <li>4. When tests begin at the specialized hospital</li> <li>5. When a definitive diagnosis is made</li> <li>6. When treatment and care commence</li> </ol>		
Items		
1	Until you received a definitive diagnosis, which of the following steps was your initial one?	
	1	Initially visited a clinic.
	2	Went directly to a specialized hospital.
	3	Other (please specify)
2	SA	Please select the emotional state you experienced when you first noticed abnormalities in your body or when you noticed abnormalities based on health examination results. Please respond on a scale from low (-5) to high (5).
	1	5 (Very positive state)
	2	4
	3	3
	4	2
	5	1
	6	0 (Normal state)
	7	-1
	8	-2
	9	-3
	10	-4
	11	-5 (Very negative state)
3	FA	At this point, what specific thoughts did you have ? Please write down any concerns or thoughts, including feelings of anxiety.
4	SA	Please respond with the emotional state you experienced during the period between visiting the clinic and going to the specialized hospital on a scale from low (-5) to high (5).
	1	5 (Very positive state)
	2	4
	3	3
	4	2
	5	1
	6	0 (Normal state)
	7	-1
	8	-2
	9	-3
	10	-4
	11	-5 (Very negative state)

5	FA	At this point, what specific thoughts did you have? Please write down any concerns or thoughts, including feelings of anxiety.
6	SA	Please respond with the emotional state you experienced when you visited the specialized hospital on a scale from low (-5) to high (5).
	1	5 (Very positive state)
	2	4
	3	3
	4	2
	5	1
	6	0 (Normal state)
	7	-1
	8	-2
	9	-3
	10	-4
	11	-5 (Very negative state)
7	FA	At this point, what specific thoughts did you have? Please write down any concerns or thoughts, including feelings of anxiety.
8	SA	Please respond with the emotional state you experienced when tests began at the specialized hospital on a scale from low (-5) to high (5).
	1	5 (Very positive state)
	2	4
	3	3
	4	2
	5	1
	6	0 (Normal state)
	7	-1
	8	-2
	9	-3
	10	-4
	11	-5 (Very negative state)
9	FA	At this point, what specific thoughts did you have? Please write down any concerns or thoughts, including feelings of anxiety.
10	SA	Please respond with the emotional state you experienced when a definitive diagnosis was made at the specialized hospital on a scale from low (-5) to high (5).
	1	5 (Very positive state)
	2	4
	3	3
	4	2
	5	1
	6	0 (Normal state)
	7	-1
	8	-2
	9	-3

	10	-4
	11	-5 (Very negative state)
11	FA	At this point, what specific thoughts did you have? Please write down any concerns or thoughts, including feelings of anxiety.
12	SA	Please respond with the emotional state you experienced when treatment and care commenced on a scale from low (-5) to high (5).
	1	5 (Very positive state)
	2	4
	3	3
	4	2
	5	1
	6	0 (Normal state)
	7	-1
	8	-2
	9	-3
	10	-4
	11	-5 (Very negative state)
13	FA	At this point, what specific thoughts did you have? Please write down any concerns or thoughts, including feelings of anxiety.
14	SA	At which point between the onset of symptoms and the definitive diagnosis did you feel the most emotionally downcast?
	1	Around the time symptoms first appeared
	2	When I first visited the clinic
	3	When I was visiting multiple clinics without understanding the cause
	4	When I visited the specialized hospital
	5	When a definitive diagnosis was made
	6	When treatment and care commenced
	7	Other
15	FA	Please also describe the reasons for your choice.
16	SA	At which point between the onset of symptoms and the definitive diagnosis did you experience the most improvement in your state?
	1	When you visited the specialized hospital
	2	When a definitive diagnosis was made
	3	When treatment and care commenced
	4	Other (please specify)
17	FA	Please also describe the reasons for your choice.