

Supplementary file 2 – Illness plots and parallel chart

2.1. Illness plot addressed to patients

We invite you to tell us about your experience of living with a hereditary retinal disorder related to the RPE65 gene (RPE65-related IRD). You can write instinctively and freely, regardless of the form and length of your narrative. Any episode you consider significant will be welcome.

Before the IRD clinical diagnosis... The first signs that something was wrong... I felt... To understand what it was about... The facilities I visited, the healthcare professionals I met... Waiting for the clinical diagnosis... When they told me that it was an IRD, I felt... The genetic test for me was... That time, with family... With others... For me, seeing was... The activities I liked to do... The activities I could not do... At school/work... Healthcare professionals and treatments were... The centre where I am treated... Healthcare professionals and treatments are... Gene therapy for me is... Between one visit and the next... With my family... With other people... For me, seeing is... The activities I like to do... The activities I cannot do... Today at school/work... Rethinking about my care pathway, I would have liked that... Thinking about tomorrow, I feel... For tomorrow, I would like to...

Thank you for your time, energy and attention. We ask you one last question: How did you feel about writing your experience?

2.2. Illness plot addressed to caregivers of patients with an RPE65-related IRD

We invite you to tell us about your experience of living next to a person with a hereditary retinal disorder related to the RPE65 gene (RPE65-related IRD). You can write instinctively and freely, regardless of the form and length of your narrative. Any episode you consider significant will be welcome.

Before the diagnosis of IRD... When we first noticed that something was wrong... I felt... She/he felt... To find out what it was... Looking at her/him I thought... The facilities we visited, the healthcare professionals we met... Before the IRD clinical diagnosis... When we were told that it was an IRD, I felt... For me, the genetic test was... At that time, she/he with the family... She/he with other people... For her/him, seeing was... The activities she/he liked to do... The activities she/he could not do... At school/work... For her/him, I wanted... Healthcare professionals and treatments were... Today I feel... Today she/he feels... The IRD is... The centre where she/he is treated... Treatments and caregivers are... Gene therapy for me is... Between one visit and the next... With family... With other people... For her/him, seeing is... The activities she/he likes to do... The activities she/he cannot do... Rethinking to the care pathway, I would have liked that... Thinking about tomorrow, I feel... For tomorrow, I would like to...

Thank you for your time, energy and attention. We ask you one last question: How did you feel about writing your experience?

2.3. Parallel chart on patients affected by an RPE65-related IRD addressed to healthcare professionals

The first time I saw this person with an IRD, I thought... The patient and her/his relatives told me... Addressing symptoms, they told me that she/he could do/not do... I felt... And I did... Waiting for the clinical diagnosis... When I had to communicate the clinical diagnosis... Proposing the genetic test was... The relationships with family and other people of the person with IRD... For her/him, seeing was... Between one visit and the next... In her/his activities at work/study/play... Today this person... With family and other people... Today this person, during work/study/play... The people next to her/him... My goal for this patient is... With her/him I feel... From the relationship with the patient, I've learned... For tomorrow, I wish that I... For tomorrow I hope she/he...

Thank you for your time, energy and attention. We ask you one last question: How did you feel about writing your experience?