

Supplementary file 3

3.1. Narrative from an underaged patient affected by an RPE65-related IRD

My mum realised that something was wrong when I was very young, about 18 months. I never felt different and was unaware of the difficulties. The professionals I met were helpful, kind, welcoming people who made me feel at home. While waiting for the diagnosis, I was very calm. When they told me that it was a hereditary retinal disease, nothing had changed for me. My eyes did not work as well as a healthy child's. The genetic test was a big step for me. The genetic test was just another test for me. At that time, we were very relaxed in my family. We had no particular problems with others. I've always seen that way. I don't know how others see. I like skating, dancing and cycling. At first, I could not ride a bike, then I did. I like school a lot, so I do not have any difficulties.

Some pills I will remember all my life because they were terrible, but the rest of the treatment was easy. Today I feel happy. The disease is stable for now, and I feel calm. I have more than one centre, and they are doing everything they can. The doctors are very nice and friendly, and I don't have any special treatment. Gene therapy is a great possibility for me because it will help keep my eyes stable, which would be very positive. Between one visit and the next, I feel calm and have no particular tension. I feel very relaxed with my family. With others, I am a sunny child. Seeing is a beautiful thing because it allows me to relate to the outside world. I like riding my bike, being with my animals, being with friends. I cannot do team sports. School is going well, and I feel at ease; I am learning to use the computer. Rethinking about the care pathway, I think everyone did what they could and what was right to do. When I think about tomorrow, I feel happy with the people who love me, and I would like everything to remain as it is now.

3.2. Narrative from an adult patient affected by an RPE65-related IRD

I don't remember a precise year, but the first signs that something was wrong were around the age of 6 or 7 when we were driving at night, and I realised that I couldn't see what my father needed to go. I could only see the light sources but not what they were illuminating. When I was 15 years old, I was driving back to the institute on Sunday afternoons; it got dark on the way, and I had a hard time walking from the station to the institute. If I had to walk together with other blind people, I would have done it with ease. I felt very uncomfortable, inappropriate, and inexplicably clumsy. In the evenings, I could not move to go out alone. If I accompanied other blind people, even two, I felt no discomfort, and the journeys went smoothly. I knew about my illness. I also met ophthalmologists who seemed to know less about it than I did. For the hope of treatment or recovery, ophthalmologists had already been consulted for my brother before I was born, or at least when I was small. While waiting for the diagnosis, I never had any expectations. When I learned that it was a hereditary disease, I was a child, and I had no reaction. I did the genetic test when I was 54 and, since I knew that there is a lot of retinitis, it was pure curiosity. Is it positive or negative to learn at 54 what exactly you have? Negative because it shows how much interest there is in such a disease: very little. It's good that research is going on, even if it's at a snail's pace. Only my mother has an attitude of some hope.

Having changed places I've gone to live [...] I have no way of comparing before and after. I never hid my problem, so they took me as I was. Seeing, even a little, even with difficulty, even when the amount of light allowed me to do things, "seeing" was, of course, more accessible. But since I knew that I would lose my sight sooner or later and that this took place over quite an extended period, I used these facts to run for cover, with the aim of not stopping. I liked cycling, which is different from riding a tandem bike, going out to look for glimpses of views, reading comics. The reading in black and the cycle rides gradually faded away. At school: I couldn't see the blackboard and do my homework alone. At work, as a teacher, I couldn't fill in the register by myself.

I only went for specific treatments for retinitis, useless but specific.

Today I feel the same as I did before. The disease has degenerated almost to the end. Functionally I am blind. Every now and then, I play the lamppost game, trying to catch the light from the lampposts as we walk down the street... in the evening.

When I go to the centre, I spend no less than 4 hours there, and 2 of them are waiting. So far, the people working there feel welcoming and helpful. So far, I've only had check-ups. I see gene therapy as an attempt to maintain the current faculties of the retina. We are still far from hoping for any kind of recovery, let alone a recovery measurable in tenths. I don't know why I've only had one visit to date where this therapy was mentioned for the first time. My family and I are on the same wavelength at the moment. So the family attends events to support my needs as they arise. I go to the swimming pool to do water gymnastics, with the others from our sports club we organise dinners in the dark. I have weekly music rehearsals with a group where only I am blind, we go to play in clubs, I go to see sculpture exhibitions if it is allowed to touch, of course. With my wife, who is also blind, we travel: when I have the chance, I like to get to know the cities, walking in their historical centres, alone. I read and listen to music. Unfortunately, I like to eat, so every opportunity is good to try a new restaurant. In everyday life, I am autonomous. Since there are many things I can do as a blind person, it seems useless to me to try at all costs to do something where sight is the only possibility. Like, for example: driving. I am autonomous in my activities; I only find difficulties when the computer aids are not adequate or modify the websites without considering the rules needed to include visually impaired users. I have been using personal assistants selected and trained by me for years in those areas where only sight works. Thinking back to my own care path, I would have liked to have had this care in the 1960s. My future is not conditioned by the presence of this care. But it seems worthwhile to me to do it: what will be, will be. We visually impaired people need civilisation. If in the behaviour of citizens, people, institutions, the observance of rules also prevails in the realisation of public and social things, we are in the right place. But in our society, this does not happen to a sufficient extent, so tomorrow will still be about making do as one can, with or without this care.

3.3. Narrative from a caregiver of a patient affected by an RPE65-related IRD

We toured the hospitals in our region. Visit after visit, the anamnesis and electrophysiological examinations were not sufficient for a diagnosis. Many signs and symptoms were confused between the different diseases affecting the retina. At six months, we realised that something was wrong with the involuntary eye movement, always searching for light, the lack of eye contact between

mother and child during breastfeeding. I felt an immense sense of absolute helplessness as a parent in front of her baby. I really needed to understand why... He is a very peaceful child; he plays, jumps, learns something new every day and knows how to give so much love. To understand what it was all about, we researched the subject because it helps us accept. I would see him and think that it's just a bad dream, with the hope of waking up to normality. We met very helpful and wonderful people.

When they told us that we needed to take a genetic test, I thought that there were no relatives with severe vision problems; it seemed so absurd. The genetic test was a simple saliva sample that allowed for greater accuracy; the genetic diagnosis was essential to know the gene that causes the disease. The wait for the diagnosis seemed like an eternity. The diagnosis, when it came, was a starting point; news like that turns your life upside down. He is a very calm child and learns every day to become more and more autonomous. When they told us that it was a hereditary retinal disease, I felt terrible because you don't expect it. It seems impossible to me to have a congenital disorder of a genetic nature in a family where there were no known cases. At that time, the environment was fundamental because I was more autonomous. At home, with the organisation of spaces, he moves on his own, and so he gets used to making do. He is friendly and loves being with other kids; he is cheerful, curious, and intelligent. There is a difference between seeing the light and not seeing it at all, so we are confident that everything has not degenerated. He likes to do everything, watch cartoons and knows some dialogues by heart. Among the activities he finds hard to do are playing football, drawing, playing basketball. As a parent, the only thing you want in life is to protect your children. It is challenging to live with this disease because I have mortifications in every area of life. We are waiting for the gene therapy to finally allow us to see the light at the end of the tunnel.

Today I feel very serene, and I never stop dreaming that after the discovery, the waiting, the hope, the light will finally come. Today he feels more peaceful, and day after day, he learns to be more vital to face his life. The disease is genetic, rare, incurable; we are healthy carriers of the defect and have passed it on to our son. The hospital that is treating us is a centre of excellence, and we have carried out the genetic test. Getting a diagnosis for a rare disease is not always easy. It is a long and tiring process. The time between checks is too long. For me, gene therapy would be the miracle we have been waiting for, as we are entering an era where diseases that were once incurable are becoming curable. Thanks to the love of those around him, he is learning to live with all the strength he needs. He is an adorable child and knows how to make others love him. He is a very healthy child who rarely gets sick. His eyesight is not yet very impaired; otherwise, he is very cheerful. Rethinking the care pathway, I would have liked to have had more information on this disease's knowledge, together with the proper psychological and educational support. If I had to imagine a service for all the people with the same disease as my son, I would think of a specialised centre for this disease, which could guarantee proper support for parents who face enormous difficulties. When I think of tomorrow, I don't know what awaits us. Still, we are very enthusiastic about the progress of science. I would like to see proper care centres and improved schooling for people with this disease in the future. Reading difficulties are essential, and there is a lack of adequate tools to deal with them.

3.4. Parallel chart on a patient affected by an RPE65-related IRD from a healthcare professional

Poor child: he is not living his life like his other healthy peers. The parents reported that they needed advice on how to make him as autonomous as possible. He could not play, run, be independent in his personal and school affairs. He could not orientate himself in space. The situation worsened from sunset onwards when the child panicked. The whole family hardly ever went out in the evening, not even for a simple dinner. I felt obliged to build a personalised rehabilitation programme to find alternative strategies to give the family tools and reassure the child to increase his self-esteem. I asked the child to tell me everything he wanted to do, everything he thought to do poorly, and his fears when he got stuck on various occasions. I asked the parents what they saw when they were with their child, their fears, their difficulties, what they wanted help with, what they hoped for. I gradually started to indicate how to organise the house according to the child's size, what light or contrast measures should be taken and how to organise the school material to make it more usable. It was not my job to communicate the diagnosis.

Other people often do not understand what and how he sees, so it ranges from denial to being overprotective. To see was not to fall, not to stumble, play football, watch television together with the family, write in the notebook without difficulty, and read without difficulty. The family was heartened and happy about the small degree of autonomy their child was able to achieve. The child began to experiment on his own without requiring the constant presence of others. When studying, the child felt frustrated because he realised that he could not write or read like the others. He felt different because he could not demonstrate his abilities and was frustrated because he could not keep up with others.

Today, he is more confident about himself, his abilities and also his limits. He has learned to set himself small goals, overcome them with his own alternative strategies and move forward. With other people, he is more present and less dependent. At school, he has found his own alternative methods to do almost the same as other peers; he participates more in the class group and verbalises his visual difficulties when he has a problem. The people around him seem more serene and confident in his potential. My aim is to make him aware of his challenges to face them with alternative strategies and overcome them even if with limitations. I feel stimulated to find with him alternative solutions to make him autonomous. I am learning from the caring relationship that there is no limit to the potential.

I would like to be able to help them even more in the future. I would like him to be aware of how extraordinary his will power is.