Joyce de Ruiter was sixteen years old when her parents had to tell her that she’d been diagnosed with Usher syndrome, an inherited disorder which deteriorates hearing and especially vision during life, often resulting in people becoming deaf and blind. “Finally, the puzzle pieces of hearing loss and night blindness had been united,” Joyce tells us. “I crossed my own limits for too long, but now I really understand the limitations.”

Joyce and her brother were hearing impaired from birth but started to experience initial symptoms of blindness around puberty, which was strange, as it didn’t run in the family. A diagnosis was lacking until her then 18-year-old brother’s vision deteriorated even further and an ophthalmologist diagnosed Usher syndrome. Joyce was diagnosed with the same disorder when she was just sixteen years old, radically changing her life and future plans. “I wanted to study work, get married, and have a family. Would that still be possible?” When I discovered the study Visual Marketing, the fire started burning again, even though the name of the study didn’t sound very useful when you have a visual handicap. Joyce made me happy and I’m convinced this enables me to do the things I want to do - despite all the limitations.

Joyce de Ruiter, Ambassador of the Usher Syndrome Foundation

INVESTING IN A POSSIBLE TREATMENT IS CHEAPER THAN LIVING WITH THE LIMITATIONS

Joyce decides to tell her personal story in the newspaper, on radio, and on TV as an ambassador for the Dutch Usher Syndrome Foundation in order to underline the importance of scientific research and development of any treatment. “I think it’s important to bring change and agility. Usher constantly confronts you with problems and changes that you have to learn to deal with. I link my personal story to insights in psychology about how to keep an agile mindset. With that, I try to be of value to other people. I think that is the most meaningful thing you can do in a human life.”

Tunnel vision, literally

Joyce is now 38. Her hearing loss is 70 decibels, but with advanced hearing aids she can still function reasonably well. Her vision is greatly reduced, though. “Just last year I still had wideopen vision, which is less than a toilet roll I can see through. My orientation on the street is becoming more and more difficult, I haven’t been able to ride a bicycle for years and I feel unsafe in crowded environments. All because of my reduced vision. This is why scientific research is so important. Investing in a possible treatment is extremely cheaper than living with the limitations. It would be fantastic if a treatment could be found that stops the deterioration and that children do not have to experience any hearing or sight restrictions at all. Of course, we hope that all patients with Usher syndrome will recover. That is why the volunteers of the Usher Syndrome Foundation are putting their heart and soul into continuing to make research into a treatment such as that at the Radboud university medical center possible.”

Restoring the function of light-sensitive cells

Usher syndrome arises from a genetic mutation which causes the light-sensitive cells in the eye progressively die as a result, and people become vision impaired or sightless. Joyce is an example of a person who has been given the hope that “the light-sensitive cells in the eye can be restored.”

Zebrafish as part of the solution

Erwin stumbled upon an unexpected animal to establish whether the genetic patch could preserve visual function. “Zebrafish happen to have an USHA2 gene and protein that’s very similar to those of humans. If a mutation is introduced into this gene, the visual function of the zebrafish is also significantly reduced. They are an excellent model for studying USHA-related visual dysfunction and for determining the effect of a therapeutic intervention. Indeed, visual function of USHA3 mutant zebrafish treated with the genetic patch was highly improved.”

INITIAL RESULTS FROM A CLINICAL STUDY IN PATIENTS ARE HIGHLY PROMISING

From discovering the gene to, hopefully, developing a therapy which will give patients a positive outlook. Erwin Van Wijk has been involved with Usher syndrome research from start to finish, which is quite unique in this field of work as it shows how rapidly developments are following up on each other. “In the most recent study, thecheapinjections stopped the progression of hearing loss. The children who had hearing loss started to hear and slowly start to lose their vision before puberty. We developed a genetic patch that may be able to limit the damage to vision, our data and the results of the trials look positive.”

“The hearing problems in Usher are not easy to treat because the underlying cause already arises during development in utero,” says Erwin. “With my research group, I have focused mainly on the visual problems. These arise during life, so there are more opportunities to do something about them at an early stage. The goal of the research was therefore to slow down or even prevent the deterioration of vision. First of all, we need to know which and how genetic defects lead to problems. Mutations in the USHA2 gene are the most common, so we took this gene as the primary starting point for developing a therapy.”

After this, the first careful steps towards the translation of the results into a use in humans could be taken. First tests in Usher patients with a mutation in the specific region of the USHA2 gene showed promising outcomes, resulting in the design of a large multicenter follow-up study in which the results of the initial clinical trial will hopefully be corroborated in a large cohort of patients. If results hold up, a request for market introduction can be submitted to the American FDA and the European EMA.”

Bringing light to the dark

Erwin van Wijk
researcher at Radboud university medical center