

Shine a Light on XLH

Heidi's story

Heidi, from Espoo, Finland, was diagnosed with X-linked Hypophosphataemia (XLH) when she was a baby; however, as Heidi has no family history of XLH, the genetic mutation that causes her disease was spontaneous. As a result, Heidi is the only person in her family with XLH.

When Heidi was eight months old, her parents noticed that she wasn't growing normally, and that her legs were bowed. They took her to the hospital for tests, where several more common diseases were ruled out, and specialists eventually diagnosed her with X-linked Hypophosphataemia (XLH).

When she was two years old, Heidi's family met Anu and her daughter Tia, who are also living with XLH and are part of the Shine a Light on XLH campaign, at a family coaching course. Anu and Tia, who is of similar age to Heidi, have been a fantastic support for her. Knowing others who suffer from XLH has made Heidi feel less alone in her condition; having a friend to confide in about her struggles with XLH has been invaluable.

Becoming a teenager was difficult for Heidi. All she wanted was to be normal, and not to be held back by her condition. But unfortunately, at 13, Heidi's XLH caused her to develop spinal stenosis, a condition that puts pressure on nerves in the spinal column, for which she required surgery. Although the surgery helped to alleviate her pain for a while, it did not stop it completely, and this has caused her to miss a lot of school. Even now, the pain she experiences on the worst days is unbearable.

Though she may appear on the surface to be a happy, social person, the past ten years have been very tough for Heidi and have taken a serious toll on her mental wellbeing, causing her to suffer from depression. As she is the only person in her family with XLH, she has always felt that her family and peers see her as being "different". The pain she experiences every day and the knock-on effects this has had on her mental health is difficult for others to comprehend, and has made living with XLH extremely challenging for Heidi. Although her family was – and continue to be – extremely supportive, Heidi often felt alone and misunderstood growing up with XLH.



"My disease doesn't show that much on the outside, it's kind of all inside me. I struggle with nerve pain, pain in my back and legs, and depression. It was a huge surprise to my family and friends that I struggle with depression and so much pain. No one else can see the pain I suffer from except me."

For the future, Heidi would like to see improvements in XLH awareness, particularly among healthcare professionals. When going to the doctor, or the emergency room, she typically spends most of her appointment explaining what XLH is. This takes precious time away from the reason she went in, to help her understand and deal with her pain.

Heidi would also like to see improvements in care for people living with XLH. Currently, Heidi takes active vitamin D supplements and pain killers to help her manage her pain. She also benefits from physiotherapy but feels that this needs to be offered to her more frequently in order to ease her pain and help her to achieve her goals in life.

About the makeup

Heidi spoke about XLH developing through her spinal cord. Using this part of her personal background, I created paintwork representing the nervous system connecting to a heart shaped design reflecting on the idea "love wins out". She has a very supportive family around her and that radiates from her energy after speaking with her. The lightwork around the face represents a veil, to show transparency of being a normal, independent person behind any of the physical traits of XLH. Heidi spoke about having her own emotions, dreams and aspirations in life aside from the limitations of living with XLH. The vibrant colours are to showcase her strength and power."

- James Mac Inerney, Makeup Artist

The Shine a Light on XLH campaign was organised and fully funded by Kyowa Kirin

Heidi would love to have her own family one day and know that if her children are diagnosed with the genetic disease, that they will not suffer as she has. She also has big ambitions to travel in her life and wants to see everything from the Grand Canyon to Mount Everest. When she is able to travel, Heidi hopes that she won't be in pain and will be able to walk around the many places she wishes to visit.

"I want younger people who suffer from XLH to know that they can still dream big. I believe that despite having XLH, the sky is the limit and you can achieve anything you want to."

