Wolfram syndrome - our journey

Wolfram syndrome (WS) is an ultra-rare condition. About 90 people in the UK are affected. Many doctors have and never will see a person with WS.

My daughter was diagnosed with WS at the age of eight following three years of investigations. The condition seems to have started when Jennifer was about two and a half years old with coughing attacks, followed by vision issues, diagnosed when she was five. At the age of six she was diagnosed with Type 1 diabetes and a neurogenic bladder. At the age of 10, she had the second of two severe choking attacks in a year, which resulted in being on a ventilator for three weeks and then a 'temporary' tracheostomy being carried out. Eight years later this is still in place.

At the age of 12 she was diagnosed with scoliosis and over the last three years she has developed short-term memory loss with her balance deteriorating so much that a wheelchair is sometimes needed.

Around the age of six, she had the first of several sleep studies that showed she had central obstructive sleep apnoea. She would de-saturate for up to 13 seconds a time, up to 20 times in an hour, but never requiring any medical intervention. We have been told she has an extreme form of WS as everything has happened within the first 10 years of her life.

Daily Life

Jennifer never had a normal childhood as time was spent out of school at hospital and invites to friend's houses and parties stopped from the age of six. Unlike her peers she can't go out and get a part time job; drive a car; go out to a bar. Over the years she has received the necessary treatment and medication for the different parts of the condition she is affected by. She has five insulin injections a day, at least three tablets a day, one twice a day, and is now also on a ventilator at night along with any additional medications as required.

In 2016, she was admitted to hospital seven times with either an upper respiratory tract infection or pneumonia. This is when the ventilator was first introduced and was used when required. In 2017, this became a permanent part of night time with a prophylactic preventative antibiotic prescribed for over the winter months. So far, this has helped. Jennifer is seen at the annual WS MDT Clinic in Birmingham and then has other medical appointments though out the year at our local hospital and hospitals in London, about 60 miles away. Since 2007, the beginning of our journey, we have seen almost every department in the hospital.

The future

We know that a 'sticking plaster' treatment is being trialed to slow down or halt the progression but we need more researchers working together, sharing information to find an actual cure. The 'sticking plaster' gives those affected and their families hope that they might live longer than the expected average age of 30-40 years.

The hardest part of the condition for those affected is the loss of vision – that is what every person with WS would like to see cured first. Other parts of the syndrome can be dealt with by medication, but vision loss is the hardest thing; no longer being able to do things easily for yourself when once you could is tough on everyone. As a parent, watching your child's health and their condition deteriorate is really tough.

I ask those working on this condition to listen to those affected. They are the ones living with the condition day in and day out. They know how they're feeling and what they are having to cope with. Don't just dismiss a symptom. See if anyone else is reporting the same, as there will be. Work with patient groups - they want to see success just as much as you. We're continuously teaching doctors when we attend appointments or get admitted to hospital. Even the specialists at our MDT clinics are learning from us each time we attend. ■

Jennifer, who has Wolfram syndrome

WRITTEN BY:
TRACY LYNCH
CEO & CO-FOUNDER,
WOLFRAM SYNDROME UK