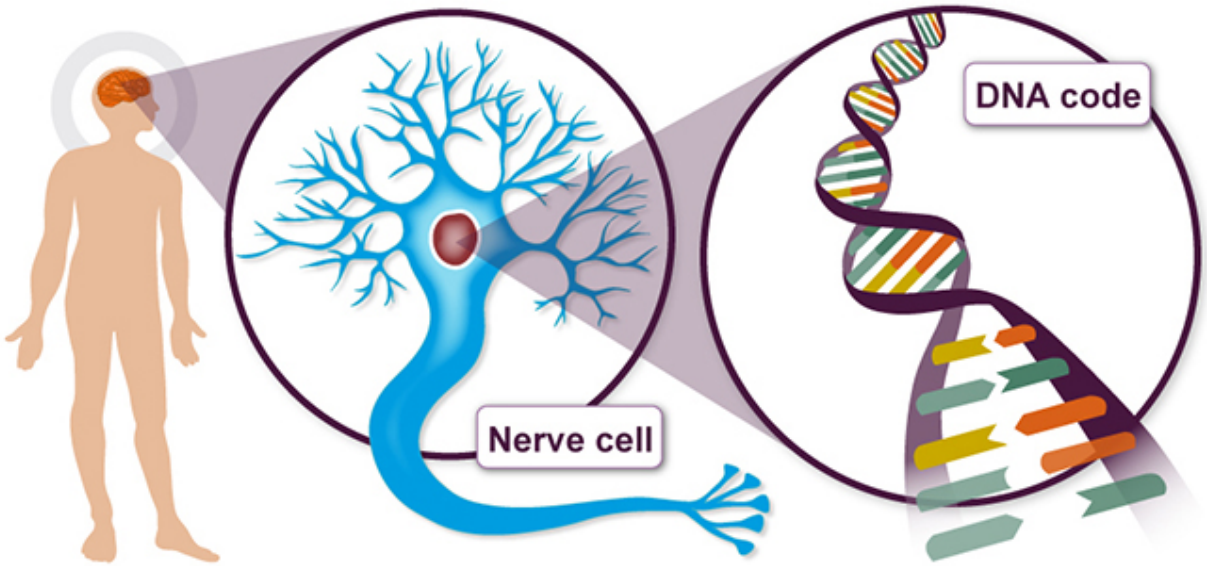
MedicalXpress

Mum in her 30s discusses realities of life with a 75% chance of Alzheimer's

August 22 2017



Credit: Alzheimer's Research UK

With a one in over 600 million chance of her parents both having the same rare, inherited gene, a young mum has spoken of the anguish of knowing she has a 75 percent risk of developing Alzheimer's in just a few years, and how she's found hope through research.

Sarah, whose name has been changed to preserve her anonymity, and her

siblings, are thought to be the only people in Europe with such a high risk of dementia.

The chances of their parents both having the faulty gene is so rare that the average person would stand a higher chance of winning the lottery 48 times with the lotto's chance being one in 14 million.

The family watched on as their father died from Alzheimer's in his early 50s, after initially showing symptoms in his mid-40s. Their mother began to show symptoms just months later.

A second blow was dealt to the siblings when they found out both their parents' diseases were caused by the same rare, inherited gene mutation – giving them an unusually high risk of Alzheimer's themselves. Only five percent of people with Alzheimer's develop the disease under the age of 65, and only 13 percent of these early-onset cases are caused by inherited gene mutations.

Now in her 30s, Sarah can't escape the thought of what's likely to come in the near future. She started a family before knowing about any genetic risk and now worries for her three young children.

But Sarah has found a way to take control back by volunteering in a unique global research study to help develop approaches that could help her children in future. Sarah's other siblings also face a future waiting to see whether they too will soon start showing the symptoms, and know their children are also at risk.

Sarah, who lives in the UK, said:

"My dad was a real joker and when he first started repeating himself he would laugh it off as a joke. We initially thought he was putting it on. We didn't know anything about Alzheimer's or the symptoms that go

with it.

"He was diagnosed with depression and various other illnesses and given medication for these. The disease hit him very hard and progressed very rapidly. He was never formally diagnosed with Alzheimer's until after his death in 2010 and we found out he had a specific gene mutation through research into his family.

"It later turned out his mother also had Alzheimer's and died in her early 50s but this was not something he'd ever spoken about.

"It was after my dad's death that we started noticing my mother's symptoms, it was as if she had given up on herself. But with her the disease progressed more slowly.

"Luckily because we knew the signs we were able to get the diagnosis and put her onto some medication to help with the symptoms. She's now in her 60s and in the later stages of the disease.

"When I found out both my parents had this extremely rare form of Alzheimer's, I was in shock. We had no idea it could happen to Mum too because her dad had previously been diagnosed with multiple sclerosis and not dementia.

"After looking at how gene inheritance works, my siblings and I could determine we had a 75 percent risk of inheriting the faulty gene that would give us the disease.

"Sometimes I feel like the unluckiest person in the world. I've won the world's worst lottery.

"But I also know I am really lucky with the life I have, apart from this dark cloud hanging over me. I try to stay as positive as I can, otherwise

you would drive yourself insane."

Both of Sarah's parents have a mutation in the presenilin 1 gene – one of three known risk genes for familial Alzheimer's.

With any parent there will be two copies of each gene. A child will inherit a copy from each parent. In most dominantly inherited cases, a child has a 50 percent chance as they could inherit the mutated gene, or the healthy gene from their affected parent.

In this case Sarah could inherit the good copies from both parents, leaving her with no risk, the mutated gene from her dad and healthy gene from her mum, the mutated gene from her mum and the healthy gene from her dad, or the mutated gene from both – the last three of the four outcomes leading to early-onset dementia, giving her a 75 percent risk.

If she has inherited the mutated gene from both parents, she has two faulty copies, meaning her children will stand a 100% chance of getting Alzheimer's if this is the case. They will stand a 50 percent of developing the disease if she has one faulty copy, but if Sarah doesn't inherit any faulty copied and is free from the disease, her children will be too.

Inherited forms of Alzheimer's account for less than one percent of all Alzheimer's cases. For the majority it is impossible to know in advance whether a person will go on to develop Alzheimer's or not.

People like Sarah are in the unique position to help scientists hunt for 'biomarkers'- biological signs of a disease that could be detected in those who haven't yet begun to show symptoms.

Biomarkers could allow for earlier diagnosis of Alzheimer's, helping to identify people with the disease before it has caused significant damage

to the brain and at a point when future treatment approaches are likely to be more effective.

Seizing the opportunity to help, Sarah, who supports Alzheimer's Research UK's work, now takes part in an international drug trial for people with familial Alzheimer's called DIAN TU, in the hope that there will one day be a treatment or cure for the disease.

She said:

"I am really hopeful that by the time my children reach adulthood there will be huge leaps in treatments, prevention, and even a cure for Alzheimer's.

"But it does break my heart that they will even have to look into this sort of stuff.

"I do everything for them and they love me so much and obviously I love them very much. The thought of one day not being able to do all I do for them is what keeps me awake at night.

"Luckily my partner is very easy going and laid back and when I am going through my hard days he is always there to listen and reassure me that he will be there for me if and when symptoms begin.

"I hope and pray for a cure for this horrible disease. I have been taking part in the drug trial for 18 months now and it will continue for another two-and-a half years. I believe that there'll be a breakthrough one day soon, and we all just have to keep doing what we can until then."

Tim Parry, Director at Alzheimer's Research UK, said Sarah's story is an extraordinary case.

He said:

"Though we knew a genetic risk such as this was possible, Sarah's story is the first case we have come across of this kind. It's extremely rare, and we feel for the family at what must be a terribly heartbreaking time for them. We can't thank them enough for allowing us to share their story to raise awareness of early-onset and familial Alzheimer's.

"Sarah's story is a perfect challenge to the prevailing misconception that dementia is just forgetfulness in old age. The condition is caused by brain diseases that turn lives and families upside down, and that no one currently survives. Sarah is doing an amazing thing by volunteering to take part in research to help create positive change from her situation, and give hope to future generations.

"The research underway with families that carry these rare inherited forms of Alzheimer's has huge potential to advance the search for new ways to diagnose and treat the disease, improving the lives of everyone affected.

"Alzheimer's Research UK is leading the fightback to bring an end to the fear, harm and heartbreak of dementia through world-class research projects across the UK and beyond."

More information: For more information about familial Alzheimer's disease and genetic risk factors, go to www.alzheimersresearchuk.org/genes

For more support, go to www.raredementiasupport.org

For more information about how to take part in dementia research, go to www.joindementiaresearch.nihr.ac.uk

Provided by Alzheimer's Research UK