PROFILE

Sandra Phair

LIVING WITH CAVERNOMA

"Sandra recognizes the importance of being connected to others that have the same condition"

In 2005 at the age 26, Sandra had her first brain haemorrhage. She was diagnosed with a bleed from a cavernoma in the brain stem. She has the rare form of cavernoma, familial cerebral cavernous malformation (FCCM), with multiple cavernomas throughout the brain, and a small benign meningioma. She has subsequently been diagnosed with the CCM3 gene.

Before her first haemorrhage, Sandra had no symptoms. She was sent home from hospital with no information, just the name 'cavernoma'. Sandra knew that she had a rare disease and had to become the expert in the disease. She was after all living with the disease every day. Sandra now knows more than most of her doctors and nurses about the disease.

Eight months after her first haemorrhage, Sandra had a second one, from the same cavernoma. She was at high risk of further haemorrhages, but surgery was not an option because of its location. Gamma knife radiosurgery was performed. A year later Sandra had brain surgery to remove two more cavernomas and a benign meniginoma in the frontal/temporal lobe.

Sandra is not just living with the risk of further haemorrhage; she is also living with a range of invisible symptoms including chronic pain, migraine, neuro-fatigue and tinnitus. Sandra in fact has an acquired brain injury.

Sandra recognizes the importance of being connected to others that have the same condition. She has been supported by Cavernoma Alliance UK, and together with Kay McGrath, has recently launched the Cavernoma Ireland Support Group.

"Knowing you are not alone, on this path. We stand together, supporting, and searching for a cure."

RARE DISEASE

About Cavernoma

A Cavernoma (also known as cavernous angioma, cavernous haemangioma or cerebral cavernous malformation) is a cluster of abnormal blood vessels found predominantly in the brain and/or spinal cord. It looks like a raspberry, and can measure from a few millimetres to several centimetres. Symptoms, which depend on size and where in the brain or spinal cord the cavernoma occur, include strokes, seizures, partial/full paralysis, sight, speech and/or hearing problems. Cavernoma can bleed at random intervals, which can make symptoms worse, and this is the most feared complication of cavernoma.

Familial cerebral cavernous malformation (FCCM) has an estimated prevalence of 1/5,000 -1/10,000. Cavernoma causing symptoms are more rare (1/12,500). A study in Scotland found that each year 1 person out of 400,000 is diagnosed with a symptomatic brain cavernoma.

FCCM has a genetic origin, with one of three genes implicated (CCM1, CCM2 and CCM3). FCCM is transmitted from parent to child with a 50% risk of inheriting the mutated gene. In others the cavernoma is sporadic. The CCM3 gene is the rarest of the genetic mutations that cause cavernomas. There are only 30-50 people in Europe who are diagnosed with the CCM3 gene and an equivalent number in the United States.