9th World Congress on

Rare Diseases and Orphan Drugs

June 17-18, 2019 | Berlin, Germany

Wyburn-Mason syndrome. Ever heard of it?

Dan Jeffries
Patient, Author and Advocate, Bristol, UK

Dan Jeffries has, and his insightful and entertaining talk explores what it's like living with one of the world's rarest medical conditions - and then finding out you have another one. Dan was diagnosed with Wyburn-Mason syndrome at four-years old. It is an exceptionally rare AVM (Artereovemous Malformation), situated around the midbrain and optic nerve. As a consequence, Dan is blind in his left eye. The AVM is untreatable and there are thought to be less than 100 reported cases in the past fifty years — worldwide. That works out to be about 1 in 70 million people. Pretty rare. And then, as he turned 30, Dan discovered that he had Acromegaly — a benign tumor in the pituitary gland. About 6 people per million are diagnosed with this unique and challenging condition that effects growth homrone and IGF-1 production, as well as impacting on vision, facial appearance, hands and feet, teeth, libido and more. Being Rare explores the early days of diagnosis, living with Wyburn-Mason syndrome and how things started to change during his twenties, leading to the bizarre discovery of Acromegaly — discovered by student doctors. Using an innovative slideshow, Dan is able to show key images, doctors notes and more, reflecting the methods used to tell his story in his published memoir, Me, Myself & Eye. Dan also explores the impact of living with rare conditions and how this can affect the individual on a day-to-day basis, as well as the empowerment it can bring too. "Dan's talk managed to be informative and entertaining, insightful and moving, educational and inspiring. You don't often find presentations that manage to combine all of those qualities! I left with a deeper understanding of what it is like to live with rare conditions, and also with a smile on my face!" - Pfizer UK.

dan@memyselfandeye.co.uk