PROFILE

Áine Lawlor

LIVING WITH 22Q11.2 DELETION SYNDROME

"Everyone was ahead in [school] work and I was always behind and nobody knew why"

Áine is 36 years of age. From the time she was very young she felt different to other children. She was frequently ill and struggled to learn in school where she was bullied and unhappy and felt like a failure. 'Everyone was ahead in work and I was always behind and nobody knew why,' she remembers.

Áine was finally diagnosed aged 15 with 22q11.2 Deletion Syndrome. The diagnosis 'explained a lot,' says Áine,

'My life changed. People started to realise I was a bit different,' she continued. 'I'm not shy anymore. I love my life even though I have 22q. It can get hard but I truly believe in myself, that's how I carry on through the days.

Áine has a Fetac Level 5 qualification in Childcare and has a part time job in a creche. She is a ten-pin bowler with Special Olympics.

These outlets keep her going. 'It's not all that bad having 22q,' she reports. A 22qYEEP member (Young Experts by Experience Panel) Áine takes part in participatory action research and speaks to various audiences about her 22q lived experience.

Áine is quick to acknowledge that she has help carrying on,

'My mother is my strength. She helps me to be strong. Without her I don't know where I'd be'

Áine's mother (Anne) co-founded www.22q11ireland.org which provides family peer support, information and regular family outings along with an annual conference. The group works in partnership with clinicians to develop integrated care pathways.

RARE DISEASE

About 22q11.2 Deletion Syndrome

22q11.2 Deletion Syndrome (DS) is caused by the deletion of a small piece of chromosome 22. The condition affects an estimated 1 in 2- 4,000 people worldwide. The features of this syndrome vary widely, even among affected members of the same family, and involve many parts of the body. Characteristic signs and symptoms include heart defects that are often present from birth, palate defects resulting in feeding, speech and language difficulties and mild differences in facial features. People with 22q11.2 DS often experience recurrent infections caused by problems with the immune system.

Aside from the varied physical health issues, many children with 22q11.2 DS have developmental delays and learning disabilities. Later in life, they are at an increased risk of developing mental illnesses such as schizophrenia, depression, anxiety, and bipolar disorder. Additionally, affected children are more likely than children without 22q11.2 to have attention deficit hyperactivity disorder (ADHD) and developmental disorders, such as autism, that affect communication and social interaction. Children with this disorder have complex care needs which require an appropriate care response tailored to each child's individual needs.