



Alanna

LIVING WITH KOOLEN DE VRIES SYNDROME (KDVS)

Her intellectual disability and her anxiety are her biggest struggle as she's aged.

“Bringing her to her potential and shaping her to be the amazing young woman she is today is my proudest achievement.”

Alanna was born in October 2000. After an uncomplicated pregnancy it was one complication after another from the time she was born. Despite being full term she only weighed 5lb 4oz. She had no suck reflex which meant she had to spend time in the special care baby unit being tube fed.

After bringing Alanna home feeding remained a huge issue for her. She could only tolerate very small feeds and as a result her weight gain was very slow. On her first birthday she weighed only 13 pounds. She failed to meet any of her developmental milestones and was referred to our local early intervention team when she was 2. She had delayed gross and fine motor skills, significant speech and language delays, hypermobile joints, low muscle tone, sleep disorder, sensory processing disorder, the list goes on...

When Alanna started school we discovered the extent of her learning disability, after having a NEPS assessment we discovered she has moderate intellectual disability and although she was attending mainstream school, the curriculum would be very difficult for her. She thrived in mainstream school, whilst

following her own individual education programs; she gained invaluable social and communication skills which she proudly carries throughout her life. She completed her 8 years in mainstream primary school before attending a special needs school until she turned 18. She traveled an hour to and from school every day but she loved it and received all the independence and life skills she would need to reach her potential.

In April 2011 when Alanna was almost 11 she was diagnosed with Koolen de Vries syndrome (KDVS). This is a small deletion on the long arm of chromosome 17 (17q21.31). It occurs in approximately 1 in 55,000 people but is extremely under diagnosed in Ireland with only about 7 known cases. Alanna was the second child in Ireland diagnosed with KDVS. After years of testing we were relieved to finally have an answer for Alanna's difficulties but we were very unprepared for the loneliness and isolation associated with a rare diagnosis. Over the next few years I made several trips with Alanna to the UK where we met other KDVS families. It was very disheartening to witness the lack of support in Ireland which eventually prompted the establishment of Rare Ireland

Family Support Network in 2017 which gives the much needed support to families dealing with a rare diagnosis, easing the isolation and giving parents of rare children a place where they belong.

Alanna is now 21, she attends a local day service for adults with intellectual disability. She struggles to do daily tasks most of us take for granted like telling time, counting money and tying her shoes. She requires the same level of care as a child aged 7 or 8. She needs round the clock supervision. Although she is 21 she can't be left home alone or go out and about by herself.

Her intellectual disability and her anxiety are her biggest struggle as she's aged. Her low muscle tone and hyper mobile joints have remained an issue for her. She tires easily when walking and has had recurring knee dislocations, particularly during growth spurts.

Getting the services Alanna needed to live a fulfilled life has been difficult at times. We have had to fight for services and supports that no parent should ever have to fight for, particularly with the mental health service to get her the help she needed to cope with her anxiety. I'm glad we knew what to do and how to fight the many systems that put obstacles in our way. Sadly this is still the reality for many rare families.

Despite all she has gone through Alanna is a very happy outgoing young lady, she loves music, singing, watching tv and getting together with her friends. She loves being the life and soul of the party.

Alanna is very skilled at computers and has recently completed a level 3 computer skills course and is preparing to move on to level 4. She has exceeded our expectations for her. Bringing her to her potential and shaping her to be the amazing young woman she is today is my proudest achievement.

Alanna has taught us and others around her more than we could ever teach her. She has the most beautiful personality and infectious sense of humour and lets nothing stop her. She is a "ray of sunshine" who is loved and remembered by everyone who meets her.