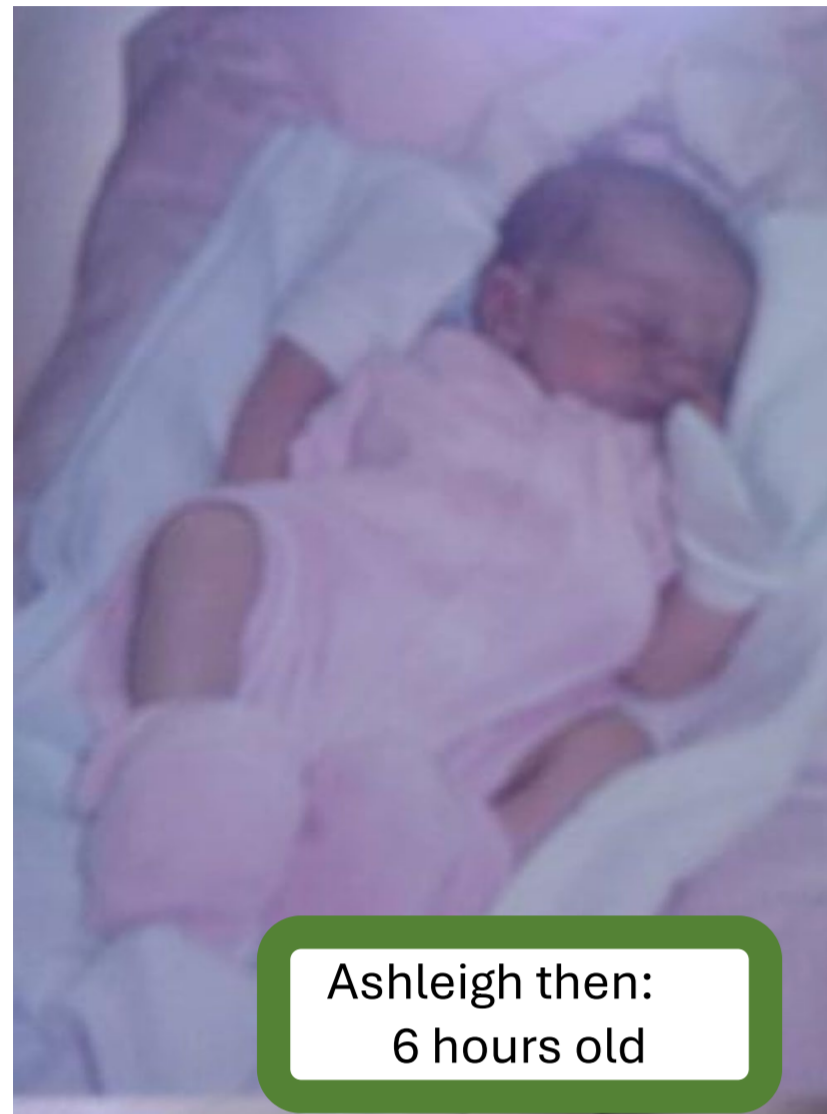


Ashleigh Kelly



Ashleigh then:
6 hours old

GRIA2 diagnosis

We didn't find out until Ashleigh was 11 years old that she had GRIA2, but it wasn't until she was 13 years old that the condition was formally diagnosed after the condition was first published. I remember that day so well! I thought all our prayers were answered! Little did I know how little was known about the condition that back then. It is only through the hard work of Curegrin and researchers at UCL and in America and Europe that we know as much as we do today! You guys are amazing!

Ashleigh (patient 2) was part of the initial study by Vincenzo Salpietro and his team at UCL. So I guess you could say she made history!

Vincenzo put me in touch with another family in America and we have kept in contact since, swapping photos and videos of our kids! We found it fascinating how similar our kids were, it was amazing to finally be in touch with people that lived similar lives to us.

With the help of the internet I came across Curegrin and Griconnect, finally a place where we could be in touch with families, researchers and clinicians all under one roof so to speak. I've made some fantastic friendships with other families like ours which is amazing!



Ashleigh now:
Age 17

Ashleigh's **main symptoms** are:

- Challenging behaviour
- Non verbal/ communication difficulties
- Sensory Proprioceptive disorder
- CVI
- Dysphagia
- Dysautonomia with dystonia (PSH)
- Sleep problems
- Double incontinent

Ashleigh's behaviour is the most debilitating part of the condition. It holds her back from being able to do every day things and interrupts her learning process.

She fluctuates from happy to crying or hyperactive at the flick of a switch. She can spend hours a day screaming (anything up to 6 hours) and then suddenly stops and then becomes extremely heightened. She can push, hit, kick, bite and try to break furniture. Transitions is a trigger, the car stopping can be a trigger but quite often there is no obvious external trigger.

Because of Ashleigh's **impulsive behaviour** she can only go out in public if she has 2 people with her to keep her safe. As you can imagine this has a huge impact on our family.



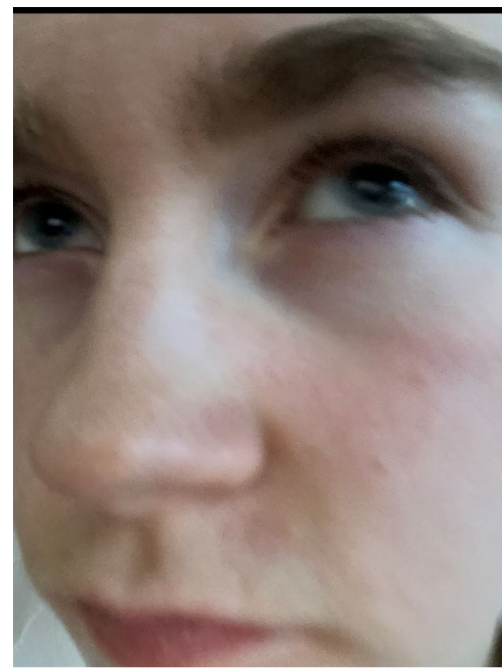
Aside from Ashleigh's issues **she is a very loving young lady.**

She can feed herself with a spoon. She is able to walk, run and manage steps independently. She can kick and catch a ball. She enjoys walks and listening to drum and bass music.

Although Ashleigh is classed as non verbal she can say Mum, Mummy and Momma and "I want it". Her understanding of spoken language is really good. She communicates mainly through body language, but is starting to use photos and pictures (pecs) to make choices, she also has a communication app called proloquo2go but hasn't seemed too interested in using that independently yet.

Ashleigh also has a fantastic sense of humour!





Part of her behaviour problems is dysautonomia with dystonia in the form of **neurostorms** (paroxysmal sympathetic hyperactivity) and although she takes medication for neurostorms it seems to have become less effective to when she started taking it.



Ashleigh Kelly



Last summer I took Ashleigh on her first ever weekend away. We went to a place in Noth Devon called **The Calvert Trust**, where they have a team of people trained to help people with additional needs access outdoor activities.

She completely blew me away taking part in every activity available to her including abseiling and the zip wire (twice).

She even waited patiently while putting on all the equipment on and again waiting patiently for her turn (waiting isn't one of her strong points).

This proves anything is possible!

As the storm unfolds you can see her skin become very red and blotchy and the blotchy parts feel hot to touch, her pupils dilate, her heart rate increases and she starts to scream. In this state she become very stiff and can have abnormal posture (dystonia).

There is absolutely nothing I can do to help her only make sure she is safe and reassure her that I'm there when she needs me, if I get to close to her while she is in this state she can become very aggressive scratching, grabbing, pulling hair and trying to bite.

She also has unusual/repetitive movements.

Current medication

- Carbamazepine
- Clonidine
- Gabapentin
- Melatonin
- Lactulose
- Vitamin D

Medication tried previously

- Risperidone: raised prolactin
- Fluoxetine: didn't help
- Aripiprazole: restless legs and made her more irritable
- Diazepam: opposite effect, made her worse

