

A day in the Life...

...of a Mum of a Child with CDKL5 by Teresa Newey

Ellie is 17 and has a genetic condition known as a CDKL5 disorder. She was diagnosed just before her 14th birthday. CDKL5 was only discovered in 2004 and consequently not a lot is known about it. I feel we are on an unknown path as we are not sure of the progression of the condition.



CDKL5 is a protein that is important in brain development and function. It affects mostly girls and most children affected have early onset, difficult to control seizures, and profound and complex learning difficulties. Most have cortical visual impairment, gastrointestinal problems, are unable to walk or talk, have some autistic tendencies and repetitive behaviours. I was asked to write about a "typical" day, but we don't have one as such and we haven't, as yet, been able to establish why Ellie's moods can be so erratic. My guess is that it has a lot to do with the types of epilepsy that CDKL5 produces, gastrointestinal problems and sleep (or lack of it), but they are all possibly interconnected.

So here we go, I will start with the positives!

Oh my, Ellie on a good day sparkles. She is relaxed, her eyes are bright, she smiles continually and when she laughs, people stop in their tracks to smile or laugh along with her. She is responsive, reactive and wants to sit on my knee and hug! A little comical since she is as big as me now! But I love these days, despite my aching back.

The manic days occur when Ellie is not sleeping for more than a couple of hours at night. Outrageous giggles, rocks in her chair and bounces on her bed. These phases are pretty exhausting. Ellie has been known to literally move her bed across the floor! Luckily the bed is "Tough" and has survived several years of Ellie crashing against the cot side, but I am so worried that one day it will fail hence, I don't sleep much during these phases.

We can then progress to the sleepy day when Ellie is very difficult to rouse. When she does wake up, she is drowsy, pale, irritable and slumps in her chair.

She can't hold her head up and is unable to be involved in very much at all.

The difficult day produces an Ellie who screams or shouts loudly to protest against any interaction and who becomes very self absorbed. She "looks" cross and can certainly make her feelings known.

Then there are the sad days and I think these may be the most difficult. Silent tears and inconsolable sobbing. They don't occur often, but when they do, they are real heartbreakers and there's no light in her eyes.

And last but not least, the poo days! Ellie has had previous operations for reflux (she loves eating!) but has been left with "slow motility". She has a tendency to regurgitate and something, referred to in the CDKL5 world as "elephant poos"! These occur when Ellie becomes constipated, something we try to avoid. But the difficulties arise when Ellie needs changing 6 or 7 times in a day, emptying several days of her GI contents (polite way of putting it!) . This is obviously stressful and limits what we can do due to lack of suitable changing facilities with hoisting equipment. So some days we just have to stay at home. if you want to read more please visit our website www.supporting-cdkl5.co.uk.

A positive in all this is that we have a new GP who has an holistic approach to her needs. She has recently had a full range of blood tests so we have a starting point to work from and has referred her to various specialists who are involved with transition from paediatric care to adult care.

So..... Keep facing the sun and the shadows of life will always fall behind you!!

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