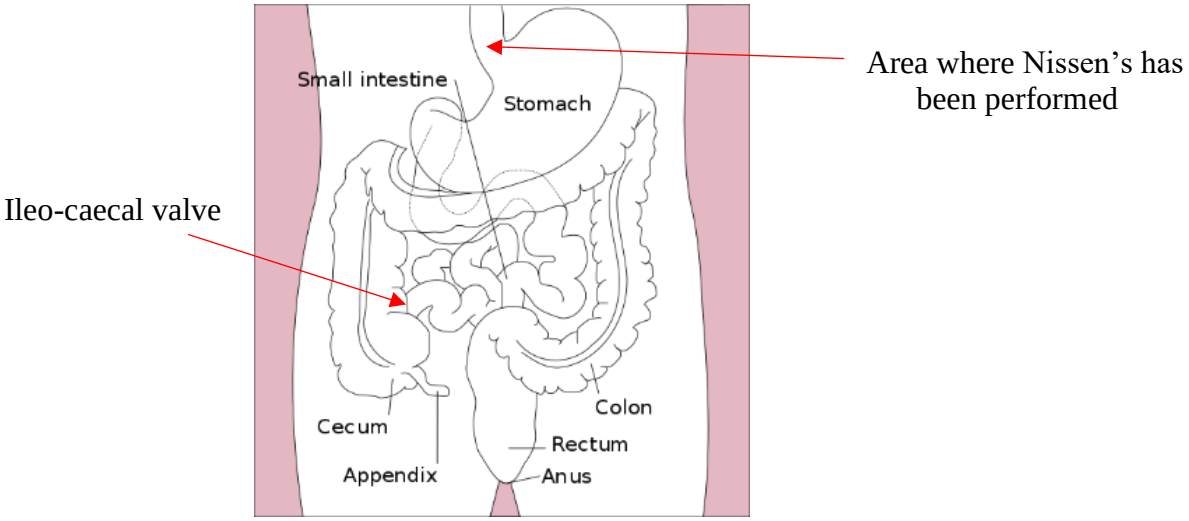
Supporting CDKL5

On the 10th March 2020, Ellie had an acute medical episode which could have had tragic consequences. Before describing what happened to Ellie, I want to give 3 pieces of background information which will help in understanding the whole episode.

Firstly, Ellie had a Nissen Fundoplication in 2005 which needed revising in 2006. Following this she was able to eat most things. Occasionally food and secretions will pool in her lower oesophagus which obviously makes her uncomfortable. She has learnt to put her fingers down the back of her throat which makes her retch. This brings her oesophageal contents back up and eases her discomfort.

Secondly, just like many individuals with CDKL5 and probably many others who have severe learning disabilities, Ellie has slow gut motility. This affects her eating but perhaps more importantly, leaves her vulnerable to constipation. Despite our best endeavors, Ellie still ends up with a loaded colon for which she needs assistance in emptying. Whether her colon gets fully emptied is a question we don't know the answer to but probably not.

Thirdly, some anatomy, the junction between the small bowel (the ileum) and large bowel (colon) is defined by something called the ileo-caecal valve. This valve limits the flow of contents of the colon back into the ileum. Previous CT scans have shown that Ellie's small bowel is slightly dilated with gas suggesting that her ileo-caecal valve is slightly incompetent.

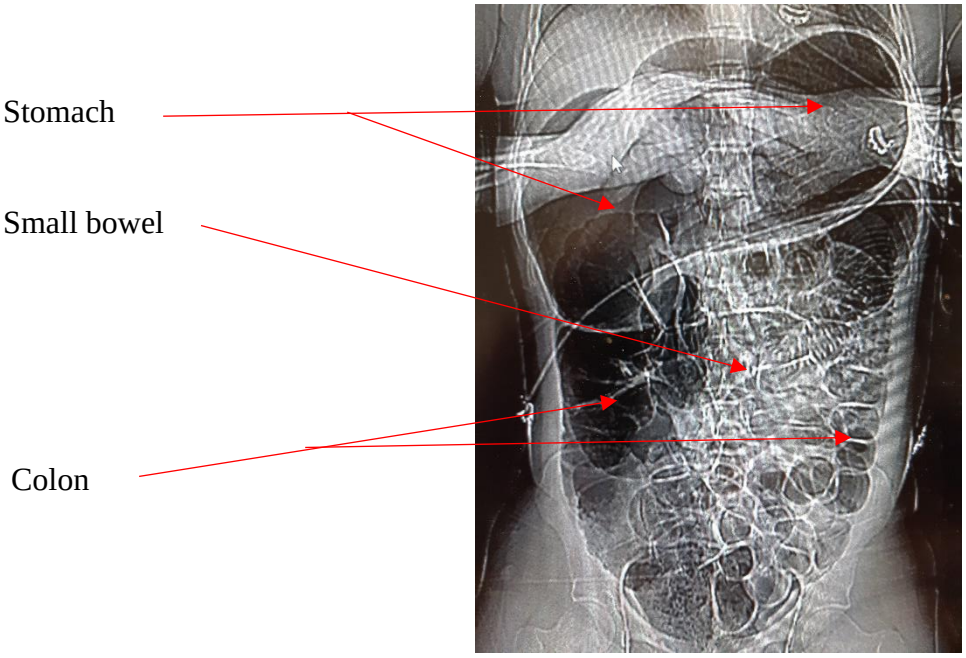


(from Wikipedia)

So, on the 10th March Ellie had had a relatively normal day. She had eaten well. She had had her bowels open the previous day. About 2 hours after her evening meal she began to retch and put her fingers down her throat. However, it was clear that this was not the usual event. Her retching became more forced without her putting her fingers down her throat. It appeared that she was trying to relieve the cause of discomfort she seemed to be experiencing.

At this stage, her tummy was soft and not tender, and we therefore wondered whether she might have something like norovirus. She had spent the previous weekend at a short-break facility and so we phoned them to see if they had heard of any of the others being ill, which they had not. However, within 20 to 30 minutes, Ellie's tummy had started to swell and was slightly tender, and she was starting to become quite agitated. Within another 30 minutes or so her abdomen had become very swollen and tense and she was starting to struggle with her breathing. She also had a 30 second tonic seizure which is something we hadn't seen for many years.

Appreciating that something serious was happening we took her to our nearest A&E department. When we arrived, Ellie looked very ill. She was grey and clammy, breathing fast with short grunting breaths and her pulse was racing. We also noticed that her lower body and legs were looking blue. She was assessed relatively quickly and had a CT scan of her abdomen. This showed that she had an acute dilatation of her stomach. Her small bowel and colon were also dilated, and she was constipated. There was no obvious sign of any type of mechanical obstruction that was causing this.



In order to treat her she urgently needed to have her stomach decompressed by inserting a naso-gastric tube. Unfortunately, this proved impossible to do as the ng tube would not pass through her Nissen's. She was therefore taken to the operating theatre where this was achieved under direct endoscopic vision. She also underwent a colonoscopy to clear her left colon.

Following this, Ellie improved although her progress has been very slow. She began to drink and eat only very small amounts at first. It was perhaps nearly 2 weeks before she was drinking adequately although at the time of writing this her food intake remains poor.

So, we have tried to make sense of all this and understand what actually happened. The alarming thing was how quickly Ellie deteriorated from having a soft, non-tender abdomen, to an abdomen that was tensely distended and the physiological distress that this produced – a time period of less than 2 hours.

There was no feature on her CT scan to suggest a mechanical type of bowel obstruction – such as a volvulus or adhesions from her previous surgery. So, although we cannot be certain, the view is that Ellie had probably developed a functional obstruction in relation to her constipation and slow gut motility. As a result, her colon started to distend. If this had continued unabated then there would have been a risk of perforation – typically of the caecum. However, because Ellie has a slightly incompetent ileo-caecal valve, the colon was able to decompress retrogradely into her small intestine which in turn decompressed into her stomach. This is when the problem really started because normally in this situation an individual would start belching and vomiting as their stomach filled up. Ellie, of course couldn't do either because of her Nissen's. Gas and fluid became trapped in her stomach which became more distended. The pressure in her abdomen built up so much that it interfered with her diaphragm affecting her breathing, and also put pressure on her major blood vessels. The pressure would have compressed her inferior vena cava bringing blood back from her legs to her heart and this is why her lower body and legs were going blue – due to venous congestion. The pressure would also have started to compress the blood supply to her stomach and intestines, which can result in serious damage to the GI tract. This situation evolved fairly rapidly and is called abdominal compartment syndrome. This is a serious condition requiring urgent treatment.

A great deal of emphasis is placed on seizure activity in CDKL5, but like us, many will be aware and familiar with the GI issues that occur – both in terms of feeding and in relation to constipation. At the time of writing this, Ellie is nowhere near back to normal and the future in terms of her GI function remains uncertain. This episode reinforces the fact that we do have to remain vigilant to all the health issues that CDKL5 throws at us, and to be prepared to act promptly when something tells us that things aren't right.

Happy Ellie on holiday in 2019

Martyn Newey
CDKL5 Dad
www.supporting-cdkl5.co.uk