

Supporting CDKL5



Proximal large bowel volvulus in children: 6 new cases and review of the literature. Journal of Pediatric Surgery 2012.

This is a review from 3 centres in the UK, Ireland and Hungary of an extremely rare condition that affects the bowel. A volvulus is a condition where the bowel twists on its mesentery (a fold of tissue that attaches it to the wall of the abdomen). This can then cause obstruction of the bowel and can even result in irreversible damage to the blood supply of the bowel. If diagnosed early enough it is possible to treat the condition without surgery although most patients do require some sort of surgical procedure. The authors review 36 cases that have previously been reported and an additional 6 cases of their own. Males and females were equally affected but 29 (69%) were described as having neurodevelopmental delay. The mean age of the group was 10 years (range 0 to 18 years). The presenting symptoms were colicky abdominal distension (100%), abdominal pain (98%), vomiting (83%) and constipation (69%). All but one of the patients with constipation had neurodevelopmental delay. All 42 patients required surgery and there were 6 (14%) deaths. The authors also noted that neurodevelopmental delay and severe chronic constipation were the commonest associated disorders. They suggest that in mentally impaired children, insufficiently treated chronic constipation results in a heavy and extremely dilated bowel that stretches the bowel mesentery and allows a volvulus to occur. They also point out that although children with a volvulus present with pain, distension and vomiting, these symptoms also frequently occur in children with neurodevelopmental delay and constipation without volvulus, and should respond to enemas and laxatives. The rarity of volvulus and the inability of a child with neurodevelopmental delay to adequately communicate their symptoms, may explain why there may be a delay in establishing the diagnosis.

Note - The first thing to say is that this is an extremely rare condition in children. The authors found only 36 reported cases in the English literature from 1965 to 2010. Although any child can be affected by this condition, it would appear that children with neurodevelopmental delay are more prone, presumably due at least in part to problems with communication - an issue which I am sure we all recognise. They do not, however, specify the nature of the neurodevelopmental delay present in the children of this study. Many of us will be only too familiar with the problems around bowel function and constipation, and must realise that it is very important that parents and carers keep a close eye on this issue, and in my view, do not delay in involving your family doctor / specialist physician if you have a concern. Also - see article below regarding GI disorders....