GASTROINTESTINAL PROBLEMS IN CHILDREN WITH A CDKL5 DISORDER: A PARENT-LED SURVEY

CDKL5 UK

Giving hope through research & support to those living with a CDKL5 Disorder

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INTRODUCTION

The CDKL5 disorder is a rare X-linked genetic condition first described in 2004. There are thought to be at least 500 - 600 individuals in the world with a known diagnosis. Because of the relative lack of information about CDKL5, a family support group was created on Facebook in 2010 in order to provide information and support to affected families. Through social networking, similarly affected families have come together, compared stories and generally learned more about the clinical aspects of CDKL5 disorder than the established medical profession might imagine.

Gastrointestinal (GI) problems are recognised in children with neurodevelopmental delay, and these are particular concerns that families of children with a CDKL5 disorder have raised. Their concerns relate both to upper GI problems of reflux and to lower GI problems of bloating and constipation, and there is a view amongst affected families that these particular issues do not receive sufficient attention from the medical profession. CDKL5 UK is new charity that provides support for affected families in the UK as well as raising funds for research. We wanted to highlight these concerns

CASE STUDIES

The first child, a female, was diagnosed with CDKL5 at about 18 months of age, having had seizures since she was 7 weeks old. She had a gastric-tube inserted at 2 ½ years of age because she lost her ability to suck and failed to thrive. She was on the ketogenic diet for her epilepsy. She always suffered with constipation, both in terms of frequency of and difficulty in passing bowel movement. She would have her bowels open usually only once every 3 to 4 days. She was treated with Miralax and her mum used to rub her abdomen. Ten days prior to her death, her mum noticed that her feeds were not flowing freely through her g-tube and indeed, it proved quite difficult at times to push them through at all. She developed bile stained vomiting, became more lethargic and developed an intermittent temperature. She then developed a very high temperature and was admitted to hospital with a presumptive diagnosis of pneumonia. On the prompting of her parents, attention was drawn to her abdomen where a subsequent diagnosis of intussusception was established through ultrasound examination. She died within 24 hours of admission while being stabilised for surgery. She was 4 years of age.

The second child was a young adult female with CDKL5. She had her first seizure at the age of 4 weeks. She had been able to walk with assistance and was not known to have any other significant medical problems. She had been on Omeprazole since about the age of 7 for reflux but could eat normally and had not undergone any related surgery. Although her bowel function appeared normal, she often seemed to be experiencing abdominal pain. She was an air-swallower and her abdomen would bloat. Although her parents were concerned about this, they found it difficult, in their own words "to get through to the doctor". Just 3 months before her death, she was started on movical to see if this would help her pain. On the day she died she had seemed well. Just prior to becoming unwell she had eaten a meal and had also had her bowels open. She then became ill and vomited once. Although she developed problems with her breathing, it was felt that her abdomen was the cause of her illness. She deteriorated rapidly and eventually had a cardiac arrest from which she did not recover. The cause of death was a volvulus. She was 19 years of age.

SURVEY RESULTS

Of about 180 families with affected children, 90 responded to our survey. Reviewing these children, there were 80 females and 10 males with an average age of 6.4 years (range 6 months to 19 years). GI problems were reported in 87% of these children, and these are summarised in table 1. Of those 44 children who were currently on medication to assist bowel function, there were 8 who still only opened their bowels less than 3 times a week and were therefore still suffering with constipation. Of those parents responding, 34 (39%) felt that they were not given adequate help from their treating physician with their child's constipation.

CONDITION / TREATMENT	NUMBER
Gastro-oesophageal reflux	68
Requires PEG feeding	23
Has had a Nissen's fundoplication	18
Constipation (bowels open < 3 times / week)	14
Medication to assist bowel function	44
Hospilalised because of bowel function	14

DISCUSSION

GI problems in children with neurodevelopmental delay are clearly reported in the literature (1-4) and in particular, the small but serious risk of lower GI problems (5, 6). Constipation itself is a common symptom in children in general and has been reported as constituting 25% of visits to gastroenterology clinics (7). We have found that parents of children who have a CDKL5 disorder continue to struggle on a day to day basis to manage related problems. There is a view amongst a significant number of parents that their concern in relation to their child's health - particularly in relation to bowel function - is not being given the attention it needs by their supervising clinician.

An issue that has arisen recently is in relation to the role (or lack) of a supervising clinician with a responsibility for the overall wellbeing of the child. Many children with a CDKL5 disorder experience significant problems with seizures and seizure control. The consequence of this is that most are primarily under the care of a neurologist. However, many of these children also have other medical issues whose management may not be the focus of attention of the neurologist, and which may lie outside the realm of a general (family) practitioner. GI problems are just one such issue and our concern is that while an emphasis is being placed on managing seizures other concomitant conditions are being overlooked to the potential detriment of the child. Our wish, therefore, is that children with a CDKL5 disorder be treated not just as sufferers of epilepsy, but managed as a whole person by a general clinician / physician, with the background and experience of the many general functional difficulties that such children will face. As the number of individuals with a rare genetic condition continues to rise, it is time that health planning recognised the issues and challenges that these individuals present, and provided the necessary resource to meet their needs and to maximise the quality of life they deserve.

www.cdkl5uk.org

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