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A NEW PEDIATRIC INTESTINAL PSEUDO-OBSTRUCTION SYNDROME BY SGOL1 MUTATION: A LATE-ONSET BUT SEVERE DIGESTIVE PHENOTYPE.

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Background: Pediatric intestinal pseudo-obstruction (PIPO) is a rare and severe gut motility disorder associated with a high morbidity and a high mortality rate. The majority of PIPO are of primary origin. Prenatal symptoms can be detected in about 20 % of children and the symptoms start by one year of age in 80%. Up to 60 - 80% of patients require parenteral nutrition. In 2014, Chetaille et al. described a new cohesinopathy affecting gut and heart rhythm by a SGOL1 mutation (Nat Gen 2014;46(11):1245-9).
Aims: We report here 4 cases of PIPO with SGOL1 mutation with an extensive description of their digestive phenotype and nutritional long term follow-up.
Methods: All data were retrospectively collected from patients' files. The ethical committees at all participating institutions approved the study, and all participants gave informed consent.
Results: The median (range) age at the end of follow-up was 18 years (14 - 19). All the patients presented initially with the classical PIPO symptoms with vomiting, abdominal distension and abdominal pain at the age of 4.5 years (2.7 - 8). Contrast studies showed massively distended small bowel and colon in the 4 patients. Antroduodenal manometry was performed in 3 patients and was abnormal in all with a neuropathic pattern and lack of response to IV erythromycin. Parenteral nutrition (PN) was started at 13 years (6.5 - 14) due to failure to thrive and intestinal failure. At initiation of PN, the median PN energy intake represented 85.5 % (53 - 100) of the resting energy expenditure and the median weight loss was of 15 percentiles (9 - 47) and of 24 percentiles (16 - 77) for height. All patients but one were on PN at the end of the follow-up. Two patients had an ileostomy at 6 years and a gastrostomy at 5 and 11 years respectively. Three patients required a pacemaker for sinusal dysfunction at the time of PIPO diagnosis.
Conclusions: Cohesinopathy with SGOL1 mutation represent a late-onset but severe PIPO etiology associated with severe bradycardia.

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