

**138****GASTROINTESTINAL MANIFESTATIONS IN PATIENTS WITH MUCOPOLYSACCHARIDOSES**

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**Objective:** To assess gastrointestinal manifestations in patients with MPS and bowel mucosa histology in a MPS I mouse model. **Methods:** Cross-sectional study with a convenience sampling strategy, including patients with a diagnosis of MPS of any type and regardless enzyme replacement therapy (ERT) status. Patients were assessed by means of a dietary record and an interview focused on gastrointestinal symptoms, and also by a set laboratory tests. Patient's DNA samples were also tested for primary lactase hypolactasia, and bowel mucosa specimens from MPS I mice underwent histological examination. **Results:** A total of 27 patients were included, with a median age of 12 (1-28) years. The most prevalent gastrointestinal symptoms were flatulence, abdominal distension, abdominal pain, and loose stools. A significant difference in the prevalence of flatulence was observed among different MPS types ( $p=0.004$ ). The prevalence of flatulence and abdominal distension was significantly higher in the non-ERT group than in the ERT group ( $p = 0.04$  and  $0.03$  respectively). Most biochemical tests performed to work up and/or rule out specific conditions were within normal limits. Histological analysis of small-bowel tissue from MPS I mice found increased cell volume indicative of some form of intracellular accumulation. On molecular testing for lactase deficiency, 58.8% of the patients had the CC genotype, which is consistent with lactose intolerance. **Conclusion:** Our results suggest that gastrointestinal manifestation, are frequent across most MPS type, and ERT plays a role in treating them. The findings in the bowel histology in analysis in the in the MPS mouse model are consistent with cellular abnormalities contributing to these manifestations. Further studies focusing on the gastrointestinal manifestations of MPS are warranted to corroborate our findings and provide a better understanding of the pathophysiological mechanisms associated with these symptoms in affected patients. **Palavra-chave:** Mucopolysaccharidoses; gastrointestinal manifestations; enzyme replacement therapy. Projeto 12-0259