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Abstract :

BMPR1A Specific Juvenile Polyposis Syndrome : A Case Series. Juvenile polyposis syndrome (JPS) is a heterogenous condition both phenotypically and genotypically, that leads to increased risk of gastrointestinal cancers and other extraintestinal manifestations. The risks of these consequences of the condition depend on the specific genetic mutation leading to the syndrome in each individual. The two major genes where mutations lead to JPS are *SMAD4* and *BMPR1A*. The majority of these cases that are well documented in literature, including the screening guidelines for colon cancer, are primarily related to *SMAD4* mutations. There has been a call for increased investigation into *BMPR1A* mutations and the potential differences in condition compression compared to *SMAD4* mutations. This case series documents four cases of pediatric patients with diagnosed JPS and have *BMPR1A* mutations. Through a deep chart review of each patient, identifying their initial symptoms, family history of polyps and other conditions, their endoscopic screening procedures to date, and their current state of health, we were able to identify a major trend for *BMPR1A* mutation JPS patients. We found that all four patients presented with symptoms secondary to their JPS and polyps identified on colonoscopy prior to the recommended screening initiation by current JPS guidelines. This further supports the fact that JPS is a heterogeneous condition with multiple genetic loci that can be the underlying cause. It also supports that *BMPR1A* mutation-possessing patients may require specific screening guidelines because of their specific phenotypes.