

Juvenile polyposis syndrome (JPS) is a rare hereditary polyposis syndrome that causes juvenile polyposis and increased risk of gastrointestinal malignancies. A diagnosis of JPS is established either by expert opinion clinical criteria and/or the presence of a *SMAD4* or *BMPR1A* mutation. Relatively limited data exist about those who present during childhood/adolescents with a clinical diagnosis of JPS, no family history, and no mutation in *SMAD4* or *BMPR1A*. It is unclear if these patients have the same cancer risks as those with *SMAD4* or *BMPR1A* mutations, which may lead to inappropriate cancer screening recommendations.

The primary aim of this study is to describe polyp history and cancer risk for patients who present as children or adolescents with at least 5 colorectal juvenile polyps, with no JPS family history or *SMAD4* or *BMPR1A* mutation. These cases will be compared to patients with one juvenile polyp in the upper and one in the lower gastrointestinal tract; any number of juvenile polyps and a family history of juvenile polyposis syndrome; and/or whom have a germline mutation in *SMAD4* or *BMPR1A*. Our **hypothesis** is that there will be differences in measured data points between children with at least 5 colorectal juvenile polyps as compared to those with mutation in *SMAD4* or *BMPR1A* and/or those meeting the above mentioned clinical criteria. Our **aim** is to determine the optimal cut point in polyp number for the diagnosis of JPS that distinguishes cancer risk.

We are inviting clinicians and researchers around the world to collaborate on this study. If you are interested in collaborating on this study, please contact Brandie Leach, LGC at leachb@ccf.org or 216-444-8114.