

## Video Case: Juvenile Polyposis Syndrome in 11- Years Old Girl

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A 11-years old Egyptian girl presented by bleeding per rectum. On endoscopic examination, multiple sigmoid colon polyps(>5) with mucosal prolapse were seen. Polypectomy was done in multiple sessions ( this video shows the first session) . Microscopic examination confirmed the diagnosis of Juvenile Polyposis . Juvenile polyposis (JP) is an autosomal dominant hamartomatous polyposis syndrome where affected individuals are predisposed to colorectal and upper gastrointestinal cancer. Forty-five percent of JP patients have mutations or deletions involving the coding regions of SMAD4 and BMPR1A[1]. Juvenile polyps have a distinctive histology characterized by an abundance of edematous lamina propria with

inflammatory cells and cystically dilated glands lined by cuboidal to columnar epithelium with reactive changes. Clinically, juvenile polyposis syndrome is defined by the presence of 5 or more juvenile polyps in the colorectum, juvenile polyps throughout the gastrointestinal tract or any number of juvenile polyps and a positive family history of juvenile polyposis[2].

### References:

1. Calva-Cerqueira D, Dahdaleh FS, Woodfield G, Chinnathambi S, Nagy PL, Larsen-Haidle J et al. Discovery of the BMPR1A promoter and germline mutations that cause juvenile polyposis. *Hum Mol Genet.* 2010 Dec 1;19(23):4654-62.
2. Brosens LA, Langeveld D, van Hattem WA, Giardiello FM, Offerhaus GJ. Juvenile polyposis syndrome. *World J Gastroenterol.* 2011 Nov 28;17(44):4839-44.