

Disease

SMAD4 associated juvenile polyposis syndrome¹

Gene

SMAD4

Inheritance	AD
Current Treatments	colonoscopy and endoscopy with polypectomy
Evidence for Treatment	guideline ²
Non-Molecular Testing Availability	no
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC066
Disease Frequency (per 100,000)	3.625 ¹
Genes Included in Disease Frequency	BMPR1A, SMAD4
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Juvenile+Polyposis&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1469/>

² https://www.nccn.org/professionals/physician_gls/pdf/genetics_colon.pdf