6-year-old male child recently immigrated to the United States presented for workup of pain upon defecation. Upon general physical examination, the patient was noted to have multiple cutaneous hypomelanotic macules. His parents also endorsed development of small nodules and plaques on the forehead and nose over the past few years.

Representative endoscopic and histologic findings are shown.
Rectal endoscopic findings
What is your diagnosis?

(hint: What germline mutation might you suspect in this patient?)
Diagnosis: Hamartomatous rectal polyps

The combination of cutaneous findings in this patient (facial nodules consistent with angiofibromas per dermatologic examination) and hamartomatous rectal polyps is suggestive of Tuberous Sclerosis Complex (TSC). Genetic sequencing ultimately confirmed a frameshift mutation in \textit{SC1}. Although not specific to TSC or included in the diagnostic criteria (Reference: PMID 24053982), hamartomatous rectal polyps have been reported in the settings of \textit{TSC1} and \textit{TSC2} mutations.