(21218) - JUVENILE POLYPOSIS SYNDROME IN TWO FAMILIAL CASES: IMPLICATIONS FOR SURVEILLANCE AND MANAGEMENT

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BACKGROUND: Juvenile Polyposis Syndrome (JPS) is a rare autosomal dominant disorder characterized by multiple hamartomatous juvenile polyps throughout the gastrointestinal tract. JPS is caused by mutations in the BMPR1A and SMAD4 genes. Most juvenile polyps are benign and either asymptomatic or present with nonspecific symptoms such as anemia or bleeding. JPS patients are estimated to have a lifetime risk of gastrointestinal cancer ranging from 9% to 50%, with colorectal cancer (CRC) being the most common. The estimated cumulative CRC risk ranges from 39% to 68%. The diagnosis of JPS is typically established when an individual meets any of the following criteria: the presence of more than five juvenile polyps in the colon or rectum; the occurrence of juvenile polyps in other parts of the gastrointestinal tract; or the presence of any number of juvenile polyps along with one or more affected family members.

METHODS: We included all patients with Juvenile Polyposis Syndrome aged over 18 years, identified in the High-Risk Digestive Tumor Consultation of our institution. The population's characteristics were collected, and statistical analysis was conducted using SPSS, version 28.

RESULTS: In our institution, we have identified 15 patients with JPS, who belong to two different families. The first family carries a mutation in the BMPR1A gene and consists of the index case (♂, diagnosed at 52 years old) and 13 family members with the identified mutation (♀5; ♂8). On the other hand, the second family carries a mutation in the SMAD4 gene, with only one patient identified with the mutation. The median age at diagnosis was 49 years. Among these, 8 patients underwent multiple polypectomies, and juvenile hamartomatous polyps were identified in 5 of them. Two patients underwent

surgery (ileocolectomy and total proctocolectomy + total gastrectomy) due to the presence of multiple polyps. All patients remain alive, without extra-intestinal manifestations, and without the development of CRC, under follow-up at our institution with endoscopic screening every 1-3 years.

CONCLUSION: In this study, the importance of clinical and endoscopic surveillance for patients with JPS is highlighted. Endoscopic examinations should be conducted every 1-3 years starting from the age of 12, with the possibility of earlier or later scheduling based on the burden and size of polyps. Clinical surveillance complements the endoscopic assessment and helps identify interval polyps in a timely manner before they become malignant. In conclusion, the incidence of colorectal cancer in juvenile polyposis syndrome is high, but early detection and regular monitoring can assist in managing this risk and improving the prognosis of patients with this condition.

Palavras-chave : Juvenile Polyposis Syndrome, endoscopic surveillance, colorectal cancer