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Original article

Phenotype Characteristics of Patients With Colonic Serrated Polyposis Syndrome: A Study of 23 Cases☆

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ABSTRACT

Introduction: Serrated polyposis syndrome (SPS) is a rare entity characterized by the presence of multiple hyperplastic polyps in the colon and an increased risk of presentation and development of colorectal cancer (CRC).
Objective: To evaluate the clinical and phenotypical characteristics of patients who present one of the 3 WHO criteria for the diagnosis of SPS diagnosed and treated at a tour hospital.
Patients and methods: Patients with the diagnosis of SPS during 2005–2012 were revised; 24.208 colonoscopies were performed during this period. Other entities include age, sex, family history of CRC (APC/MYH), proximal/mixed/distal phenotype, indication for colonoscopy, number, size, location of the hyperplastic polyps, presence of mixed/adenomatous polyps, CRCI, follow-up and endoscopio/surgical treatment.
Results: A total of 23 cases were included (19 male). The median age was 51. 34% of the cases had a prior family history of CRC or polpyps. Distal phenotype was more frequent (48%). Another 73% presented synchronous adenomatous polyps, and 26% a CRC. 57% of them were asymptomatic. Surgery was performed in 9 cases (6 for cancer and 3 for polyposis), and 14 were treated by polypectomy and observation. Eleven patients (47%) presented recurrent/persistent lesions after initial surgical/endoscopic treatment.
Conclusion: SPS is a heterogeneous syndrome that is variable in the type, size, distribution and number of polyps, and is more common in male smokers with a distal phenotype. The majority of patients also present synchronous adenomatous polyps. These patients require an organized multidisciplinary evaluation.

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Características fenotípicas de los pacientes con síndrome de poliposis serrada de colon: estudio de 23 casos

Palabras clave:
Poliposis serrada
Pólipos hiperplásicos
Síndrome de poliposis serrada
Fenotipo

RESUMEN

Introducción: El síndrome de poliposis serrada (SPS) es una entidad rara caracterizada por la presencia de múltiples pólipos de histología hiperplásica en el colon y un riesgo aumentado de presentar y desarrollar cáncer colorrectal (CCR).

Objetivo: Evaluar las características clínicas y fenotípicas de los sujetos que reúnen alguno de los 3 criterios de la OMS para el diagnóstico de SPS, diagnosticados y seguidos en nuestro hospital.

Pacientes y métodos: Se revisan los pacientes con SPS durante 2005-2012, periodo en el que se realizan 24.208 colonoscopias. Se analizan edad, sexo, historia familiar de CCR (APC/MYH), fenotipo proximal/mixto/distal, indicación de colonoscopia, número, tamaño, localización de los pólipos hiperplásicos, presencia de pólipos mixtos/adenomatosos, CCRI, seguimiento y tratamiento endoscópico/quirúrgico.

Resultados: Se han recogido 23 casos (19 hombres). El promedio de edad fue 51 años. El 34% presentaba antecedentes familiares de CCR o pólipos. El fenotipo distal (48%) fue más frecuente. El 73% presentaba pólipos adenomatosos sincrónicamente, y el 26% un CCR. El 57% eran pacientes asintomáticos. Se realizó cirugía en 9 casos (6 por cáncer y 3 por poliposis, y 14 con polipectomías sucesivas y observación). Un total de 11 pacientes (47%) presentaron lesiones recurrentes/persistentes tras el tratamiento quirúrgico/endoscópico inicial.

Conclusión: El SPS es un síndrome heterogéneo, variable en tipo, tamaño, distribución y número de pólipos, siendo más frecuente en varones fumadores con fenotipo distal. La mayoría de los pacientes presentan además pólipos adenomatosos de manera sincrónica. Estos pacientes requieren una evaluación organizada multidisciplinar.

Introduction

Colorectal cancer (CRC) affects 6% of the general population. Incidence forecasts for 2015 indicate that 30 230 people in Spain will be affected making it the most frequent type of cancer, far more than lung and breast cancer.[1] Currently, it is the second cause of cancer-related mortality in Spain,[2] accounting for 12.7% of deaths in males and 15% in females.[3]

The adenoma–carcinoma sequence is considered the main pathway for colorectal carcinogenesis. Adenomatous polyps can evolve into a malignancy through the ''traditional'' pathway of carcinogenesis, namely characterized by loss of heterozygosity or APC and P53 gene allelic loss and the resulting chromosomal imbalance. This is the carcinogenesis pathway for 70%–80% of CRC.[4,5]

Hyperplastic polyps are presumed to be benign, malignancy-free lesions; however, in the last 2 decades, subtypes of these polyps have been reported. Today, they are known collectively as serrated polyps, due to the ''saw-toothed'' infolding of the crypt epithelium, which do pose a cancer risk. Two serrated pathway subtypes are described[6,7]: (a) sessile serrated adenoma through the serrated pathway; it occurs typically in the proximal colon, and its initial molecular change is MAP kinase signalling pathway activation by BRAF proto-oncogene mutation leading to CpG island methylator phenotype (CIMP-high); this is defined by methylation and subsequent silencing of the tumour gene promoter region, such as MLH1, and ensuing microsatellite instability (MSI); (b) the ''alternate serrated pathway,'' likely through traditional serrated adenoma, although less well defined than the former and characterized by KRAS gene mutation, leading to low-level methylator phenotype (CIMP-low) and silencing of the MGMT promoter region. Serrated pathways may be the cause of up to 30% of CRC.[6,7]

Hyperplastic polyposis syndrome, or rather, serrated polyposis syndrome (SPS) is rare and poorly understood entity; its genetic base is unknown, although it shows characteristics distinctive of diseases with genetic predisposition[8–10] such as colonic serrated polyps, early diagnosis, and greater prevalence conferred to CRC family history. Patients affected by this syndrome carry a high risk (25%–40%) of developing CRC[10–13]; sporadic as well as hereditary cases have been reported.[9–11] A great rate of extracolonic cancer seems to exist.[14–16] SPS likely includes a variety of patient types having different phenotypes; however, to this date, no major phenotype-related risk of developing CRC has been described.[15,16]

This study aims to assess the phenotype and clinical characteristics of patients meeting SPS criteria.

Materials and Methods

Table 1 lists the clinical criteria for SPS diagnosis reported by Burt and Jass and approved by the WHO in 2000[17]; they were