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Familial Adenomatous Polyposis (FAP) in 9 Hispanic women

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Background

Familial adenomatous polyposis (FAP) is a rare hereditary colorectal cancer syndrome estimated to account for about 1% of colorectal cancers. While there is variation in the FAP phenotype amongst individuals and families with mutations, it is characterized by a striking phenotype of colonic polyposis and other distinctive features such as desmoids and gastric fundic gland polyps. It is estimated that about 30% of APC mutations are de novo. APC mutations have been reported worldwide across different ethnic and racial groups. We report on the features of FAP seen in 9 Hispanic women with colonic polyposis, identified over 18 months.

Methods

Individuals were referred for cancer risk assessment. Genetic analysis of the APC gene, including sequencing and rearrangement studies, was conducted after counseling and informed consent.

Results

All of the individuals referred were women; the majority was originally from Mexico (67%) with the remainder

Table 1

Country of Origin*	Diagnosis		Polyps	Extra-colonic Findings	Family History**	Gene Analysis	
	Cancer	Age					exon
MX	Cholangio	48	>100	gastric polyp	mom-co-44, sis-co-32+pan-64, sis-co-38, cousin-co-38	Q1062X	15
MX	Rectal	35	>100		sis-co-50	IVS3-1G>A	
MX	Rectal	35	>100	2 mesenteric desmoids; gastric polyps	mom died at 54 of a "tumor between heart & lungs"	3709delCA	15
GU	None	39	>100		maun-co mass, not ca-49	del exon 6-15	
MX	Rectal	26	Polyposis, # unknown	abdominal desmoid; 2 pilomatrixomas	mom-ut-45, mgm-GI ca-75	3927del5	15
HO	Sigmoid descending colon	42	>100	gastric polyps	mgm-ut-35	E268X	7
MX	Rectal	33	>100		pun-"some polyps"	3183del5	15
HO	Tubular adenoma high-grade dysplasia+	42	>100	gastric polyps; duodenal polyps	maun-br-20	pending	
MX	None	35	>100	gastric polyps; duodenal polyp	none	pending	

*MX=Mexico; HO=Honduras; GU=Guatemala; +Surgery pending

**sis=sister; co=colon cancer; pan=pancreatic cancer; ut=uterine cancer; br-breast cancer; maun=maternal aunt; pun=paternal uncle; mgm=maternal grandmother; GI=gastrointestinal (not otherwise specified)

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from Central America. The average age at identification of polyposis was 37.2 years and 5 had concomitant colorectal cancer (average age 34.2 years). The most common site of cancer was the rectum and the most common extra-colonic finding was gastric polyps. The majority of women reported either no family history or cancer history inconsistent with FAP, suggesting de novo mutations. All individuals, for whom results are available, were found to have APC gene mutations. Results are found in Table 1.

Conclusions

These Hispanic women with FAP demonstrate a phenotype consistent with the existing understanding of this syndrome. Of interest, is the lack of males presenting with polyposis and the apparent overrepresentation of de novo mutations. Both of these observations may disappear as cohort size increases. However, there are other factors such as reduced access to regular and diagnostic medical services in other countries, communication barriers within families, and cultural and gender differences that might be at play.

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