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# Characteristics of Lynch syndrome in 13 Hispanic Families

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## Background

While the incidence of colorectal cancer is lower in Hispanics than in non-Hispanic Caucasians, it is the second most common cancer in this diverse ethnic population. Emerging data indicate hereditary colon cancer syndromes contribute to cancer burden regardless of race and ethnicity. These data derive from research and cohorts where Hispanics are underrepresented. Here we report on 14 individuals from 13 Hispanic families with Lynch syndrome.

## Methods

After referral for cancer risk assessment, immunohistochemical staining was performed for the four MMR gene products and/or analysis of the appropriate gene(s) was initiated.

## Results

Clinical features of the 14 individuals identified with Lynch syndrome are detailed below (Table 1). Of the 13 families, 9 (71 %) are from Mexico, 4 from Central

**Table 1**

| | Country of Origin** | 1st Cancer Diagnosis | | 2nd Cancer Diagnosis | | Family History Classification | IHC-Proteins unexpressed | Gene Analysis | |
|---|---|---|---|---|---|---|---|---|---|
| | | | Age | | Age | | | Gene | |
| 1 | ES | Cecal | 37 | Transverse | 37 | Amsterdam I | hMLH1/hPMS2 | MLH1 | S698X |
| 2 | GU | Sigmoid | 34 | | | Amsterdam I | hMSH2/hMSH6 | MSH2 | S142X |
| 3 | MX | Sigmoid | 33 | | | Amsterdam I | | MLH1 | 1105insT+ |
| 4 | MX | Gastric | 31 | Rectal | 32 | Amsterdam I | | MLH1 | R226X |
| 5 | MX | Uterine | 48 | | | Amsterdam II | hMLH1/hPMS2 | MLH1 | Q409X |
| 6 | MX | Cecal | 47 | Descendng | 47 | Bethesda | hMLH1/hPMS2 | Declined | |
| 7 | GU | Cecal | 45 | | | Bethesda | hMLH1/hPMS2 | MLH1 | ICS3-2A>G+ |
| 8 | MX | Sigmoid | 40 | | | Bethesda | hMHS2/hMSH6 | None detected[1] | |
| 9 | MX | Cecal | 36 | | | Bethesda | hMSH2/hMSH6 | MSH2 | Q593X |
| 10 | ES | Cecal | 31 | | | Bethesda | hMLH1/hPMS2 | MLH1 | K618del |
| 11 | MX | Transverse | 45 | RCC-clear cell | 45 | Bethesda | hMLH1/hPMS2 | MLH1 | del exon 2-3 |
| 12 | MX | Splenic Flexure | 32 | | | Bethesda | Pending | MSH2 | 2179delCinsAG+ |
| 13* | MX | Cecal | 44 | | | Bethesda | hMSH2/hMSH6 | MSH2 | Q76X |
| 14* | MX | Gastric | 37 | Cecal | 47 | Bethesda | hMSH2/hMSH6 | Declined | |

*Individuals 13 and 14 are brothers; **MX=Mexico; ES=El Salvador; GU=Guatemala

[1]MLH1 and MSH6 sequencing (MSH6 VUS 4071ins4), MSH2/MLH1 rearrangement studies

+not reported in Leiden Variation Open Database or the Human Gene Mutation Database at the Institute of Medical Genetics in Cardiff

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America. Seventy-two percent presented first with colon cancer (64% right-sided); 14% presented first with gastric cancer; and 1 individual with uterine cancer. The average age at first cancer diagnosis was 38.6 years; 38.5 years for colon cancer. One third had two primaries, 3 synchronous. Five of the 13 families met Amsterdam I or II criteria, while 8 met Bethesda guidelines. Two were single-case indicators, two had multiple family members affected in only One generation, and the remainder had contributory family history but too distant to meet Amsterdam. Twelve individuals underwent genetic testing; 8 MLH1 mutations and 3 MSH2 mutations were detected. Pathology was thoroughly reviewed in 11 colon tumors; 2 were well differentiated, 6 were well to moderately differentiated and 3 were focally poorly differentiated, 3 exhibited tumor infiltrating lymphocytes, 3 showed Crohn's reaction, 9 demonstrated pushing borders, 3 contained dirty necrosis, and 4 were mucinous carcinoma (>50%) with an additional 4 cases showing mucinous features (<50%).

## Conclusions

As the U.S. Hispanic population grows and access to cancer genetics services increases, the contribution of Lynch syndrome must be understood. In our cohort of Hispanic families, the early-onset right-sided colon cancer is consistent with known Lynch syndrome features. Of interest are the young age at diagnosis and the high number of families meeting only Bethesda criteria. Another area to explore is the frequency of gastric cancer in Hispanic families with Lynch syndrome, given the higher incidence of gastric cancer in Latin America.

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