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Common mutations identified in the *MLH1* gene in familial Lynch syndrome

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Abstract

Lynch syndrome (Hereditary Non Polyposis Colorectal Cancer, HNPCC) is one of the most common hereditary familial colorectal cancers (CRC) with an autosomal dominant pattern of inheritance. It accounts for 2-5% of the total CRCs reported worldwide. Although a lower incidence for CRCs have been observed in India, the last decade has shown a remarkable increase of CRC incidences (2-4 %).

Features of Lynch syndrome associated colorectal cancer include early age of cancer onset, accelerated carcinogenesis of adenomas into carcinomas, and predilection to cancer of the proximal colon (about 70% of Lynch syndrome–related colorectal cancers occur in the right colon, which is not accessed through sigmoidoscopy). Mutations in the DNA mismatch repair genes (MMR) MLH1, MSH2, MSH6 or PMS2 cells are known to be responsible for Lynch syndrome. Almost 70% of the mutations in Lynch syndrome are seen in the MLH1 and MSH2 gene.

In this study we identified three families with Lynch syndrome from a rural cancer center in western India (KCHRC, Goraj, Gujarat), where 70-75 CRC patients are seen annually. DNA isolated from the blood of consented family members of all three families (8-10 members/family) was subjected to NGS sequencing methods on an Illumina HiSeq 4000 platform. We identified unique mutations in the *MLH1* gene in all three HNPCC family members. Two of the three unrelated families shared a common mutation (154delA and 156delA). Total 8 members of a family were identified as carriers for 156delA mutation of which 5 members were unaffected while 3 were affected (age of onset: 1 member <30yrs & 2 were>40yr). The family with 154delA mutation showed 2 affected members (>40yr) carrying the mutations.LYS618DEL mutation found in 8 members of the third family showed that both affected and unaffected carried the mutation. Thus the common mutations identified in the *MLH1* gene in two unrelated families had a high risk for lynch syndrome especially above the age of 40.

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