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**hMSH2 is the most commonly mutated MMR gene in a cohort of Greek HNPCC patients.**

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**Source**

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**Abstract**

Germline mutations in genes encoding proteins involved in DNA mismatch repair are responsible for the autosomal dominantly inherited cancer predisposition syndrome hereditary nonpolyposis colorectal cancer (HNPCC). We describe here analysis of hMLH1 and hMSH2 in nine Greek families referred to our centre for HNPCC. A unique disease-causing mutation has been identified in seven out of nine (78%) families. The types of mutations identified are nonsense (five out of seven) (hMLH1: E557X, R226X; hMSH2: Q158X, R359X and R711X), a 2 bp deletion (hMSH2 1704\_1705delAG) and a 2.2 kb Alu-mediated deletion encompassing exon 3 of the hMSH2 gene. The majority of mutations identified in this cohort are found in hMSH2 (77.7%). Furthermore, four of the mutations identified are novel. Finally, a number of novel benign variations were observed in both genes. This is the first report of HNPCC analysis in the Greek population, further underscoring the differences observed in the various geographic populations.