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Screening for Lynch Syndrome in young Saudi colorectal cancer patients using microsatellite instability testing and next generation sequencing

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Background: Individuals with Lynch Syndrome (LS) have germline mutations in DNA mismatch repair genes that confer a greatly increased risk of colorectal cancer (CRC), often at a young age. Identification of mutation carriers has been demonstrated to increase their survival through improved surveillance. We previously identified 33 high risk cases for LS in the Saudi population by screening for microsatellite instability (MSI) in the tumour DNA of 284 young CRC patients.

Aim: The aim of the present study was to identify germline mutations in this cohort of patients.

Methods: Peripheral blood DNA was obtained from 13 individuals who were at high risk of LS due to positive tumour MSI status and young age (<60 years). Next generation sequencing, Sanger sequencing and Multiplex Ligation-dependent Probe Amplification were used to screen for germline mutations in the MLH1, MSH2, MSH6 and PMS2 DNA mismatch repair genes. Variants were cross-referenced against several mutation databases including the International Society for Gastrointestinal Hereditary Tumours Incorporated database.

Results: Germline mutations were identified in 8/13 (62%) high risk cases, comprising 4 mutations in MLH1 and 4 in MSH2. All mutation carriers had a positive family history for CRC or endometrial cancer.

Conclusions & Significance: Next generation sequencing is an effective strategy for the identification of young CRC patients who are at high risk of LS by positive MSI status. We estimate that 7% of CRC patients aged <60 years in Saudi Arabia are due to LS, potentially involving more than 50 new cases per year.

Biography

Masood Alqahtani has his expertise in Histocompatibility and Immunogenetics Clinical Laboratory. His role is to evaluate and monitor the graft survival during solid organ and bone marrow transplantation to provide renal failure or leukemic patients with better quality of life. The cancer incidence in some transplanted patients creates new pathway which is translational cancer research to improve healthcare. He has focused on colorectal cancer as it ranked the first cancer among Saudi male and second among female according to the latest cancer registry report. The goal that he set is to create surveillance program for familial cancer patients through genetic screening, family history and health awareness.

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