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Update in genetic colorectal cancer syndrome

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Introduction: Colorectal cancer is the third common cancer in the world. About 3-5% of the patients are carrier of genetic syndrome with high risk of colorectal cancer (CRC) and others malignancy. 20-30% of the patients with new diagnosed colorectal cancer had a family history of colorectal cancer. The most common hereditary syndrome is Lynch Syndrome (HNPCC hereditary non-polyposis colorectal cancer). Other syndromes with increased number of polyps include Familial adenomatous polyposis (FAP), attenuated FAP and MUTYH associated Polyposis (MAP).

Genetics: lynch syndrome is characterized by a germline mutation at a defective DNA mismatch repair (MMR) genes, with a high level of microsatellite instability. The most common genes involved in the syndrome are MLH1, MSH2, MSH6, PMS2 and EpCAM. FAP caused by APC gene defects and MAP caused by a defect in the MUTYH gene. Lynch syndrome and FAP are inherited autosomal dominant, while MAP inherited autosomal recessive. Diagnosis is made by genetic investigation, founder mutation and gene sequencing.

Cancer risk: Mutation carrier of the different types of the syndromes has increased risk of colonic and extra-colonic neoplasm. The lifetime CRC risk is estimated to be 50-80% in HNPCC and about 100% in FAP. The risk of the malignancy development is depending on mutation and gene.

Clinical setting: Amsterdam criteria and revised Bethesda criteria were developed to identify persons and families with high risk form Lynch syndrome. Patients with FAP are characterized by thousands of polyps and MAP patients by 10-100 of polyps.

Universal screening for lynch syndrome: should patients with colorectal cancer or endometrial cancer undergo screening by immunohistochemistry (IHC) or microsatellite instability (MSI) for lynch syndrome? Yes, several recommendations include the universal screening for all diagnosed patients under age 70 years. The Surveillance recommendation and treatment with aspirin or cox2 will be discussed. All the above points will be updated and discussed during the lecture.

Speaker Biography

Naim Abu-Freha received his MD from the Tuebingen University, Germany at 2005 before becoming resident at internal medicine and then completed his gastroenterology residency at the Soroka Medical Center at 2014. He received his master degree MHA from Ben-Gurion University, Beer-Sheva, Israel. He researched different topics in gastroenterology/Hepatology and different issues regarding the Bedouin Arab minority in southern Israel. He is one of the founders groups of the Arab Medical Associations in the Negev (AMAN) and the first Chairman of the Associations since 2015.

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