

## Update in Genetic Colorectal Cancer Syndrome

Naim Abu Freha

Soroka University Medical Center, Israel

Email: abufreha@yahoo.de

### Abstract :

**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.

**INTRODUCTION COLONRECTAL SYNDROME** a typically printed as he development of cancer from the colon or part (parts of

the massive intestine). A cancer is that the abnormal growth of cells that have the flexibility to invade or unfold to completely different components of the body. part cancer (CRC), in addition said as organ cancer, carcinoma, or part cancer, Most part cancers unit owing to maturity and modus vivendi factors, with exclusively a little vary of cases owing to underlying genetic disorders part cancer begins once healthy cells among the liner of the colon or part modification and grow out of management, forming a mass said as a tumor. A tumor a typically cancerous or benign. A cancerous tumor is malignant, which means it'll grow and unfold to completely different components of the body. A {benign tumor |benign growth | nonmalignant tumour |non-malignant neoplasm |tumor ,|tumour |neoplasm} means the neoplasm can grow but will not unfold. These changes generally take years to develop. every genetic and environmental factors can cause the changes. However, once a personal has Associate in Nursing uncommon transmitted syndrome changes can occur in months or years. part cancer most often begins as a polyp, a noncancerous growth that may develop on the inner wall of the colon or part as If not treated or removed, a polyp can become a most likely severe cancer. Finding and removing metastatic tumor polyps can forestall part cancer. regarding 100% of colon polyps unit flat and exhausting to go looking out with a scrutiny unless a dye is utilized to spotlight them. These flat polyps have a high risk of turning into cancerous, nonetheless their size. Hyperplastic polyps could develop among the colon and part. they are not thought-about metastatic tumor. large intestine cancer is termed "hereditary" or "inherited" once several generations of a family have bowel cancer. several issue mutations, or abnormalities, that cause bowel cancer and allow it to be transmitted to members of the family, are found. an element could also be a block of deoxyribonucleic acid that

Extended Abstract

holds the ordering, or directions, for producing proteins vital to our bodily functions.

**TYPES ;**

The two commonest hereditary bowel cancer syndromes are hereditary nonpolyposis bowel cancer (HNPCC) and Familial adenomatous polyposis (FAP). They will have an impact on men and ladies, and conjointly the kids of people who carry these factors have a 5 hundredth probability of hereditary the disease-causing sequence. Nonpolyposis bowel cancer, HNPCC, is that the most typical variety of hereditary malignant neoplastic disease. People with HNPCC even have a more robust risk of developing completely different kill syndrome-related cancers, like viscus, ovarian, uterine, renal, pelvis, intestine, and abdomen cancer

**CAUSES ;**

Among the genes found to be concerned in large intestine cancer are: MSH2 and MSH6 each on body two and MLH1, on body three. Normally, the macromolecule merchandise of those genes facilitate to repair mistakes created in DNA replication as the causes of genetic large intestine cancer. A awfully tiny portion of large intestine cancers are caused by heritable sequence mutations. Several of those DNA changes and their effects on the expansion of cells are currently celebrated. For example: Familial adenomatous polyposis (FAP), attenuated FAP (AFAP), and Gardner syndrome are caused by heritable changes within the APC sequence.

**SYMPTOMS**

Signs and symptoms of carcinoma include:

- A persistent modification in your gut habits, together with diarrhoea or constipation or a modification within the consistency of your stool
- Rectal trauma or blood in your stool
- Persistent abdominal discomfort, like cramps, gas or pain
- A feeling that your gut does not empty utterly
- Weakness or fatigue
- Unexplained weight loss

**RISK FACTORS :**

Factors that will increase your risk of carcinoma include:

- Older age. carcinoma may be diagnosed at any age, however a majority of individuals with carcinoma are older

than fifty. The rates of carcinoma in folks younger than fifty are increasing, however doctors are not certain why.

- African-American race. African-Americans have a larger risk of carcinoma than do folks of different races.
- A personal history of large intestine cancer or polyps. If you have already had carcinoma or noncancerous colon polyps, you have got a larger risk of carcinoma within the future.
- Inflammatory enteral conditions. Chronic inflammatory diseases of the colon, like inflammatory bowel disease and inflammatory bowel disease, will increase your risk of carcinoma.

**TREATMENT :**

Include Surgeries like Laparoscopic surgery, ostomy for body part cancer, Radiofrequency ablation (RFA) or cryoablation, therapy like External-beam therapy, Stereotactic therapy, Therapies victimization medication embody

The types of general therapies used for large intestine cancer include:

- Chemotherapy
- Targeted medical care
- Immunotherapy

**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