

Microsatellite instability pathway and molecular genetics of colorectal cancer

Juel Chowhdury, Khatja Batool, Sidra Mubasher, Humera Batool Hani Pharaon

Microsatellite instability (MSI) is a form of DNA damage that occurs when the length of a microsatellite sequence (a short, repetitive DNA sequence) is altered. This can happen due to errors in DNA replication or repair. In the context of colorectal cancer, MSI is a key feature of the microsatellite instability pathway, which is a type of DNA mismatch repair (MMR) defect. The MMR pathway is responsible for identifying and correcting errors in DNA replication, such as mismatches between base pairs. When this pathway is defective, errors are not corrected, leading to MSI. This is particularly common in colorectal cancer, where it is associated with a specific set of genetic mutations, including those in the MMR genes MLH1, MSH2, MSH6, and PMS2. The presence of MSI in colorectal cancer is associated with a distinct clinical course and prognosis, and it is a key factor in the classification of colorectal cancer into microsatellite stable (MSS) and microsatellite unstable (MSI) subtypes. The MSI pathway is also a key feature of the Lynch syndrome, a hereditary cancer predisposition syndrome. The molecular genetics of colorectal cancer, including the role of MSI, is a complex and rapidly evolving field, and it is essential for understanding the pathogenesis and treatment of this disease.

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