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Molecular basis of hereditary colorectal cancer and current research status

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On the basis of genetics, colorectal cancer is classified with two types as sporadic cancer and hereditary colorectal cancer, which mainly includes familial adenomatous polyposis (FAP) and hereditary non-polyposis colon cancer (HNPCC). They have different molecular modules of coloretal carcinogenesis. FAP has clear clinical features, including 100s to 1000s of adenomatous polyps throughout colon & rectum, 100% penetrance without surgery and very early age of onset. FAP is an autosomal dominant disease which mainly attributed to the germline mutations of APC gene, which located at chromosome 5q21.3. HNPCC has obscure clinical features and is diagnosed mainly by family history investigation, such as Amsterdam Criteria. HNPCC is also an autosomal dominant disease which mainly attributed to the germline mutations of mismatch repair (MMR) genes, which mainly include hMLH1 and hMSH2. Systematic mutation screening methods, including denaturing high performance liquid chromatography (DHPLC), multiplex ligation-dependent probe amplification (MLPA) and DNA sequencing are the powerful tools to detect different types of mutations. DHPLC is a method chromatography for the detection of base substitutions, small deletions or insertions at the DNA. MLPA is a variation of the multiplex polymerase chain reaction that permits multiple targets to be amplified with only a single primer pair. MLPA is one of the only accurate, time-efficient techniques to detect genomic deletions and insertions, which are frequent causes of HNPCC. With these techniques, we have screened mutations of APC and MMR genes in identified FAP and HNPCC pedigrees and novel mutations were discovered. The genetic testing helps us to screen out the high risk family members and offer clinical treatment.

Biography

Chen Senqing has completed his PhD in 2008 from Nanjing Medical University. He is presently the Director of Laboratory of Genetics and Molecular Biology, Jiangsu Institute of Cancer Research. He has published more than 60 papers in reputed journals, including 15 SCI papers.

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