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Discovery of genetic defects in unexplained colorectal cancer syndromes

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Discovery of genetic defects in unexplained Lynch Syndrome colorectal cancer syndromes

colorectal cancer
familial adenomatous polyposis
PhD
variant
CRC susceptibility
APC
homopolymer
pathogenic
FFPE
Leiden University Medical Center
mosaicism
MSI
somatic
MSH6
patients
adenoma
Whole Gene Capture
MMR-deficiency
MLH1 promoter hypermethylation
next-generation sequencing
microsatellite instability
pseudogenes
RNA
DNA
field cancerization
POLE
nonsense mediated decay
mismatch repair
splice site
Pathology
POLD1
MLH1
leukocyte
PMS2
exon
Human Genetics

Anne Jansen