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Discovery of genetic defects in unexplained colorectal cancer syndromes

Lynch Syndrome

Whole Gene Capture

MLH1 promoter hypermethylation

next-generation sequencing

field cancerization

POLE

mismatch repair

splice site

CRC susceptibility

unexplained

MMR-deficiency

mismatch repair

pathogenic

leukocyte

somatic

adenoma

DNA

RNA

MSH6

Patients

Pathology

leukocyte

exon

familial adenomatous polyposis

CRC susceptibility

Human Genetics Leiden University Medical Center

variant

FFPE

Pathology

familial adenomatous polyposis

Lynch Syndrome

MSI

microsatellite instability

pseudogenes

nonsense mediated decay

MLH1

PMS2

POLE

splice site

POLD1

msi

homopolymer

adenoma

DNA

RNA

pathogenic

leukocyte

exon

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