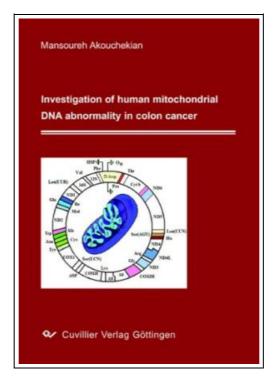
Investigation of human mitochondrial DNA abnormality in colon cancer



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(Ms. Kellie O'Hara I)

INVESTIGATION OF HUMAN MITOCHONDRIAL DNA ABNORMALITY IN COLON CANCER



Cuvillier Verlag Okt 2008, 2008. Taschenbuch. Condition: Neu. Neuware - The appearance of mtDNA abnormalities including deletions, duplications, insertions or combinations of them in colorectal cancer (CRC) patients wasinvestigated in this work. Our result showed that 11 of 90 CRC patients had a 8.7kb mtDNA deletion (12.2%) while this deletion was not found in 33 healthycontrols. (P=0.035)We conclude that the 8.7 kb deletion can be a secondary effect of the cancerprocess, which is not inherited. This is the first report on large scale deletions inCRC.We also analyzed the correlation between cancers and mitochondrial haplogroups.The relationship between CRC and each of 9 major mitochondrial haplogroupswere examined in Iranian CRC patients. This is the first study to trace mtDNAHVSI variants in CRC patients of the Persian population. Our data showed thatpatients with CRC have a significantly (P=0.001) higher frequency of haplogroupK (9.5%) when compared with controls (0%). The mitochondrial polymorphisms in haplogroup K might play a causative role in predisposing to CRC.Variations in the D-loop region were found in both CRC patients and healthypeople but the frequency of SNPs in CRC patients was higher than in controlsamples. We found 13 new polymorphisms that had not been recorded in themitochondrial database. We also detected one T C transition at np16519 in 28out of 40 patients (70%). In a recent study researchers reported that the T16519Cvariation worsen the outcome of pancreatic cancer patients, possibly because it isinvolved in altering cellular metabolism. Because we found this mutationfrequently in our healthy controls (43%) and could not find any differentiationamong our patients with respect to the alleles at this mutation, we conclude thatthe functional significance of this mutation needs further investigation.We also analyzed somatic mutations in mitochondrial genes encoding subunits ofthe respiratory complex I. Seven somatic mutations in the MT-ND1 gene werefound. Six of these mutations were synonymo

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