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(54) Title: CONVERTING DIPLOIDY TO HAPLOIDY FOR GENETIC DIAGNOSIS

(57) Abstract: Detection of mutations associated with hereditary diseases is complicated by the diploid nature of mammalian cells. Mutations present in one allele are often masked by the wild-type sequence of the other allele. Individual alleles can be isolated from every chromosome within somatic cell hybrids generated from a single fusion. Nucleic acids from the hybrids can be analyzed for mutations in an unambiguous manner. This approach was used to detect two cancer-causing mutations that had previously defied genetic diagnosis. One of the families studied, Warthin Family G, was the first kindred with a hereditary colon cancer syndrome described in the biomedical literature.