Detection of microsatellite instability (MSI) is the standard part of mutational analysis in hereditary nonpolyposis colorectal cancers (HNPCC). Characteristic phenotypic feature of MSI indicates loss of mismatch repair (MMR) in tumor cells.

We studied MSI in 205 tumors from 152 patients with HNPCC. Of these, 37 patients fulfilled Amsterdam criteria, 72 patients were familial and 43 were sporadic cases. We used methods of fragmentation analysis on polyacrylamide gel and/or with fluorescent labelled primers (ABI Prism 310 Genetic Analyzer). Three mononucleotide (BAT-RII, BAT-25, BAT-26) and five dinucleotide (D2S123, D3S1029, D5S346, D17S250, D18S58) repeat loci were analysed. We detected 75 tumors with high degree of MSI (MSI-H), 12 tumors with low degree of MSI (MSI-L) and 118 tumors with stable microsatellites (MSS). In 44 of these, loss of heterozygozity (LOH) was found.

In 30 patients with MSI-H tumors a mutation in one of mismatch repair genes was detected. Microsatellite analysis was positively correlated with immunohistochemical detection of MLH1 and MSH2 proteins.