Detection method for the 3'*EPCAM* genomic deletion and its frequency in Polish HNPCC patients

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Lynch syndrome is a frequent, autosomal, dominantly-inherited cancer predisposition syndrome caused by various germline alterations that affect DNA mismatch repair genes, mainly *MLH1*, *MSH2* and *MSH6*. *EPCAM* (Epithelial Cell Adhesion Molecule) large rearrangements, localized upstream of *MSH2* on chromosome 2, have been recently described as the one of the genetic factors associated with the Lynch syndrome occurrence. 3'*EPCAM* genomic rearrangements may be a cause of mismatch repair deficiency in some Lynch syndrome families. The aim of the study was to develop cost-effective screening tool for the rapid detection of 3'*EPCAM* genomic rearrangements, along with its validation, and determination of the 3'*EPCAM* genomic rearrangements status in our group of HNPCC patients. We applied developed by us C-HRM method enable to detect CNVs of the 3'EPCAM and simultaneously screen for small mutation in two exons of the *MLH1* gene containing small mutation hot-spots. In our group of 250 Lynch syndrome probants we detected 2 cases of 3'*EPCAM* genomic rearrangement and 4 small mutations within the *MLH1* gene.