

Incidence of breast cancer is continuously increasing in the Czech Republic. Tumor development is a result of gene alterations' accumulation, particularly associated with genes involved in regulation of cell growth and division. Hereditary carcinomas account for approximately 5-10% of all breast tumors and in 60-80% cases are caused by a germline mutation in the major predisposition genes BRCA1 and BRCA2. Nevertheless, other genes, mostly of lower penetrance, may play a role in breast pathogenesis such as the ATM tumor-suppressor gene.

ATM is the apex of the repair pathway of DSB. This protein kinase activates through phosphorylation of its substrates cell cycle checkpoints, which leads either to the delay of the cell cycle progression until DSB are repaired or to the promotion of apoptosis.

To sum up, the ATM gene seems to have a role in breast cancer development in a minority of the high-risk families in our population which is significantly lower compared to BRCA1/2 and it also seems to be involved in pathogenesis of sporadic breast cancer. Despite the ATM gene's length, we do not perform the preventive screening of this gene in breast cancer high-risk families. Nevertheless, we offer the molecular diagnostics of ATM to ataxia telangiectasia patients.