Summary:

Background: An increased risk for development of hereditary breast cancer is associated with germline mutations in *BRCA1/2* and the influence of *NBN* mutations is also supposed. The aim of this study is to specify the frequency of recurrent mutations in *BRCA1/2* in unselected breast cancer patients and the frequency of most common pathogenic mutations in *NBN* in Czech republic, to assess current criteria for genetic testing and to consider the addition of *NBN* to the tested genes. **Methods:** Screening for recurrent mutations 5382insC and 300T>G in *BRCA1* was performed by RFLP, screening for mutations in exon 11 of *BRCA1* was performed by PTT, screening for mutations in a selected region of exon 11 of *BRCA2* by DHPLC, and screening for mutations in exon 6 of *NBN* by HRMA. All the mutations were confirmed by direct sequencing. **Results:** In 679 unselected breast cancer patients 7 carriers of 5382insC, 3 of 300T>G, and 4 of other mutations in *BRCA1* were identified. 2 locally prevalent mutations were found in *BRCA2*. In 730 controls only one 5382insC *BRCA1* mutation was identified. Out of 5 *NBN* mutations found in 600 high-risk patients two were 657del5 and one R215W. A total of 8 *NBN* mutation carriers were identified among 703 breast cancer patients, 2 of them 657del5 carriers and three R215W carriers. In 915 controls 9 *NBN* mutations were detected, of which two 657del5 and four R215W. **Conclusion:** *BRCA1/2* mutation frequency of 2,4% was observed in our series of unselected breast cancer patients. 10 of 16 *BRCA1/2* patients would not meet any of currently used criteria for genetic testing. Screening of all breast cancer patients might detect more carriers prior to surgery and thus improve the decision making regarding therapy. *NBN* mutation frequency is very low in Czech republic and did not differ significantly among all three studied groups. Routine screening for *NBN* mutations in Czech unselected breast cancer population cannot be recommended.