Results

In total, a pathogenic mutation was identified in 79 of the 297 families (26.6%) analyzed. Clinically significant mutations were identified in 17 of the genes included in the panel. The most commonly mutated genes in the Romanian population were BRCA1 and BRCA2, accounting for 50% of the mutations identified, followed by PALB2 (12%), CHEK2 (9.4%) and ATM, NBN and RAD50 which accounted for 3.5% of the mutations each. Of note is that 7 of the 79 affected families (8.8%) carried clinically significant mutations in different two genes.

Conclusions:
Our results support the use of a panel of genes involved in hereditary cancer predisposition. In this series of patients, analysis of this panel allowed for the identification of 14% additional pathogenic variants. This is especially true in those cases where more than one pathogenic variant was identified.

References: