

## **Prevalence of germline TP53 mutations among early-onset breast cancer patients from Polish population.**

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The objective of this study was to determine spectrum and prevalence of germline mutations in the TP53 gene among Polish women with early-onset BC. The study was conducted in two independent cohorts: a) 100 patients with breast cancer diagnosed  $\leq 30$  years old, b) 1,880 consecutive women diagnosed with BC  $\leq 45$  years and 2,000 healthy women, as a control group. Four heterozygous pathogenic mutations were detected in a group of 100 patients with early-onset breast cancer – TP53 c.844C>T (p.Arg282Trp, rs28934574), TP53 c.818G>A (p.Arg273His, rs28934576), TP53 c.733G>A (p.Gly245Ser, rs28934575), TP53 c.659A>G (p.Tyr220Cys, rs121912666). On the basis of software prediction and available literature data, all these variants can be defined as pathogenic. None of these TP53 variants were detected among 1,880 breast cancer patients and 2,000 healthy controls.

In order to improve the treatment and surveillance screening the search for TP53 germline mutations should be recommended in all female BC cases diagnosed  $\leq 30$  years old.