Germline variants in ACD, TERF2IP and POT1 genes in patients with melanoma in the Polish population.

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Malignant melanoma (MM) accounts for about 2% of all cancers.
In the recent years, the frequency of incidents of malignant melanoma the frequency of melanoma, increased about 60% in Poland which gives up to 3,000 new cases each year. Germline mutations CDKN2A, the major melanoma determinant were found only in 6% of Polish families.

The goal of the project was to determine the frequency and spectrum of POT1, ACD, TERF2IP gene mutation in Polish population.

The study was conducted in two cohorts of patients: 60 patients (45 females with mid age of 53, range 29-91 and 15 males with average age of 58, range 32-85) b) 1500 unselected MM patients (women aged 15-92 (mean age 59.9) and men aged 18-84 (mid age 55) and control group of 1500 healthy adults without cancer diagnosis in family history in first and second degree relatives.

We found one potentially pathogenic “missense” mutations in POT1 (rs116916706), one “missense” variant in TERF2IP (rs4888444) and one mutation in splice region in ACD gene (rs571116752) using Sanger sequencing. In the next step using Real- time PCR (Taqman) we detected POT1 variants in two patients out of 1500 melanoma patients, one patients in 1500 healthy controls. In TERF2IP in first cohort 40 CMM patients exhibits mutation variant and 140 in the second one. None of ACD variants were detected among 1500 melanoma patients and 1500 healthy controls.

In conclusion germline mutation POT1, ACD, TERF2IP genes are infrequent in Polish Malignant Melanoma patients.