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High cumulative risks of cancer in patients with PTEN hamartoma tumour syndrome

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Résumé en anglais	BACKGROUND: PTEN hamartoma tumour syndrome (PHTS) encompasses several clinical syndromes with germline mutations in the PTEN tumour suppressor gene, including Cowden syndrome which is characterised by an increased risk of breast and thyroid cancers. Because PHTS is rare, data regarding cancer risks and genotype-phenotype correlations are limited. The objective of this study was to better define cancer risks in this syndrome with respect to the type and location of PTEN mutations. METHODS: 154 PHTS individuals with a deleterious germline PTEN mutation were recruited from the activity of the Institut Bergonie genetic laboratory. Detailed phenotypic information was obtained for 146 of them. Age and sex adjusted standardised incidence ratio (SIR) calculations, cumulative cancer risk estimations, and genotype-phenotype analyses were performed. RESULTS: Elevated SIRs were found mainly for female breast cancer (39.1, 95% CI 24.8 to 58.6), thyroid cancer in women (43.2, 95% CI 19.7 to 82.1) and in men (199.5, 95% CI 106.39 to 342.03), melanoma in women (28.3, 95% CI 7.6 to 35.4) and in men (39.4, 95% CI 10.6 to 100.9), and endometrial cancer (48.7, 95% CI 9.8 to 142.3). Cumulative cancer risks at age 70 were 85% (95% CI 70% to 95%) for any cancer, 77% (95% CI 59% to 91%) for female breast cancer, and 38% (95% CI 25% to 56%) for thyroid cancer. The risk of cancer was two times greater in women with PHTS than in men with PHTS ($p<0.05$). CONCLUSIONS: This study shows a considerably high cumulative risk of cancer for patients with PHTS, mainly in women without clear genotype-phenotype correlation for this cancer risk. New recommendations for the management of PHTS patients are proposed.
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