Human Genomics

CORRECTION

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Correction: Two recurrent pathogenic/likely pathogenic variants in *PALB2* account for half of *PALB2* positive families in Slovenia

Vita Andreja Mesarič[1,2], Ana Blatnik[3,2], Kristina Drusany Starič[1,2], Ksenija Strojnik[3], Vida Stegel[3,2], Simona Hotujec[3], Vita Šetrajčič Dragoš[3,2], Petra Škerl[3], Srdjan Novaković[3,2] and Mateja Krajc[3,2*]

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In this article [1], the Abstract, Keywords, and Ethical approval sections were missing. It has been updated as given below.

The original article has been corrected.

Abstract

Background The prevalence and spectrum of *PALB2* pathogenic/likely pathogenic variants (PV/LPVs) may vary across different regions, and these have not yet been analysed and reported in Slovenian HBOC families.

Methods We performed a retrospective analysis of all 5099 consecutively tested individuals from 4610 families who fulfilled national criteria for HBOC-panel testing from January 2015 to January 2022. After genetic counselling, genetic testing with next generation sequencing was performed for all probands and cascade testing was offered to their blood relatives.

Results Among all probands tested 0.9% (40/4610) were *PALB2* PV/LPV carriers. 13 different *PALB2* PV/LPVs were detected, one of them was novel. Four PV/LPVs were found to be recurrent in Slovenian population with two most frequent being c.509_510del and c.1451T > A. Altogether, 60 individuals from 40 *PALB2* positive families were identified, 42 being cancer patients. 28.6% *PALB2*-positive cancer patients were diagnosed with more than one malignant tumour. We identified three double heterozygote carriers with additional PV/LPVs in *ATM*, *CHEK2* and *BRCA1*.

Discussion This report provides the first comprehensive description of molecular and clinical characteristics of *PALB2* carriers in Slovenia. The frequency of *PALB2* pathogenic variants in the Slovenian HBOC accounts for 0.9% of all individuals tested for PVs in HBOC-related genes. Our study adds a novel recurrent mutation, which is unique to the Slovenian context and one PV/LPVs, which had not been reported in the literature so far. The results of our study add information on genotype and phenotype in *PALB2*-positive patients and may be used for population specific assessment.

Keywords HBOC, *PALB2*, Breast cancer, Ovarian cancer, Risk-reducing salpingoophorectomy, Recurrent pathogenic variants

Ethics approval and consent to participate: The present study was approved by the National Ethics Committee and the Institutional Ethics Committee of the Institute of Oncology Ljubljana (0120-591/2020/3 on the 20th of January 2021).

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*Correspondence:
Mateja Krajc
mkrajc@onko-i.si

[1]Ljubljana University Medical Centre, Ljubljana, Slovenia
[2]University of Ljubljana, Ljubljana, Slovenia
[3]Institute of Oncology Ljubljana, Ljubljana, Slovenia

Reference

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