

# DETEKSI POLIMORFISME RS249954 DAN RS16940342 GEN *PALB2* PADA WANITA DENGAN KANKER PAYUDARA DI RSUD DR. MOEWARDI SURAKARTA

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## INTISARI

Kanker payudara merupakan tumor ganas pada jaringan payudara yang berasal dari transformasi neoplastik epitel duktus ataupun lobulus payudara. Kanker payudara memiliki angka insidensi, tingkat mortalitas, dan tingkat morbiditas tertinggi di Indonesia. Mutasi pada gen *PALB2* dapat meningkatkan risiko terjadinya kanker payudara. Mutasi ini dapat berupa *single nucleotide polymorphisms* (SNPs) seperti halnya rs249954 dan rs16940342. Oleh karena itu, penelitian ini bertujuan untuk mendeteksi keberadaan polimorfisme rs249954 dan rs16940342 gen *PALB2* pada wanita dengan kanker payudara di RSUD Dr. Moewardi Surakarta. Selain itu, penelitian ini dilaksanakan untuk mengetahui jumlah subjek yang membawa polimorfisme rs249954 dan rs16940342 gen *PALB2*. Subjek penelitian ini adalah wanita dengan kanker payudara yang melakukan pemeriksaan di RSUD Dr. Moewardi Surakarta yang memenuhi kriteria inklusi dan bersedia ikut serta dalam penelitian. Deteksi polimorfisme rs249954 dan rs16940342 dari gen *PALB2* dilakukan dengan metode PCR dan sekuensing. Isolat DNA diperoleh dari sampel darah *whole blood*. Analisis data dilakukan dengan observasi pita hasil elektroforesis, penajaran hasil sekuensing, dan analisis kromatogram melalui program *Benchling*. Hasil penelitian ini menunjukkan bahwa polimorfisme rs249954 dan rs16940342 gen *PALB2* dapat dideteksi pada wanita dengan kanker payudara di RSUD Dr. Moewardi Surakarta. Polimorfisme rs249954 gen *PALB2* ditemukan pada 1 dari 8 individu dan polimorfisme rs16940342 gen *PALB2* ditemukan pada 1 dari 8 individu. Hasil penelitian ini mengindikasikan potensi deteksi polimorfisme gen *PALB2* sebagai biomarker untuk prediksi risiko kanker payudara pada level genetik dan molekuler.

**Kata kunci :** kanker payudara, gen *PALB2*, SNPs, rs249954, rs16940342

## **DETECTION OF POLYMORPHISM RS249954 AND RS16940342 *PALB2* GENE IN WOMEN WITH BREAST CANCER AT RSUD DR. MOEWARDI SURAKARTA**

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### ***ABSTRACT***

Breast cancer is a tumor malignancy in breast tissue originating from the neoplastic transformation of the ductal epithelium of breast lobules. Breast cancer has the highest incidence, mortality, and morbidity rates in Indonesia. Mutations in the *PALB2* gene are known to increase the risk of breast cancer in women. This mutation can be in forms of single nucleotide polymorphisms (SNPs), such as rs249954 and rs16940342. Therefore, the objective of this study was to detect polymorphisms rs249954 and rs16940342 *PALB2* genes in women with breast cancer at RSUD Dr. Moewardi Surakarta. This study also aimed to determine the number of subjects which carry polymorphisms rs249954 and rs16940342 *PALB2* genes. The subjects of this study were women with breast cancer at RSUD Dr. Moewardi Surakarta who met the inclusion criteria and were willing to participate in the research. The detection of polymorphisms rs249954 and rs16940342 of the *PALB2* gene was carried out by PCR and sequencing methods. DNA isolates were obtained from the whole blood samples. Data analysis was carried out by observing the electrophoresis bands results, the DNA sequence alignment, and the aligned chromatogram results using the Benchling program. The results of this study showed that polymorphisms rs249954 and rs16940342 in the *PALB2* gene were detected in women with breast cancer at RSUD Dr. Moewardi Surakarta. Polymorphism rs249954 was detected in 1 out of 8 individuals and rs16940342 was detected in 1 out of 8 individuals. The results of this study indicated that the detection of the *PALB2* gene polymorphism can potentially be used as a biomarker for the prediction of breast cancer risk at the genetic and molecular level.

**Keywords :** breast cancer, *PALB2* gene, SNPs, rs249954, rs16940342