

DETEKSI POLIMORFISME GEN *PTPNI* DAN *HNF4A* PADA PASIEN TALASEMIA DI BANYUMAS

ABSTRAK

Talasemia adalah penyakit hereditas akibat berkurangnya produksi rantai globin sehingga pasien memerlukan transfusi darah berulang. Salah satu komplikasi transfusi darah yaitu diabetes melitus. Diabetes melitus diduga dipengaruhi oleh lingkungan serta faktor genetik seperti gen *PTPNI* dan *HNF4A*. Tujuan penelitian ini adalah mengetahui data epidemiologi polimorfisme *PTPNI* -1023 C>A dan *T130I HNF4A* pada pasien talasemia di Banyumas. Penelitian ini menggunakan studi deskriptif dan pengambilan sampel *consecutive sampling* pada pasien talasemia yang sudah terdaftar dalam *database* YTI Banyumas. DNA pasien talasemia diproses dengan PCR-RFLP untuk mengetahui polimorfisme gen *PTPNI* dan *HNF4A*. Data polimorfisme disajikan dalam bentuk proporsi. Total subjek pada penelitian adalah 102 sampel DNA pasien talasemia. Polimorfisme *PTPNI* -1023C>A dengan genotipe CC 90 sampel (69%), CA 30 sampel (29%), AA 2 sampel (2%). Polimorfisme *T130I HNF4A* dengan genotipe CC 81 sampel (79%), CT 16 sampel (16%), TT 5 sampel (5%). Proporsi mutan heterozigot sebanyak 29% dan mutan homozigot 2% pada polimorfisme *PTPNI* -1023C>A. Proporsi mutan heterozigot 16% dan mutan homozigot 5% pada polimorfisme *T130I HNF4A*.

Kata Kunci : diabetes melitus tipe 2, polimorfisme *PTPN* -1023C>A, polimorfisme *T130I HNF4A*, talasemia

**DETECTION OF PTPN1 AND HNF4A GENE POLYMORPHISM IN
THALASSEMIA PATIENTS IN BANYUMAS**

ABSTRACT

Thalassemia is a hereditary disease due to reduced globin chain production so patients need repeated blood transfusions. One of the complications of blood transfusions is diabetes mellitus. Diabetes mellitus is related to the environment by genetic factors such as PTPN1 and HNF4A. The purpose of this study was to determine the epidemiological data of PTPN1 -1023 C> A and T130I HNF4A polymorphisms in thalassemia patients in Banyumas. This study used descriptive studies and sequential sampling in thalassemia patients who were registered in the YTI Banyumas database. The DNA of thalassemia patients was processed by PCR-RFLP to determine the PTPN1 and HNF4A gene polymorphisms. Polymorphism data is presented in the form of contributions. The total subjects in the study were 102 DNA samples of thalassemia patients. PTPN1 -1023C>A polymorphism with 90 samples of CC (69%), CA 30 samples (29%), AA 2 samples (2%). T130I HNF4A polymorphism with 81 CC genotype samples (79%), 16 CT samples (16%), 5 TT samples (5%). The proportion in PTPN1 -1023C>A polymorphism is 29% of heterozygous mutants and 2% of homozygous mutants. The proportion of T130I HNF4A polymorphism is 16% of heterozygous mutants and 5% of homozygous mutants.

Keywords: type 2 diabetes mellitus, PTPN -1023C>A polymorphism, T130I HNF4A polymorphism, thalassemia