

# Pengaruh mutasi Southeast Asian Ovalocytosis (SAO) dan thalassemia &#945; (alfa) terhadap penderita defisiensi enzim G6PD (Glucose-6-Phosphate-Dehydrogenase) = Southeast Asian Ovalocytosis (SAO) and &#945; (alfa) thalassemia mutation effect on people with G6PD (Glucose-6-Phosphate-Dehydrogenase) enzyme deficiency

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## Abstrak

[Southeast Asian Ovalocytosis (SAO), thalassemia &#945;; serta defisiensi enzim G6PD (Glukosa-6-Fosfat Dehidrogenase) merupakan kelainan sel darah merah yang terjadi akibat adanya mutasi. Kelainan sel darah merah tersebut banyak ditemukan pada daerah endemik malaria. Hal tersebut diduga terkait dengan adanya mekanisme proteksi terhadap parasit malaria. Penelitian bertujuan untuk mengetahui frekuensi penderita SAO, thalassemia &#945;; G6PDd/SAO dan G6PDd/thalassemia &#945;; Metode deteksi yang dilakukan yaitu dengan pengamatan morfologi eritrosit, perhitungan sel darah total (CBC), serta biologi molekuler menggunakan Polymerase Chain Reaction (PCR). Berdasarkan hasil PCR, didapatkan frekuensi penderita SAO sebesar 11,4%, thalassemia &#945; sebesar 15,2%, G6PDd/SAO sebesar 0,8% , serta penderita G6PDd/thalassemia &#945; sebesar 0,32%. Manifestasi klinis dapat dilihat dari nilai perhitungan sel darah total (CBC) penderita G6PDd/SAO dan G6PDd/thalassemia &#945; yang cenderung rendah.

;Southeast Asian Ovalocytosis (SAO), &#945; thalassemia, and G6PD (Glucose-6-Phosphate-Dehydrogenase) enzyme deficiency are red blood cell disorders that occur due to mutations in the DNA. These red blood cell disorders are commonly found in malaria endemic areas. That lead to the assumption that may provide protection against malaria parasite. The research done in order to determine the frequency of SAO, &#945; thalassemia, G6PDd/SAO, and also G6PDd/ &#945; thalassemia. Detection method used by erythrocytes morphological observation and also from haematological profile. In addition, for more accurate result used molecular method detection by Polymerase Chain Reaction (PCR). Based on PCR, result showed frequency for SAO 11,4%, &#945; thalassemia 15,2%, G6PDd/SAO 0,8% , and for G6PDd/ &#945; thalassemia 0,32%. Haematological profile from suffered showed tend to be lower.

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