ABSTRACT

Southeast Asian Ovalocytosis (SAO) is a hereditary genetic disorder caused by a 27 bp deletion in the *SLC4A1* gene, leading to a disrupted folding at the extracellular face of the SLC4A1 protein in the red blood cell (RBC) membrane. SAO is an autosomal dominant disorder where heterozygous individuals are asymptomatic and homozygous SAO are considered embryonic lethal. SAO can be observed microscopically on peripheral blood smears because of its characteristic oval-shaped RBC that is sometimes accompanied by a slit in the middle. In this study, microscopic detection of RBC morphology is compared with molecular genotyping using PCR to validate SAO diagnosis using samples taken from Malinau, North Kalimantan along with finding the prevalence of the disease in the area. From this, it was found that the prevalence of SAO in Malinau is 1.1% out of 178 patients. Along with this, statistical analysis of microscope screening characteristics showed that the apparatus is highly sensitive (100%), but not specific enough (65%) to fully replace PCR as a main diagnostic tool for SAO. This study highlights the importance of the employment of a properly trained microscopist and the optimization of blood smearing and fixation techniques in the utilization of a microscope for SAO diagnosis.

Keywords: Southeast Asian Ovalocytosis (SAO), diagnosis, microscopy, red blood cell, genotyping, *SLC4A1*