

INTISARI

Albinisme merupakan kelainan genetik yang ditimbulkan akibat gangguan biosintesis melanin pada kulit, rambut maupun bagian mata. Albinisme dibedakan menjadi dua tipe berdasarkan ciri fenotipe, yaitu *Ocular Albinism* (OA) dan *Oculocutaneous Albinism* (OCA). *Oculocutaneous Albinism* (OCA) memiliki karakteristik hipopigmentasi pada kulit, rambut dan mata, sementara *Ocular Albinism* (OA) hanya jalur penglihatan yang terpengaruh. *Oculocutaneous Albinism* (OCA) terbagi menjadi 7 tipe yaitu OCA tipe 1, 2, 3, 4, 5, 6, 7. OCA tipe 2 disebabkan oleh mutasi gen *OCA2* (gen *P*). Gen *OCA2* mengkode protein P yang diprediksi berperan dalam proses transfer tirosin dan protein melanosomal lain yang berhubungan dengan TYRP1 di dalam membran sel. Individu albinisme di Rejang Lebong memiliki ciri-ciri rambut, alis dan bulu mata berwarna coklat, kuning jahe dan kekuning-kuningan, yang merupakan ciri individu albinisme *OCA2*. Mutasi yang sering ditemukan pada individu *OCA2* di wilayah Asia terletak pada ekson 13, 14 dan 22. Tujuan penelitian yaitu mengetahui ada tidaknya mutasi pada ekson 13, 14, dan 22 gen *OCA2* pada keluarga albinisme di Rejang Lebong serta menentukan letak dan jenis mutasi apabila ditemukan. Sebagai bagian dari pendekatan molekuler dalam penelitian ini, dilakukan analisis genetik terhadap sampel biologis untuk mengidentifikasi kemungkinan mutasi pada gen target. DNA diekstraksi dari sampel saliva anggota keluarga dengan albinisme. Fragmen DNA ekson 13, 14, dan 22 gen *OCA2* diamplifikasi menggunakan metode PCR. Hasil amplifikasi digunakan sebagai *template* sekuensing. Selanjutnya, data sekuensing dianalisis menggunakan program *GeneStudio* dan *Mega11*, serta dilanjutkan dengan analisis menggunakan *Benchling* dalam deteksi mutasi pada gen *OCA2* ekson, 13, 14, dan 22. Hasil analisis menunjukkan bahwa tidak terdapat variasi nukleotida pada ekson 13, dan 14 gen *OCA2*. Namun, mutasi terdeteksi pada seluruh ekson 22 di seluruh subjek albinisme. Jenis mutasi subjek A1 dan A2 adalah mutasi *missense* c.2249C>T (p.Pro750Leu) dan diklasifikasikan sebagai *benign*. Sementara pada subjek A3 ditemukan mutasi sebagai mutasi *silent* c.2328T>C (Ala776=).

Kata kunci: albinisme, *OCA2*, tirosinase, mutasi

ABSTRACT

Albinism is a genetic disorder caused by disruption of melanin biosynthesis in the skin, hair and eyes. Albinism is divided into two types based on phenotypic characteristics, namely *Ocular Albinism* (OA) and *Oculocutaneous Albinism* (OCA). *Oculocutaneous Albinism* (OCA) has the characteristics of hypopigmentation of the skin, hair and eyes, while *Ocular Albinism* (OA) only affects the visual pathway. *Oculocutaneous Albinism* (OCA) is divided into 7 types, namely OCA types 1, 2, 3, 4, 5, 6, 7. OCA type 2 is caused by a mutation in the *OCA2* gene (*P* gene). The *OCA2* gene encodes the P protein which is predicted to play a role in the transfer process of tyrosine and other melanosomal proteins related to TYRP1 in the cell membrane. Albinism individuals in Rejang Lebong have the characteristics of brown, ginger yellow and yellowish hair, eyebrows and eyelashes, which are the characteristics of *OCA2* albinism individuals. Mutations frequently found in *OCA2* individuals in Asia are located in exons 13, 14, and 22. The purpose of this study was to determine whether there were mutations in exons 13, 14, and 22 of the *OCA2* gene in albinism families in Rejang Lebong and to determine the location and type of mutations if found. As part of the molecular approach in this study, genetic analysis was performed on biological samples to identify possible mutations in the target gene. DNA was extracted from saliva samples of family members with albinism. DNA fragments of exons 13, 14, and 22 of the *OCA2* gene were amplified using the PCR method. The amplification results were used as templates for sequencing. The sequencing data were analyzed using the *GeneStudio*, and *Mega11* software, followed by further analysis using the *Benchling* software to detect mutations in exon 13, 14, and 22 of the *OCA2* gene. The result revealed mutation in exon 22 of the *OCA2* gene in three albinism subjects exhibiting the *OCA2* phenotype. The mutation type in subjects A1 and A2 was a *missense mutation*, c.2249C>T (p.Pro750Leu), which is classified as *benign*. In subjects A3, a *silent mutation* c.2328T>C (p.Ala776=) was identified.

Keywords: albinism, *OCA2*, tyrosinase, mutation