Abstrack

Background: The incidence of cutaneous melanoma is increasing worldwide, with the highest incidence in Caucasians. The prognosis of melanoma is generally poor because it has a high risk of metastasis and more often causes death. BRAF mutation is one of the prognosis factors for melanoma. The presence of BRAF mutations stimulates the MAPK pathway (mitogen-activated protein kinase) so that cell proliferation increases. Research on BRAF mutations in melanoma in Indonesia is still very rare. This study aims to identify BRAF mutations in primary melanoma cases and their relationship to age, sex, location, and lymph node metastasis.

Objective: Researched paraffin blocks from cases of primary melanoma from the Department of Pathology Anatomy Dr. Sardjito and Dr. Hospital Soeradji Tirtonegoro. The status of the BRAF V600 mutation is detected using qReal Time PCR. The relationship of BRAF V600 mutation status with age, sex, anatomical location, and lymph node metastasis was analyzed by Fisher's Exact Test.

Methods: Formalin-fixed paraffin-Embedded (FFPE) tissue of primary melanoma cases will be collected from the Department Anatomical Pathology of Dr. Sardjito and dr. Soeradji Tirtonegoro Hospital. BRAF mutation status will be detected using real-time PCR. The existence of these mutations is then analysed and to know its association with associated with age, sex, anatomic location, and lymph node metastasis.

Result: From 43 samples, there were 4 (9.3%) samples with the BRAF V600 mutation. Mutations were obtained 3 (30%) in the central / non-extremity region and 1 (3.03%) in the extremity region, the number of male patients was the same as women (2 each), 3 patients were young ≤ 65 years and 1 patient age> 65 years, the number of patients with and without lymph node metastasis is the same (2 each). The results of the statistical analysis showed a relationship ($p = 0.034$) between the anatomical location of melanoma and the BRAF V600 mutation, the highest location in the central /non-extremity region. There was no association between age, sex and lymph node metastasis with the BRAV V600 mutation.

Conclusion: BRAF V600 mutations were found in 9.3% of patients with cutaneous melanoma. BRAF V600 mutations were significantly associated with an anatomic location and did not associate significantly with age, sex, and lymph node metastasis.

Keywords: primary cutaneous melanoma, BRAF V600 mutation, age, sex, anatomic location, lymph node metastasis

Abstrak

Latar Belakang: Insiden melanoma kutaneus meningkat di seluruh dunia, dengan insiden tertinggi pada ras Kaukasia. Prognosis melanoma umumnya buruk karena memiliki risiko metastasis yang besar dan lebih sering menyebabkan kematian. Mutasi BRAF merupakan salah satu faktor prognosa melanoma. Adanya mutasi BRAF menstimulasi jalur MAPK (mitogen-activated protein kinase) sehingga proliferasi sel meningkat. Penelitian tentang mutasi BRAF pada melanoma di Indonesia masih sangat jarang. Penelitian ini bertujuan untuk mengidentifikasi mutasi BRAF pada kasus melanoma primer dan hubungannya dengan usia, jenis kelamin, lokasi dan metastasis limfonodi.

Metode: Diteliti blok parafin dari kasus-kasus melanoma primer dari Departemen Patologi Anatomi Dr. Sardjito dan Rumah Sakit Dr. Soeradji Tirtonegoro. Status mutasi BRAF V600 dideteksi dengan menggunakan *qReal Time* PCR. Hubungan status mutasi BRAF V600 dengan usia, jenis kelamin, lokasi anatomi dan metastasis limfonodi dianalisis dengan *Fisher's Exact Test* .

Hasil: Dari 43 sampel, didapatkan 4 (9,3%) sampel dengan mutasi BRAF V600. Mutasi didapatkan 3 (30%) di regio sentral/non ekstrimitas dan 1 (3,03%) di regio ekstrimitas, jumlah pasien laki-laki sama dengan perempuan (masing-masing 2), 3 pasien usia muda ≤ 65 tahun dan 1 pasien usia > 65 tahun, jumlah pasien dengan dan tanpa metastasis limfonodi sama (masing-masing 2). Hasil analisis statistik menunjukkan adanya hubungan (p=0,034) antara lokasi anatomi melanoma dengan mutasi BRAF V600, lokasi terbanyak pada regio sentral/non ekstrimitas. Tidak didapatkan hubungan antara umur, jenis kelamin dan metastasis limfonodi dengan mutasi BRAV V600.

Kesimpulan: Mutasi BRAF V600 didapatkan pada 9,3% pasien melanoma kutaneus. Mutasi BRAF V600 berhubungan bermakna antara lokasi anatomi dan tidak berhubungan secara bermakna dengan umur, jenis kelamin, dan metastasis limfonodi.

Kata kunci: melanoma kutaneus primer, mutasi BRAF V600, Usia, Jenis kelamin, lokasi anatomi, metastasis limfonodi