

INTISARI

Latar belakang: Glioma adalah bentuk tumor otak primer yang paling sering ditemukan di Indonesia di antara keganasan otak lainnya. Penderita glioma mempunyai angka harapan hidup yang rendah karena sering mengalami kekambuhan dan resistan terhadap terapi. Angka harapan hidup penderita glioma dipengaruhi beberapa faktor yaitu usia, stadium, jenis histopatologis, Karnofsky Performance Scale, luas pembedahan, defisit neurologis, dan modalitas terapi. Karnofsky Performance Scale merupakan penilaian untuk membantu klasifikasi pasien berdasarkan gangguan fungsionalnya. Skala pemeriksaan ini dapat digunakan untuk menilai prognosis pasien secara individual. Sejak 2008, *sequencing* pada glioma telah mengidentifikasi mutasi pada gen isositrat dehidrogenase 1 dan 2 (IDH1 dan IDH2). Mutasi IDH ini dikaitkan dengan umur yang lebih muda dan peningkatan *survival* pada pasien glioma sehingga didapatkan nilai KPS yang lebih tinggi. Namun, penelitian mengenai hubungan mutasi gen IDH dengan nilai Karnofsky Performance Scale pada pasien glioma belum pernah dilakukan di Indonesia.

Tujuan: Tujuan penelitian ini adalah untuk mengetahui hubungan mutasi gen IDH1/2 dengan nilai Karnofsky Performance Scale pada pasien glioma di Indonesia.

Metode: Penelitian ini merupakan studi *cross sectional*. Subjek penelitian adalah pasien glioma yang datang ke RSUP DR. Sardjito yang memenuhi kriteria inklusi dan eksklusi diikutsertakan dalam penelitian dengan jumlah 37 subjek. Data klinis dan demografis subjek penelitian dikumpulkan dari rekam medis yang terdaftar di Instalasi Catatan Medik dan jaringan glioma subjek penelitian diambil dari preparat FFPE (*Formalin Fixed Paraffin Embedded*) di Bagian Patologi Anatomi RSUP Dr. Sardjito. Sampel jaringan kemudian dikirimkan ke Laboratorium Biologi Molekuler Fakultas Kedokteran Universitas Gadjah Mada untuk dilakukan analisis DNA. Hasil analisis DNA kemudian dikumpulkan dan dilakukan uji statistika untuk menilai hubungan antara mutasi Gen IDH1/2 dengan nilai Karnofsky Performance Scale pada pasien glioma.

Hasil: Dari total 37 subjek penelitian glioma yang diambil dari preparat FFPE, didapati sebanyak 9 subjek memiliki profil IDH1 *mutant* dan sebanyak 28 subjek memiliki profil IDH1 *wild-type*. Tidak ditemukan mutasi pada IDH2. Berdasarkan hasil analisis statistik, diketahui tidak terdapat perbedaan signifikan pada nilai Karnofsky Performance Scale antara kelompok IDH *mutant* dengan IDH *wild-type* ($p=0,562$).

Kesimpulan: Adanya mutasi gen IDH1/2 tidak memiliki hubungan yang signifikan dengan nilai Karnofsky Performance Scale pada pasien glioma di Indonesia.

Kata kunci: Glioma, Mutasi Isocitrate Dehydrogenase, Karnofsky Performance Scale.

ABSTRACT

Background: Gliomas are the most common form of brain tumor found in Indonesia among other brain malignancies. Glioma patients have a low life expectancy because they often experience recurrence and resistance to therapy. Survival rate of glioma patients are affected by several factors like age, stadium, histologi type, Karnofsky Performance Scale, extent of surgery, neurological deficit, and modality of therapy. Karnofsky Performance Scale is an assessment to give patient's classification based on the fungtional disorder. This scale can be used to assess patient's prognosis individually. Since 2008, sequencing in glioma had identified mutation of gene Isocitrate Dehydrogenase 1 and 2 (IDH1 and IDH2). IDH mutation was associated with younger age and better survival in glioma patients, giving the higher KPS score. Nevertheless, there has been no study linking IDH gene mutation with Karnofsky Performance Scale score in Indonesia.

Objective: The purpose of this study was to determine the association of IDH1 / 2 gene mutations with Karnofsky Performance Scale score in glioma patients in Indonesia.

Methodes: This is a cross sectional study. Research subjects were glioma patients who came to RSUP DR. Sardjito who met the inclusion and exclusion criteria were included in the study with a total of 37 subjects. Clinical and demographic data of the research subjects were collected from the medical records listed in the Instalasi Catatan Medik RSUP DR. Sardjito. The Glioma tissues of the research subject were taken from the FFPE preparation (Formalin Fixed Paraffin Embedded) in the Anatomical Pathology Department RSUP Dr. Sardjito. The tissue samples were then sent to the Molecular Biology Laboratory of Gadjah Mada University's Medical Faculty for DNA analysis. The DNA analysis was then collected and statistically tested to assess the relationship between the IDH1/2 gene mutation and the Karnofsky Performance Scale score in glioma patients.

Results: From a total of 37 FFPE samples of glioma's research subject, 9 subjects had IDH1 mutant profiles and 28 subjects had wild-type IDH1 profiles. No mutations were found on IDH2. Based on statistical analysis, there is no significant difference in Karnofsky Performance Scale score between IDH mutant group and IDH wild-type group ($p = 0,562$).

Conclusion: The presence of IDH1/2 gene mutations has no significant association with Karnofsky Performance Scale score in glioma patients in Indonesia.

Keywords: Glioma, Isocitrate Dehydrogenase Mutation, Karnofsky Performance Scale.