

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. Sejak pertama kali ditemukan pada tahun 2008, mutasi gen Isocitrate Dehydrogenase (IDH) memiliki banyak peranan penting dalam mengembangkan pengetahuan mengenai kasus glioma. Berdasarkan penelitian yang dilakukan oleh Sanson et al, disimpulkan bahwa mutasi gen IDH1 kodon 132 dan gen IDH2 kodon 172 memiliki keterkaitan dengan profil genomik tumor dan diketahui bahwa mutasi gen IDH merupakan faktor prognostik independen yang kuat dengan overall survival yang lebih baik pada pasien glioma. Berdasarkan penelitian yang dilakukan oleh Brazil et al, penggunaan Barthel Index sebagai alat pengukuran disabilitas fungsional pada pasien glioma memiliki berbagai kelebihan seperti; mudah digunakan, memiliki tingkat reliabilitas tinggi, dapat menggambarkan prognosis pasien, dan lebih objektif jika dibandingkan dengan penilaian Karnofsky Performance Scale (KPS). Diketahui bahwa mutasi gen IDH memiliki insidensi tinggi dan keterkaitan erat dengan kasus glioma. Di sisi lain penilaian Barthel Index pada pasien glioma dapat menggambarkan kemampuan fungsional pasien. Selain itu diketahui juga, baik penilaian barthel index maupun pengecekan mutasi IDH pada kasus glioma dapat memberikan gambaran terhadap prognosis pasien glioma. Meskipun begitu hingga saat ini masih belum ada penelitian yang menghubungkan mutasi gen IDH dengan nilai Barthel Index.

Tujuan: Tujuan penelitian ini adalah untuk mengetahui hubungan mutasi Gen IDH1/2 dengan nilai Barthel Index 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 RSUP DR. Sardjito. 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 dengan menggunakan perangkat lunak SPSS untuk menilai hubungan antara mutasi Gen IDH1/2 dengan nilai Barthel Index pada pasien glioma.

Hasil: Dari total 37 subjek penelitian glioma, 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 Barthel Index antara kelompok IDH *mutant* dengan IDH *wild-type* ($p=0,235$).

Kesimpulan: Adanya mutasi gen IDH 1/2 tidak memiliki hubungan yang signifikan dengan nilai Barthel Index pada pasien glioma di Indonesia.

Kata kunci: Glioma, Mutasi Isocitrate Dehydrogenase, Barthel Index.

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. Since it was first discovered in 2008, Isocitrate Dehydrogenase (IDH) gene mutations have many important roles in developing knowledge about glioma cases. Based on a study conducted by Sanson et al, it was concluded that the gene mutations IDH1 codon 132 and IDH2 gene codon 172 were associated with tumor genomic profiles and it was found that the IDH gene mutation was a strong independent prognostic factor with better overall survival in glioma patients. Based on research conducted by Brazil et al, the use of Barthel Index as a functional disability measurement tool in glioma patients has many advantages such as; easy to use, has a high degree of reliability, can describe the patient's prognosis, and more objective when compared with the assessment Karnofsky Performance Scale (KPS). It is known that IDH gene mutations have a high incidence and are closely related to glioma cases. On the other hand, the assessment of Barthel Index in glioma patients can describe the patient's functional ability. Also known, both barthel index assessment and IDH mutation checks in glioma cases can provide an overview of the prognosis of glioma patients. Nevertheless, until now there has been no study linking IDH gene mutation with Barthel Index score.

Objective: The purpose of this study was to determine the association of IDH1 / 2 gene mutations with Barthel Index scores in glioma patients in Indonesia.

Methods: 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 using SPSS software to assess the relationship between the IDH1/2 gene mutation and the Barthel Index score in glioma patients.

Results: From a total of 37 subjects of glioma research, 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 Barthel Index score between IDH mutant group and IDH wild-type group ($p = 0,235$).

Conclusion: The presence of IDH 1/2 gene mutations has no significant association with Barthel Index score in glioma patients in Indonesia.

Keywords: Glioma, Isocitrate Dehydrogenase Mutation, Barthel Index