ANALISIS PROFIL KADAR PROTEIN *SURVIVAL MOTOR NEURON* (SMN) DALAM DARAH PADA PASIEN ANAK DENGAN *SPINAL MUSCULAR ATROPHY* (SMA) DI RSUP DR. SARDJITO

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INTISARI

Latar belakang: *Spinal Muscular Atrophy* (SMA) adalah salah satu penyakit genetik yang menyerang sistem neuromuskular. Individu dengan SMA mengalami degenerasi neuron motor pada cornu anterior medulla spinalis, menyebabkan atrofi otot dan kelemahan anggota tubuh yang progresif. Pada sebagian besar kasus SMA, ditemukan adanya penurunan ekspresi protein *Survival Motor Neuron* (SMN), yaitu polipeptida bermassa 38 kDa yang tersusun atas 294 asam amino dan dikodekan oleh gen *SMN1*. Sekitar 95% kasus SMA disebabkan oleh mutasi delesi homozigot gen *SMN1* yang diwariskan secara resesif autosomal. Penelitian terdahulu menunjukkan bahwa terdapat asosiasi antara kadar protein SMN dengan tipe SMA. Akan tetapi, studi yang menggambarkan kadar protein SMN pada anak dengan masing-masing tipe SMA masih sangat terbatas.

Tujuan: Mengetahui profil kadar protein SMN dalam darah pada pasien anak dengan SMA dari masing-masing tipe SMA, serta mengetahui ada atau tidaknya asosiasi antara tingkat keparahan (tipe SMA) dengan kadar protein SMN dalam darah.

Metode: Kadar protein SMN dalam sampel darah pasien anak dengan SMA dan pasien non-SMA diperiksa menggunakan *enzyme-linked immunosorbent assay* (ELISA).

Hasil: Pemeriksaan kadar protein menggunakan teknik ELISA, melibatkan 21 subjek yang terdiri atas 8 pasien non-SMA dan 13 pasien positif SMA, terdiri atas 11 pasien SMA tipe II dan 2 pasien SMA tipe III. Kadar protein SMN dalam darah yang dinilai menggunakan metode *enzyme-linked immunosorbent assay* (ELISA) pada pasien anak dengan SMA tipe II dan tipe III secara berurutan adalah 135.24 ± 74.08 pg/mL dan 201.60 ± 29.70. Nilai ini tidak berbeda signifikan dengan kelompok kontrol (orang sehat).

Kata kunci: *Spinal Muscular Atrophy, Spinal Motor Neuron, ELISA*.

ANALYSIS OF BLOOD SURVIVAL MOTOR NEURON (SMN) PROTEIN LEVEL IN CHILDREN WITH SPINAL MUSCULAR ATROPHY AT RSUP. DR. SARDJITO

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ABSTRACT

Latar belakang: Spinal Muscular Atrophy (SMA) is a genetic disorder that impacts the neuromuscular system. Individuals with SMA experience the degeneration of motor neurons in the anterior horn of the spinal cord, resulting in progressive muscle wasting and limb weakness. In the majority of SMA cases, there is a reduction in the production of the Survival Motor Neuron (SMN) protein, a 38 kDa polypeptide composed of 294 amino acids, which is encoded by the SMN1 gene. Approximately 95% of SMA cases are caused by a homozygous deletion mutation in the SMN1 gene, inherited in an autosomal recessive pattern. Prior studies have indicated a link between SMN protein levels and the different types of SMA. However, research examining SMN protein levels in children with each specific SMA type is still quite limited.

Objective: Determining the profile of SMN protein levels in the blood, assessed using the enzyme-linked immunosorbent assay (ELISA) method, in pediatric patients with SMA.

Methods: Peripheral Blood Mononuclear Cells (PBMC) were isolated from whole blood samples of pediatric patients with SMA and non-SMA patients. The SMN protein levels in the patient blood samples were then measured using the enzyme-linked immunosorbent assay (ELISA) method.

Result: The SMN protein levels in the blood, assessed using the ELISA, in children with SMA type II and type III were 135.24 ± 74.08 pg/mL and 201.60 ± 29.70 pg/mL, respectively. Meanwhile, the SMN protein levels in the blood of non-SMA children, assessed using the ELISA, were 147.13 ± 46.77 pg/mL.

Conclusion: The total protein levels, SMN protein levels, and the SMN/total protein ratio in children with SMA type II and III were not significantly different from those in the control group (non SMA).

Keyword: Spinal Muscular Atrophy, Spinal Motor Neuron, ELISA.