

PENGARUH TERAPI OBAT ANTI EPILEPSI DAN INTERVENSI PSIKOSOSIAL TERHADAP KUALITAS HIDUP ANAK DENGAN TUBEROSKLEROSIS

Rima Hemawati R¹, Elisabeth Siti Herini², Mei Neni Sitaresmi³

¹Mahasiswa MS-PPDS Ilmu Kesehatan Anak Fakultas Kedokteran, Kesehatan Masyarakat dan Keperawatan, Universitas Gadjah Mada Yogyakarta / RSUP Dr. Sardjito, Yogyakarta

^{2,3}Departemen Ilmu Kesehatan Anak, Fakultas Kedokteran, Kesehatan Masyarakat dan Keperawatan, Universitas Gadjah Mada Yogyakarta / RSUP Dr. Sardjito, Yogyakarta

INTISARI

Latar Belakang: Manifestasi klinis *tuberous sclerosis complex* (TSC) sangat beragam dan terus berkembang seumur hidup pasien. Karenanya diagnosis kelainan ini sering sulit dan terlambat ditegakkan. Banyak penderita tuberosklerosis dengan gejala ringan tanpa epilepsi yang tidak terdiagnosis. Penanganan awal dapat segera dilakukan agar membantu mencegah komplikasi dan meningkatkan kualitas hidup pasien.

Metode: Pengamatan kasus longitudinal merupakan penelitian observasional dengan metode kohort prospektif untuk mengamati perjalanan penyakit subjek dengan TSC dengan Epilepsi *general tonic clonic*, disabilitas intelektual ringan, gangguan pemusatan perhatian hiperreaktivitas, temper tantrum. Penelitian ini mengamati luaran klinis yaitu kualitas hidup, status pertumbuhan dan gizi, epilepsi tidak relaps, EEG, efek samping obat, fungsi kognitif, fungsi adaptif, GPPH remisi, keterlibatan organ lain pada subjek yang diberikan intervensi berupa obat anti epilepsi, dan intervensi psikososial.

Hasil: Epilepsi tidak relaps, EEG membaik, tidak didapatkan efek samping obat antiepilepsi, status nutrisi pasien gizi baik, fungsi kognitif dan adaptif memburuk, GPPH membaik, gangguan sistem organ lain tidak didapatkan perburukan manifestasi klinis, kualitas hidup pasien membaik.

Simpulan: Pemberian obat antiepilepsi memperbaiki kualitas hidup pasien TSC dengan epilepsi. Manifestasi klinis timbul seiring bertambahnya usia dari pasien TSC. Oleh karenanya diperlukan monitoring pada pasien TSC, monitoring dapat dilakukan sesuai dengan rekomendasi *International Tuberous Sclerosis Complex Consensus Group*.

Kata Kunci: Tuberosklerosis, epilepsi, kualitas hidup



ANTIEPILEPTIC MEDICATION AND PSYCHOSOCIAL INTERVENTION
OUTCOME ON QUALITY OF LIFE IN A PATIENT WITH TUBEROUS
SCLEROSIS COMPLEX

Rima Hemawati R¹, Elisabeth Siti Herini², Mei Neni Sitaresmi³

¹ MS-PPDS Pediatric Residency Program, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada / Dr. Sardjito General Hospital, Yogyakarta, Indonesia

^{2,3}Department of Pediatrics, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada / Dr. Sardjito General Hospital, Yogyakarta, Indonesia

ABSTRACT

Background: Tuberous sclerosis complex (TSC) is a genetic disorder characterized by multisystem involvement and highly variable clinical manifestations that may progress throughout life. This heterogeneity often results in delayed diagnosis, particularly in patients with mild symptoms. Early diagnosis and appropriate management are crucial to prevent complications and improve quality of life.

Method: This is a longitudinal case observation using a prospective cohort approach in a patient diagnosed with TSC presenting generalized tonic-clonic epilepsy, mild intellectual disability, attention-deficit/hyperactivity disorder (ADHD), and behavioral disturbances. The patient received antiepileptic drug therapy combined with psychosocial interventions. Clinical outcomes assessed included quality of life, growth and nutritional status, seizure recurrence, electroencephalography (EEG) findings, adverse drug effects, cognitive and adaptive functions, ADHD status, and involvement of other organ systems.

Result: During follow-up, no seizure relapse occurred, and EEG findings showed improvement. No adverse effects related to antiepileptic drugs were observed. The patient maintained good nutritional status. Cognitive and adaptive functions declined, whereas ADHD symptoms improved. No progression of clinical manifestations in other organ systems was identified. Overall, the patient's quality of life improved.

Conclusion: Antiepileptic drug therapy contributes to improved quality of life in pediatric patients with TSC and epilepsy. Clinical manifestations of TSC may evolve with age, thus long-term and structured monitoring is essential, in accordance with the recommendations of the *International Tuberous Sclerosis Complex Consensus Group*.

Kata Kunci: tuberous sclerosis complex, epilepsy, quality of life.