

INTISARI

Latar Belakang Tuberosklerosis (TSC) adalah kelainan bawaan autosom dominan yang mempengaruhi berbagai sistem organ. Kelainan ini memiliki beban penyakit yang signifikan dan berdampak besar pada kualitas hidup. Kami melakukan pengamatan selama 18 bulan pada pasien dengan TSC untuk mengevaluasi terapi dengan obat anti-epilepsi (OAE) dan intervensi psikososial terhadap kualitas hidup pasien.

Deskripsi kasus Seorang anak lelaki berusia 13 tahun telah mengalami kejang berulang selama lima tahun terakhir. Kejang memburuk meskipun telah mendapat pengobatan dengan tiga OAE yang berbeda. Pasien juga memiliki lesi kulit yang dikenal sebagai angiofibroma pada wajah, *shagreen patch*, dan makula hipomelanotik. Pasien didiagnosis sebagai TSC berdasarkan temuan > 2 kriteria utama Konsensus TSC 2012. Selama periode pengamatan, terdapat perbaikan frekuensi kejang dan perbaikan EEG dengan penggunaan OAE asam valproat (60mg / kg / hari), karbamazepin (18 mg / kg / hari), dan topiramate (3 mg / kg / hari). Namun, jumlah dan ukuran lesi kulit meningkat, dan kami juga menemukan fibroma kuku pada saat usia pasien 14 tahun. Beberapa *tuber* dan nodul subependimal tampak pada MRI otak. Tidak ada komplikasi pada sistem organ lain. Selain medikamentosa, pasien juga dikelola dengan manajemen psikososial *self management support*, suatu supervisi kesehatan melibatkan tim multidisipliner, yang terdiri dari : *overarching care processes, informational support, peer support, coaching support, informational and technological support, and family daily management support*. Pada akhir pengamatan, kualitas hidup pasien meningkat, dinilai dari Skor PedsQL pasien yang menunjukkan peningkatan dibandingkan dengan sebelum pengamatan.

Kesimpulan Manajemen OAE yang optimal dan program *self management support* untuk pasien TSC dengan *intractable* epilepsi meningkatkan kepatuhan terhadap pengobatan, perbaikan klinis, dan kualitas hidup.

Kata kunci: tuberosklerosis, *intractable epilepsy*, *self management support*, kualitas hidup

ABSTRACT

Background Tuberous Sclerosis Complex (TSC) is an autosomal dominant inherited disorder affecting multi organ system. It is associated with significant disease burden and considerable impact on quality of life. We conducted an 18-month follow-up of a patient with TSC to determine the effect of anti-epileptic drugs (AEDs) and psychosocial intervention on quality of life of tuberous sclerosis patients.

Case summary A 13-years-old boy had experienced recurrent seizures for the past five years. The seizures worsened despite treatment with three different AEDs. He also had skin lesions that were recognized as facial angiofibromas, shagreen patches, and hypomelanotic macules. He was diagnosed as TSC based on the presence of >2 major criteria from the 2012 TSC Consensus. During the observation period, there was resolution of seizures and EEG improvement with a regimen of valproic acid (60mg/kg/day), carbamazepine (18 mg/kg/day), and topiramate (3 mg/kg/day). However, the skin lesions increased in number and size, and we found unguinal fibromas at the age of 14 years. Multiple tubers and subependymal nodules were seen on brain MRI. There were no complications in other organ systems. The patient also obtained self-management support, health provisions involving a multidisciplinary team to support children with chronic diseases, delivering overarching care processes, informational support, peer support, coaching support, and informational and technological support, and family daily management support. This support system was included in our psychosocial intervention program to improve quality of life. The patient's PedsQL score showed improvement after, compared to before the follow-up period.

Conclusion Optimal AEDs treatment and self-management support for children with intractable epilepsy due to TSC improves compliance to medication, clinical outcome, and quality of life.

Keywords: *tuberous sclerosis, intractable epilepsy, self-management support, quality of life*



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HIDUP ANAK DENGAN
TUBEROSKLEROSIS**

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