INTISARI

Latar Belakang *Tuberosklerosis* atau disebut *Tuberous Sclerosis Complek* (*TSC*) adalah kelainan genetik *autosomal dominan* dan mempunyai karakteristik multisistem yang menyebabkan kelainan pada otak dan organ lainnya seperti jantung, mata, paru-paru, ginjal, dan kulit yang menyebabkan mortalitas dan morbiditas.
Tujuan Untuk melakukan pemantauan dan intervensi jangka panjang pada anak dengan *TSC*. intelectual disability , dan Epilepsi belum terkontrol.
Metode Pemantauan dan intervensi jangka panjang terkait luaran anak dengan *TSC* dengan kohort prospektif. Anak 11 tahun perempuan, di RSUP Dr. Sardjito dari Desember 2015 sampai November 2017.
Hasil Pemantauan selama 24 bulan, pertumbuhan anak (berat badan, tinggi bada, lingkar kepala) normal. Status gizi baik, manifestasi pada otak (cortical tubers dan sub ependymal nodul), kulit (angiofibroma, *macula hypomelanotic*, *dan shagreen patch*), epilepsi terkontrol, gangguan perilaku, gangguan perkembangan, dan disabilitas intelektua. Tidak didapatkan gangguan pada gigi, mata, telinga, jantung, paru-paru, dan ginjal.
Kesimpulan Diagnosa dini dan intervensi (multi disiplin) dini mempengaruhi kualitas hidup anak.
Kata kunci: laporan kasus, *tuberous sclerosis complex*, Yogyakarta

ABSTRACT

Background *Tuberosclerosis* or well known as *Tuberous Sclerosis Complex* (*TSC*) is an autosomal dominant genetic disorder characterized by multisystem organ disorder including brain, heart, eyes, lung, kidney, and skin which causes morbidity and mortality.
Objective To conduct long-term monitoring and intervention towards children with TSC, intellectual disability, and uncontrolled epilepsy.
Methods We conducted long-term monitoring and intervention cohort prospective study in an 11 years old girl in Dr .Sardjito hospital from December 2015 until November 2017.
Results Following 24 months monitoring, we found normal growth (body weight, height, head circumference). She was in good nutritional state and there were some manifestations on the brain (cortical tubers and subependymal nodule) and skin (angiofibroma, *macula hypomelanotic*, *and shagreen patch*) and the epilepsy was controlled. However, there were behavioral disorder, maturation disorder, and intellectual disability. There was no disorder on the lips, teeth, eyes, ears, heart, lungs, and kidneys.
Conclusions Early diagnostic and multidisciplinary intervention affect their quality of life.
Keywords: a case report, tuberous sclerosis complex, yogyakarta