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Tuberous sclerosis complex

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Background & Aim: Tuberous sclerosis or tuberous sclerosis complex (TSC) is a genetic disorder, caused by mutations on either of two genes TSC1 on chromosome 9q34 (hamartin) and TSC2 on chromosome 16p13 (tuberin). First described by Desiree Magloire Bourneville in 1880. The prevalence of one in 9000 live birth, affecting both sexes and all ethnic groups. Patients develop hamartomas of the brain, kidneys, heart, lungs, skin and eyes. The important oral manifestations include oral mucosal angiofibromas and dental enamel pits. It is a multisystem disorder which becomes perceptible only in late childhood, limiting the expediency for early diagnosis in infancy. 60-90 % presents with seizures, usual onset 1 year. The management of these patients is often multidisciplinary involving specialists from various fields. Aim is to identify clinical features of signs, symptoms and characteristic of tuberous sclerosis complex.

Method: Observational descriptive study of patient's medical records. Case report is of 11-year-old boy child presented to the Emergency Department of Zainoel Abidin Hospital with history of repeated episodes of generalized tonic-clonic seizure associated with loss of consciousness. In past, he had multiple hospital admissions for the same reason and was on antiepileptic drug since the age of 3 months with poor control. Antenatal, natal history was normal. Cerebral palsy history since 3 months old. There was no history of seizure in family members

Findings: He had multiple adenoma sebaseum at regio fasialis, and fasial angiofibroma. Dental enemal pits. Detailed CNS examination revealed increase deep tendon of both upper and lower limbs reflexes and bilateral positive Babinski's sign. Other systemic examination and fundus examination revealed no abnormality Investigations showed sub ependymal nodules and Cortical glioneuronal tubers in computed tomography (CT) scan of head and show other test like haemoglobin, complete blood count, renal and liver function tests were normal. During the hospital stay the dose of sodium valproate was increased after which the seizure frequency decreased though he had short seizures in-between.

Conclusion: The prognosis of TSC depends on the severity or multiplicity of organ involvement. About a quarter of severely affected infants are thought to die before the age of 10% and 75% before 25 years. However, in the case of individuals diagnosed late in life with few cutaneous signs, prognosis depends on the associated internal tumors and cerebral calcifications.

Biography

Hastuti Sri is a Lecturer and Neurologist in Aceh Province, Indonesia. She worked as Faculty of Medicine at Syiah Juala Universit. She completed her Specialist Education at the University of Indonesia. She worked as a Neuropediatric Fellow at University Malaya Medical Center, Kuala Lumpur. She is a Staff of Neurology Department, Zainoel Abidin Hospital, Banda Aceh, Indonesia, Staff of Neuropediatric Division in Neurology Department, Zainoel Abidin Hospital, Banda Aceh Indonesia and Lecturer of Medical Faculty, Syiah Kuala University, Banda Aceh, Indonesia.

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