Luaran Anak Laki-Laki dengan Hipotiroid Kongenital, Anemia Defisiensi Besi, Dermatitis Atopik, Disabilitas Intelektual, *Severely Stunted*

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Abstrak

Hipotiroidisme kongenital merupakan penyebab penting stunting karena hormon tiroid mengatur pertumbuhan linier dan perkembangan neurokognitif di awal kehidupan. Tanpa diagnosis dan pengobatan yang cepat, hipotiroidisme kongenital akan menyebabkan gangguan pertumbuhan fisik dan keterbelakangan mental. Kami melaporkan pasien dengan diagnosis hipotiroidisme kongenital. Seorang anak laki-laki berusia 9 tahun dirujuk dengan keluhan perawakan sangat pendek. Anak tampak sangat pendek (HAZ -8.60) dan mengalami kesulitan belajar di sekolah. Pada pemeriksaan fisik anak tampak kurang aktif, pucat, bengkak, rambut jarang, makroglosia, dan kulit tampak kering. Dia juga mengalami obesitas. Pemeriksaan laboratorium menunjukkan kadar T4 bebas rendah (0,43 ng/dl), TSH tinggi (>100 uIU/ml) dan anemia defisiensi besi. Ultrasonografi tiroid menunjukkan hipoplasia tiroid. Usia tulangnya setara dengan bayi berusia 6 bulan. Dia memiliki disabilitas intelektual, Intelligent Quotient (IQ)-nya 40, sedangkan Social Quotient adaptifnya adalah 42,25. Pasien didiagnosis sebagai hipotiroidisme kongenital primer dan anemia defisiensi besi. Dia diberi levothyroxine, suplementasi zat besi dan asam folat. Setelah 17 bulan masa tindak lanjut, pasien menunjukkan perbaikan klinis yang signifikan, kecuali fungsi kognitifnya. Tingginya bertambah 16,2 cm, kecerdasan sosial adaptif dan kualitas hidupnya meningkat. Namun, IQ-nya relatif sama.

Kata kunci: hipotiroidisme kongenital; keterbelakangan mental; stunting

Outcome of Patient with Delayed Diagnosis Congenital Hypothyroidism : A Case Report

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Abstract

Congenital hypothyroidism is an important cause of stunting because thyroid hormone regulates linear growth and neurocognitive development early in life. Without prompt diagnoses and treatment, congenital hypothyroidism will lead to impaired physical growth and mental retardation. We reported outcome of delayed diagnosis congenital hypothyroidism. A 9 years old boy was referred with complaints of a very short stature. The child was very short (HAZ 8.60) and had severe learning difficulty at school. On physical examination, the child looked less active, pale, puffy, sparse hair, macroglossia, and his skin looked dry. He was also obese. Laboratory testing showed low free T4 level (0.43 ng/dl), high TSH (>100 uIU/ml) and iron deficiency anemia. Thyroid ultrasonography showed thyroid hypoplasia. His bone age was equivalent to that of a 6 months old infant. He had intellectual disability, his Intelligent Quotient (IQ) was 40, while his adaptive Social Quotient was 42.25. The patient was diagnosed as primary congenital hypothyroidism and iron deficiency anemia. He was given levothyroxine, iron supplementation and folic acid. After 17 months of follow up, the patient showed significant clinical improvement, except for his cognitive function. He gained 16.2 cm in height, his adaptive social quotient and quality of life improved. However, his IQ was relatively the same.

Keywords: congenital hypothyroidism; mental retardation; stunting