



KASUS LONGITUDINAL SINDROM BECKWITH-WIEDEMANN PADA ANAK: KOMPLEKSITAS KLINIS DAN PERAN PENDEKATAN MULTIDISIPLIN

INTISARI:

LatarBelakang: Sindrom Beckwith–Wiedemann (Beckwith–Wiedemann Syndrome/BWS) merupakan kelainan overgrowth genetik akibat gangguan regulasi epigenetik pada kromosom 11p15.5 yang ditandai oleh variasi manifestasi klinis luas serta peningkatan risiko tumor embrional. Heterogenitas fenotipik menyebabkan diagnosis sering menjadi tantangan klinis dan membutuhkan evaluasi komprehensif serta pendekatan multidisiplin dalam tata laksana jangka panjang.

Presentasi Kasus: Dilaporkan seorang anak perempuan usia 17 bulan dengan keluhan utama makroglosia sejak lahir. Pasien lahir post-term dengan makrosomia dan mengalami hipoglikemia neonatal yang memerlukan perawatan di NICU. Pemeriksaan klinis menunjukkan makroglosia, pertumbuhan lateral asimetris, dan riwayat hipoglikemia dengan skor klinis BWS sebesar 5 sehingga diagnosis ditegakkan secara klinis. Evaluasi lanjutan menemukan hipertrofi ventrikel kiri, gangguan bahasa ekspresif-reseptif, obesitas, serta defisiensi vitamin D. Pasien mengalami kesulitan menyusu pada awal kehidupan dan menjalani reduksi lidah bertahap dengan tindakan trakeostomi untuk mempertahankan jalan napas. Pemantauan longitudinal dilakukan melalui pendekatan multidisiplin meliputi nutrisi anak, kardiologi, bedah plastik, tumbuh kembang, terapi wicara, dokter gigi anak, serta surveilans tumor berkala. Faktor keterbatasan ekonomi dan kurangnya dukungan ayah biologis menjadi tantangan tambahan dalam kepatuhan perawatan jangka panjang.

Kesimpulan: Kasus ini menegaskan bahwa BWS merupakan kondisi multisistem yang memerlukan deteksi dini, pemantauan komplikasi berkelanjutan, serta koordinasi tata laksana multidisiplin. Pendekatan family-centered care dan dukungan sosial yang adekuat berperan penting dalam mengoptimalkan pertumbuhan, perkembangan, serta luaran klinis jangka panjang pasien dengan BWS.

Kata kunci: Beckwith–Wiedemann syndrome, makroglosia, pendekatan multidisiplin, keterlambatan perkembangan, laporan kasus anak



LONG CASE REPORT

BECKWITH-WIEDEMANN SYNDROME IN CHILDREN: CLINICAL COMPLEXITY AND THE ROLE OF A MULTIDISCIPLINARY APPROACH

ABSTRACT

Background: Beckwith–Wiedemann Syndrome (BWS) is a genetic overgrowth disorder caused by epigenetic dysregulation at chromosome 11p15.5 and characterized by heterogeneous clinical manifestations along with an increased risk of embryonal tumors. The wide phenotypic variability often complicates diagnosis and necessitates comprehensive evaluation and long-term multidisciplinary management.

Case Presentation: We report a 17-month-old female presenting with macroglossia since birth. The patient was born post-term with macrosomia and developed neonatal hypoglycemia requiring neonatal intensive care admission. Clinical findings included macroglossia, lateralized growth, and a history of neonatal hypoglycemia, resulting in a Beckwith–Wiedemann clinical score of 5, establishing the diagnosis clinically. Further evaluation revealed left ventricular hypertrophy, expressive–receptive language disorder, normoheight obesity, and vitamin D deficiency. Early feeding difficulties were observed, and staged tongue-reduction surgeries with temporary tracheostomy were performed to maintain airway patency. Longitudinal follow-up involved multidisciplinary management including pediatric nutrition, cardiology, plastic surgery, developmental monitoring, speech therapy, dentist, and routine tumor surveillance. Limited socioeconomic resources and absence of paternal support represented additional challenges affecting long-term care adherence.

Conclusion: This case highlights BWS as a multisystem disorder requiring early recognition, continuous complication surveillance, and coordinated multidisciplinary care. A family-centered approach and adequate social support are essential to optimize growth, developmental outcomes, and long-term prognosis in children with BWS.

Keywords: Beckwith–Wiedemann syndrome, macroglossia, multidisciplinary care, developmental delay, pediatric case report