



Luaran Anak dengan Hiperplasia Adrenal Kongenital, Epilepsi dan Gangguan Bahasa Ekspresif

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Abstrak

Latar Belakang

Hiperplasia adrenal kongenital (HAK) merupakan kondisi defisiensi enzim yang diperlukan untuk steroidogenesis, sehingga menyebabkan defisiensi kortisol dan aldosteron, serta peningkatan androgen. Peningkatan androgen berhubungan dengan keterlambatan maturasi neuron yang mengganggu perkembangan kognitif, salah satunya kemampuan berbahasa. Beberapa pasien HAK mengalami epilepsi, terkait kondisi krisis adrenal di awal kehidupan serta tingginya paparan *corticotropin-releasing factor* sejak intrauterin.

Tujuan

Studi ini bertujuan untuk mengamati luaran pasien HAK selama 12 bulan, meliputi komplikasi, kepatuhan terapi, perkembangan neurodevelopmental, dan kualitas hidup.

Metode

Studi observasional dilakukan selama 12 bulan di RSUP Dr.Sardjito pada seorang perempuan 2 tahun, dengan diagnosis HAK, epilepsi general tonik-klonik, dan gangguan bahasa ekspresif.

Hasil

Studi ini mendapatkan perbaikan pada perkembangan bahasa setelah dilakukan terapi wicara rutin dan partisipasi di sekolah. Akan tetapi, krisis adrenal dan kejang masih berulang sekali selama perawatan, dicetuskan oleh gastroenteritis. Laju pertumbuhan BB dan TB normal sesuai usia. Kepatuhan terapi dan kualitas hidup pasien tercapai optimal selama pengamatan.

Kesimpulan

Kasus menarik ini menunjukkan korelasi HAK dengan luaran neurodevelopmental. Tata laksana yang sesuai dan konsisten diperlukan untuk mencapai luaran kualitas hidup yang optimal.

Kata kunci

Hiperplasia adrenal kongenital, epilepsi, gangguan bahasa ekspresif, luaran, anak

Child Outcomes with Congenital Adrenal Hyperplasia, Epilepsy, and Expressive Language Disorder

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Abstract

Introduction

Congenital adrenal hyperplasia (CAH) characterized by a deficiency in enzymes required for steroid biosynthesis, leads to insufficient production of cortisol and aldosterone, along with an increase in androgen. Elevated androgen levels may delay the maturation of nerve cells, potentially impacting cognitive development, including language skills. Some CAH patients also develop epilepsy as sequelae of early adrenal crises or exposure to *corticotropin-releasing factor* since fetal development.

Objectives

This study aims to monitor outcomes in patients with CAH over a 12-month period, including complication, treatment compliance, neurodevelopmental outcomes, and quality of life.

Methods

We conducted a 12-month observational study in Dr. Sardjito General Hospital of two-year-old female patient diagnosed with CAH, generalized tonic-clonic epilepsy, and expressive language disorder.

Results

The study found that regular speech therapy and school participation led to improvements in language development. However, there was one episode of seizures and an adrenal crisis during observation, triggered by gastroenteritis. The patient's weight and height progressed within the normal range. Furthermore, both treatment adherence and the patient's quality of life reached optimal levels within observation.

Conclusion

This interesting case showed relation between CAH and neurodevelopmental outcomes. Appropriate and consistent management can lead to an optimal quality of life for patients with CAH.

Keywords

Congenital adrenal hyperplasia, epilepsy, expressive language disorder, outcome, child