



INTISARI

Latar belakang

Hiperplasia adrenal kongenital (HAK) merupakan kelainan genetik akibat gangguan pada salah satu dari 5 tahap enzimatis yang diperlukan untuk biosintesis steroid di kelenjar adrenal, menyebabkan defisiensi hormon kortisol dan aldosteron serta peningkatan hormon androgen sebagai bentuk kompensasi.¹ Secara umum, HAK adalah penyebab tersering genital ambigu pada neonatus dengan angka kejadian sebesar 1:10.000 sampai 1:15.000

Kasus:

Seorang anak laki-laki, usia 3 bulan dirujuk RSUD Kebumen karena hiponatremia berat dan pasien selalu muntah setiap menetek. Gejala muntah setiap habis menetek sekitar 10-20 cc warna kuning dimulai sejak usia 20 hari yang semakin berat saat anak usia 3 bulan disertai berak cair 3x per hari tanpa disertai lendir, darah dan pasien mulai malas menetek. Anak tampak lemah, mata cekung dan cenderung tidur. Hasil pemeriksaan laboratorium saat itu menunjukkan jumlah leukosit 8.090/ μ L, jumlah trombosit 279.000/ μ L, kadar hemoglobin 12,3 g/dL, hematokrit 35%, kadar natrium 105 mmol/L, kalium 6.15 mmol/L, clorida 97 mmol/L, sedangkan hasil urin rutin dalam batas normal. Pasien didiagnosis gastroenteritis akut oleh karena alergi susu sapi dengan dehidrasi tak berat, dengan komplikasi hiponatremia berat dan hiperkalemia. sehingga dipikirkan kemungkinan terjadi Hiperplasi Adrenal Kongenital (HAK) dengan diagnosis banding Asidosis tubular renal.

Kesimpulan: Muntah berulang, tampak lemah dan hipnatremia berat pada anak diawal kehidupan dapat disebabkan oleh hyperplasia adrenal kongenital dengan faktor risiko tertentu. Kombinasi hidrolortison dan fludrokortison dengan intervensi multidisiplin dapat membuat prognosis anak dengan HAK menjadi lebih baik.

Kata kunci: HAK, Hiponatremia, Muntah berulang

ABSTRACT

Background

Congenital adrenal hyperplasia (CAH) is a genetic disorder caused by a disturbance in one of the 5 enzymatic stages required for steroid biosynthesis in the adrenal glands, causing a deficiency of the hormones cortisol and aldosterone and an increase in androgen hormones as a form of compensation. 1 In general, CAH is the most common cause of genital ambiguous in neonates with an incidence rate of 1:10,000 to 1:15,000.

Cases Presentation

A boy, aged 3 months was referred to the Kebumen Hospital because of severe hyponatremia and the patient always vomited every time he suckled. Symptoms of vomiting every time after sucking about 10-20 cc of yellow color begin at the age of 20 days which get worse when a child aged 3 months is accompanied by watery stools 3x per day without mucus, blood and the patient begins to be lazy to suckle. Children look weak, sunken eyes and tend to sleep. Laboratory test results at that time showed a leukocyte count of 8.090/ μ L, a platelet count of 279.000/ μ L, a hemoglobin level of 12.3 g/dL, a hematocrit of 35%, a sodium level of 105 mmol/L, a potassium level of 6.15 mmol/L, a chloride of 97 mmol/L. L, while routine urine results were within normal limits. The patient was diagnosed with acute gastroenteritis due to cow's milk allergy with mild dehydration, complicated by severe hyponatremia and hyperkalemia. so that the possibility of congenital adrenal hyperplasia (CAH) is considered with the differential diagnosis of renal tubular acidosis.

Conclusion

Recurrent vomiting, weakness and severe hyponatremia in early life may be due to congenital adrenal hyperplasia with certain risk factors. The combination of hydrocortisone and fludrocortisone with multidisciplinary interventions can improve the prognosis of children with CAH.

Keywords

HAK, Hyponatremia, Recurrent vomited