INTISARI

Latar belakang

Sindrom Bartter adalah kelainan bawaan pada tubular ginjal yang diwariskan secara resesif autosom dan merupakan kelainan yang heterogen secara genetik dan jarang ditemukan. Sindrom ini pertama kali dijelaskan oleh Federic Bartter pada tahun 1962 sebagai kombinasi dari hiperplasia kompleks juxtaglomerular, hiperaldosteronisme, dan alkalosis metabolik dengan hipokalemia. Insiden Sindrom Bartter sampai saat ini tidak diketahui secara pasti, tetapi diperkirakan insidennya sebesar 1,2/1.000.000 kelahiran di dunia.[1,2,3]

Anak-anak dengan Sindrom Bartter dapat bertahan hidup sampai usia dewasa namun data komprehensif terkait luaran jangka panjang pada populasi ini terbatas.[3,4] Meskipun pemeriksaan genetik telah meningkat, penegakan diagnosis Sindrom Bartter masih sangat bergantung pada gambaran klinis. Gambaran klinis polihidramnion idiopatik awitan dini dan gagal tumbuh intrauterine harus dipertimbangkan sebagai Sindrom Bartter neonatal. Kemudian adanya gagal tumbuh, polidipsi, dan poliuria pada anak-anak usia dini merupakan ciri khas Sindrom Bartter klasik.[4,5,6]

Kasus

Pemantauan jangka panjang dan intervensi multidisiplin dilakukan terhadap anak laki-laki berusia 4 tahun 4 bulan dengan diagnosis *syndrome of inappropriate antidiuretic hormone secretion* (SIADH) DD/ *cerebral salt wasting syndrome* (CSWS) karena terdapat poliuria dan hiponatremia. Penelitian ini mengamati luaran jangka panjang anak dengan Sindrom Bartter selama 12 bulan. Kemudian tujuan khusus kasus panjang ini antara lain mengamati gangguan elektrolit, kepatuhan terapi, tuli sensorineural, status pertumbuhan dan perkembangan dengan target tidak terjadi gangguan metabolik, epilepsi terkontrol, kualitas hidup anak meningkat, tumbuh kembang anak dapat optimal sesuai dengan potensi genetiknya.

Selama 12 bulan pengamatan dan intervensi telah dilakukan secara prospektif pada seorang anak laki-laki dengan Sindrom Bartter, Disabilitas Intelektual, Gangguan Spektrum Autisme (GSA), epilepsi belum terkontrol dan *stunted*. Pendekatan multidisiplin yang telah diterapkan meliputi terapi farmakologi, pemantauan gangguan elektrolit, pencegahan komplikasi, rehabilitasi medik, pendampingan psikolog anak, pemantauan tumbuh kembang anak serta efek samping obat.

Selama pengamatan longitudinal, anak mendapatkan luaran yang baik yaitu epilepsi berhasil terkontrol, fungsi adaptif membaik, keparahan klinis GSA membaik, tuli sensorineural dan *stunted* teratasi sehingga kualitas hidup anak membaik. Namun terdapat luaran yang belum dicapai, yaitu tidak terjadi nefrokalsinosis berdasarkan hasil USG renal.

Kesimpulan

Sindrom Bartter merupakan kelainan bawaan pada tubular ginjal yang diwariskan dan kombinasi dari hiperplasia kompleks juxtaglomerular, hiperaldosteronisme, dan alkalosis metabolik dengan hipokalemia yang menyebabkan berbagai komplikasi pada anak. Pendekatan multidisiplin yang telah diterapkan meliputi terapi farmakologi, pemantauan gangguan elektrolit, pencegahan komplikasi, rehabilitasi medik, pendampingan psikolog anak, pemantauan tumbuh kembang anak serta efek samping obat dapat memperbaiki keluaran perkembangan pasien.

Kata kunci

Sindrom Bartter, disabilitas intelektual, gangguan spektrum autisme, epilepsi, luaran

ABSTRACT

Background

Bartter syndrome is a congenital renal tubular disorder that is inherited in an autosomal recessive manner and is a genetically heterogeneous and rare disorder. This syndrome was first described by Federic Bartter in 1962 as a combination of juxtaglomerular complex hyperplasia, hyperaldosteronism, and metabolic alkalosis with hypokalemia. The incidence of Bartter Syndrome is currently not known with certainty, but it is estimated that the incidence is 1.2/1,000,000 births in the world.[1,2,3]

Children with Bartter Syndrome can survive into adulthood but comprehensive data regarding long-term outcomes in this population are limited.[3,4] Despite improvements in genetic testing, the diagnosis of Bartter Syndrome still relies heavily on the clinical picture. The clinical features of early-onset idiopathic polyhydramnios and intrauterine failure to thrive should be considered neonatal Bartter Syndrome. Then the presence of failure to thrive, polydipsia, and polyuria in early childhood are characteristic of classic Bartter Syndrome.[4,5,6]

Case

Long-term monitoring and multidisciplinary intervention were carried out on a boy aged 4 years 4 months with a diagnosis of syndrome of inappropriate antidiuretic hormone secretion (SIADH) DD/ cerebral salt wasting syndrome (CSWS) due to polyuria and hyponatremia. This study looked at the long-term outcomes of children with Bartter Syndrome for 12 months. Then the specific objectives of this long case include observing electrolyte disturbances, therapy compliance, sensorineural deafness, growth and development status with the target of no metabolic disorders, controlled epilepsy, improved child quality of life, optimal child growth and development in accordance with their genetic potential.

For 12 months observation and intervention were carried out prospectively on a boy with Bartter Syndrome, Intellectual Disability, Autism Spectrum Disorder (ASD), uncontrolled epilepsy and stunted. The multidisciplinary approach that has been implemented includes pharmacological therapy, monitoring electrolyte disorders, preventing complications, medical rehabilitation, assistance from child psychologists, monitoring child growth and development and drug side effects.

During longitudinal observations, children had good outcomes, namely epilepsy was successfully controlled, adaptive function improved, the clinical severity of ASD improved, sensorineural deafness and stunting resolved so that the child's quality of life improved. However, there are outcomes that have not been achieved, namely that nephrocalcinosis does not occur based on the results of renal ultrasound.

Conclusion

Bartter syndrome is a congenital renal tubular disorder that is inherited and a combination of juxtaglomerular complex hyperplasia, hyperaldosteronism, and metabolic alkalosis with hypokalemia that causes various complications in children. The multidisciplinary approach that has been implemented includes pharmacological therapy, monitoring electrolyte disorders, preventing complications, medical rehabilitation, child psychologist assistance, monitoring child growth and development and drug side effects can improve patient development outcomes.

Keywords

Bartter syndrome, intellectual disability, autism spectrum disorder, epilepsy, outcome