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Congenital nephrogenic diabetes insipidus with basal ganglia calcifications - A case report

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Abstract

A male toddler aged 33 months presented with recurrent episodes of fever, polyuria, polydipsia, dehydration, excessive cry, seizures and failure to thrive since early infancy. Developmental milestones were normal. Clinical examination was essentially normal except for malnutrition (IAP Grade 2). Investigations revealed water deprivation test positive for Nephrogenic Diabetes Insipidus. Serum ADH level was raised at 20.76 pmol/l (N -0.00-13.00 pmol/l). Renal function tests and other biochemical parameters were normal except for persistent hypernatremia ((155-168mEq/L). Ultrasound abdomen did not reveal any abnormality. Computed tomography of the head showed bilateral basal ganglia calcification. He was treated with hydrochlorothiazide (2.0 mg/kg/d) and amiloride (0.2mg/kg/d) along with a salt and protein restricted diet. Following treatment there was a marked improvement in his condition as evidenced by reduction of polyuria by 40%, normalizing of serum sodium state and a steady weight gain of 2.0kg over a period of six months. This child had a pathognomic presentation of Congenital Nephrogenic Diabetes Insipidus but the condition escaped detection due to a multitude of common symptoms as well as seizures. We therefore report a rare entity of a CNDI with bilateral basal ganglia calcifications.

Keywords: Polyuria, Polydipsia, Seizures, Basal Ganglia Calcification, Nephrogenic Diabetes Insipidus

Introduction

Congenital Nephrogenic Diabetes Insipidus (CNDI) is a hypotonic polyuric state which manifests in the neonatal period but is usually detected later. Resulting from renal insensitivity to arginine vasopressin (AVP), it is a rare disorder with common symptomatology making it an often missed condition.(1) We report a rare entity of a case of CNDI which was associated with seizures and bilateral basal ganglia calcifications.

Case Report

A male child aged 33 months, the product of a non consanguineous marriage was hospitalised with history of recurrent episodes of fever, dehydration, excessive cry and seizures since early infancy. He presented with fever, polyuria (urine output >4000ml/day), polydipsia (water intake>5000ml/day), poor feeding, dehydration and failure to thrive. The perinatal period, family history and developmental milestones were normal. The child was alert, pyrexic (99.5-101^{0}F) and dehydrated. His height was below 3rd percentile and he had malnutrition (IAP Grade 2). Systemic examination revealed a normal cardiovascular and respiratory examination. There was no neurological deficit and abdominal examination did not reveal any organomegaly.

Investigations revealed water deprivation test positive for Nephrogenic Diabetes Insipidus. Serum ADH level was raised at 20.76 pmol/l (N -0.00-13.00 pmol/l). Renal function tests and other biochemical parameters were normal except for persistently raised serum sodium levels (155-168mEq/L). Ultrasound abdomen was normal. Computed tomography of the head showed bilateral basal ganglia calcifications (Fig. 1). In view of clinical features of polyuria, polydipsia, dehydration, persistent hypernatremia, positive water deprivation test and raised serum ADH levels, symptomatic since infancy, a diagnosis of CNDI was made.

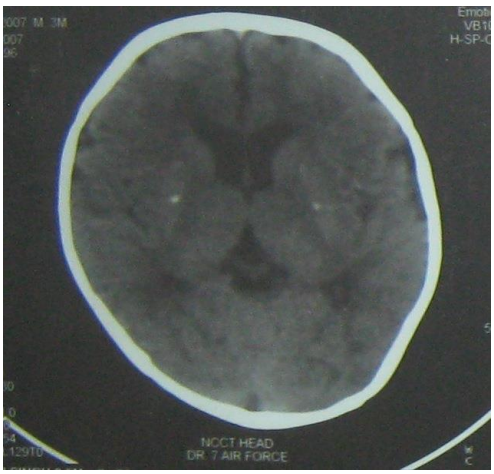

Fig. 1: NCCT Brain

He was started on therapy with hydrochlorothiazide (2.0 mg/kg/d), amiloride (0.2mg/kg/d) and a salt and protein restricted diet. He showed marked improvement as evidenced by reduction of polyuria by 40%, normalizing of serum sodium state and a steady weight gain of 2.0kg over a period of six months.

Discussion

CNDI, a rare disorder though manifesting in the early neonatal period is often diagnosed late due to common symptomatology. It is commonly inherited as an X-linked recessive trait that is caused by V2R mutation, and occasionally as an autosomal recessive form due to mutation in AQP2 water channel gene.[2] The mutations in the V2 receptor gene or the AQP-2 water channel gene can induce resistance to AVP which is a pathophysiologic characteristic of NDI.[3]

Usually clinical symptoms begin within the first week of life but are unrecognized. Later recurrent episodes of severe hypernatremia due to dehydration, and non-specific symptoms such as anorexia, nausea, and fever occur as in this case. Unless recognized and treated appropriately, the recurrent episodes of dehydration can lead to growth disturbance and mental retardation .Intracranial calcification and seizures have been reported in a few cases of CNDI particularly during infancy.[4-6] The hypothesis suggested is that CNDI leads to recurrent hyperosmolar dehydration which damages the endothelial cells and calcium phosphate and other substances are deposited within or around the walls of small vessels.[7] The degree of psychomotor retardation is said to be proportional to the degree of calcification. Our child had a normal developmental assessment, which corroborated with the mild calcification seen on the computed tomography. Growth retardation as present in our index case is frequently noted finding in children with NDI. This is probably related to caloric deprivation or may be an inherent character of the disease.[8,9] Treatment focuses on the reduction of polyuria and hypernatremia to avoid dehydration and thereby preventing complications. Low sodium diet is recommended to reduce the osmotic load that the kidney has to excrete in addition to liberal fluid intake, particularly during febrile illness. Hydrochlorothiazide alone or in combination with indomethacin was used earlier for therapy of NDI. Hypokalemia, renal, hemopoietic and gastro-intestinal complications were seen with these drugs. Long term treatment with hydrochlorothiazide and amiloride has resulted in normal growth and mental development along with better tolerance.[7,10]

Conclusion

Nephrogenic Diabetes Insipidus in childhood is a rare entity. This child had a pathognomic presentation but the condition escaped detection due to a multitude of common symptoms. An interesting and rather unusual feature in this child was the presentation with seizures and the basal ganglia calcifications. Appropriate management provided gratifying results.

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