

Association of Mucopolysaccharidosis Type 4A and Bartter Syndrome

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Keywords.

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A 6-year-old Syrian boy presented with complaints of facial dysmorphism and difficulty of walking. He had coarse face, macrocephaly, pectus carinatum, x-bain deformity, kyphosis, corneal clouding, and claw hand deformity. Galactose-6 sulphatase enzyme level was 0.1 nmol/mg.17 h (reference range, > 68 nmol/mg.17 h), compatible with Morquio syndrome. On laboratory examinations, potassium level was 2.9 mmol/L (reference range, 3.5 mmol/L to 5.1 mmol/L), sodium level was 130 mmol/L (reference range, 135 mmol/L to 148 mmol/L), and chloride level was 92 mmol/L (reference range, 101 mmol/L to 109 mmol/L). Blood pH was 7.5 and bicarbonate level was 31 mEq/L. Urine sodium and chloride levels were high. Arterial blood pressure was normal and these findings were consistent with Bartter syndrome. This is the first report of a patient with the association of Bartter syndrome and mucopolysaccharidosis type 4A, which was thought to be coincidental.

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INTRODUCTION

Mucopolysaccharidosis (MPS) type 4A (Morquio syndrome) is a rare autosomal recessive lysosomal storage disease caused by the deficiency of N-acetyl-galactosamine-6-sulfatase (Galactose-6 sulphatase). The clinical manifestations are muscular atrophy, pectus carinatum, odontoid hypoplasia, short stature, dorsilumbar kyphoscoliosis, corneal opacity, deafness, hypermobility of joints, cardiac abnormalities, quadriplegia, and respiratory paralysis.¹ Bartter syndrome is a rare autosomal recessive disorder caused by a defect in one of the transporters involved in distal tubule transport of sodium and chloride.² It is characterized by hyperreninemia, hiperaldosteronism, hypokalemia, metabolic alkalosis, and normal blood pressure. Small children typically present with growth failure, muscle weakness, constipation, polyuria, and dehydration.³ Morquio syndrome and also Bartter syndrome have been associated with various disorders. Here, we present the first patient with the

association of Bartter syndrome and MPS type 4A.

CASE REPORT

A 6-year- and 3-month-old Syrian boy presented with complaints of facial dysmorphism and difficulty of walking. He was born after an uneventful pregnancy by spontaneous vaginal delivery. He had an elder brother with similar findings. His weight was 9.6 kg (below the 3rd percentile) and his height was 71 cm (below the 3rd percentile). He had coarse face, macrocephaly, pectus carinatum, x-bain deformity, kyphosis, corneal clouding, and claw hand deformity. He had the findings of dysostosis multiplex in direct radiographies.

The patient was initially thought to have mucopolysaccharidosis and lysosomal enzyme levels were studied. Galactose-6 sulphatase enzyme level was 0.1 nmol/mg.17 h (reference range, > 68 nmol/mg.17 h), compatible with Morquio syndrome. He had a borderline intelligence level

in his intelligence quotient test.

On laboratory examinations, potassium level was 2.9 mmol/L (reference range, 3.5 mmol/L to 5.1 mmol/L), sodium level was 130 mmol/L (reference range, 135 mmol/L to 148 mmol/L), and chloride level was 92 mmol/L (reference range, 101 mmol/L to 109 mmol/L). Blood pH was 7.5 and bicarbonate level was 31 mEq/L. Bartter syndrome was considered in this patient because of metabolic alkalosis, hypopotassemia and hyponatremia. Serum calcium, alkaline phosphatase and magnesium levels were within normal limits. Urine sodium level was 151.6 mg/dL (reference range, 27 mg/dL to 147 mg/dL), chloride level was 213 (reference range, 73 mg/dL to 167 mg/dL). Renal ultrasonography was normal and arterial blood pressure values were within normal limits. Renin level was 60 ng/mL/h (reference range, < 5 ng/mL/h) and aldosterone level was 242 mg/dL (reference range, 3.7 mg/dL to 31 mg/dL), consistent with Bartter syndrome. Indomethacin and oral potassium supplementation were started. Serum electrolytes were normal after treatment. Molecular genetic analysis was not performed because of familial reasons. This case report was written after receiving informed consent from the family.

DISCUSSION

Lysosomes are very important organelles for malignant cells because of their carcinogenic effects. This situation occurs due to complex metabolic pathways which involve lysosomal proteases and various proteins.⁴ Lysosomal storage diseases are a rare group of inborn errors of metabolism that result from lysosomal dysfunction. Mucopolysaccharidoses are a group of lysosomal storage diseases that are important for cancer formation and metastasis of malignancies with epithelial origin. Morquio syndrome has been associated with bone cancer and advanced stage gastric carcinoma.⁵

Both of Morquio and Bartter syndrome can rarely be seen with other disorders. Pseudotumor cerebri is rarely coexist with Morquio syndrome. Chan reported Morquio syndrome and pseudotumor cerebri in a female with reduction of vision.⁶

Ertekin and colleagues reported Bartter syndrome with empty sella in a 12-month-old boy who presented with gastrointestinal symptoms and edema.⁷ Also, Bartter syndrome has been associated

with vasculitis and periodic paralysis.^{8,9}

A search of the literature revealed no case of Morquio syndrome coexist with Bartter syndrome. It is interesting to see that two different autosomal recessive diseases together. However, this is thought to be coincidental.

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CONFLICT OF INTEREST

None declared.

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