Alder–Reilly Anomaly in Hurler’s Syndrome in a Neonate: A Rare Case Report

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Abstract A 22-days-old male newborn baby presented with persistence of neonatal jaundice since birth. On clinical examination he had coarse facial features, a prominent forehead, enlarged tongue, icterus, hepatosplenomegaly, skeletal deformities and bilateral inguinal hernia. On investigation the peripheral smear revealed Alder–Reilly anomaly in the neutrophils suggesting mucopolysaccharidosis. Mucopolysaccharide excretion spot test of the urine was positive; and an assay for glycosaminoglycans in the urine was also high, which confirmed the clinical diagnosis of Hurler’s syndrome. We present this rare case to highlight the association of Alder–Reilly anomaly and bilateral inguinal hernia in Hurler’s syndrome even in neonates.

Keywords Alder–Reilly anomaly · Bilateral inguinal hernia · Mucopolysaccharidosis (MPS) type 1 · Hurler’s syndrome

Introduction

Hurler’s syndrome is an autosomal recessive disorder of mucopolysaccharide metabolism caused by a deficiency of the enzyme α -L-iduronidase [1]. The inability to degrade these macromolecules, which are ubiquitous, results in their storage in a variety of tissues including the liver, spleen, heart, connective tissue and others, resulting in premature death, usually by 10 years of age. Hurler’s syndrome represents the classical prototype of a mucopolysaccharide disorder, with a very low prevalence of 1:150,000 births [2]. The diagnosis is usually made between the age of 6 and 24 months with evidence of coarse facial features, prominent forehead with large tongue, hepatosplenomegaly, corneal cloudiness, skeletal deformities, joint stiffness, short stature, acquired cardiomyopathy, and progressive lenticular enlargement with increased intracranial pressure caused by communicating hydrocephalus. We present an extremely rare case of Hurler syndrome associated with Alder–Reilly anomaly in the granulocytes and bilateral inguinal hernia in a neonate.

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Case Report

A 22-days-old male newborn baby was admitted to the newborn unit of Department of Pediatrics, S.C.B. Medical College, Cuttack, Orissa, India with complaints of persistence of jaundice since birth. The antenatal and natal histories were non-contributory. This baby was born of a non-consanguineous marriage and was the second child of the couple. There was no family history of a similar disorder or neonatal deaths and miscarriages.

On general examination the newborn baby had coarse facial features, a prominent forehead, enlarged tongue,

icterus, hepatosplenomegaly, and bilateral inguinal hernia (Fig. 1). The respiratory and cardiovascular systems were normal. Chest radiograph revealed oar-shaped ribs (Fig. 2).

Hematological examination revealed normocytic normochromic anemia in RBC series. The WBC series showed prominent cytoplasmic granules called Alder–Reilly anomaly suggesting Hurler syndrome (Fig. 3). The biochemical profile showed raised conjugated bilirubin (3.11 mg%) and raised enzyme markers (SGPT-270 IU/L, SGOT-470 IU/L, Alkaline phosphatase-1,316 IU/L) suggesting neonate cholestasis. Routine urine examination was normal, but a spot test for mucopolysaccharide excretion in urine was positive. Urine assay for glycosaminoglycans showed a high value of 5 mg/mmols.

Fig. 1 Neonate with coarse facial features and bilateral inguinal hernia

Fig. 2 Chest radiograph showing oar-shaped ribs

Fig. 3 Peripheral blood smear showing Alder–Reilly anomaly in the neutrophils. ($\times 400$ Leishman stain)

Discussion

Hurler's syndrome is an autosomal recessive disorder that has been described in families with a history of consanguinity [1]. Our patient was born of a nonconsanguineous marriage and without a family history of similar disease in the past two generations. The usual age of presentation is between 6 and 24 months. However, it was observed at 22 days in our case.

Besides the classical clinical features, the association of bilateral inguinal hernia with Hurler syndrome in a neonate, a rare event was also observed in our case. Infants with the hurler phenotype usually appear to be normal at birth, although very few cases, have inguinal or umbilical hernia [3]. Our patient had most of the features of Hurler's syndrome except for its cardiovascular manifestation and corneal clouding. The diagnosis was confirmed by a urine mucopolysaccharide excretion spot test.

The peripheral blood smear evaluation of excessive granulation of circulating neutrophils help distinguish several of the storage diseases. Alder–Reilly anomaly is one such condition. Alder's anomaly was first described by Alder in 1939 and by Reilly in 1941; accordingly, it is known also as Alder–Reilly anomaly [4]. It is an autosomal recessive inherited disorder characterized by the presence of large azurophilic and basophilic granules in cells of the myeloid and lymphocytic series.

Hematopoietic stem cell transplantation is the treatment of choice for a child younger than 2 years of age suffering from Hurler's syndrome with no or minimal central nervous system disease. Enzyme replacement therapy with α -L-iduronidase is recommended for mild to moderate disease and for patients with neurological impairment [5]. To prevent mental retardation, a bone marrow transplant probably needs to be performed at a very early age. As

bone marrow transplantation and enzyme replacement therapy were not possible in our set up, we advised regular follow-up and symptomatic treatment.

Conclusion

Advances in stem cell transplantation and the use of recombinant enzymes necessitates early diagnosis in newborns with Hurler's syndrome before the onset of irreversible tissue and organ damage.

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