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Case Report

Angiokeratoma in Hurler syndrome: a rare pediatric case report

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ABSTRACT

Mucopolysaccharidosis type I (MPS I H, Hurler syndrome) is caused by alpha-L-iduronidase deficiency, leading to glycosaminoglycan (GAG) buildup and severe cognitive and physical impairments, typically presents with coarse facial features and usually manifests as cognitive developmental delay, corneal clouding and characteristic musculoskeletal manifestations. An interesting case of a three-year-old girl with a combination of skeletal, neurological, ophthalmologic, and radiological findings along with Mongolian spots present on back and widespread asymptomatic small, multiple, red skin lesions with rapid progression in the number and size of the lesions with MPS I- (Hurler syndrome) has been presented here in this case report. Diagnosis was confirmed with urinary investigation for GAG and serum levels of alpha-L-iduronidase. Angiokeratomas are bluish-red, hyperkeratotic papules caused by dilated blood vessels in the upper dermis which are typically seen in Fabry disease and rarely in Hurler syndrome. Their occurrence in a 3-year-old with classic Hurler features highlights the importance of considering uncommon dermatologic signs when evaluating patients with metabolic disorders.

Keywords: Mucopolysaccharidosis type I, Hurler syndrome, Angiokeratoma

INTRODUCTION

Mucopolysaccharidosis (MPS) I result from inherited loss of α-L-iduronidase activity, leading to progressive accumulation of glycosaminoglycans (GAGs) such as dermatan and heparan sulfate in various tissues. The severe form, Hurler syndrome (MPS I-H), appears in early childhood with progressive neurodegeneration and multisystem involvement. Without timely diagnosis and treatment, affected children face fatal complications within the first decade of life. The genetic defect is mapped to chromosome band 4p16.3 and results from deficient lysosomal hydrolase activity.[1,2] However, atypical presentations may delay diagnosis and treatment, leading to severe complications. We report a unique case of hurler syndrome with widespread asymptomatic small, multiple, red skin lesions with rapid progression in the number and size of the lesions, emphasizing the need to recognize such unusual presentations.

CASE REPORT

A 3-year-old female child presented with the complaints of being unable to walk without support and unable to speak in sentences, abdominal distension, snoring while sleeping and persistent nasal discharge widespread asymptomatic small, multiple, red skin lesions that were distributed on the face, neck and over the thoracic region with rapid progression in the number and size of the lesions. Antenatal and birth histories were unremarkable. However, the family history revealed a significant detail: the child is 8th born to 3rd degree consanguineous marriage couple, with 3 other female siblings in the family had similar facial dysmorphism, expired before 1 year of age, but exact cause is unknown.

On examination the child has coarse facial features, including macroglossia, a prominent forehead, depressed and broad nasal bridge with flaring nostrils, prominent supraorbital ridges, ocular hypertelorism, ptosis, thick

eyelids, full lips, and dolichocephaly with marked macrocephaly and frontal bossing (Figure 1).

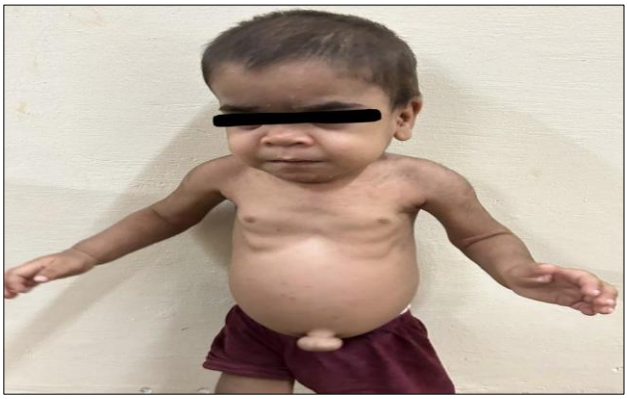

Figure 1: Clinical photograph demonstrating coarse facial features with a protuberant abdomen and a visible umbilical hernia.

Notable findings also include a short neck, hirsutism, stubby and clawed fingers, joint stiffness, kyphosis, and stunted growth (Figure 2). The abdomen is grossly distended with an everted and herniated umbilicus, and the abdominal girth measures 43 cm. On palpation, hepatosplenomegaly is observed: the liver is enlarged (4 cm below the right subcostal margin) with a span of 9 cm, firm to hard in consistency, non-tender, and has a smooth surface with a sharp border. The spleen is palpable 4.5 cm below the left subcostal margin, also non-tender, firm, smooth, and with a palpable notch.

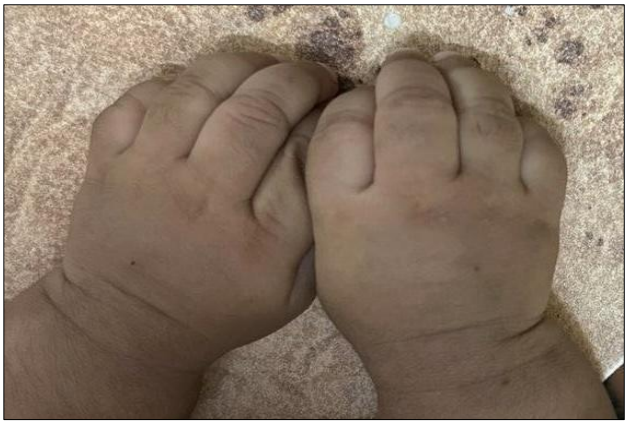

Figure 2: Hand deformities (clinical photograph demonstrating bilateral clawing deformity of the fingers).

Additional findings include widespread small, hyperkeratotic, multiple, non-tender, red papules, accompanied by bleeding due to the avulsion of a lesion, which subsequently regrew at the same location, distributed on the face and neck, extending over the thoracic and abdominal regions, and Mongolian spots on the back (Figures 3 and 4). Radiographic examination revealed dysostosis multiplex. X-ray of the hand and wrist demonstrated bullet-shaped phalanges, along with proximal tapering of the second through fifth metacarpal bones. The chest X-ray (posteroanterior and lateral views) revealed oar-shaped ribs, characterized by narrowing near the spine and widening toward the sternum (Figures 5-7).

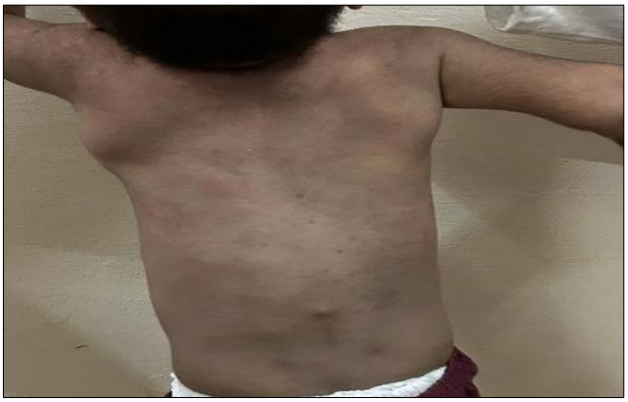

Figure 3: Cutaneous pigmentation (clinical photograph demonstrating dermal melanocytosis (Mongolian spots) over the back).

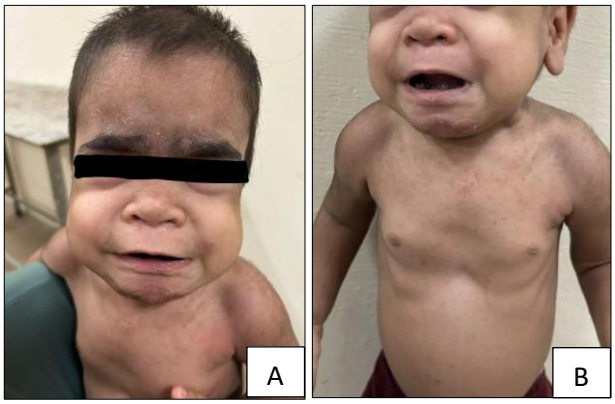

Figure 4 (A and B): Cutaneous lesions (clinical photograph demonstrating multiple erythematous, hyperkeratotic papules involving the face, neck, thorax, and abdomen).

Initial investigations, including complete blood count (CBC) and peripheral blood smear (PBS) revealed microcytic hypochromic anemia, renal function tests (RFT), liver function tests (LFT), thyroid function tests (TFT) were unremarkable, but abdominal ultrasound revealed hepatosplenomegaly (Table 1).

Urine analysis indicated elevated levels of glycosaminoglycans (GAGs), and a fluorometric blood test revealed significantly low alpha-L-iduronidase activity (0.09 nmol/hr/mg) in white blood cells. Ophthalmologic evaluation confirmed b/l corneal opacity and fundus examination that showed b/l corneal haze is present, DISC: 0.2CDR clear disc margin and mild disc pallor was present. Considering the clinical presentation, imaging results, increased urinary GAGs, and enzyme

deficiency, a diagnosis of Hurler syndrome (MPS I H) was established.

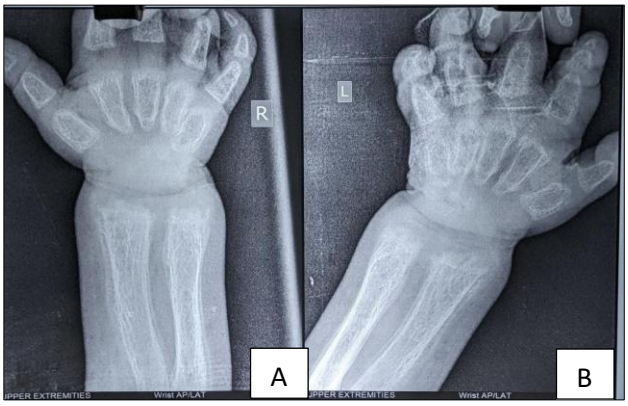

Figure 5 (A and B): Skeletal abnormalities (bilateral hand radiograph (anteroposterior view) demonstrating bilateral bullet-shaped phalanges).

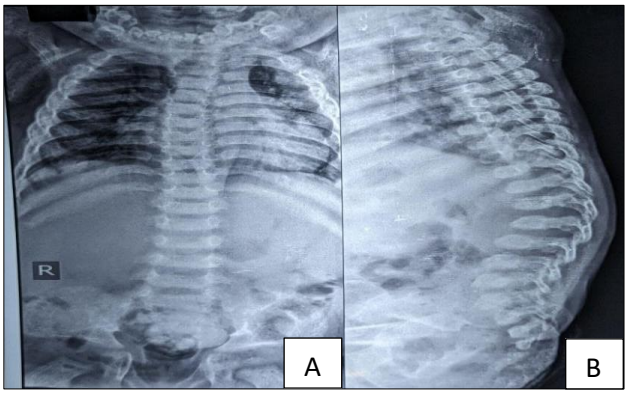

Figure 6 (A and B): Thoracic skeletal and cardiac abnormalities (chest radiograph demonstrating oar-shaped ribs, cardiomegaly, and thoracolumbar kyphotic deformity).

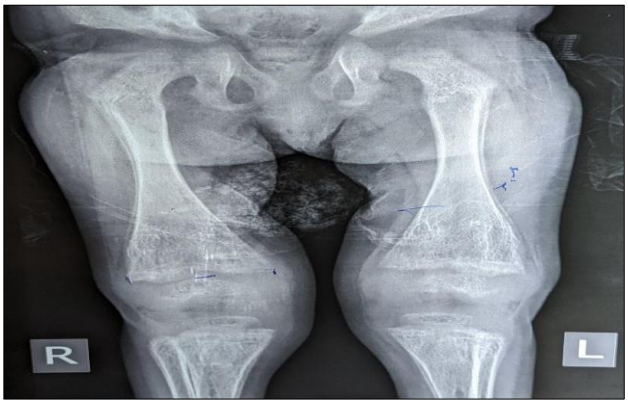

Figure 7: Pelvic skeletal abnormality (anteroposterior pelvic radiograph demonstrating bilateral acetabular dysplasia).

Dermatology examination revealed small, multiple, hyperkeratotic, erythematous papules of varying sizes. The lesions were present on the face, neck and extending up to the thoracic and abdominal region with sizes varying from 0.5×0.5 mm to as large as 1×1 mm in diameter. A skin biopsy revealed features consistent with angiokeratomas, including dilated blood vessels in the papillary dermis, hyperkeratosis, acanthosis, and elongated rete ridges. Cryotherapy was suggested.

Table 1: Investigations in the patient at presentation.

Parameter	Patient's value	Reference value
Haemoglobin (g/dl)	9.9	12.5-16.1
Total leukocyte count ($\times10^9$ cells/l)	12800	4.0-10.5
Platelet count ($\times10^9$ cells/l)	3.8	150-400
Blood urea (mg/dl)	27	3-12
Serum creatinine (mg/dl)	0.7	0.31-0.88
Serum sodium (mEq/l)	137	135145
Serum potassium (mEq/l)	4.57	3.3-4.6
Serum calcium (mg/dl)	8.6	8.8-10.8
Serum albumin (g/dl)	4.0	3.5-5.6
Serum uric acid (mg/dl)	3.97	2.3-5.4
Serum total bilirubin (mg/dl)	0.6	<1.0
ALT, SGPT (U/l)	15	5-45
AST, SGOT (U/l)	28	10-40
Thyroid profile	T3: 1.48 ng/ml, T4: 6.34 ug/dl, TSH: 1.47 uIU/ml	
Peripheral blood smear	Microcytic hypochromic anemia	
USG whole abdomen	Hepatosplenomegaly is seen	
Fluorometric blood test of alpha-L-iduronidase activity in white blood cells	0.09 nmol/hour/mg	

DISCUSSION

Hurler syndrome, or MPS I, is a rare autosomal recessive lysosomal storage disorder caused by a deficiency of the enzyme α-L-iduronidase, leading to progressive accumulation of glycosaminoglycans such as dermatan and heparan sulfate in tissues.[1] This accumulation results in multisystem involvement with characteristic features including coarse facial features, macroglossia, corneal clouding, hepatosplenomegaly, skeletal deformities, and developmental delay, typically presenting within the first few years of life.[1] The 3-year-old female described in this case displayed these classical features, consistent with early-onset Hurler syndrome.[2] Similar phenotypic variations have also been reported across the MPS I spectrum, such as in Hurler-Scheie syndrome, emphasizing the importance of early recognition and intervention.[3] A unique aspect of this case was the presence of angiokeratomas, vascular lesions rarely reported in MPS I but more commonly associated with Fabry disease.[4] These lesions may arise independently in

children without systemic disease and can spontaneously resolve, suggesting that their presence in this case might represent either a coincidental finding or an underrecognized feature of MPS I.[5]

Treatment for Hurler syndrome primarily involves hematopoietic stem cell transplantation (HSCT), provides the best chance of stabilizing neurological decline when performed early, while enzyme replacement therapy (ERT), primarily benefits the somatic disease and improves quality of life.[1] Management of angiokeratomas depends on the severity and symptoms; while often benign and requiring no treatment, options include laser therapy, cryotherapy, or electrocautery when lesions are symptomatic or cosmetically distressing.[4,5] The coexistence of angiokeratomas in this classical Hurler case may represent an underrecognized cutaneous manifestation or incidental finding, underscoring the need to include dermatologic evaluation in the clinical assessment of MPS I and expanding its phenotypic spectrum.[1,4]

CONCLUSION

This case of classical Hurler syndrome in a 3-year-old female with coexisting angiokeratomas underscores a potentially underrecognized cutaneous manifestation of mucopolysaccharidosis type I. While angiokeratomas are more commonly associated with other lysosomal storage disorders, their presence in this patient broadens the clinical spectrum of MPS I and highlights the importance of thorough dermatological evaluation in such cases. Early diagnosis and timely management with enzyme replacement therapy and hematopoietic stem cell transplantation remain critical to improving outcomes. Further studies are needed to clarify the significance of angiokeratomas in Hurler syndrome and their possible role in diagnosis or prognosis.

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