Horseshoe kidney in a patient with Bloom's syndrome

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Summary

Bloom's syndrome is a rare autosomal recessive disorder. Small body stature, erythematous, sun sensitive telangiectatic facial skin and increased tendency to develop malignancy in early life due to inherent genetic predisposition and tendency to develop infections due to immunodeficiency are characteristic features in this condition. We report a 22 month old girl with typical features of Bloom's syndrome, with a horseshoe kidney. Horseshoe kidney as a possible association of Bloom's syndrome has not been described before. As the risk of renal malignancy is higher in horseshoe kidneys, when it occurs in a patient with Bloom's syndrome the relative risk of malignancy could be higher.

Key words: Bloom's syndrome, renal involvement, fused kidneys

Introduction

Bloom's syndrome(BS) is an autosomal recessive disorder which involves multiple systems. It is characterized by short stature, telangiectasia, facial erythema, photosensitivity and increased tendency to develop haematological and other malignancies[1-2]. Telangiectases usually develop several months after birth, especially when exposure to sun occurs. At a cellular level, there are breaks in the chromosomes making them vulnerable to malignant transformation[2-5]. BS with renal abnormalities is extremely rare according to a literature search.

Case report

A 1 year 10 month old girl was brought to the Dermatology Clinic at the Base Hospital Panadura with a history of development of 'scar like marks' on the face in the previous 8 months, and general growth retardation. The birth weight of the child had been 1.78 kg only. It was the first child of a mother of 25 years. There was no parental consanguinity or family history of similar illness.

The weight gain since birth had been poor despite giving breast milk for four months and subsequent weaning foods as recommended by her paediatric clinic. Developmental milestones such as smiling, sitting, walking has not been delayed. Recommended immunizations had been carried out on schedule. The child had not had a major illness to account for the stunting of growth. There was no history of haematuria, dysuria or increased frequency of micturition. There was one episode of history suggestive of mild gastroenteritis and one episode of folliculitis of the scalp which had been effectively treated by the local practitioners.

On clinical examination she was an active child, but very stunted in growth (Figure 1), with multiple telangiectatic, atrophic, speckled skin lesions on the face; concentrated on the sun exposed areas of the cheeks (Figure 2). She had numerous café au lait spots scattered on the trunk and limbs. The weight was 5.5 kg and the height was 72 cm. The head circumference was 39 cm. The weight, height and head circumference were all below the 5th percentile for her age. The hair was thin and lusterless and brownish black, compared to the healthy black hair of her parents. Finger and toe nails were normal. She was not pale or icteric. There was no arthropathy. There was no swelling of legs or eyelids. The cardiovascular, respiratory and central nervous system were clinically normal. Abdomen was soft, liver was just palpable under the costal margin. Spleen was impalpable. Kidneys could not be assessed properly as the patient was uncooperative.

A photograph taken on the first birthday, brought by the mother, did not reveal any obvious marks on the face, indicating that most changes on the face had occurred after the first year.

A diagnosis of Bloom's syndrome was made, based on the characteristic facies, features of facial telangiectasia and atrophic lesions on the sun exposed areas, stunted growth and multiple café au lait spots. In the differential diagnosis, Werners's syndrome and Rothmund Thompson syndrome were considered, but the classical clinical features and the natural history of disease progression was highly suggestive of BS.

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The following investigations were carried out. Haemoglobin was 11.1 g%, packed cell volume was 39%, white cell count was 9900 with 52% neutrophils, 32% lymphocytes and 16% eosinophils. Platelet count was 240,000/mm^2. The ESR was 20 mm in the first hour. Serum IgM was 66.5 mg/dl(normal 14-114 mg/dl), IgA was 115.9 mg/dl (normal 19-119 mg/dl) and IgG was 2245.2 mg/dl (normal 258 -1393 mg/dl). Serum C 3 complement level was slightly elevated; 145.3 mg/dl (normal 55-120 mg/dl). Serum creatinine level was normal. The urinary deposits microscopy did not reveal any abnormalities.

High resolution ultra sound examination of the abdomen showed a fused, horseshoe kidney (Figure 3). The ultra sound examination was repeated on another day to re-evaluate, and that too confirmed the same findings. There were no other tumours or cysts. Intravenous urogram was not done, as excessive radiation could be detrimental to Bloom's syndrome patients. Chromosomal studies to detect sister chromatid breakages were not carried out due to non-availability of facilities.

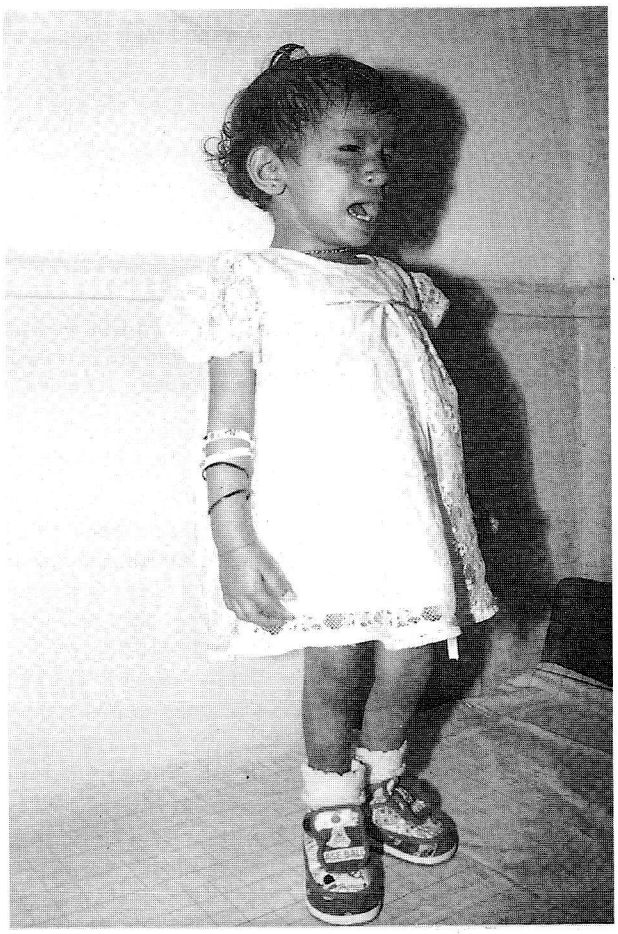

Figure 1. *Twenty two month old child with Bloom's syndrome weighing 5.5 kg and measuring 72 cm in height, indicating gross growth retardation.*

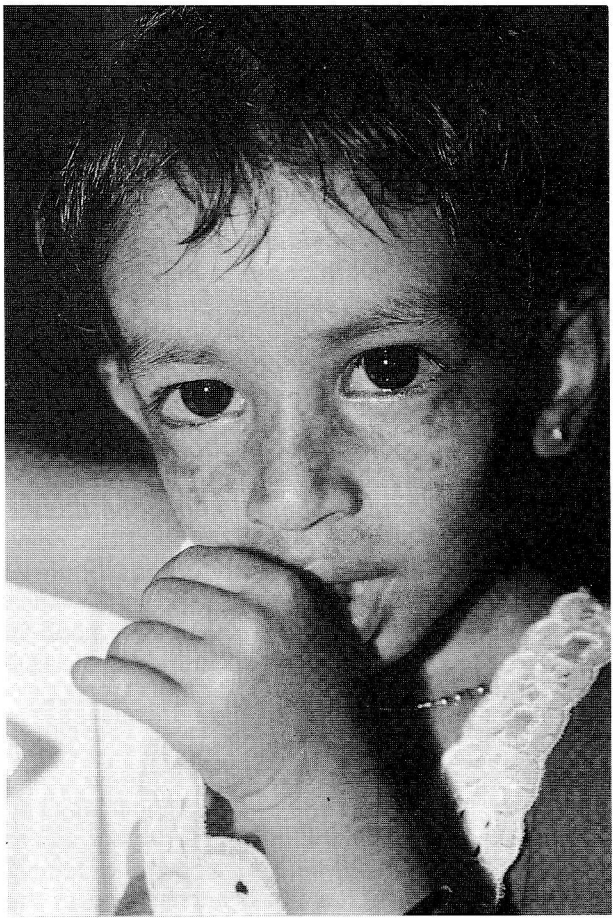

Figure 2. *Telangiectatic skin lesions on sun exposed areas of the face.*

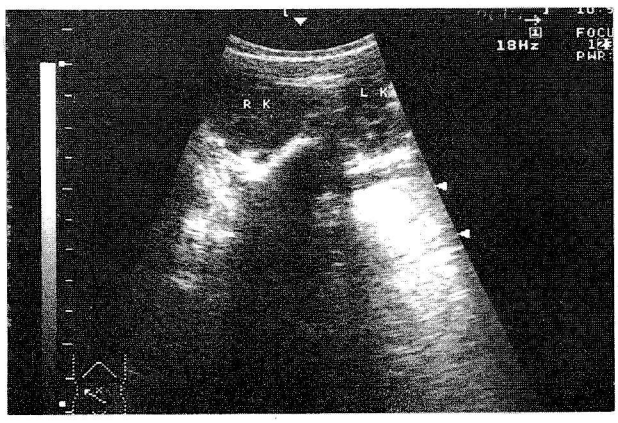

Figure 3. *High resolution ultra sound examination still; depicting fusion of the kidneys in the lower segments giving the 'horseshoe kidney' appearance.*

Discussion

Bloom's syndrome is an autosomal recessive condition which involves multiple systems. It is characterized by short stature (dwarfism), telangiectasia, photosensitivity and increased tendency to develop haematological and other malignancies[1,2]. The telangiectasia usually develop several months after after birth especially when exposure to sunlight occurs. At a cellular level, there are breaks in the chromosomes making them more vulnerable to malignant transfornmation[2-5]. BS with renal abnormalities is extremely rare according to a literature search of publications in the last two decades.

Horseshoe kidney may occur as a congenital anomaly in 1 in 400 cases[6]. Many patients are symptomatic. A variety of tumours including renal carcinoid tumour, Wilm's tumour, transitional carcinoma and adenocarcinoma have been reported in patients with horse shoe kidneys[6-9]. Many authors believe that certain renal malignancies more commonly occur in horse shoe kidneys compared to normal kidneys[6-8].

Bloom's syndrome has not been documented in Sri Lanka before. However the first author has seen another case with typical features of BS in Sri Lanka previously. This is the first instance a renal anomaly was detected in a patient with BS in Sri Lanka, and to our knowledge, the association of a horseshoe kidney in a patient with BS has not been reported in the English medical literature. It is difficult to ascertain whether the occurrence of horseshoe kidney is a chance occurrence, or an actual association, in this patient with BS. However, it is relevant because there is a greater risk of malignancy occurring in both these conditions even considering them as two separate entities. When both risks are combined, there would be a much greater risk of malignancy occurring in the kidney.

In our patient there was no evidence of any haematological, renal or other malignancies. An unexpected finding in this patient was the normal IgM levels; which is usually low in many BS patients. However, low IgM is not an essential feature for a diagnosis of BS.

The child is kept under surveillance for early detection of malignancies if they occur. Sun protection has been advised; with advice to keep out of sun, use appropriate clothes and use of sun block.

It would be useful to carry out high resolution ultra sound examination of the abdomen, as a routine investigation in all BS patients. A larger study would be necessary to ascertain how frequently horseshoe kidney occurs in patients with BS.

References

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