Computerized tomography scans revealed dissection of aorta Type B (Stanford Classification) without renal artery involvement.

Echocardiogram done revealed LV dysfunction. Aortic root was dilated to 45mm. There was mitral and aortic regurgitation. Serology for antinuclear antibody, anti-cardiolipin antibody and lupus anticoagulant was negative and serum creatinine was normal.

Examination revealed trauma-induced phthisis bulbi of the right eye and recurrent retinal detachment (rhegmatus). There was also evidence of coarse chorioretinal atrophy on fundus examination of left eye suggestive of myopic disease. There was no positive family history or evidence of dural ectasia on MRI examination of the spine. Based on these, a diagnosis of Marfans syndrome was made in our patient.[10] Our patient satisfied the diagnosis of Marfans syndrome proposed by Rimoin et al in the absence of a positive family history by the presence of two major criteria (aortic dissection and aortic root dilatation) and the presence of more than two minor criteria (myopia, retinal detachment, mitral valve prolapse, skeletal deformities).[13]

Urinary was negative for homocysteine. Screening for other inborn errors of amino acid metabolism like cystinuria, dibasic aminoaciduria and Hartnup disease was negative. Urine ninhydrin test for amino acids was positive and quantitative urine amino acid analysis revealed generalized aminoaciduria of the physiological type. Urine for glucose was negative. The 24h urine collection revealed proteinuria of 800 mg/24h (Normal < 150 mg/24h) and phosphaturia of 1100 mg/24h (Normal 10-15 mg/kg/24h). All other solutes in the urine like uric acid, glucose, calcium, sodium and potassium were normal. There was no past history of exogenous intoxications with metals, organic compounds or drugs to suggest the cause of proximal tubular dysfunction. Her serum calcium, magnesium, phosphate and arterial blood gas analysis was normal. X-ray was normal with no osteopenia.

Renal involvement in Marfans syndrome is distinctly rare.[23] There have been reports of reno-vascular hypertension and glomerular involvement. We report here a case with aminoaciduria and phosphaturia, a forme fruste of Fanconi syndrome. The patient was investigated for other evidence of Fanconi syndrome, which was found to be negative. The workup for other causes of aortic root dilatation like Ehlers-Danlos syndrome Type 4 (EDS) and familial aortic aneurysms was not done in view of the patient satisfying the criteria for Marfans syndrome convincingly, with no other suggestive features of these syndromes and also that EDS Type 4 only occasionally causes aortic dissection.

Fanconi syndrome is characterized by aminoaciduria, glucosuria and phosphaturia as cardinal features, while acidosis, hypouricemia, hypercalcemia, hypokalemia, polyuria and proteinuria are also commonly associated features.[6] In addition there are incomplete forms of Fanconi syndrome in which at least one of the cardinal features are missing.

The cause of aminoaciduria in Marfans syndrome as a forme fruste of Fanconi syndrome is very difficult to speculate. Probably, if the renal artery was involved in dissection, that could explain the findings as due to ischemia. But the renal artery was not involved in our case to explain the association. Probably, mutations in the fibrillin gene in Marfans syndrome produce defective binding regions in the epidermal growth factor-like domains, which could cause defective arrangement of transporters in the renal tubule cells. Probably, the proximal tubular cells perform the bulk of reabsorption of the glomerular filtrate, while the distal cells have less of these functions and so are not evident in an overt manner.

This is a new observation. There have been a few sporadic reports in some older papers,[7,8] but the real diagnosis of Marfans syndrome in these cases are in doubt. The clinical significance of this observation requires long-term follow-up and experimental studies.

Udayakumar N, Sivapraeksh S*, Rajendiran C* Department of Internal Medicine, University of Cincinnati, Ohio, USA. *Institute of Internal Medicine, Madras Medical College and Govt. Gen. Hospital, Chennai - 3, India

Correspondence:
Dr. Udayakumar N, E-mail: udhaykumar81@yahoo.co.in

References:

Vitamin D deficiency rickets with Lamellar ichthyosis

Sir,
A five-year-old boy presented with history of multiple fractures involving both upper and lower limbs and progressive bony deformities leading to inability to walk over the past two years.
He was the first child of non-consanguinous parents and was born normally. He was noticed to have thick skin right from birth. He was immunized appropriately and his dietary intake (including that of vitamin D) was adequate. There was no history suggestive of malabsorption, renal disorder or decreased exposure to sunlight.

His weight was 13 kg (< 50th centile), height 87 cm (< 3rd centile) and head circumference 50 cm. He had signs of rickets in the form of frontal bossing of the skull, widening of the wrists, rachitic rosary and protuberant abdomen without organomegaly. His upper arm and lower limb bones showed marked lateral bowing. Both fontanelles were closed. He had generalized thickening and hyperpigmentation of the skin with scaling.

As shown in Table 1, investigations confirmed the presence of rickets. The serum level of 25 OH Vitamin D3 was less than 5 ng/ml (Normal: 9-37.6 ng/ml). Serum creatinine levels and arterial blood gas analysis were within normal limits. Radiographs of the wrists also corroborated the diagnosis of rickets as they showed cupping and fraying of the distal ends of the radius and ulna with growth plate widening [Figure 1A]. Radiograph of upper arms and lower limbs showed bowing and shortening with multiple fractures. The skin biopsy showed a markedly thickened stratum corneum and epidermal thickening consistent with lamellar ichthyosis.

It was postulated that the patient’s vitamin D deficiency was secondary to lamellar ichthyosis as a result of poor penetration of skin by ultraviolet rays. The child showed clinical, radiological and biochemical response [Table 1, Figure 1B] to oral vitamin D3 granules 30,000 units daily. He was advised regular application of liquid paraffin over the body. He underwent bilateral femoral closed wedge osteotomy and bilateral tibial osteotomy with rush pin nailing of both tibia for correction of deformities. With the aid of calipers, he was able to walk [Figure 2]. His height velocity was 6cm/year over the next two years of follow-up. He was advised to continue vitamin D3 supplementation lifelong to prevent recurrence of the deficiency and development of further complications.

Cutaneous hyperproliferative states like ichthyosiform dermatoses are uncommon causes of rickets in children.[1] Lamellar ichthyosis is an autosomal recessive disorder that is apparent at birth and is present throughout life. The following factors are proposed for development of rickets in skin disorders, (i) alterations in epidermal cholesterol metabolism possibly involving vitamin D receptors, (ii) increased keratinocyte proliferation resulting in poor or no penetration of skin by sunlight, (iii) associated vitamin D dependent rickets and (iv) limited sun exposure to prevent sunburn and sunstroke.[2] Milstone et al reported elevated parathyroid hormone and low-to-normal 25-hydroxyvitamin D values in patients with various disorders of keratinization, including three adult patients with lamellar ichthyosis.[3]

In our case vitamin D deficient rickets is most likely to be due to poor penetration of skin by sunlight resulting from increased keratinocyte proliferation. A low serum 25-hydroxyvitamin D3 level in the absence of other causes of vitamin D deficiency supported our diagnosis. This boy showed marked improvement with vitamin D supplements, clinically and biochemically. Although the ichthyosis did not improve with resolution of
vitamin D deficiency and rickets, one of two children treated with topical calcipotriene showed improvement in the treated areas of skin. Calcipotriene does not seem to be effective in reversing systemic vitamin D deficiency but can be effective in improving the severity of skin disease in children with ichthyosis. Children with vitamin D deficiency secondary to skin disorders need lifelong supplementation with vitamin D to prevent its deficiency and consequences.

Sathish Kumar T, Scott XJ, Simon A, Raghupathy P
Department of Child Health, Christian Medical College, Vellore - 632 004, India

Correspondence:
Dr. T. Sathish Kumar, E-mail: sathishkumar@cmcvellore.ac.in

References

Does penis radiological shadow indicate the side of hip fracture?

Sir,

Hip fractures are common injuries in the elderly. Standard radiographic evaluation of the hip includes an anteroposterior (AP) view of the pelvis. For this additional purpose, radiograph of the pelvis is one of the commonest prescribed radiographs in emergency departments. Usually we notice the fractured hip side and the type of fracture, whenever we are looking at an anteroposterior pelvis radiograph. But do we not question if this radiograph represents a true anteroposterior view.

We have noticed that the radiological shadow of the penis almost always turns to the side of the hip fracture in male patients and especially in displaced fractures. Many surgeons have tried to explain this observation.

So we would like to test the hypothesis that the radiological shadow of the penis indicates the side of the hip fracture [Figure 1].

In our retrospective study, 712 male patients with hip fracture were included. In order to test our hypothesis we studied their first pelvis radiograph on admission, before operation. Intertrochanteric hip fracture was noticed in 475 cases and subcapital hip fracture in 237 cases. All the patients had no history of previous hip fracture, hip osteoarthritis, spinal cord disorders, hernia or congenital penis disorders. The mean age of the patients was 78.6 (range 68-83) years. Statistical analysis was performed by chi-square analysis.

In 457 (96.2%) displaced intertrochanteric fractures and in 221 (93.2%) displaced subcapital fractures, radiological shadow of the penis turned to the fractured side ($\chi^2 = 17.1, P<0.01$). Furthermore in 18 (3.8%) undisplaced intertrochanteric and 16 (6.8%) undisplaced subcapital fractures, the radiological shadow of the penis was in midline position. We concluded that the radiological shadow of the penis indicates the side of the fracture when a displaced hip fracture occurs.

Discussion

The position of patients during the anteroposterior pelvis radiograph may explain this observation. Patients with a displaced hip fracture exhibit the classic presentation of a shortened and externally rotated extremity. We believe that the patients in order to avoid further external rotation of their leg, turn their pelvis and body to the fractured side, so as to immobilize the fracture and to reduce pain. This means that the penis follows the fracture because the pelvis turns to the fractured side.

So the anteroposterior view of the pelvis is in fact an oblique view, which shows the fracture and the penis shadow on the same side.

Mouzopoulos GJ, Stamatakos MK, Mouzopoulos DJ
Orthopedic Department of Evangelismos Hospital, Athens, Greece

Correspondence:
Dr. Mouzopoulos George, E-mail: gmouzop@yahoo.gr