Unusual association of diseases/symptoms

Osteogenesis imperfecta with right renal artery occlusion

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Summary

We here report a case of osteogenesis imperfecta who presented with severe hypertension and left ventricular failure and had right renal artery occlusion. The case is very interesting as renal artery occlusion has not been reported earlier in osteogenesis imperfecta.

BACKGROUND

Osteogenesis imperfecta (OI) is an inherited disorder of connective tissue resulting in osteoporosis, clinically manifested by increased bone fragility with liability to fracture. It results from mutations affecting either the COL1A1 or COL1A2 gene of type I collagen.^{1 2} The incidence of OI is 0.4 per 10 000 live births, about half of which (0.19 per 10 000) represent type II disease³ as per the classification of OI devised by Silience¹ in 1979 and modified in 1986. The important characteristics in this classification include blue sclera, the severity of the disorders and the mode of inheritance, although the accurate classification is difficult because of phenotypic overlap.² We report here a case of OI with severe hypertension resulting from renal artery occlusion. This association has not been reported earlier.

CASE PRESENTATION

A 35-year-old man was admitted with complaint of sudden onset of breathlessness for last 7 days. There was no history of chest pain, cough, fever or wheezing. The patient had similar complaints 2 years back when he was detected to be hypertensive but did not take treatment regularly. The patient also had a history of multiple fractures since childhood. The patient neither smoked nor drank alcohol. There was no history of repeated fractures in other family members.

On general examination, the patient was short statured (height-120.6 cm) with a relatively large head (head circumference 53.3 cm). His upper limbs were apparently normal with an arm span of 153.6 cm. The lower limbs were bowed (figure 1). The ratio of the upper segment to lower segment of body was 0.88. There was mild pallor, sclera was blue and dentition was normal. There was kyphosis of thoracic spine. Pulse was 110/min, regular; blood pressure was 270/130 mm Hg in the right upper limb in supine position on admission. Jugular venous pressure was normal and no oedema was present. Precordial examination showed apex beat in left sixth intercostal space lateral to the mid-clavicular line, forceful and well sustained. On auscultation S3 gallop was present, no murmurs heard. Respiratory examination revealed diffuse rhonchi and basal crepts. Abdomen and nervous system examinations were within normal limits.

INVESTIGATIONS

On investigation – haemoglobin—9.5 gm/dl, total leucocyte counts—8600/ μ l with differential counts of polymorphs-70, lymphocyte-23, eosinophils-5, monocytes-2, platelet count 2.35 lacs/ μ l, red blood cells (RBCs) microcytic hypochromic. Urine examination showed albumin 10 mg/dl, occasional pus cells, no RBCs, casts or crystals. Random blood sugar—110 mg/dl, Serum Na⁺ 135 mEq/l, K⁺ 3.2 mEq/l, blood urea 35 mg/dl, serum creatinine—0.9 mg/dl, serum calcium (total)—9.11 mg/dl, ionic calcium 1.11 mEq/l and serum alkaline phosphatase 110 IU/l. Chest x-ray showed mild cardiomegaly, ECG showed evidence of left ventricular hypertrophy. On ultrasonography of the abdomen right

Figure 1 In front view, a case of osteogenesis imperfecta (type IV) with multiple bony deformities.

Figure 2 Renal angiography showing complete (100%) occlusion of the right renal artery origin (arrow).

kidney was small (6.5×3 cm) and left kidney was normal (9.5×5 cm). No other abnormality was detected. Renal angiography showed 100% proximal occlusion of right renal artery while left renal artery was normal (figure 2). Pelvis x-rays showed generalised osteopenia, triradiate pelvis and protrusio acetabuli (figure 3). Lower limbs' x-rays revealed generalised osteopenia and pseudo-fractures. There was anterior and medial bowing of tibia and fibula (figure 4).

DIFFERENTIAL DIAGNOSIS

Increased prevalence of renal artery stenosis has been reported in other inherited collagen disorders like Marfan's syndrome.⁴ Ehler Danlos syndrome⁵ and tuberous sclerosis.⁶

Figure 3 Pelvis x-ray showing triradiate pelvis and protrusio acetabula.

Figure 4 Leg x-ray elucidates anterior and medial bowing of tibia and fibula.

TREATMENT

The patient was treated for left ventricular failure. He was given ACE inhibitors, calcium channel blockers and diuretics.

OUTCOME AND FOLLOW-UP

He rapidly improved with the above treatment and was discharged after few days with blood pressure of 130/80 mm Hg. He has been doing well since then.

DISCUSSION

OI is usually diagnosed on the basis of clinical features. The musculoskeletal findings in the case are highly suggestive of OI type IV.¹ OI type IV is a mildly severe form and is similar to type I. The sufferers need crutches and braces to walk. The patient, however, primarily came to us for severe hypertension and left ventricular failure. On investigation he was found to have right renal artery occlusion that was probably responsible for his hypertension.

Renovascular hypertension is caused by narrowing of a renal artery. Complete occlusion of a renal artery usually does not cause hypertension as the kidney generally gets infarcted. If the kidney, however, survives as a result of collateral circulation following renal artery occlusion, then it may lead to considerable elevation of the blood pressure.^{7,8} The renal vein renin levels have been found to be high in such cases.^{8,9} This was probably responsible for hypertension in our case. This is not a common presentation and only few such cases have been reported.⁷⁻⁹ It is important to keep this possibility in mind as nephrectomy will be curative.

The other point in this case is regarding the relationship between renal artery occlusion and OI. Are these cases more prone to develop such complications? There are no data regarding this in the literature. This is probably the first case where the two have been found in the same patient. There are, however, a number of other anomalies that have been described in OI such as — spinal

deformity, thoracic deformity, pelvic deformity, multiple fractures, bowing of weight bearing bones,¹⁰ hearing loss and abnormal dentition. Increased prevalence of renal artery stenosis has been reported in other inherited collagen disorders like Marfan's syndrome,⁴ Ehler Danlos syndrome⁵ and tuberous sclerosis.⁶ OI is also an inherited disorder of collagen synthesis and therefore these cases may also be prone to develop such complication, but we cannot definitely say this till more data are available. However, in future it may worth observing such cases for any such manifestation.

Learning points

- Renal artery stenosis, an unusual complication can develop in patients of osteogenesis imperfecta (OI), although this manifestation has not been reported earlier, but the casual association is likely because in other inherited connective tissue disorders cases of renal artery stenosis have been reported.
- Renovascular hypertension is due to unilateral narrowing of the renal artery, occasional severe hypertension may occur with complete occlusion of renal artery.
- Such life-threatening complication may herald the prognosis in patient of OI where life expectancy is close to normal or completely normal.

Competing interests None.

Patient consent Obtained.

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