Unusual chest radiograph

A newborn twin with unusual chest radiograph

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A 7-day-old (term small for gestational age) newborn female, first of a twin delivery, presented to the paediatric admissions unit, with a 2-day history of coryza, cough, shortness of breath and poor feeding. The twins were born at term by normal delivery (twin 1 weighed 1.89 kg and twin 2 2.58 kg) and they did not require any resuscitation at birth. The parents were of Asian origin and were first cousins. The mother had three previous normal deliveries and the children were healthy. The pregnancy had been complicated by gestational diabetes mellitus requiring insulin treatment. There was no other major family history of note.

On examination, the baby had signs of upper respiratory tract infection, tachypnoea, mild respiratory distress, a chest clear on auscultation and normal oxygen saturations in air. Systemic examination was otherwise unremarkable. The baby was admitted for observation and nasogastric feeding, with a diagnosis of acute bronchiolitis on admission. The nasopharyngeal aspirate was negative for common respiratory viruses, including respiratory syncytial virus. Full blood count showed haemoglobin 18.9 g/dl, white cells $19.5 \times 10^9/l$, neutrophils $8.9 \times 10^9/l$, lymphocytes $6.6 \times 10^9/l$ and platelets $190 \times 10^9/l$. The levels of urea, creatinine, electrolytes and capillary blood gas were normal.

A chest radiograph (fig 1) was taken at admission.

QUESTIONS

1. What abnormality does the chest radiograph (fig 1) show?
2. What further investigations would you consider to confirm the diagnosis?
3. What further investigations/assessments would you consider?
4. What are the treatment options and what is the prognosis of this condition?
5. What other conditions may have a similar radiological appearance?

1. What abnormality does the chest radiograph show?

The skeletal system on the chest radiograph (fig 1) shows sclerosis, loss

of corticomedullary differentiation and some fragmentation of both humeral metaphyses, but of the right side in particular. The appearance is suggestive of sclerosing skeletal dysplasia such as osteopetrosis. The cardiac shadow and lung fields appear normal.

2. What further investigations would you consider to confirm the diagnosis?

Skeletal survey.

The radiograph of the left radius and ulna as part of the skeletal survey (fig 2) showed generalised sclerosis of bones with obliteration of marrow cavity, suggestive of osteopetrosis. The skeletal survey findings were identical in twin 2.

3. What further investigations/assessments would you consider?

Ophthalmological assessment (including visual evoked potentials and electroretinography), hearing assessment, echocardiogram, tissue typing of child and family, feeding assessment, neurological/developmental assessment and neuroimaging.¹ Children with classical congenital osteopetrosis should not have central nervous system involvement. The neurodegenerative condition associated with osteopetrosis is rapidly progressive and generally carries a poor prognosis. This may constitute a contraindication to bone marrow transplantation. It is thus mandatory to carry out a thorough clinical, radiological (magnetic resonance

Figure 1 The skeletal system on the chest radiograph showing sclerosis, loss of corticomedullary differentiation and some fragmentation of both humeral metaphysis, but of the right side in particular. The appearance is suggestive of sclerosing skeletal dysplasia such as osteopetrosis.

imaging of the brain) and neurophysiological examination on all patients with osteopetrosis.¹

4. What are the treatment options and what is the prognosis of this condition?

The only treatment that has a noticeable favourable effect on the disease course is bone marrow transplantation.¹ Corticosteroids, high-dose calcitriol and interferon γ have all been reported to be of variable benefit in the treatment of osteopetrosis.²⁻⁴ Management involves multidisciplinary supportive treatment of associated problems such as anaemia, recurrent infections, failure to thrive, hypocalcaemia, fractures and dental problems. Severe autosomal recessive malignant osteopetrosis has a high mortality in the first 2 years of life. Death in infancy is not uncommon for those with severe visual impairment and bone marrow failure before 3 months of age.¹ Children with associated neurodegenerative conditions generally carry a poor prognosis. Children who are alive at 2 years and are not transfusion dependent have a relatively better prognosis.

Figure 2 Radiograph of the left radius and ulna as part of the skeletal survey showing generalised sclerosis of bones with obliteration of marrow cavity, suggestive of osteopetrosis.

5. What other conditions may have a similar radiological appearance?

Pyknodysostosis, fluorosis and heavy-metal poisoning.

DISCUSSION

Osteopetrosis, in its autosomal recessive malignant form, is a rare congenital disorder caused by the failure of osteoclasts to resorb immature bone.⁵ Mutations in several genetic loci that govern osteoclast development and function could be responsible; recently, mutations in the gene coding for an osteoclast-specific vacuolar pump have been described in a subgroup of affected children.⁶

Pathologically, persistence of primary spongiosa and abnormal remodelling of bone result in "brittle" bone that is prone to fracture.⁷ Classical features include bone marrow failure, consequences of bony narrowing of cranial nerve foramina and fractures.¹ Clinical presentation is commonly in the form of parental concern regarding the child's vision or as an incidental finding of distinctive sclerotic bony changes on x rays taken for another reason. This condition often presents in early

infancy, and frequently within the first 3 months of life as in our patient.⁸ Less common presenting features include anaemia, failure to thrive, recurrent infections, hepatosplenomegaly, hypocalcaemic seizures, easy bruising, fractures, nasal congestion and a dysmorphic facies.¹ Skeletal survey usually confirms the diagnosis in presence of classical clinical features.

This case clearly highlights the need for clinicians to consider unusual medical conditions presenting as incidental findings on radiography carried out for common medical illnesses. The prognosis depends on early diagnosis and effective intervention.

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