CASE REPORT

A rare combination of amniotic constriction band with osteogenesis imperfecta

Krupa Hitesh Shah,¹ Hitesh Shah²

¹Department of Obstetrics and Gynaecology, Melaka Manipal Medical College, Manipal University, Manipal, Karnataka, India

²Department of Orthopaedics, Kasturba Medical College, Manipal University, Manipal, Karnataka, India

Correspondence to

Dr Hitesh Shah,
hiteshshah12@gmail.com

Accepted 26 October 2015

SUMMARY

Amniotic constriction bands and osteogenesis imperfecta are disorders arising from a collagen defect. We report a rare association of amniotic bands with osteogenesis imperfecta in a child. The child was born with multiple amniotic bands involving the right leg, both hands and both feet. Multiple fractures of long bones of lower limbs occurred in childhood due to trivial trauma. Deformities of the femur and tibia due to malunion with osteopenia and blue sclerae were present. The patient was treated with z plasty of constriction band of the right tibia and bisphosphonate for osteogenesis imperfecta. This rare association of both collagen diseases may provide further insight for the pathogenesis of these diseases.

BACKGROUND

Amniotic constriction bands and osteogenesis imperfecta are disorders arising from a collagen defect.^{1,2} Incidences of isolated osteogenesis imperfecta along with amniotic band syndrome are rare.^{3,4} Only two reports have described the combination of osteogenesis imperfecta and amniotic band syndrome. The earlier reported cases showed amputation with multiple amniotic bands on limbs associated with multiple fractures of long bones and ribs. Both reported cases were during the perinatal period and were lethal (spontaneous respiration was not established in one; while the other died after 7 days). To the best of our knowledge, a non-lethal combination of both has not been reported before. We report a rare association of amniotic band syndrome with a non-lethal type of osteogenesis imperfecta in a child.

CASE PRESENTATION

A newborn male infant presented with deformities of both upper and lower limbs. He was born following an uncomplicated pregnancy and caesarean delivery. He was the first child of non-consanguineous parents. His birth weight was 2.5 kg and height was 50 cm. Both parents (age 29 and 25 years) were healthy with normal stature. The boy's other family members, including grandparents, were normal.

The parents noticed multiple constriction bands involving all limbs with absence of a few fingers at birth. The constriction of the right leg with swelling of the right foot was marked. The boy was treated with z plasty of the right tibia for severe swelling of the right foot at 6 months of age. His height was 64 cm and weight was 6 kg at 6 months of age. He attained developmental milestones

normally. The child started walking at 13 months of age. At 2 years of age, his height was 77 cm (Z score -2.8), upper segment; lower segment ratio was 35:32 and his weight was 8.6 kg. He had multiple fractures of both femurs and both tibias (6 fractures) due to trivial trauma. He was treated with a hip spica cast for both femurs and above knee cast for the fracture tibias. The child was treated for long bone fractures by orthopaedic surgeons. Serial radiographs were taken to assess healing of the fractures. The boy was sent to the paediatric orthopaedics department for evaluation of the unusual higher frequency of long bone fractures.

The boy presented at 4 years of age with deformities of both femurs and both tibias. On examination, generalised ligamentous laxity with blue sclera was noted (figure 1). His general health was good and intellect was normal. His height was

Figure 1 Clinical photographs of the eyes showing blue sclerae.

Figure 2 Both hands showing classic features of amniotic band syndrome with amputation of fingers.

CrossMark

To cite: Shah KH, Shah H. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2015-212400

Figure 3 Both legs and feet showing scars of z plasty of lower third right leg with multiple constriction bands with auto amputation of right side with syndactyly of left side toes.

90 cm (Z score -3), upper segment; lower segment ratio was 46.5:43.5 and his weight was 12 kg. Multiple constriction bands involving both hands were noted. Auto amputation of both side distal index fingers through proximal phalanx and left side little fingers was noted (figure 2). Multiple constriction bands involving both feet with syndactyly of left first, second and third toes of the left foot were noted. A healed z plasty scar on the right tibia with swelling of the right foot was observed. Auto amputations with rudimentary toes were observed on the right side (figure 3). No deformity of knees and ankles was noted. Alignment of both upper limbs and lower limbs was normal. No limb length discrepancy was noted.

The spine was normal. Skin, hair, teeth and genitals were all normal. Ultrasound examinations of the abdomen and pelvis were normal. Ophthalmological, cardiological and otorhinolaryngological examinations were normal.

INVESTIGATIONS

The radiological examination of earlier available radiographs clearly depicted multiple fractures of both femurs and both tibias (figure 4). Radiographs of the lower limbs showed mal-united fractures (femurs and tibias) with cortical bone thinning and excessive trabecular bone transparency (figure 5).

Figure 4 Radiographs of both femurs and tibias of patient at 3 years of age showing fractures of both femurs.

Deformities of the femurs and tibias with osteopenia were marked on radiographs at 4 years of age. The rest of the skeletal system was visualised as normal.

TREATMENT

The boy was medically treated with bisphosphonate. Oral alendronate (35 mg/week) was administered with 240 mL of water, 30 min before food intake. He was instructed to remain in an upright position for 30 min after taking the alendronate. He did not have any complications 6 months following the oral bisphosphonate treatment. Surgical correction of the deformity of both femurs is planned.

DISCUSSION

Amniotic band syndrome is a rare collagen disorder.³ It commonly involves the distal part of extremities. Common manifestations are constriction rings, equinovarus deformity, amputation and acrosyndactyly. It is also associated with non-classic manifestations such as cleft lip, cleft palate and imperforate anus. Many theories are postulated and debated for the pathogenesis of amniotic band syndrome.³ The earlier literature reported association of amniotic band syndrome with Ehlers-Danlos and the lethal variety of osteogenesis imperfecta.^{1,2}

Osteogenesis imperfecta is a disorder arising from a quantitative and qualitative defect of type I collagen.⁴ Most reported cases (90%) are due to mutations in the COL1A1 and COL1A2 genes with dominant inheritance. Mutation in more than 10 genes are identified with recessive inheritance. There is a wide variation in presentation of osteogenesis imperfecta, ranging from a mild phenotype to a perinatal lethal variety. Brittle bone disease is a clinical diagnosis.⁵ Blue sclerae, generalised osteoporosis and dental abnormalities are major criteria for diagnosis of osteogenesis imperfecta. Wormian bones, ligamentous laxity, deafness, easy bruisability and excessive sweating are non-specific minor criteria. This child had multiple fracture with trivial trauma, osteopenia with blue sclerae, which confirmed brittle bone disease. Bisphosphonate therapy reduces the incidence of fracture and bone pain.⁶

We report a non-lethal variety of osteogenesis imperfecta with amniotic constriction bands. The child was thought to have a Sillence type IV form of osteogenesis imperfecta, as he had moderate deformity, no congenital fractures, moderate growth

Figure 5 Radiographs of both femurs and tibias of patient at 4 years of age showing malunion of the femurs and tibias, with marked osteopenia.

retardation and no hearing loss. A postnatal severity grading scale showed moderate osteogenesis imperfecta according to Van Dijk and Sillence.⁴

Currently, medical management with bisphosphonate has been started. The fracture shafts of both femurs were treated with a hip spica cast application. The boy got appropriate treatment for the previous fractures, as fractures in osteogenesis imperfecta heal well with a short duration of immobilisation. The displacement and angulation in younger children get remodelled very well in children.⁷ The fractures of both femurs were malunited. Future surgery will rectify the misalignment of both femurs. Surgical correction of the deformities with intramedullary rodding are indicated to reduce the frequency of long bone fractures and improve the quality of life.^{8,9}

There is no consensus on the duration of bisphosphonate and the exact timing of surgical intervention for deformity correction in relation to bisphosphonate therapy in the literature. However, osteotomy for correction of the deformities showed delayed healing following bisphosphonate therapy.¹⁰ In our case, the bisphosphonate treatment will be temporarily stopped at the time of surgical intervention. The medical treatment will be resumed after the union of the osteotomy.

Learning points

- Osteogenesis imperfecta with amniotic band contracture is a rare combination.
- The lethal type of osteogenesis imperfecta has been reported to be associated with amniotic bands.
- Amniotic band contracture can also be seen in the non-lethal type of osteogenesis imperfecta.
- Osteogenesis imperfecta with amniotic band contracture can be effectively managed with z plasty of the amniotic band syndrome followed by management of brittle bone disease.

The combination of both disorders has rarely been reported.^{1,2} Collagen provides tensile strength of bone and amnion. A qualitative and quantitative defect in collagen can lead to defective bone, amnion and skin, which can produce the perinatal lethal phenotype. The compromised strength of the amnion can lead to its earlier rupture. Early rupture of the amnion with weak bone due to poor collagen during the perinatal period leads to the severe lethal phenotype. However, the non-lethal type of osteogenesis with amniotic bands has not been reported earlier. In our case, the defect in collagen may have been milder in nature. Further detailed investigation will be required to know the exact nature of the defect in collagen in our case. The next generative sequencing tool will help to identify the exact molecular nature of the case.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 van der Rest M, Hayes A, Marie P, *et al.* Lethal osteogenesis imperfecta with amniotic band lesions: collagen studies. *Am J Med Genet* 1986;24:433–46.
- 2 Young ID, Lindenbaum RH, Thompson EM, *et al.* Amniotic bands in connective tissue disorders. *Arch Dis Child* 1985;60:1061–3.
- 3 Goldfarb CA, Sathienkijkanchai A, Robin NH. Amniotic constriction band: a multidisciplinary assessment of etiology and clinical presentation. *J Bone Joint Surg Am* 2009;91(Suppl 4):68–75.
- 4 Van Dijk FS, Sillence DO. Osteogenesis imperfecta: clinical diagnosis, nomenclature and severity assessment. *Am J Med Genet A* 2014;164A:1470–81.
- 5 Falvo KA, Root L, Bullough PG. Osteogenesis imperfecta: clinical evaluation and management. *J Bone Joint Surg Am* 1974;56:783–93.
- 6 Harrington J, Sochett E, Howard A. Update on the evaluation and treatment of osteogenesis imperfecta. *Pediatr Clin North Am* 2014;61:1243–57.
- 7 Wallace ME, Hoffman EB. Remodelling of angular deformity after femoral shaft fractures in children. *J Bone Joint Surg Br* 1992;74:765–9.
- 8 Mulpuri K, Joseph B. Intramedullary rodding in osteogenesis imperfecta. *J Pediatr Orthop* 2000;20:267–73.
- 9 Wilkinson JM, Scott BW, Clarke AM, *et al.* Surgical stabilisation of the lower limb in osteogenesis imperfecta using the Sheffield Telescopic Intramedullary Rod System. *J Bone Joint Surg Br* 1998;80:999–1004.
- 10 Munns CF, Rauch F, Zeitlin L, *et al.* Delayed osteotomy but not fracture healing in pediatric osteogenesis imperfecta patients receiving pamidronate. *J Bone Miner Res* 2004;19:1779–86.

Copyright 2015 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow