Proximal femoral focal deficiency as a manifestation of Antley-Bixler syndrome: a case report

AR Sulaiman, H Nawaz, I Munajat, AY Sallehudin
Department of Orthopaedics, School of Medical Sciences, Universiti Sains Malaysia, Kubang Kerian, Kota Bharu, Kelantan, Malaysia

INTRODUCTION

Proximal femoral focal deficiency (PFFD) is a congenital defect in which the proximal end of the femur does not grow normally during embryological development. It may be associated with growth retardation of the tibia and fibula. Other associated abnormalities are ulna hypoplasia, clubfoot, unusual facies, and Pierre Robin syndrome. Bilateral PFFD has never been reported as a manifestation of Antley-Bixler syndrome (ABS).

CASE REPORT

In May 2004, an 11-year-old girl presented with right thigh pain and swelling after a fall at school. Clinical and radiological assessment revealed that she had a right clubfoot, a left radiohumeral synostosis, and bilateral PFFD (Fig). Her right femur was fractured at the proximal third level. Apart from skeletal...
abnormalities, she had bilateral ear hypoplasia, cleft palate, tongue tie, and missing teeth. Other abnormalities included congenital heart disease (arterial septal defect and patent ductus arteriosus) and a pelvic kidney with hydronephrosis. The fractured femur was managed with traction for 2 weeks followed by a hip spica. There was no delay in fracture healing and unprotected walking was allowed after 2 months.

She was the youngest of 6 siblings from a non-consanguineous marriage. All other siblings were normal. Her mother had an intravenous urogram during the first trimester and was found to be pregnant 2 weeks later. The mother decided to continue with the pregnancy and had a lower segment caesarean section for breech presentation at term. The child underwent multiple surgical procedures to correct her congenital abnormalities since birth. She started walking at the age of 5. Her social development was slow and she had to attend special school. She could walk without aid with a short limb gait. Her left elbow was fixed in 90° flexion with a mid-pronated forearm. She used her left shoulder movement to compensate for the disability. Her wrist and hand functions were normal.

DISCUSSION

The incidence of PFFD has been reported as 1:50,000 live births. Its aetiology is multi-factorial, comprising cellular malnutrition, drug ingestion, vascular abnormality, and intra-uterine arterial compression. The severity varies from a minor ossification defect of the femoral head to absence of the hip joint with only a small fragment of the femur developed. PFFD can be unilateral or bilateral and is associated with musculoskeletal defects distal to the femur such as fibula hemimelia and clubfoot. It may be associated with Pierre Robin syndrome, characterised by micrognathia and glossoptosis; some may have cleft palate.

The manifestations of ABS include craniofacial defects (craniosynostosis and brachycephaly, midface hypoplasia with choanal stenosis or atresia, a pear-shaped nose, dysplastic ears, abnormal dentition), musculoskeletal disorders (radiohumeral synostosis, multiple joint contractures, femoral bowing, synostosis of the carpal bones and tarsal bones, long hands and fingers, clubfoot, camptodactyly, vertebral anomalies), mental retardation, and occasionally, urogenital or cardiac defects.

Our patient has many features of ABS along with bilateral PFFD. To our knowledge, PFFD has never been reported as a manifestation of Antley-Bixler syndrome. The multiple skeletal anomalies seen in ABS may lead to early orthopaedic consultation. Surgeons should be aware of the associated problems, especially choanal atresia as it can lead to respiratory obstruction and early death in infancy.

Radiation is the main cause of the mutations that develop in utero. Over 80% of absorbed radiation arises from the natural environment, such as food and ambient air, with large geographical variations. It is difficult to determine whether a particular deformity or genetic defect in an individual is due to background or medical radiation. Foetal mutagenic risk is dependent on the gestational age and radiation dose. Limb bud formation and organogenesis peak during the 4th to 8th weeks of gestation. When radiation doses exceed an estimated threshold of 150
mGy, teratogenic mutations (potentially attributable to medical X-rays) may occur. Such mutations only occur during the 2nd to 15th weeks of gestation.\(^9\) It is estimated that 2 radiographs of the pelvis deliver a 4 mGy dose to the foetus.\(^7\) An intravenous urogram requires at least 6 pelvic radiographs. Therefore, as a foetus, our patient may have absorbed 12 mGy when her mother had an intravenous urogram during the first trimester. Nonetheless, her exact gestational age and the radiation dose she was exposed to are uncertain. A patient with ABS and disordered steroidogenesis was born to a mother virilised by luteomas of pregnancy.\(^10\) Exposure to radiation has not been implicated before.

REFERENCES

1. Pappas AM. Congenital abnormalities of the femur and related lower extremity malformations: classification and treatment. J Pediatr Orthop 1983;3:45–60.
2. Graviss ER, Monteleone PA, Wampler LR, Silberstein MJ, Brodeur AE. Proximal femoral focal deficiency associated with the Robin anomalad. J Med Genet 1980;17:390–2.
3. Bottero L, Cinalli G, Labrune P, Lajeunie E, Marchac D, Renier D. Antley-Bixler syndrome. Description of two new cases and review of the literature. Prognostic and therapeutic aspects. Ann Chir Plast Esthet 1997;42:48–55.
4. Rumball KM, Pang E, Letts RM. Musculoskeletal manifestations of the Antley-Bixler syndrome. J Pediatr Orthop B 1999;8:139–43.
5. Tsuchiya Y, Sueishi K, Yatabe K, Yamaguchi H. A case of Antley-Bixler syndrome with severe skeletal Cl. III malocclusion. Bull Tokyo Dent Coll 2004;45:87–93.
6. Hosalkar HS, Shah HS, Gujar PS, Shaw BA. The Antley-Bixler syndrome: two new cases. J Postgrad Med 2001;47:252–5.
7. Mann FA, Nathens A, Langer SG, Goldman SM, Blackmore CC. Communicating with the family: the risks of medical radiation to conceptuses in victims of major blunt-force torso trauma. J Trauma 2000;48:354–7.
8. Gamble JG. Development and maturation of the neuromusculoskeletal system. Lovell and Winter's pediatric orthopaedics, 4th ed. Philadelphia: Lippincott-Raven; 1996.
9. Wagner LK, Lester RG, Saldana LR. Exposure of the pregnant patient to diagnostic radiations: a guide to medical management, 2nd ed. Madison: Medical Physics Publishing; 1997.
10. Roth C, Hinney B, Peter M, Steinberger D, Lakomek M. Features of Antley-Bixler syndrome in an infant born to a mother with pregnancy luteoma. Eur J Pediatr 2000;159:189–92.