Case Reports

CHONDRODYSPLASIA PUNCTATA

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Introduction

Chondrodysplasia Punctata (CDP) is a rare congenital skeletal dysplasia with many clinical variants [1]. The radiological hallmark of CDP is varying degrees of punctate stippling of the cartilages of the axial and appendicular skeleton [2-4]. We report a newborn with CDP, presenting with polyhydramnios on routine antenatal ultrasonographic scan (USG). The antenatal USG scans, plain X-ray films of the newborn along with the differential diagnosis of this rare disorder is presented in this report.

Case Report

A 24-year-old lady, third gravida with two normal children, presented for routine antenatal USG on 12 Dec 96. A booked and immunised patient; she had no history of consanguinity, any antenatal illness or drug intake. USG scans revealed a gestational age of 35 weeks based on biparietal diameter and head circumference, whereas the femoral length and abdominal circumference corresponded to a gestational age of 30 weeks and 5 days. Placenta was normal. There was polyhydramnios indicated by a four quadrant amniotic fluid index of 33.

Detailed USG examination of the foetus revealed prominent hands with both fists open (Fig 1). There was evidence to suggest

dysplasia of the thoracolumbar spine (Fig 2) with associated disproportionate growth between the head and the limbs. No other fetal anomaly was noted. The ossification centres at upper end of both humeri and lower ends of both femora were not visualised.

Admitted as a high-risk pregnancy, she delivered a live male baby on 19 Dec 96. Birth weight was 1800 g, length 41.0 cm, and head circumference 30.5 cm. The baby had bilateral parietal prominences, coarse scalp hair, saddle nose, anteverted nares, bilateral choanal atresia and bilateral low set ears. Both hands revealed increased gap between second and other fingers, with associated lowset thumbs.

The baby was markedly hypotonic, the heart rate was 60-70 per minute, and respiratory effort consisted of occasional gasps. Immediate endotracheal intubation was done and bagging with 100% oxygen was instituted along with intravenous fluids and care under a radiant warmer. The newborn had two cardiac arrests in quick succession and despite all resuscitative measures, the baby died within 2 hours of birth.

Plain X-rays films of the baby taken soon after birth revealed multiple, punctate, fine stippling of the cartilage ossification centres of the entire vertebral column (Fig 3), both ends of both humeri, both femora, and all bones of the feet. Both hands showed stubby, splayed fingers with features of soft tissue oedema correlating with the antenatal USG findings. The entire vertebral col-

Fig. 1 : Ultrasonography (USG) scans of the fetus revealing prominent open hands with separation of the fingers

Fig. 2 : USG scans of the thoracolumbar spinal column showing features suggestive of dysplasia with possible fragmentation

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Fig. 3 : Plain X-ray film of the newborn pelvis showing punctate, fine stippling of the cartilage ossification centers of the lumbosacral column and both femoral heads

umn showed features of 'coronal clefting' (Fig 4). There were no Wormian bones, abnormalities of skull shape, rhizomelia or situs inversus. The X-ray findings of the classical features of Chondrodysplasia Punctata clinched the diagnosis.

Discussion

The reported incidence of Chondrodysplasia Punctata is 1 in 1,10,000 [1,4]. It occurs in two forms, the autosomal dominant form also known as Conradi Hunermann Syndrome and the severe autosomal recessive form [1,2]. Babies with the latter form commonly have rhizomelia of the limbs [2,5]. Both these types show X-ray features of multiple, small foci of punctate calcification at knees, humerus, spinal column and soft tissues of laryngeal cartilages and trachea as was seen in this case [2,3,5,6].

Clinically, the babies with this disorder may have dysmorphic features in the form of prominent forehead, saddle nose with anteverted nares, hypertelorism, cataracts, excematous dermatitis with hyperkeratosis and erythema of skin in infancy, short stature, kyphoscoliosis, contractures of elbows, knees and hips. It is important to understand that not all features are present in a given case of Chondrodysplasia Punctata [1,3,6].

A hallmark in plain X-ray films of Chondrodysplasia Punctata is 'coronal clefting' of the vertebral bodies [7]. This is due to abnormal development and fusion at the two ossification centres at each vertebral body preventing the formation of a normal rectangular block of vertebra. This results in vertebrae with a gap in the body, seen clearly in the coronal plane. This feature was present in our patient.

Other conditions with stippling of the epiphyses include Zellweger syndrome, Warfarin embryopathy and trisomy 18 [1,2]. In Zellweger syndrome (Cere-

Fig. 4 : Plain X-ray film (lateral view) of the newborn spinal column showing 'coronal clefting' in each of the vertebral bodies

brohepatorenal syndrome) important features seen include severe hypotonia with poor reflexes in a baby with a round face with a high bulging forehead, brachycephaly and micrognathia. Hypertelorism, glaucoma, cataracts and Brushfield spots may be associated. None of these features were observed in our patient. Moreover, the calcific stippling in Zellweger Syndrome preferentially involves the knees and patella unlike the generalised distribution of stippling seen in CDP.

Warfarin embryopathy is another rare condition which is characterised by a baby with hypoplastic nose, deformed nasal cartilage, anteverted nares, microphthalmia, cataract, optic atrophy, blindness and macroglossia. Additionally, plain X-ray films in these cases show stippling limited to the epiphyses in the lumbosacral area and trochanters of femurs in association with hypoplasia of terminal phalanges and proximal phalanx of index finger. In our patient, neither did the mother have a history of warfarin intake, nor did the baby have the characteristics described above.

Trisomy 18 is a rare disorder with features of dolicocephaly, prominent occiput, microphthalmia, epicanthal folds, short palpebral fissures, small mouth, short upper lip, micrognathia, low set dysplastic ears, short neck, shield chest, short sternum, clenched hands, flexed and overlapping index fingers, limited movements of abduction at hip joints, short dorsiflexed hallux, and talipes calcaneovalgus. Hypotonia followed by hypertonia is a feature of this trisomy. Our patient did not have most of the above features.

The classical radiological picture in our patient and the rarity of this condition prompted us to report this case.

REFERENCES

1. Spranger JW, Langer W, Weidmann HR. Bone dysplasia: An atlas of congenital disorders of skeletal development. Philadelphia: WB Saunders Co Ltd 1974;342-53.
2. Goodman RM, Gorlin RJ. Chondrodysplasia Punctata syndromes: The malformed infant and child. An illustrated guide. New York. Oxford University press 1983:336-7.
3. Chapman S, Nakielny R. Irregular or stippled epiphysis. In: Aids to Radiological Differential Diagnosis, 3rd edn. Bangalore, WB Saunders Co Ltd. Prism Books Pvt. Ltd., 1995, p 47.
4. Camera G, Mastroiacovo P. Birth prevalence of skeletal dysplasias in the Italian Multicentre monitoring systems for birth defects. In: Papandatos CJ, Bartssokes CS, ed. Skeletal dysplasias. New York. Alan R Liss 1982:441-9.
5. Gilbert EF, Opitz JM, Spranger JW, et al. Chondrodysplasia Punctata-Rhizomelic form: Pathological and Radiological studies of three infants. Eur J of Paediatrics 1976;123:89-93.
6. Mason RC, Kozlowski K. Chondrodysplasia Punctata. A report of 10 cases Radiology 1973;109:145-7.
7. Fielden P, Russel JGB. Coromal cleft vertebrae. Radiology 1970;21:167-73.