Antenatal diagnosis of camptomelic dysplasia

Sir,

Campomelic dysplasia (CD, also known as camptomelic dysplasia) is a rare skeletal dysplasia characterized by the radiological features of short and bowed femora and tibiae, absent or hypoplastic fibulae, hypoplastic scapulae and pelvic bones, and nonmineralized thoracic pedicles. Clinically, an affected infant exhibits bowed lower limbs with pretilial skin dimpling, sex reversal (75%), dolichocephaly, micrognathia, cleft palate, a flat nasal bridge, low set ears, talipes equinovarus, and congenital dislocation of the hips. Patients usually succumb in the neonatal period owing to respiratory insufficiency.\(^1\) In almost all the cases of CD described in literature, accurate diagnosis was made postnatally. In the present paper, we describe the antenatal diagnosis.

A 22-year-old lady, normotensive, gravida 2, para 1, and living 1 with 28 weeks of gestation was referred for an ultrasound examination (USG), because clinically, the uterine size was large for gestational age. The USG revealed bowing of the long bones (Figures 1 and 2), club feet, reduced thoracic diameter, hypoplastic scapulae, hydronephrosis and polyhydramnios, consistent with the diagnosis of camptomelic dysplasia. The neonate expired within 1 hour of the birth owing to severe respiratory distress. It had ambiguous genitalia with a vaginal opening and a phallus (Figure 3). Babygram (Figure 4) confirmed the skeletal abnormalities seen on antenatal sonography. The parents refused an autopsy or karyotyping of the baby.

In our case, the first abnormality picked up was short and bent femora with hydramnios. Short limbs associated with hydramnios, mentioned frequently in the literature, is a useful marker for lethal skeletal dysplasia.\(^2\) On further screening, hypoplastic scapulae were seen, leading to the diagnosis of CD. Hypoplasia of the scapula is an important diagnostic feature of this condition, irrespective of the presence or absence of camptomelia of the femora.\(^3\) The prenatal differential diagnosis of this condition includes asphyxiating thoracic dysplasia, hypophosphatasia, osteogenesis imperfecta types 2 and 3, and unclassified varieties of congenital bowing of the long bones.\(^4\) All these differential diagnoses were excluded by the absence of facial anomalies, bell-shaped thorax, hypoplastic scapulae, and sex reversal, which can be assessed using an ultrasound.\(^4\) There is significant literature in the pathogenesis of this condition. It is now believed that mutations or translocations in the gene SOX9 on the long arm of chromosome 17, result in the syndrome of CD.\(^1,5,6\) These mutations only affect a single allele of SOX9, suggesting a dominant mode of inheritance for this syndrome.\(^1\) SOX9 plays an important role in chondrogenesis and testogenesis. Three important domains on it, HMG

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Figure 1: Bowing and shortening of femur in the fetus on antenatal ultrasound (arrow)

Figure 2: Bowing of tibia and fibula on antenatal ultrasound (arrow)

Figure 3: Clinical photograph of the abortus showing ambiguous genitalia (upper arrow), bowed lower limbs, and club feet (lower arrow)
domain, proline/glutamine/serine-rich C-terminal transcription activation domain, and dimerization domain, are needed for its function. Mild forms of CD, non-lethal type, or acampomelic dysplasia are most often associated with chromosome 17 rearrangements, resulting in decreased, rather than absent, SOX9 activity, or they may be owing to incomplete penetration or mosaicism of the mutant gene. Histological analysis of chondro-osseous tissues of the long bones of the lower extremities shows hypoplasia and scant organization of the chondrocytes in the columnar cartilage of the growth plate with increased vascular degeneration and scarce cytoplasmic calcification. Bony trabeculae are thin and poorly defined. Though we diagnosed CD at 28 weeks of gestation (first USG), the clear visibility of femora at 13–14 weeks should enable us to make the diagnosis earlier and help in counseling of the couple and further obstetric management.

Nagar AM, Sangle PM, Morani AC, Rajpal ND
Department of Radiology, KEM Hospital, Acharya Dhonde Marg, Parel, Mumbai-400012, India

Correspondence: Dr. Ajay Morani, E-mail: ajaycmorani@yahoo.com

References

Transient global amnesia following coronary angiography

Sir,

Transient global amnesia (TGA) is a memory disorder, which is characterized by an episode of amnesia and bewilderment lasting for several hours. A few cases have been reported in literature following coronary angiography. In this paper, we present the findings of a case and briefly discuss the various etiologies which are responsible for TGA.

A 32-year-old man was admitted for coronary angiography, having experienced an acute anteroseptal myocardial infarction 5 days earlier. He had a history of diabetes mellitus and was on regular treatment with oral glipizide. He was also receiving atenolol, aspirin, and sorbitrate. There was no past or family history of transient ischemic attack, epilepsy, migraine, or stroke. Physical examination was unremarkable. The electrocardiogram showed a qs pattern in the anteroseptal leads. His left ventricular ejection fraction was 35% on the echocardiogram. He complained of class III angina (Canadian classification) for which coronary angiography was performed. No sedation was used and 15cc of 1% lignocaine was infiltrated in the groin. The left ventriculogram was not performed. His blood pressure and arterial oxygen saturation was 170/90 mmHg and 95%, respectively, and his blood glucose was 153 mg/dl. The coronary angiogram revealed a recanalized left anterior descending artery and the other arteries were normal. A total of approximately 50 ml of omnipaque [Iohexol 350 mg iodine per ml] was used. As the artery had recanalized, no angioplasty was performed.

Three hours later, the patient complained of headache and started retching. He was talking irrelevantly and enquiring about his whereabouts. He had total disorientation in space and time. A neurology opinion was sought which did not reveal any clinical focal deficit. A CT scan of the brain done immediately (without contrast injection) was normal. Nine hours later the patient’s orientation was normal, however, he had retrograde amnesia. He had no recollection about the procedure he had undergone. Electroencephalogram done on the next day was normal. His blood sugar during the episode was 172 mg/dl. He was discharged on the third day without any neurological deficit.

TGA is the name applied by Fisher to a particular type of memory disorder, which is characterized by amnesia and memory disorder lasting for several hours. Transient dysfunction of the medial temporal lobe and the