Discussion

Chondrodysplasia punctata is characterized by stippled epiphysis and by growth disturbances in affected structures. Though heterogeneity exists among the three varieties described in the literature, few differences help in differentiating amongst them. Spranger et al. (5) described the two forms—rhizomelic and Conradi Hunermann syndrome. The rhizomelic type is a severe form of disease with autosomal recessive inheritance, characterized by symmetrical rhizomelic shortening, flexion contractures (60%), cataract (70-100%), ichthyosiform erythroderma (33%) and facial dysmorphism. Whereas the Conradi Hunermann type—an autosomal dominant verity is more heterogenous in its expression. The facial dysmorphic features and stippled epiphysis are the hall marks of the disease. Asymmetrical rhizomelic shortening is an usual feature. Cataracts and contractures are less frequently seen. Manzke et al. (6) described a third type known as X linked dominant form in female children with characteristic skin lesions and radiological findings, indistinguishable from Conradi disease. Prognosis of a patient with full blown picture is poor as most of them die in infancy. However some of them were noted to have lived up to 7 to 8 years.

Clinical conditions like warfarin embryopathy, Zellweger syndrome, trisomy 18 and 21 and cretinism needs to be entertained in differential diagnosis.

The classical clinical and radiological features in the present case are diagnostic of Chondrodysplasia punctata—Conradi Hunermann syndrome. However, no ocular changes were detected in the present case.

REFERENCES


Tolbutamide: Teratogenic Effects

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It is difficult to predict in a malformed infant of a diabetic mother whether the congenital deformity is due to disease per se or a consequence of drugs received

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during the pregnancy. However, the involvement of the drugs in the evolution of these malformations cannot be completely ignored. We present 2 such cases where the same drug used during pregnancy, perhaps, caused skeletal malformations.

Case Report

Case I: A boy was born to a 25-year-old primigravida mother, at term through a vaginal delivery and weighed 3.2 kg. The baby had an Apgar score of 6, 8 and 8 at 1, 5 and 10 minutes, respectively. There was no consanguinity. The mother was a known diabetic and was being treated by a general practitioner with oral hypoglycemics (tolbutamide). She continued the drug throughout her pregnancy without regular monitoring of blood sugar. There was no other relevant antenatal history and she had no formal antenatal check up.

On examination, the child had a length of 51 cm, head circumference 33 cm and upper-lower segment ratio 1.7:1. Both the upper and lower limbs were malformed and few digits were missing. There was syndactyly of the hands (Fig. 1). The power in the limbs was normal without any neurological deficit. The general physical and systemic examinations beside this were normal.

The laboratory parameters were normal. The radiological examination revealed the absence of phalanges of middle finger with syndactyly in left hand, while the right hand showed absence of 2nd and 3rd metacarpals as also the adjacent phalanges (Fig. 2). In the lower limbs, the left foot showed absence of all small bones except for one centre of calcaneum. The right foot had one calcaneal centre while the 3rd, 4th and 5th metatarsal bones and adjacent phalanges were absent. The stump of 2nd metatarsal was visualized (Fig. 3). The child had an uneventful stay in the nursery but was lost to follow up after discharge.

Case II: A boy, a full term infant, was born to a primigravida mother aged 28 years. The mother was being treated for diabetes mellitus for the past 2 years by a general practitioner with an oral hypoglycemic (tolbutamide). She visited the antenatal clinic of this hospital in her 2nd trimester when she was taken off tolbutamide and put on insulin. The diabetes during pregnancy remained under control.

The neonate suffered birth asphyxia
Fig. 2. Upper extremities—Left: Absent middle finger phalanges with syndactyly.
Right: Absent 2nd and 3rd metacarpals and adjacent phalanges.

Fig. 3. Lower extremities—Left: Absent small bones; only one calcaneal centre is visualized. Right: Missing 3rd, 4th, 5th metatarsal and adjacent phalanges.
and was transferred to the Neonatal Unit. On examination, the baby weighed 2.8 kg with a length of 49.5 cm and upper lower segment ratio of 1.7 : 1. The child had respiratory distress. The general physical examination revealed malformation of the upper limbs.

Investigations revealed normal hemogram, platelet count and biochemical parameters (serum calcium, blood urea, and sugar). The radiological examination showed the absence of radii in both upper limbs and bronchopneumonia (Fig. 4).

Discussion

Congenital malformations are known to occur in infants of diabetic mothers. The teratogenic effects of tolbutamide are also well known. But neither of these two are known to produce the kind of bony malformations that we encountered.

No exact cause of malformations in diabetes is known although a patient with uncontrolled diabetes mellitus during pregnancy has a higher risk of having malformed fetuses(1).

Other(2,3) have reported the caudal regression anomaly and congenital spinal anomalies which included sacral agenesis, vertebral spinal cord agencies, pelvic deformity, femoral hypoplasia, talipes equinus varius and muscular atrophy of lower extremity. The common malformations of the bone reported by Eriksson et al., in their study on congenital malformations in diabetic pregnancy were micrognathia and sacral dysgenesis(4).

Smithberg et al.(5) studied the teratogenic effects of hypoglycemic treatment in inbred strains of mice and found that 44% subjects developed anomalies when subjected to tolbutamide. The common anomalies reported were those of rib and vertebra. Exencephaly was another malformation found with this drug. However Belisle et al., had contrasting findings in their study on treatment of pregnant mice with tolbutamide. They found no gross anomalies with the use of this drug, while thrombocytopenia and petechiae were seen in most subjects(6). Menon et al., found a correlation between fetal macrosomia and concentration of animal insulin in cord serum; however, this effect leading to skeletal malformation has not been documented(7).

None of the workers have reported the skeletal malformations that we encoun-
tered in our two patients. Whether the malformations can be attributed to the same drug used by the two mothers or, to the disease per se is difficult to say. However, this drug should not be used during pregnancy and with utmost caution in young women who are likely to conceive because any accident may be disastrous for the fetus.

REFERENCES


Zygomycosis of Colon

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Zygomycoses (mucormycoses) refers to invasive human fungal infections caused by member of the phylum zygomata. They usually occur in presence of predisposing conditions like diabetes mellitus, lymphomas, leukemias and other immunosuppressed states(1). One third of the patients with zygomycoses have been infants or children. In these malnutrition and concomitant infection have been the most commonly encountered predisposing states(2). Rhinocerebral and cutaneous zygomycoses are the most common forms of infection; and gastrointestinal involvement is infrequently encountered(3). We report here a case of gastrointestinal mucormycoses in a child, perhaps for the first time from this part of the world.

Case Report

A 14-month-old malnourished child (weight 7.5 kg; expected 10.5 kg) was

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