Discussion

Carter et al., reported the prevalence of myositis ossificans progressiva to be less than 0.1 per million in the UK population(3). Review of Indian literature did not reveal any similar case report. The condition was first described by Patin in 1692(3) and since then many more cases have been described(3-6). Robbins(1) described the pathologic basis which is characterized by replacement of muscles, tendons, ligaments, fascia and aponeurosis by bone. It is invariably associated with some or other congenital anomalies such as absence of digit, teeth, hallux valgus, short big toe and thumb, and the disease may later involve heart and lungs leading to death(1). The condition is also known as fibrodysplasia ossificans progressiva as it has got no connection with myositis ossificans traumatica(7). Ours was an advanced case, there were multiple heterotrophic new bone formation taking shape of thick bands of bone in the neck, trunk, axilla and hip. The vital capacity of patient was markedly reduced indicating involvement of chest and respiratory muscles. Thornton et al. reported even viable pregnancy in a patient with myositis ossificans progressiva(6).

REFERENCES


Chondrodysplasia Punctata—Conradi Hunermann Syndrome

T. Redgeppa
K.S. Rao
S. Ramachandra
V.S. Gurunadh
V. Datta

Chondrodysplasia punctata formerly known as Chondrodystrophy calcificans congenita, was described to have transient calcifications in skeletal and respiratory cartilages. It is a rare disease (1:500,000), first described by Conradi(1) in 1914. So far 150 cases have been described in world literature, of which only seven cases were reported from India(2-4).

From the Departments of Pediatrics and Dermatology, Military Hospital, Agra Cantonment 282 001.

Received for publication November 13, 1990; Accepted December 10, 1990
Prior to 1970, the three major forms presently recognized as rhizomelic, Conradi Hunermann and X linked dominant form were not well differentiated. Conradi Hunermann syndrome is characterized by stippling of ischio-pubis and epiphysis of long bones (mushrooming of lower ends of femur and humerus), asymmetric rhizomelic shortening, flexion contractures, ichthyosiform erythroderma, congenital cataract and dysmorphic facial features. This communication describes one such case.

Case Report

A female infant aged four months, product of non-consanguinous marriage, born to a second gravida by normal vaginal delivery at home has been brought to the hospital with the complaints of failure to thrive and dry scaly skin. There was no maternal history of consuming anticoagulant drugs like Warfarin. The mother did not show any skin lesions. The first sib is alive and healthy.

Clinical examination revealed the infant to have rhizomelic shortening of both upper and lower limbs, flexion contractures involving both hips and knees, ichthyosiform erythroderma over trunk and lower limbs, depressed nasal bridge, prominent forehead, low set ears and short neck. No ocular changes were detected (Fig. 1).

Routine laboratory investigations did not reveal any abnormality. Radiological examination revealed stippling of the ischiopubis and epiphysis of the femur and tibia (mushrooming), including para articular calcifications around knee joint. Characteristic asymmetric rhizomelic shortening of left femur was observed (Fig. 2).

Fig. 1. Clinical photograph showing ichthyosiform erythroderma and dysmorphic facial features.

Fig. 2. Radiograph showing characteristic epiphyseal stippling.
Discussion

Chondrodysplasia punctata is characterized by stippled epiphysis and by growth disturbances in effected 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


Toibutamide: Teratogenic Effects

A. Saili
M.S. Sarna

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

From the Neonatal Division, Kalawati Saran Children's Hospital, New Delhi 110 001.
Reprint requests: Dr. Arvind Saili, 963, Vikas Kunj, New Delhi 110 018.
Received for publication November 23, 1990; Accepted May 3, 1991