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A Case of Rhizomelic Chondrodysplasia Punctata in Newborn

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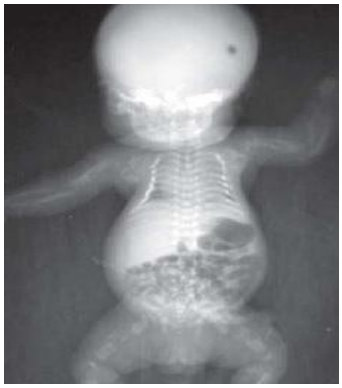
I. Introduction

Rhizomelic chondrodysplasia punctata (RCDP) is a rare disorder of peroxisomal metabolism, with an estimated incidence 1□ :□100,000. There are 3 genetic subtypes. RCDP type 1, caused by mutations in the PEX7 gene, is the most common type. RCDP type 2 and 3 are single enzyme deficiencies in the plasmalogen biosynthesis pathway. The main features of the disease are shortening of the proximal long bones, punctate calcifications in the metaphysis and epiphysis of long bones and the thoracic and lumbar vertebrae, dysmorphic face, and severe growth retardation.

II. Case Report

The term infant (Fig1) was admitted to the neonatology department because of its atypical facial appearance and extremity anomalies on Day1. The female infant, at 38 weeks of gestation was the 2nd order of a healthy 22-year-old mother. She did not have any chronic disease and there was no history of drug exposure. On physical examination, birth weight was 2.3kg, height was 50□ cm, head circumference was 34□ cm, and there was depressed nasal bridge and a highly arched palate. There was shortness of the upper extremities and flexion contractures in all extremities. The infant's systemic examination was otherwise normal.Xray revealedproximal boneshortness, thick and short diaphyses, and large and irregular metaphyses in the long bones and normal fingers. The radiological findings of the patient were compatible with CDP with punctate calcifications in the epiphyses. Complete blood count, biochemical parameters, and abdominal ultrasonography were all normal.



III. Discussion

Chondrodysplasia punctata (CDP) is a rarely occurring skeletal dysplasia characterized by stippled, punctuate calcifications around joints and within cartilages [1]. CDP is associated with a number of disorders, including inborn errors of metabolism, involving peroxisomal and cholesterol pathways, embryopathy, and chromosomal abnormalities [2–7]. Several classification systems of the different types of CDP have been suggested earlier. More recently, the biochemical and molecular basis of a number of CDP syndromes has recently been elucidated and a new aetiological classification has emerged [2]. Rhizomelic chondrodysplasia punctata is a disorder caused by abnormal peroxisomal function which can be mediated both through disorders of biosynthesis, for example, peroxisomal assembly (RDCP1), and by single enzyme defects, affecting plasmalogen synthesis (RCDP2, RCDP3). Clinically, RCDP1, RCDP2, and RCDP3 are characterized by rhizomelic shortening, mainly affecting the humerus, facial dysmorphism, seizures, cataracts, and joint contractures. Growth and development are severely restricted. Life expectancy is considerably reduced [1, 2, 8]. Pathognomonic finding for RCDP is a reduced level of plasmalogens with normal very long chain fatty acids (VLCFA) [1]. The following situations should be considered in the differential diagnosis of CDP: peroxisomal diseases (Zellweger Syndrome, adrenoleukodystrophy, and infantile Refsum disease), maternal disease, and exposure to warfarin, Smith-Lemli-Opitz Syndrome, and foetal alcohol syndrome [2, 9, 10]. There was no history of maternal drug or alcohol use and no symptoms or positive laboratory test that indicated autoimmunological disease in the mother.

Prenatal diagnosis of RCDP is possible from the first trimester onwards by demonstration of peroxisomal dysfunction in cultured chorionic villous or amniotic fluid cells [3]. Genetic counseling was given to parents of our case.Since only few cases have been reported in literature,we find it worthwhile to report this case.

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