SPOT DIAGNOSIS (IMAGE GALLERY)

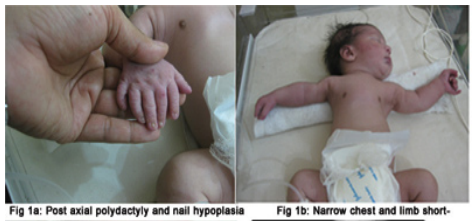

Fig 1a: Post axial polydactyly and nail hypoplasia

Fig 1b: Narrow chest and limb short-

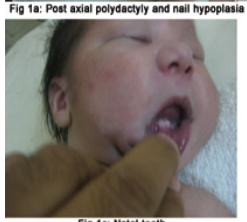

Fig 1c: Natal teeth

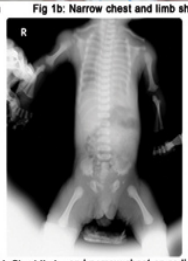

Fig 1d: Short limbs and narrow chest on radiogram

DISPROPORTIONATE SHORT STATURE WITH POLYDACTYLY

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We report a male newborn, born to consanguineously married couple by caesarean section. Newborn was referred to us for respiratory distress with meconium aspiration. He was treated with oxygen by hood for 7 days, IV antibiotics, IV fluids and was discharged on 10th day of life against medical advice on oral diuretics for his impending cardiac failure due to congenital heart disease. His physical examination showed narrow chest, postaxial polydactyly in both hands, nail hypoplasia, lip tie, natal teeth, disproportionate dwarfism. (Figure 1) Antenatal ultrasound done at 30 weeks showed absent inter-atrial septum. His x-ray of extremities showed short long bones (Figure 1). On family history his elder sibling, male child, died at age of 1 month due to respiratory problems. He had 7 fingers in both hands and 6 in each foot. In family, few members are dwarf adults. His cousin also expired in neonatal period and had polydactyly.

What is the diagnosis?

Ellis-van Creveld syndrome. It is also termed as chondroectodermal dysplasia and is an autosomal recessive disorder characterised by the clinical features of postaxial polydactyly, disproportionate dwarfism, narrow chest, lip tie and natal teeth, nail hypoplasia and congenital heart anomaly usually single atrium. The incidence of this syndrome is 1:60000 in general population with increased incidence among persons of old order Amish descent. Being autosomal recessive disorder, varied phenotypical presentation is the rule. Thoracic dysplasia leads to respiratory insufficiency and cardiac anomalies lead to death in infancy in 50 percent of patients. Patients who survive infancy have a normal life expectancy. Developmentally, most patients have had intelligence in the normal range

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