Short Rib Polydactyly Syndrome Type I (Saldino-Noonan Syndrome)

A full term female newborn, weighing 2 Kg, was born with severe shortening of limbs with bilateral postaxial polydactyly. Chest was very narrow. There was polysyndactyly of right and left toe (*Fig. 1*) and anthropometry revealed a length of 40 cms, upper segment 28 cm, arm span 35 cm and chest circumference 21 cm. The neonate had respiratory distress since birth and died after 5 hrs. A clinical diagnosis of short-rib polydactyly syndrome was made. The skeletal skiagram revealed short ribs, small pelvis, short tubular bones *(Fig. 2*).

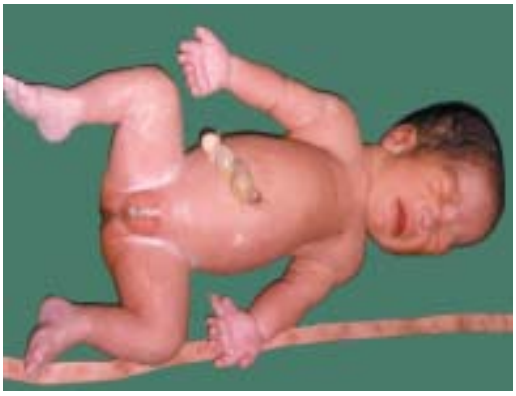

Fig. 1 Clinical photograph of a neonate with SRPS.

Fig. 2. Skeletal skiagram of a newborn with SRPS.

Short rib-polydactyly syndrome is a descriptive category for a group of lethal skeletal dysplasias characterized by a hypoplastic thorax, short ribs, short limbs, polydactyly and visceral abnormalities. The four established variants are SRPS I (Saldino-Noonan type), SRPS II (Majeviski type), SRPS III (Verma-Naumoff type) and SRPS IV (Beemer-Langer type). All the variants are thought to be inherited as an autosomal recessive pattern. SRPS type I is characterized by narrow thorax, short limbs and post axial polydactyly. The base of the ileum is hypoplastic and the vertebrae are rounded sometime with coronal cleft. The ends of the long bones are either pointed or have a convex central area of ossification with lateral metaphyseal spikes. Associated genitourinary and anorectal and cardiac malformations are frequently seen. Prenatal diagnosis by ultrasound can be made by detecting the triad of micromelic dwarfism, short and horizontal ribs with narrow thorax and polydactyly.

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