

Grebe syndrome antenatally because of history of previous two siblings affected with the same disorder and by comparing BPD and length of limb bones. In the affected male fetus the BPD was on the 50th percentile, whereas all the other long bones measured were less than 5th percentile(5,6), thus indicating limb reduction anomaly in the fetus. The family history of 2 previously affected sibs was an important pointer for the diagnosis. This only emphasizes the importance of good family history and study of members of family before attempting prenatal diagnosis by ultrasonography.

Ultrasonological features noted in our case included shortening of all 4 limbs, lower limb bones shorter than upper limb bones, the severity of shortening increasing distally with short bulbous fingers. The high incidence of premature deaths accounting for 11% of stillbirths and 38% of infant mortality(7) and the extreme disability it produces in an affected individual, may warrant early detection by ultrasound and possibly termination of pregnancy.

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## Lethal Forms of Short Limb Dwarfism

**K.S. Srinath**  
**B. Vishnu Bhat**  
**M. Ravi Kumar**

Short limb dwarfism is part of tubular bone chondrodysplasia syndromes. It

can manifest both in lethal and non-lethal forms(1). Short rib-polydactyly

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*From the Department of Pediatrics (Neonatal Division), Jawaharlal Institute of Postgraduate Medical Education and Research, Pondicherry 605 006.*

*Reprint requests: Dr. B. Vishnu Bhat, Professor of Pediatrics and Incharge of Neonatal Division, Department of Pediatrics, JIPMER, Pondicherry 605 006.*

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syndromes (SRP), thanatophoric dwarfism, achondrogenesis, osteogenesis imperfecta-type II are some of the lethal forms. Asphyxiating thoracic dystrophy and Ellis-Van Creveld syndrome are generally considered nonlethal but a few cases of lethal forms have been described(2-4). Five cases of lethal short limb dwarfism including one family with three affected siblings seen among 21,891 births over six years period are being reported.

### Case Reports

*Case 1:* A male baby weighing 2.06 kg was born to a 19 years old primigravida at 36 weeks gestation by Cesarean section for breech presentation. There was no significant antenatal illness and the parents were nonconsanguinous. The baby had an Apgar of 7/10 at 1' and 8/10 at 5'. He had a small chest with short limbs, spade like hands with short fingers, dysmorphic features, a length of 35 cm, span of 37.5 cm and head circumference of 33.5 cm. He developed respiratory distress at birth and required oxygen, intravenous fluids and antibiotics. His condition deteriorated and he expired on the 5th day of life. Radiograph revealed short ribs and limbs with telephone receiver type of long bones, flat vertebrae and large cranium suggestive of thanatophoric dysplasia (*Fig. 1*).

*Case 2:* A female baby weighing 3 kg was born at term gestation to a 20 year old primigravida mother by Cesarean section for fetal distress. There was history of drug intake on and off during pregnancy for headache. The mother was father's first cousin. The baby had an Apgar of 4/10 at 1' and 2/10 at 5' and 10'. She died after 15 min. The baby had short limbs, depressed nasal bridge,

hypertelorism, postaxial polydactyly of all the four limbs, abdominal distension and hepatosplenomegaly. The plain radiograph (*Fig. 2*) showed short limbs with hypoplastic ilia and short ribs suggestive of short rib polydactyly syndrome. Autopsy revealed hypoplastic lungs, malrotation of intestine, accessory spleen and patent ductus arteriosus in addition to the external features described.

Cases 3-5: These three babies were siblings born to uncle-niece consanguinous parents. The mother had polyhydramnios during all the three pregnancies. She had congenital aortic stenosis and her karyotyping revealed a



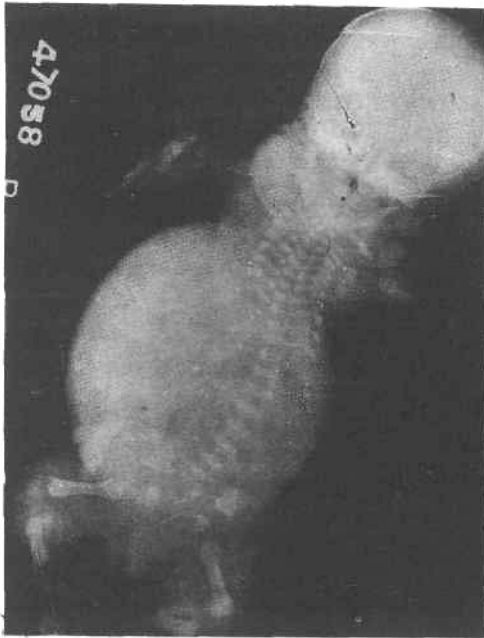
*Fig. 1.* Radiograph of Case 1 showing telephone receiver type of long bones, flat vertebrae and large cranium.

mosaic pattern of 46/xx, 45/xx with D to L translocations. There was no history of infection or exposure of teratogens during the antenatal periods. The features observed among the three siblings are shown in *Table I*. The radiological findings characteristic of short rib polydactyly syndrome (SRP), *viz.*, short ribs and limbs with poorly developed pelvis, are shown in *Fig. 3*.

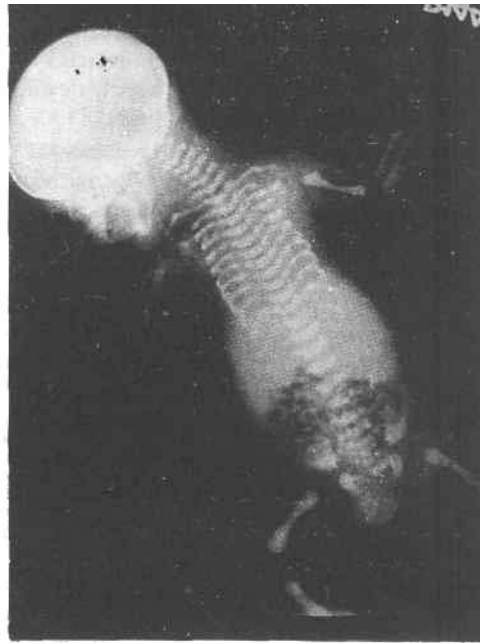
### Discussion

Lethal forms of short limb dwarfism are rare and most of these conditions are inherited as autosomal recessive(1). Thanatophoric dysplasia is the most commonly observed lethal form of short limb dwarfism. Apart from short limbs, they have prominent forehead, small pear shaped chest and hypotonia. Pres-

ence of telephone receiver type of long bones, flat vertebrae and large cranium as observed in *Case 1* are characteristic. The condition is inherited as autosomal dominant(5,6). Thanatophoric dwarfism can rarely be associated with clover leaf skull (Kleeblattschadel syndrome) deformity (7). The majority of reported cases have died within a few hours after birth or were still births and only one case lived for 25 days(8). Thanatophoric dwarfism should be differentiated from achondrogenesis. Radiologically, babies with achondrogenesis have thin ribs with multiple fractures, poorly ossified skull and vertebral bodies. This condition is transmitted as autosomal recessive(9).



*Fig. 2.* Skiagram of Case 2 revealing short ribs, poorly formed iliac bones and short limbs.



*Fig. 3.* Radiograph of Case 5 showing short limbs with polydactyly in the upper limbs, short ribs and poorly formed pelvic bones.

**TABLE I-Features of SRP Syndrome Among the Siblings**

Features	Case number		
	3	4	5
Sex	Female	Male	Male
Birth weight (Kg)	1.15	2.2	2.4
Gestation	36 weeks	37 weeks	Term
Survival	15 min	10 min	Still birth
Short limbs	+	+	+
Short ribs	+	+	+
Polydactyly	4 limbs	4 limbs	Upper limbs only
Natal teeth	+	+	-
Anal atresia	+	-	-
Hypoplastic nails	-	-	+
Absence of eye brows and lashes	-	-	+
CTEV	-	-	+
Radiography	Short ribs & limbs with poorly developed pelvis	Same as Case 3	Fig. 3 showing characteristic features
Autopsy	Hypoplastic lungs with anal atresia	-	-

Short rib-polydactyly syndromes are further classified into four types, *viz.*, SRP I (Saldino-Noonan), SRP II (Majewski), SRP III (Verma. Naumoff) and SRP IV (Beemer-Langer)(10-14). SRP I and III have similar morphological features with high incidence of cloacal involvement. They are differentiated based on the extent of rib involvement and short cranial base seen in SRP III. SRP II and IV have associated cleft lip and palate with minimal pelvis involvement. SRP IV has no polydactyly which distinguishes it from SRP II(14). Asphyxiating thoracic dystrophy has narrow chest and sometimes polydactyly in

the upper limbs. Ellis-Van-Creveld syndrome (chondroectodermal dysplasia) is associated with ectodermal abnormalities like absent hair, hypoplastic nails and natal teeth. These two conditions are generally nonlethal but lethal forms have been described(1-4).

Case 2 had features of SRP I but the presence of malrotation of gut and accessory spleen and absence of cloacal abnormalities are unusual. All the three siblings (Cases 3-5) had natal teeth while the third sibling also had hypoplastic nails, absent eye brow and eye lashes along with talipes equinovarus.

Anorectal malformation was present in the first sibling only. The third sibling had polydactyly in the upper limbs only. The clinical and radiological features suggest SRP I. Presence of ectodermal abnormalities and absence of cloacal defects in the later two siblings are unusual and point towards the possibility of Ellis-Van-Creveld syndrome which is often nonlethal. Polydactyly is almost always seen in SRP syndromes and chondroectodermal dysplasia and less often in asphyxiating thoracic dysplasia (ATD). It is possible that SRP and Ellis-Van-Creveld syndromes are different manifestations of the same condition since the varying features were observed in the same family(10-12).

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