

## Case Report

# Rare cases of skeletal dysplasia: 1) Campomelic dysplasia, 2) Radial ray defect

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### Introduction

Fetal skeletal dysplasia mainly occurs in a sporadic or autosomal recessive pattern. Majority of the cases with skeletal dysplasia are associated with polyhydramnios, fetal structural anomalies, or positive family history.

### Campomelic dysplasia

#### Case report: 1

A 30 year old 4<sup>th</sup> gravida with 8 months amenorrhea came for routine antenatal checkup on 8<sup>th</sup> January 2004. She had two female babies aged nine and six years. Her one male child who had short limbs died within 1 hour of birth three years ago. Nothing was contributory in her personal history, family history, general examination and systemic examination. Her routine sonography showed a single intrauterine pregnancy with biparietal diameter, 8.1cm (33w, 2d), femur length 3.9 an (21w, 5d), tibia 4cm, fibula 3.9cm, humerus 4.2cm, radius 3.7cm, ulna 4.4 cm (Figure 1).

All these long bones were showing bowing of variable degrees. The fetus also showed bilateral talipes equinovarus. These features were suggestive of normocephalic campomelic dysplasia (rhizomelic and mesomelic dysplasia). She was counseled following which she asked for a termination of pregnancy. Induction was done with dinoprostone gel. She delivered a stillborn male child weighing 1.7 kg on 12<sup>th</sup> January 2004. An x-ray of the baby showed bowing of bones in the lower extremities and absence of radius and fibula on left side (Figure 2).

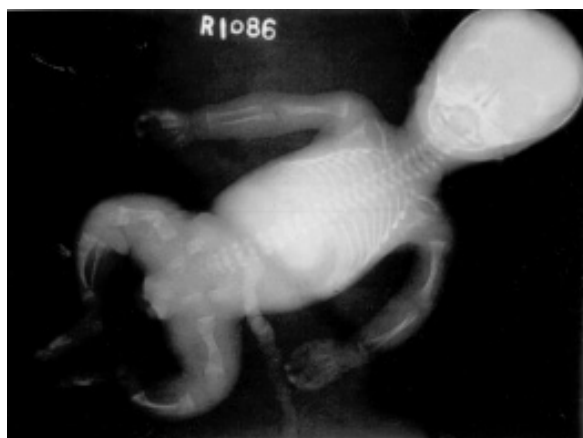


**Figure 1.** Sonography showing bowing of left femur case 1.

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**Figure 2.** X-ray of baby of campomelic dysplasia (case 1) showing bowing of bones in the lower extremities and absence of radius and fibula on left side

## Discussion

The incidence of campomelic dysplasia is about 1 in 150,000 births. The features of campomelic (“bent bone”) dysplasia include ventral bowing of shortened tibia and femora, severe talipes equinovarus, absent or hypoplastic fibulas, extremely small scapulae, narrow thorax and hypoplastic pedicles of the thoracic vertebrae<sup>1</sup>. The bowing of the tibia and femora probably results from a primary shortness of the calf and hamstring musculature<sup>1</sup>. Campomelic dysplasia is a fatal condition with few exceptions of survivors for shorter duration.

## Radial ray defect

### Case report 2

A 35 year old G<sub>4</sub> P<sub>3</sub> was referred as a case of eclampsia with history of 9 months amenorrhea with severe anemia. She was unregistered and unsupervised. Labor was induced. She delivered a stillborn male child weighing 2.5 kg. The baby had only three fingers on left hand with thumb and index finger absent. The baby was also having imperforate anus. X-ray showed absence of radius, thumb, index finger, corresponding metacarpals, fibula on the left hand (Figure 3).



**Figure 3.** X-ray of baby of radial ray defect (case 2) showing absence of radius, thumb, index finger, corresponding metacarpals, and fibula on the left side.

## Discussion

There are several degrees of radial defects, from the mildest form of hypoplasia of the 1<sup>st</sup> metacarpal and thumb to the most severe form of absence of both radius and ulna<sup>3</sup>. The incidence among live births is estimated at 1/30,000<sup>4</sup>. Radial hypoplasia or aplasia is seen in many syndromes and conditions, including Holt-Oram syndrome, thrombocytopenia-absent radius (TAR) syndrome, VATER association (Vertebral and Vascular anomalies, Anorectal atresia, Tracheo-esophageal atresia, and radial limb anomalies), trisomy 18, Roberts-SC phocomelia, and exposure conditions such as fetal valproate syndrome<sup>4</sup>. Therefore, prenatal detection of an absent or abnormal radius requires further evaluation of all fetal limbs and of the fetal anatomy.

## References

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