

Picture Story

## Short thorax and disproportionate dwarfism due to Kniest dysplasia

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*Sri Lanka Journal of Child Health*, 2003; 32: 25-6

(Key words: Kniest dysplasia, short stature)

A breastfed male infant aged six weeks presented with constipation and weight loss. He was the third born to a 33 year old mother in a non-consanguineous marriage. Both siblings were normal.

Abnormalities observed were: short length (crown-to-heel 46cm), disproportionate body proportions, short barrel shaped chest (Figure 1), a relatively large head, kyphoscoliosis, enlarged knees and elbows with limited range of active and passive movements, bilateral inguinal herniae, flat facies, prominent eyes and a wide posterior cleft palate. Cardiovascular, respiratory, abdominal and neurological examinations were normal.



Figure 1. Infant with short chest, large joints and inguinal hernia.

X rays showed vertebral clefts in thoracic spine (Figures 2 & 3), flared metaphyses and large epiphyses in femur and tibia (Figure 4).

Expressed breast milk fed using a long teat corrected weight loss and constipation. Cleft palate repair was planned for nine months of age. Herniotomy was performed. On follow up at four months he had satisfactory weight gain and normal development.

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(Received on 23 Jan 2003)



Figure 2. X rays spine (AP view) – short thorax with vertebral clefts.



Figure 3. X ray spine (lateral view) – short barrel chest and vertebral anomalies.



Figure 4. Large epiphyses and flared metaphyses of femur and tibia.

### Discussion

The absence of respiratory difficulty and lung hypoplasia made asphyxiating (Jeune) thoracic dysplasia unlikely to be the cause of short thorax. The skeletal abnormalities characteristic of spondyloepiphyseal dysplasia congenita (i.e. delayed mineralisation of epiphyses and narrow vertebral discs without vertebral clefts) were not found.

The diagnosis of Kniest dysplasia was based on characteristic dysmorphology and typical radiological features<sup>1,2</sup>. Children with this particular osteochondrodysplasia have normal intelligence and good prognosis. Short stature and orthopaedic problems are likely<sup>1,3</sup>. The definitive diagnosis brightened the outcome. Correcting the feeding difficulty was the simple intervention necessary for significant improvement.

### References

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