Correspondence

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Thanatophoric dysplasia in identical twins

SIR,

The report of thanatophoric dysplasia in identical twins that recently appeared in your pages1 is of interest. The diagnosis of thanatophoric dysplasia must be questioned, however, because the authors state: “No other external or internal abnormalities were found at necropsy.” Megalencephaly and highly characteristic temporal lobe malformations invariably are present in thanatophoric dysplasia and other abnormalities in central nervous system topography are frequently apparent by microscopy.2,3 Varieties of lethal short limbed platyspondylic dysplasia that closely resemble thanatophoric dysplasia externally and by radiographical criteria are recognised.4 To my knowledge, central nervous system abnormalities have not been identified in infants with these disorders. Might not this pair of twins actually represent a form of lethal short limbed dwarfism other than thanatophoric dysplasia?

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References


This letter was shown to Drs Young and Lamont who reply as follows.

Dr Knisely may well be correct in suggesting that the absence of observed CNS abnormalities raises doubt about the diagnosis of thanatophoric dysplasia in these twins. However, based on radiological criteria we feel that this is by far the most plausible diagnosis.

Anteroposterior and lateral radiographs of these twins showed the classical and distinctive hallmarks of thanatophoric dysplasia, that is, U/H shaped vertebral bodies on AP view with narrowing of the mid portion and anterior rounding visible on lateral projection, short bowed femora with metaphyseal spurs, hypoplasia of the lateral aspect of the proximal tibiae, and disproportionate widening of the fibulae. In contrast, babies with variant forms of lethal short limbed platyspondylid dwarfism1,2 tend to show extreme platyspondyly of the vertebral bodies, straighter and more tubular long bones, and absence of proximal lateral tibial hypoplasia. These points are illustrated in the figure which shows

FIGURE Whole body radiograph of a baby with a variant form of lethal short limbed platyspondylid dysplasia showing wafer thin vertebral bodies and short femora with minimal bowing.
the radiograph of an infant (unpublished) with a variant form of lethal short limbed platyspondylic dysplasia.

We agree entirely that careful neuro- and histopathological study is indicated in every case of lethal short limbed platyspondylic dwarfism if the undoubted heterogeneity in this group of disorders is ever going to be fully delineated. Meanwhile, we would like to draw attention to a recently published study from Spain in which a parental age effect was documented for thanatophoric dysplasia, thereby providing further evidence that babies with this disorder are likely to represent new autosomal dominant mutations.

References
Correspondence

I D Young and A C Lamont

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