Thanatophoric dysplasia in identical twins

Sir,

The report of thanatophoric dysplasia in identical twins that recently appeared in your pages 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. Varieties of lethal short limbed platyspondylus dysplasia that closely resemble thanatophoric dysplasia externally and by radiographical criteria are recognised. 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.

FIGURE Whole body radiograph of a baby with a variant form of lethal short limbed platyspondylus dysplasia showing wafer thin vertebral bodies and short femora with minimal bowing.