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

- ¹ Young ID, Patel I, Lamont AC. Thanatophoric dysplasia in identical twins. *J Med Genet* 1989;26:276-9.
- ² Wongmongkolrit T, Bush M, Roessmann U. Neuropathological findings in thanatophoric dysplasia. *Arch Pathol Lab Med* 1983;107:132-5.
- ³ Ho K-L, Chang C-H, Yang SS, Chason JL. Neuropathologic findings in thanatophoric dysplasia. *Acta Neuropathol (Berl)* 1984;63:218-28.
- ⁴ Horton WA, Rimoin DL, Hollister DW, Lachman RS. Further heterogeneity within lethal neonatal short-limbed dwarfism: the platyspondylic types. *J Pediatr* 1979;94:736-42.

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 platyspondylic dwarfism^{1,2} 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 platyspondylic dysplasia showing wafer thin vertebral bodies and short femora with minimal bowing.