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Prenatal cortical hyperostosis (Caffey disease)

Received: 29 August 2001 Accepted: 11 April 2002 Published online: 4 July 2002 © Springer-Verlag 2002

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We read with interest the letter from Basel et al. [1] documenting the case of a 27-week-old aborted fetus with an unusual congenital bone dysplasia and requesting additional opinion regarding diagnosis. The major radiographic findings reported in the fetus comprise marked cortical hyperostosis with consequent periosteal 'cloaking' of the long bones, which were poorly modelled, and asymmetrical bowing of the bones of the lower extremity.

These appearances are consistent with the diagnosis of 'prenatal cortical hyperostosis (Caffey disease)'. This condition appears to be distinct from the infantile form of cortical hyperostosis described by Caffey [2] in which the onset of pathology is postnatal.

We have previously presented clinical, radiographic and histological findings of nine new cases of this prenatal form of cortical hyperostosis [3]. It appears that prenatal onset, polyhydramnios, bowing of long bones and generalised, symmetrical involvement of the skeleton help

distinguish this condition from the 'classic' postnatal form of Caffey disease. The condition appears to be inherited as an autosomal recessive trait [3], which has important implications for genetic counselling in the family of the fetus reported by Basel et al.

In conclusion, we agree with the second differential diagnostic possibility put forward by Basel et al. [1] that this fetus has "affey's disease in its severe congenital form" that we have termed 'prenatal cortical hyperostosis'.

References

- Basel D, Beighton P, Kozlowski K (2001). Help! Unusual X-ray appearances of a congenital bone disease of unknown aetiology. Pediatr Radiol 31:212
- 2. Caffey J, Silverman W (1945). Infantile cortical hyperostosis, preliminary report on a new syndrome. AJR 54:1–16
- Cormier-Daire V, Savarirayan R, Wilcox WR et al. (1999). Prenatal cortical hyperostosis (Caffey disease): clinical, radiographic and chondroosseous findings in 9 new cases. Am J Hum Genet 65[Suppl A7]:28