
Exclusion of retinoic acid receptor and a cartilage matrix protein in non-syndromic CL(P) families

We read with interest the report of Vintiner et al. 12 exclusion linking to RARA (17q21), D17S2473, D17S2476, D17S2475, and D17S2478 RARA to the X chromosome. The mutation which was located within the SH2 domain, and the mutation in our patient is a new missense mutation within the SH2 domain.

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Further report of a patient with humeroulnar synostosis and hydronephrosis

A case of humeroulnar synostosis with lambdoid synostosis was published recently in this journal. We report on a male baby with clinical and skeletal abnormalities very similar to those previously reported. 1–4

The proband was the first child of young and healthy non-consanguineous parents. He was born at term following a normal delivery because of cephalopelvic disproportion. Birth weight and length were 3490 g and 51 cm, respectively. Apart from the upper limb defects and a midline capillary haemangioma, no other abnormality was found on physical examination.

At 11 months he measured 73.5 cm (50th centile), head circumference was 47 cm (50th centile), and weight was 8300 g (10th centile). Psychomotor development has been normal. Both upper limbs were short, the left one more markedly than the right. Both shoulders had normal range of movement. The left upper limb was shorter than the right and kept in a fixed position; there were two digits joined to TCTE, which included the HLA region. 5

Our findings and those of Vintiner et al. suggest that RARA, CRTL1, and F13A1 do not have a major causal role in the aetiology of CL(P) in the 18 families tested. Therefore, as previously suggested, we cannot dismiss the possibility that RARA plays a modifying role in the aetiology of CL(P). 3

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