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The etiologic factor of cleidocranial dysplasia

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To the Editor:

We read with great interest the April article by Kelkar et al¹ reporting a humoral immunodeficiency associating cleidocranial dysplasia and von Willebrand's disease in a family cluster. There is an issue concerning the chromosomal location of cleidocranial dysplasia that we would like to raise.

Recently, the etiologic factor of cleidocranial dysplasia, the CBFA1 gene, was mapped on the short arm of chromosome 6 (OMIM 119600).² CBFA1 is considered a master gene in the formation of bone and dental tissue.

This discovery opens the door for phenotype-genotype correlations, allowing actual intensive studies on the gene function in developmental and repair mechanisms.³ Furthermore, minor cases can now be diagnosed independently from the clinical expression.

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