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Robertsonian translocations in Paget's disease of bone.

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Dear Editors:

Several concerns have arisen after reading the short communication "Robertsonian Translocations in Paget's Disease of Bone" by B. G. Mills et al. (*J Orthop Res* 15:477–481, 1997). The authors reviewed the karyotypes of 14 patients with Paget's disease of bone. Eight of the 14 patients had chromosomal translocations in the D and C groups. Thus, they concluded that 57% of their patients with Paget's disease had Robertsonian translocations. Using age and sex-matched controls and on the basis of the low prevalence of Robertsonian translocations in the general population, they concluded their data suggested that an environmental factor introduced during the lifetime of the patient affected gene replication and induced the development of Paget's disease.

This is an important topic, to our knowledge, no cytogenetic analysis of Paget's disease of bone has been undertaken. However, on careful review of the manuscript three important issues are raised.

- a. What is the correlation between abnormal karyotypes identified in leukocytes and abnormal karyotypes detected in pathologic osteoblasts? This cytogenetic study was performed on whole-blood analyses of patients with Paget's disease of bone. How was the diagnosis of Paget's disease made? Was there any tissue confirmation? How are chromosome changes in leukocytes correlated with bone pathology?
- b. Figure 1 does not uniformly demonstrate Robertsonian translocations. For example, chromosomes 13/15, 14/15, 15/22, and 22/22 may in fact represent telomeric associations. The results and conclusions are greatly affected if the findings for the chromosomes are due to telomeric associations and not to Robertsonian translocations.
- c. Additional studies should have been performed to characterize the chromosome abnormalities such as C banding, argyrophilic nucleolar organizer region staining, and molecular cytogenetics by *in situ* hybridization with alpha centromeric probes. Without these confirmatory studies, one cannot conclude that whole arm translocations occurred.

An alternative explanation for their findings is proposed. Telomeric associations are a more common and more anticipated cytogenetic finding. Telomeric associations have been identified in giant-cell tumor of bone and in other musculoskeletal neoplasms (1–4).

Without addressing these important issues, the finding of the manuscript cannot be verified.

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