

Mapping of the human tuftelin (TUFT1) gene to Chromosome 1 by fluorescence in situ hybridization

D. Deutsch,^{1,2} A. Palmon,¹ M.F. Young,² S. Selig,³ W.G. Kearns,⁴ L.W. Fisher²

¹Dental Research Unit, Hebrew University Hadassah School of Dental Medicine, P.O. Box 1172, Jerusalem, Israel 91010

²Bone Research Branch, National Institute of Dental Research, National Institutes of Health, Bethesda, Maryland 20892, USA

³Department of Cellular Biochemistry, Hebrew University, Hadassah Medical School, Jerusalem, Israel 91010

⁴John Hopkins University School of Medicine, Center for Medical Genetics, Baltimore, Maryland 21205, USA

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Tuftelin is a conserved novel acidic protein (Deutsch et al. 1991a) found in developing and mature extracellular enamel (Deutsch et al. 1991b), a unique and highly mineralized ectodermal tissue covering vertebrate teeth. Tuftelin is thought to play a major role in the mineralization and structural organization of enamel (Termine et al. 1980; Deutsch 1989).

We have recently cloned (Deutsch et al. 1992) and partially sequenced (four exons) the tuftelin gene from a human genomic library contained in Lambda Fix (purchased from Stratagene), employing the bovine tuftelin cDNA probe (Deutsch et al. 1991b). This human tuftelin genomic clone was used as a probe for fluorescence in situ hybridization on human metaphase chromosomes. Metaphase chromosome preparations were prepared from phytohemagglutinin (PHA)-stimulated peripheral blood lymphocyte cultures according to standard cytogenetic protocol. The tuftelin genomic clone was biotinylated by nick translation (Kievits et al. 1990), hybridized, and detected by two layers of Fluorescein Avidin DN (Vector Laboratories; Gartner et al. 1993). The chromosomes were counterstained with propidium iodide (0.05 µg/ml) and DAPI 4',6-diamidino-2-phenylindole (0.05 µg/ml) in antifade medium (Johnson and de Noguera 1981; Shimozawa et al. 1992). The slides were viewed in a Leitz Aristoplan epifluorescent microscope equipped with a double band pass filter (FITC and Texas Red) and a DAPI filter. Fifty-one metaphase spreads were analyzed, and 32/51 (62.7%) showed labeling in both chromatids of each long arm of Chromosome (Chr) 1 (Fig. 1). These data suggest that the tuftelin gene localizes to the long arm of Chr 1 approximately at bands q21–q31. The localization of the human tuftelin gene to a well-defined cytogenetic region may be important in understanding the etiology of the autosomally inherited Amelogenesis Imperfecta disease (AI), the most common heredity disease of enamel.

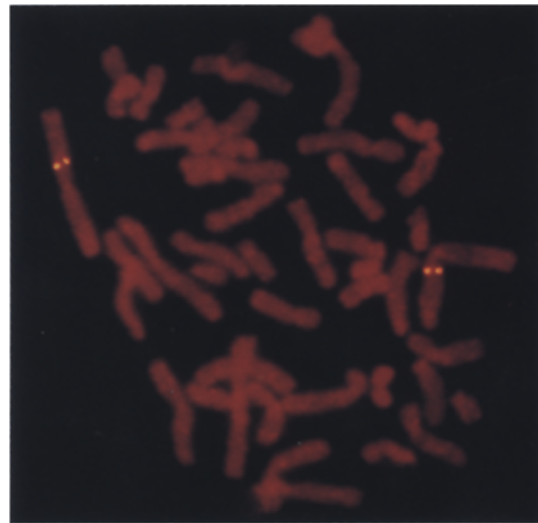


Fig. 1. Localization of the TUFT1 locus by fluorescent in situ hybridization. Metaphase chromosomes are counterstained with propidium iodide and the TUFT1 gene localization is by FITC fluorescent signals.

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