Hum Genet (1993) 92:631

human genetics

DNA variants

An *Eco*RI RFLP in the 5′ region of the human NF1 gene

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Received: 18 March 1993 / Revised: 4 May 1993

Abstract. Von Recklinghausen neurofibromatosis or type 1 neurofibromatosis (NF1), is one of the most common autosomal dominant disorders. NF1 is characterized by neurofibromas, café-au-lait spots and Lisch nodules of the iris. The NF1 gene is located in 17q11.2. The restriction fragment length polymorphism reported here will be useful in linkage analysis in NF1 families.

Description. GE2 is a 550-bp fragment of the 5′ region of the neurofibromatosis type 1 (NF1) cDNA (Marchuk et al. 1991).

*Polymorphism. Eco*RI detects a two-allele polymorphism with fragments of 7.3 kb (A1) and 4.2 kb (A2) (Fig. 1).

Frequency. The frequency of the alleles was studied in 50 unrelated Caucasians, and was shown to be: A1 = 0.71 and A2 = 0.29.

Chromosomal localization. The NF1 gene has been assigned to 17q11.2.

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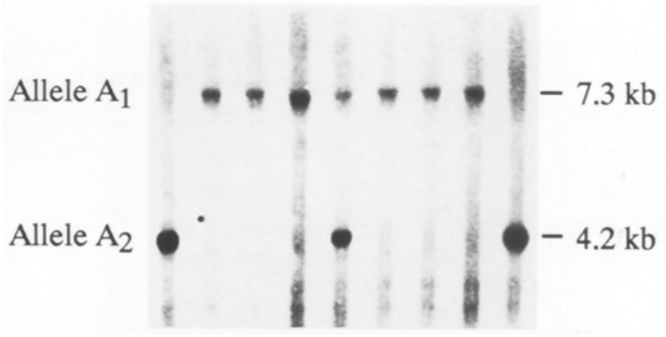

Fig. 1. Detection of the two-allele polymorphism by *Eco*RI. Fragments are: 7.3 kb *(Allele A1)* and 4.2 kb *(Allele A2)*

Mendelian inheritance. Co-dominant segregation was observed in 4 families (41 individuals).

Availability. Contact F. S. Collins.

Reference

Marchuk DA, Saulino AM, Tavakkol R, Swaroop M, Wallace MR, et al. (1991) cDNA cloning of the type 1 neurofibromatosis gene: complete sequence of the NF1 gene product. Genomics 11:931–940