

A *Stu*I RFLP in the human β -spectrin gene (SPTB)

Tang K.Tang*, Yu-Lin Ko¹ and Wen-Pin Lien¹

Institute of Biomedical Sciences, Academia Sinica, Taipei 11529, Taiwan and ¹National Taiwan University Hospital, Taipei, Taiwan, China

Source/Description: β -28 is a 2.7 kb partial cDNA clone which encodes human β -spectrin gene (Winkelmann *et al.*, 1990).

Polymorphism: In addition to the *Hind*III polymorphic fragments of 17 kb (allele C1) and 14 kb (allele C2) (Costa *et al.*, 1990; Tse *et al.*, 1990), *Stu*I identifies another two allele polymorphism with bands at 8 kb (D1) and 4.8 kb and 3.2 kb (D2).

Frequency: Studied in 25 unrelated Chinese.

D1: 0.34

D2: 0.66

Not Polymorphic For: *Eco*RI, *Bam*HI.

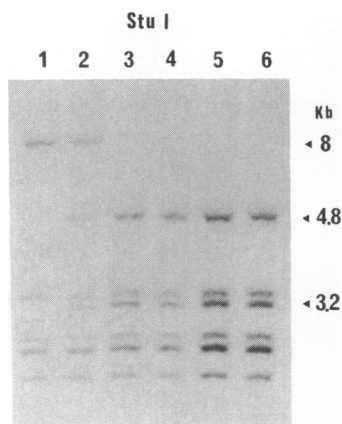
Chromosome Localization: Assigned to chromosome 14q23-q24.2 by in situ hybridization (Fukushima *et al.*, 1990).

Mendelian Inheritance: Co-dominant segregation shown in two families, 38 individuals.

Probe Availability: Available from Dr. Bernard G.Forget.

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References: 1)Winkelmann *et al.* (1990) *J. Biol. Chem.* **265**, 11827–11832. 2)Costa *et al.* (1990) *N. Engl. J. Med.* **323**, 1046–1050. 3)Tse *et al.* (1990) *J. Clin. Invest.* **86**, 909–916. 4)Fukushima *et al.* (1990) *Cytogenet. Cell Genet.* **53**, 232–233.



* To whom correspondence should be addressed

PCR detection of an *Mbo*I polymorphism in the ERBB2 (HER2; NEU) gene on chromosome 17q11.2-q12

Jeff M.Hall and Mary-Claire King*

School of Public Health, University of California, Berkeley, CA 94720, USA

Polymorphism: We have detected a polymorphic *Mbo*I site in a 1.1 kb amplification fragment from an intron in the ERBB2 locus (MIM 164870; identical to NEU and HER2) (1). Amplification with the two primers listed below followed by *Mbo*I digestion yields a constant band of 550 bp and two alleles of 520 bp (A1) and 500 bp (A2). Due to the close proximity of the 550 bp constant band to the two polymorphic bands, optimal resolution is obtained after a double digest with *Mbo*I and *Pvu*II. (*Pvu*II cleaves the 550 bp constant band into two constant bands at 330 and 220 bp, with no change in the two polymorphic bands).

PCR Primers:

Sense oligo: 5'CTGGAATGGGAAGCA

Antisense oligo: 5'GCCAGCAAAGAAATCTTAGACGT

Frequency: Allele frequencies were calculated from 106 unrelated Caucasians.

f(A1) = 0.70

f(A2) = 0.30

Not Polymorphic For: *Msp*I, *Taq*I, *Pst*I, *Pvu*II, *Hinf*I, *Rsa*I, *Hae*III.

Chromosomal Location: ERBB2 has been localized to 17q11.2-q12 (2).

Mendelian Inheritance: Codominant segregation of the two alleles was demonstrated in 12 informative, three-generation CEPH families.

PCR Conditions: PCR was carried out in a total volume of 50 μ l containing 100 ng genomic DNA, 50 pmoles of each primer, 2 mM *MgCl*₂, 100 μ M dNTPs, 50 mM *KCl*, 10 mM *Tris*, pH 8.3. Amplification was performed for 40 cycles with the following parameters: one minute at 92°C, one minute at 56°C, three minutes at 72°C. DNA was sequentially digested with *Pvu*II followed by *Mbo*I. DNA fragments were resolved on a 3% Nusieve GTG + 1% Seakem GTG agarose gel.

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* To whom correspondence should be addressed