A BamHI polymorphism at the fibrillin (FBN) locus

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Source and Description of Clone: pCLM8 is a 3.3 kb EcoRI fragment of fibrillin cDNA in the EcoRI site of pTZ18U (1–3).

Polymorphism: BamHI identifies a two-allele polymorphism with A1 = 18 kb and A2 = 11 + 7 kb. Constant bands are at 9, 6, 3.5, 1.4, and 1.3 kb.

Frequency: Estimated in 44 unrelated individuals:

A1 = 0.83

A2 = 0.17

Not Polymorphic For: EcoRI, HaeIII, PstI, BclI, RsaI, and MspI.

Chromosomal Localization: 15q21.1, by fluorescent in-situ hybridization (1). Linkage studies in 3 informative CEPH families showed no recombination with D15S48 (CRI-L442) giving a LOD score of 6.32 at $\theta = 0$. This confirms localization of FBN to chromosome 15.

Mendelian Inheritance: Co-dominant inheritance was demonstrated in 6 informative CEPH families with a total of 48 children.

Availability: Available for collaboration, contact C.Maslen or L.Sakai.

Other Comments: Fibrillin is a candidate gene for Marfan syndrome. (1–3) Final stringency wash of $0.1 \times \text{SSC}/0.1\%$ SDS at 55–60°C.

Acknowledgements: Supported by NIH grant HG00022 to M.L. and by a grant from the Shriner's Hospital for Crippled Children to S.L.M. and L.Y.S.

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NcoI RFLP in the human prothrombin (F2) gene

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Source/Description: Two primers (PT1 and PT2) were used to amplify a 418 bp long fragment spanning exon 5 and 6 of the human prothrombin gene (1).

Primers:

PT1 = AATAAGTCCCCAGGCTCCAA

PT2 = TGGTCATGGGTCGCCCACT

Polymorphism: After PCR amplification (30 cycles: 1 min at 92°C, 1 min at 65°C, 2 min at 72°C), the amplified fragments were digested by NcoI restriction endonuclease. In addition to one NcoI site was present in the published sequence (1), another site was demonstrated in exon 6. The allele B1 designates originally reported allele with two fragments of 177 and 241 bp. The allele B2 is the newly found allele with 3 fragments of 101, 140, and 177 bp.

Frequency: Estimated in 20 unrelated Japanese

Heterozygosity = 35.0%

Allele B1: 0.425

Allele B2: 0.575

Chromosomal Localization: The human prothrombin gene has been localised on chromosome 11p11-q12 (2).

Mendelian Inheritance: Co-dominant segregation was shown in 4 families.

Other Comments: It was shown by the sequence analysis that the polymorphism described above is a result from a substitution of T for C at position 4203 of the human prothrombin gene, which changes Thr to Met at position 122 of the polypeptide chain (1).

Acknowledgements: This study was supported in part by the grant from Otsuka Pharmaceutical Factory Inc. for Otsuka Department of Clinical and Molecular Nutrition, School of Medicine, The University of Tokushima.

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Figure 1. Lane 1: B1B2; Lane 2: B2B2; Lane 3: B1B1; Lane 4: B1B2.

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