Isolation and characterization of a hypervariable region [D4S163] on chromosome 4

John Neuweiler, Vivian Ruvolo, Howard Baum, Karl-Heinz Grzeschik¹ and Ivan Balazs
Lifecodes Corporation, Saw Mill River Road, Valhalla, NY 10595, USA and ¹Institute of Human Genetics, University of Marburg, Marburg, FRG

Source/Description: A 7 Kb genomic fragment was isolated from a partial EcoRI human genomic library in Charon 30 (LILA5). A 0.92 Kb HaeIII fragment, was further subcloned into Bluescript.

Polymorphisms: Human DNA samples digested with PstI identify a VNTR-type of polymorphism. The screening of random individuals revealed a continuous distribution of DNA fragment sizes varying from approximately 5 to 18 Kb. The same polymorphism can be studied with HaeIII with DNA fragments in the range of 2.5 to 9 Kb.

Heterozygosity: With PstI, 90% heterozygosity was observed in 72 unrelated Caucasians.

Chromosomal Localization: Chromosomal localization to chromosome 4 was established by hybridization to DNA from a panel of human-mouse hybrid cell lines.

Mendelian Inheritance: Co-dominant segregation of the DNA polymorphism was observed in 19 C.E.P.H. families.

Probe Availability: Contact I.B.

Other Comments: This genomic fragment also cross-reacts with constant DNA fragments (PstI-digested human DNA) at approximately 4.0, 1.4 and 1.3 Kb.

A Sac I RFLP is detected with the 5-HT1a serotonin receptor probe G21

M.A.R.Khan, K.E.Isenberg, H.Aschauer¹ and E.J.Devor*

Department of Psychiatry, Washington University School of Medicine, St. Louis, MO 63110 USA and ¹Psychiatrische Universitätsklinik Wein, Währinger Gürtel 18–20, A-1090 Vienna, Austria

Source and Description G-21 is a 1.7 kb genomic fragment containing the entire coding sequence of the intronless 5-HT1a receptor gene plus ~300bp of 5' non-coding sequence cloned in the Xba I-Bam HI sites of pSP64 (Kobilka *et al.*, 1987).

Polymorphism: SacI (GAGCT/C) identifies a two allele polymorphism with bands at 1.83 kb (A1) and 1.80 kb (A2) (see Figure).

Frequency: Studied in 35 unrelated Caucasians (17 male, 18 female):

1.83 kb allele (A1) = 0.87

1.80 kb allele (A2) = 0.13

Not polymorphic for: BamHI, BclI, BglII, DraI, EcoRV, HindIII, MspI, PvuII, RsaI, SspI, TaqI, XbaI in a panel of seven unrelated Caucasians.

Chromosomal Localization: G-21 has been assigned to chromosome 5q11.2-q13 by *in situ* hybridization (Kobilka *et al.*, 1987).

Mendelian Inheritance: Co-dominant segregation demonstrated in two informative families (see Figure).

Probe Availability: Contact Dr Brian Kobilka (see reference).

Other Comments: RFLP resolution is best in high concentration agarose gel (~1.3%), normal stringencies are sufficient for hybridization and washes.

Reference: Kobilka, B.K., Friele, T., Collins, S., Yang-Feng, T., Kobilka, T.S., Franke, U., Lefkowitz, R.J. and Caron, M.G. (1987) *Nature* **329** 75–79.

Acknowledgements: We are grateful to Dr Brian Kobilka for providing us with the G-21 probe. This work was supported by NIMH Grant MH31302.

*To whom correspondence should be addressed at Department of Psychiatry, Washington University School of Medicine, St. Louis, MO 63110, USA