Radboud Repository



PDF hosted at the Radboud Repository of the Radboud University Nijmegen

The following full text is a publisher's version.

For additional information about this publication click this link. http://hdl.handle.net/2066/23455

Please be advised that this information was generated on 2017-12-05 and may be subject to change.

SHORT COMMUNICATION

A Pst I Restriction Fragment Length Polymorphism near the MAO locus on Xp

P. V. SALENGER¹, P. HUEBER¹, P. J. SPELLER¹, G. VAN DUIJNHOVEN², R. R. HOOPES, JR. 1, R. V. THAKKER³, W. BERGER⁴ AND S. J. SCHEINMAN¹

¹ Department of Medicine, SUNY Health Science Center, Syracuse, NY, USA ² Department of Human Genetics, University Hospital Nijmegen, Nijmegen, The Netherlands ³MRC Molecular Endocrinology Group, Royal Postgraduate Medical School-Hammersmith Hospital

⁴ Max-Planck Institut fur Molekulare Genetik, Berlin, Germany

(Received 18.4.96. Accepted 28.5.96)

We report a Pst I polymorphism located within the peri-centromeric region of the short arm of the X-chromosome (Xp).

A Restriction Fragment Length Polymorphism (RFLP) with fragment sizes of 5·1 kb (allele 1) and 3.8 kb (allele 2) was detected in human DNA samples subjected to digestion with Pst I. individuals. The frequency of the 5.1 kb allele The plasmid PD8/27 (DXS1702), generated by was 0.347, and the frequency of the 3.8 kb allele Alu-PCR on radiation-reduced cells hybrids was 0.652. Mendelian segregation was noted in (Berger et al. 1992a), was used as the probe in the three three-generation families and one fourhybridization of Southern blots containing Pst I- generation family studied. digested human DNA.

5.1 kB (Allele 1) 3.8 kB (Allele 2)

Fig. 1. The Pst I polymorphism. DNA (5 µg) from each individual was digested with Pst I and electrophoresed on a 20 cm 0.8% agarose gel at 40 V for 21 hours.

The location of the PD8/27 probe was previously refined to a 640 kb YAC that also contains the DXS7, MAOA and MAOB loci (in the Xp 11·4-11·2 region) (Berger et al. 1992b).

The frequency of occurrence of the Pst 1 alleles was examined in 69 chromosomes from unrelated

This work was supported by a Grant-in-Aid from the American Heart Association and from Dialysis Clinics, Inc., and by a Collaborative Research Grant from NATO.

REFERENCES

BERGER, W., MEINDL, A., DE LEEUW, B., DE ROOS, A., VAN DE POL, T. J. R., MEITINGER, T., VAN DER VELDE-VISSER, S.D., ACHARZ, H., VAN KESSEL, A.G., CREMERS, F. P. M. & ROPERS, H. H. (1992a) Generation and characterization of radiation reduced cell hybrids and isolation of probes from the proximal short arm of the human X chromosome. Hum. Genet. 90, 245-246.

BERGER, W., MEINDL, A., VAN DE POL, T.J., CREMERS, F.I.P., ROPERS, H. H., DOERNER, C., MONACO, A., BERGEN, A.A., LEBO, R. & WARBURGH, M. (19926) Isolation of a candidate gene for Norrie disease by positional cloning. Nature Genet. 1, 199-203.