Physical mapping of D8S7 and D8S11 to the band 8p23.1 of the short arm of chromosome 8

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A family, containing members presenting with multiple dysplasia and severe mental retardation (to be described elsewhere), was analysed by high resolution chromosome banding techniques. A balanced translocation was found with a breakpoint in the short arm of chromosome 8, t(8;19)(p23.1;q13.43), and affected members of the family were deleted for the region distal to 8p23.1, Fig1. EDTA blood was obtained from both affected and unaffected family members and DNA extracted by standard methods.¹ Separate Southern blots were carried out on 2µg samples digested with *Hind*III, *Rsa*I or *Hinf*I, the concentration of DNA used being controlled by measuring the absorption at 260nm. The blots were hybridised with the probes D8S7² (*Hind*III) and D8S11³ (*Rsa*I) which had been labelled with [³²P]dCTP by the random primed labelling reaction⁴.

The resultant autoradiographs reveal a single band of 3.1kb in DNA digested with *Hind*III and hybridised with D8S7 (Fig 2a) and a single band of 1.70kb when digested with *Rsa*I and hybridised with D8S11 (Fig 2b). They also demonstrate that unaffected family members, carrying a balanced translocation (lanes 1, 3 & 4) or a normal chromosome complement (lane 2) have a band of higher intensity when compared with affected family members, deleted for the band 8p23.1 (lanes 5-8). Lanes 9 & 10 contain DNA from unrelated control individuals. These results physically locate the probes D8S7 and D8S11 distal to this band on the short arm of chromosome 8.

Fig1. The arrow points to the breakpoint at the band 8p23.1

Fig2. Autoradiographs of DNA hybridised with a) D8S7 and b) D8S11

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