

**P004** A heterochromatic-like knob at the boundary of the centromere repressive effect for human chromosome 21q  
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Meiotic recombination is repressed at distance from the centromere of eucaryotic chromosomes. The transition between the proximal repressed domain and the distal active region is poorly characterised in mammals. Few chromosome-specific markers have been designed until recently within juxta-centromeric regions, true patchworks of contiguous segmental duplications. We used paralogous sequence variants as chromosome-specific anchors to develop microsatellite markers, single nucleotide polymorphisms and sequence tagged sites (STSs) along the most proximal 1.3 Mb region of human chromosome 21q. Linkage analysis in pedigrees shows that recombination is highly repressed within the proximal interval in both genders whereas only female recombination is active in the distal interval. Accordingly, linkage disequilibrium analyses reveals large haplotype blocks within the proximal region and relatively short ones within the distal one. The chromatin flavour of the region was assayed by real-time quantification of STSs immunoprecipitated with antibodies raised against histone modifications associated with either euchromatin or constitutive heterochromatin. A sharp peak of heterochromatic marks colocalises with the transition between the proximal repressed domain and the distal active domain for recombination. Our results, together with transcriptional repression data along this region, suggest that this heterochromatic-like knob is the boundary of the centromere repressive effect.