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Sequence organization of the human Y chromosome

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A comparison of restriction patterns of human male and female DNA after digestion with Hae III reveals two bands which are present only in male DNA and which are produced by cleavage of repetitive sequences found only on the human Y chromosome (Cooke 1976). Repetitive Y specific sequences can also be detected by exhaustive DNA/DNA hybridization (Kunkel, Smith & Boyer 1976).

When DNA from one of these repetitive sequences is isolated as a fragment 3300 bases long from a Hae III digest of male DNA this material can be used as a probe for related sequences in male and female DNA. In both male and female DNA there is DNA which does not contain Hae III sites, which is complementary to this sequence and probably represents related tandem repeats. However, in male DNA fragments which are multiples of 3300 bases long are present showing that this sequence is tandemly repeated.

Further digestion of the 3300-base repeat from male DNA with Mbo I gives a cleavage pattern in which the DNA is found in fragments of multiples of 550 bases. This is consistent with the sequence data presented by Cooke (1976). A fragment of 3300 bases is also produced by digestion of male DNA with Eco RI, and on digestion with Mbo I a similar pattern is generated. Digestion of male DNA with both Eco RI and Hae III does not increase the amount of material found in this size class, and it is therefore concluded that the RI and Hae III sites are close together on the same molecules with Mbo I sites frequently present at 550 base pair spacing.

The human Y chromosome is highly polymorphic in the size of the fluorescent part of the long arm. When DNA from individuals with different sizes of Y chromosome is analysed in terms of the amount of the 3300-base fragment no significant differences are observed with most samples. This is most probably due to the insensitivity of the method of analysis, since one individual with a fluorescent part of the long arm which is five times the cleavage length has about 2–3 times as much of the 3300-base sequence. A likely reason for the nonlinear relation between Y size and amount of this sequence is that the fluorescent part of the Y contains other sequences (as is known (Gosden *et al.* 1975)) and that this sequence is at the centromeric end of the fluorescent region. Thus, if the DNA distal to the fluorescent region were amplified this would not increase the amount of the 3300-base fragment.

References (Cooke)

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