

DISSERTATION SUMMARY

Isolation, cloning and characterization of satellite DNA families in rabbit

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The pericentromeric heterochromatin of mammalian chromosomes is composed of tandemly repeated satellite DNAs, frequently forming arrays of several million nucleotides. Satellite compositions of centromere regions are variable, indicating that it is epigenetic modification, rather than the particular sequence of nucleotides, what supports centromere function. The rabbit satellites described below are arrays of medium-sized monomer units with family-specific features. They have not only different lengths (~375 and ~585 bp), but completely different sequences, as well. Individual Rsat I sequences showed differences in the 5.3–21.8% range (78.2–94.7% identities), and that of Rsat II/III was 8.0–22.2% (77.8–92%). The rabbit satellites appear to be more homogenous than their human counterparts, owing to the less extensive higher-order repeat organization. FISH experiments with Rsat I, Rsat II and Rsat III showed, that the satellite sequences identified so far did not cover the complete chromosome complement of rabbit, although signals were detected with Rsat I on 11, with Rsat II to 12, and with Rsat III to 2 chromosome pairs. It is remarkable that both Rsat I and Rsat II were found on nine chromosomes, but Rsat I was undetectable on the two chromosome pairs (#9 and #16) with Rsat III. Apparently, the two main satellite families have always been, or have become so different that they could coexist without much influence on each other. Probably, these two sequences are equally compatible with centromere function. The fact that the Rsat III-bearing chromosomes still carry Rsat II supports the view that probably the former arose from the local amplification of the latter. Processes in accordance with the „library” hypothesis of satellite DNA sequence evolution appear to be at work in rabbit in two forms. First, both of the two main families, Rsat I and Rsat II, were detected together in varying ratios on a number of

chromosomes, forming thus major components of the library. Second, the emergence of Rsat III on chromosomes #9 and #16 is the result of a relatively recent branching from Rsat II, thus creating new „volumes”. However, the catalogue of the rabbit satellite „library” is still incomplete, since there are still nine chromosomes (seven somatic pairs and the sex chromosomes) for which no satellite has been characterized so far. The possibility that these chromosomes had some variant of either Rsat I or Rsat II seems unlikely, since no FISH signal was detected with any of these probes under low-stringency conditions. Although both the Rsat II and Rsat III arrays are based on similar, but diverged repeat units, only Rsat II showed signs of higher-order periodicity. Therefore, it is plausible that the Rsat III arrays of these chromosomes formed via relatively recent large-scale amplification events of diverged repeat units, and consequently, they are expected to be more homogenous. Since the Southern hybridization pattern of Rsat III is more regular than that of Rsat II, such putative saltatory amplification events could indeed have occurred. Taken together, it is plausible that Rsat III was derived from divergent Rsat II units, and not vice versa. The use of an rDNA hybridization probe showed that in addition to the three known NOR chromosomes, there were signals at 21q(ter). This locus is as a useful cytological marker at least in some species of the Leporidae. While attempts are made to identify the missing satellites, those present on the NOR chromosomes could become components of rabbit satellite-based artificial chromosomes.

References

- Ékes C, Csonka E, Hadlaczky G, Cserpán I (2004) Isolation, cloning and characterization of two major satellite DNA families of rabbit (*Oryctolagus cuniculus*). *Gene* 343:271-279.