

P003 "Birth-and-Death" evolution of the mammalian beta-defensin loci on chromosome 20

E.J. Hollox and J.A.L. Armour

Institute of Genetics, University of Nottingham

In humans, there are three main beta-defensin clusters at genomic regions 8p23.1, 20p13 and 20q12. We investigated the evolution of the beta defensin genes on chromosome 20 by using both sequence analysis and analysis of synteny in different mammalian species. We show that two chromosome 20 clusters were generated from a single cluster by pericentric inversion in primates, and that the gene cluster itself is syntenic across human, chimpanzee, mouse, rat and dog, and therefore at least 70 million years old. Several genes show conservation across different lineages, and four genes show evidence of positive selection along certain lineages. However, the dominant evolutionary patterns have been loss of genes along certain lineages by formation of pseudo-genes or drastic change of protein sequence by small deletions in the genome. We also discuss the functional consequences of this in relation to the evolution of the other beta-defensin genes at 8p23.1.