
The authors present a high-resolution physical map of the euchromatic, centromeric, and heterochromatic regions of the human Y chromosome. The methods of constructing the physical map of the human Y chromosome included genomic clone subtraction and dissection of sequence family variants. Of the map’s 758 DNA markers, 136 have multiple locations in the human Y chromosome, indicating its unusually repetitive sequence composition.—Hans E. Grossniklaus

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The authors constructed a high-resolution map of human chromosome 12. The map is different from previous low-density clone maps, genetic linkage maps, and radiation hybrid maps. The map is based on large bacterial clones that include bacterial artificial chromosomes, P1 artificial chromosomes, and a few cosmids. The map contains 3,090 STS markers, and consists of 18 contigs that are not linked by PCR or sequence data. The map has 14 gaps, excluding the centromere, in its bacterial clone coverage.—Hans E. Grossniklaus

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The authors report the tiling path of approximately 650 clones covering more than 99% of human chromosome 14. Clone overlap information to assemble the map was derived by comparing fully sequenced clones with a database of clone end sequences. The authors selected homogeneously distributed seed points using an auxiliary high-resolution radiation hybrid map comprising 1,895 distinct positions. The high long-range continuity and low redundancy of the tiling path indicates that the sequence tag connector approach compares favorably with alternative mapping strategies.—Hans E. Grossniklaus

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The group used the whole-genome random shotgun method with subsequent assembly of the sequenced segments. A 2.91-billion base pair (bp) consensus sequence of the euchromatic portion of the human genome was generated. The 14.8-billion bp DNA sequence was generated from 27,271,853 high-quality sequence reads (5.11-fold coverage of the genome) from both ends of plasmid clones made from the DNA of five individuals. Two assembly strategies were used, each combining sequence data from Celera and the publicly funded human genome project. The two assembly strategies yielded very similar results that largely agree with independent mapping data. More than 90% of the genome is in scaffold assemblies of 100,000 bp or more and 25% of the genome is in scaffolds of 10 million bp or larger. Analysis of the genome sequence revealed 26,588 protein-encoding transcripts in which there was strong corroborating evidence and an additional approximately 12,000 computationally derived genes with mouse match or other weak supporting evidence. Although gene-dense clusters are obvious, almost half of the genes are dispersed on low G+C sequence separated by large tracts of apparently noncoding sequence. Only 1.1% of the genome is spanned by exons, whereas 24% is in introns, with 75% of the genome being intergenic DNA. Duplications of segmental blocks, ranging in size up to chromosomal lengths, are abundant throughout the genome and reveal a complex evolutionary history. Comparative genomic analysis indicates vertebrate expansions of genes associated with neuronal function, with tissue-specific developmental regulation and the hemostasis and immune systems. DNA sequence comparisons between the consensus sequence and publicly funded genome data provided locations of 2.1 million single-nucleotide polymorphisms (SNPs). A random pair of human haploid genomes differed at a rate of 1 bp per 1250 on average, but there was marked heterogeneity in the levels of polymorphism across the genome. Less than 1% of all SNPs resulted in variation in proteins, but the task of determining which SNPs have functional consequences remains an open challenge. The whole genome sequence approach is in contrast to the BAC by BAC method. The authors found a lower than expected number of human genes, human DNA sequence variation distribution across the genome, and genome complexity.—Hans E. Grossniklaus

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The complexity of the apoptotic system is described in detail. The apoptosis-associated system components and their homologs encoded in the human genome are compared with those in the fly and worm genomes. There is a major increase in complexity in