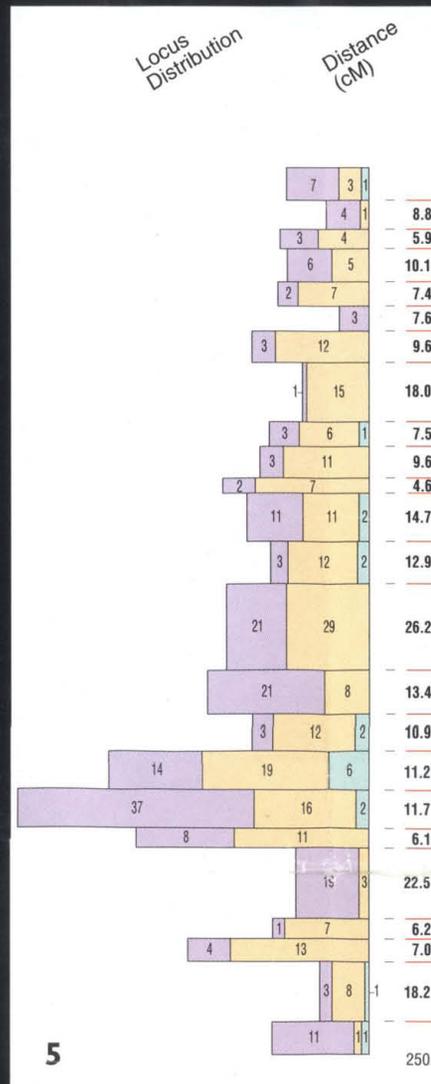
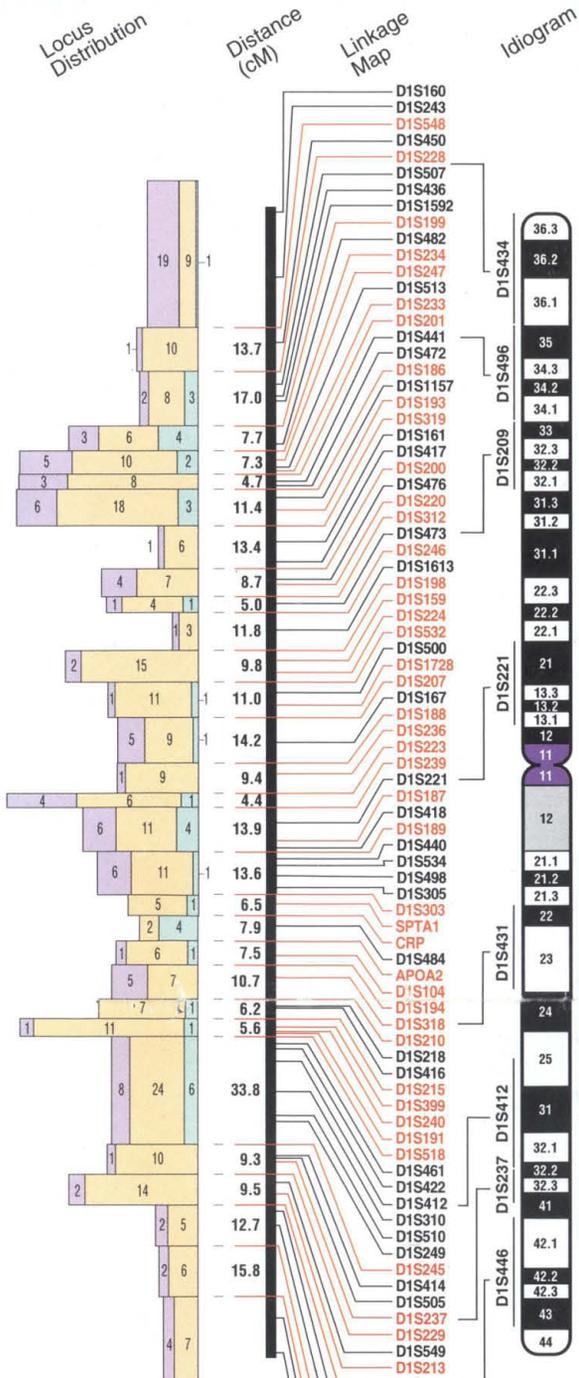


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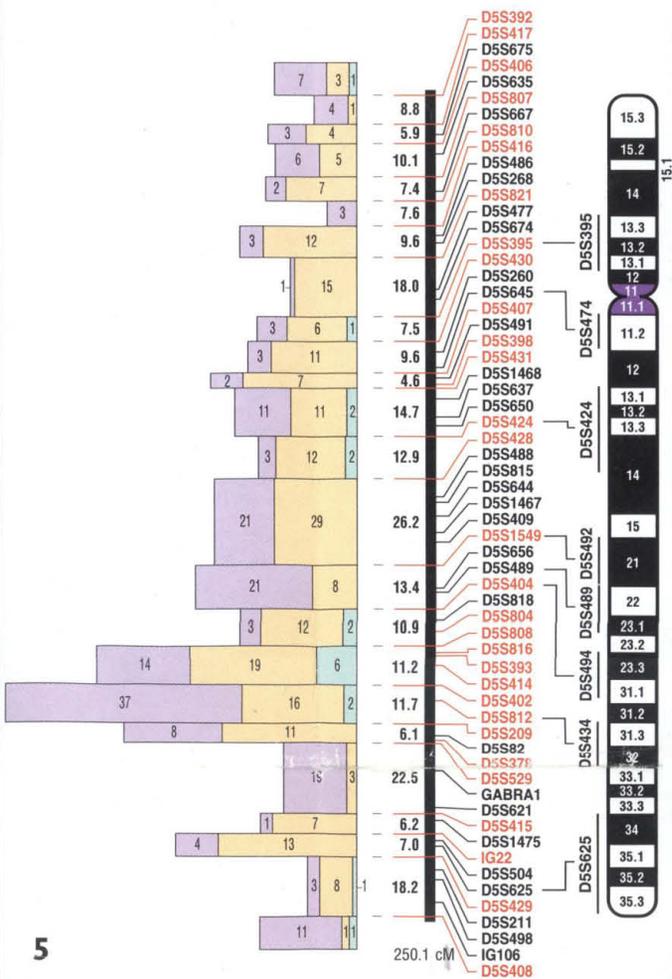
SCIENCE

HUMAN GENOME

GENOME

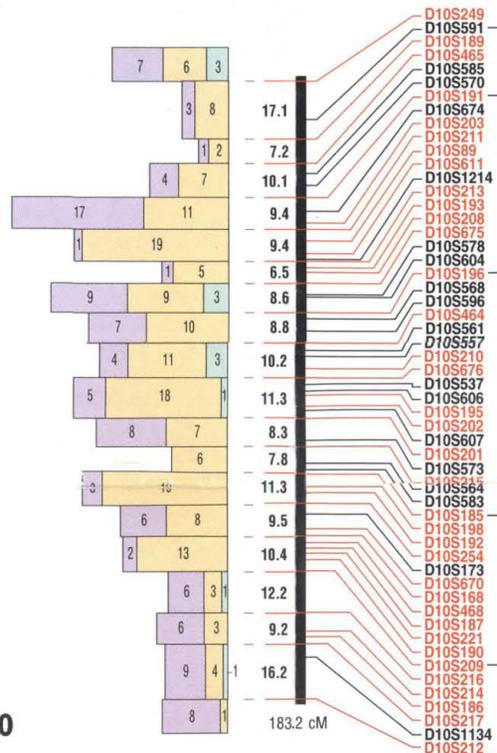


Locus Distribution Distance (cM) Linkage Map Idiogram



5

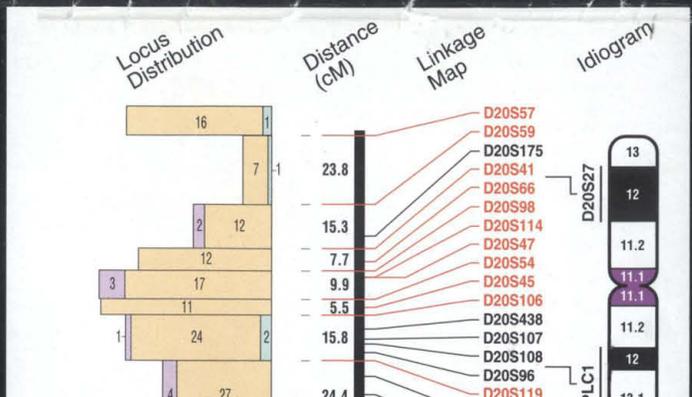
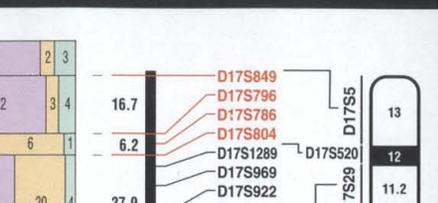
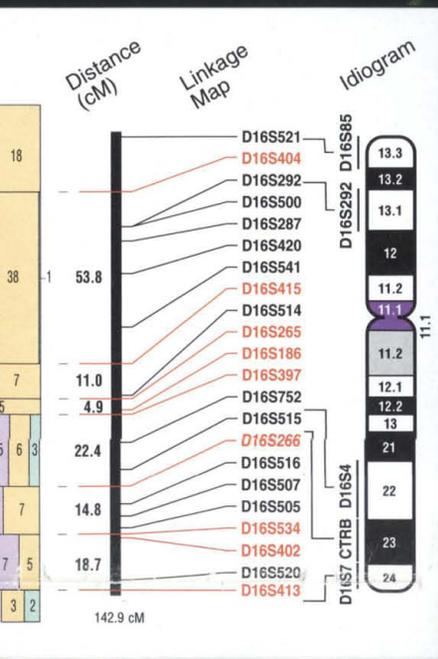
Locus Distribution Distance (cM) Linkage Map

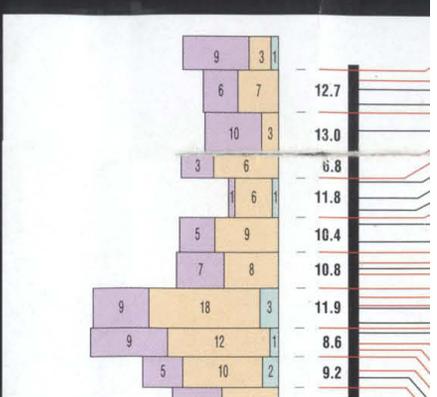
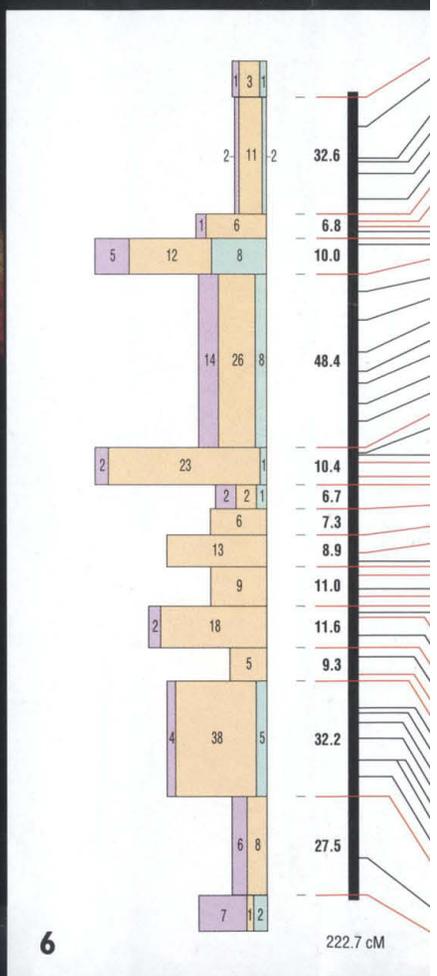
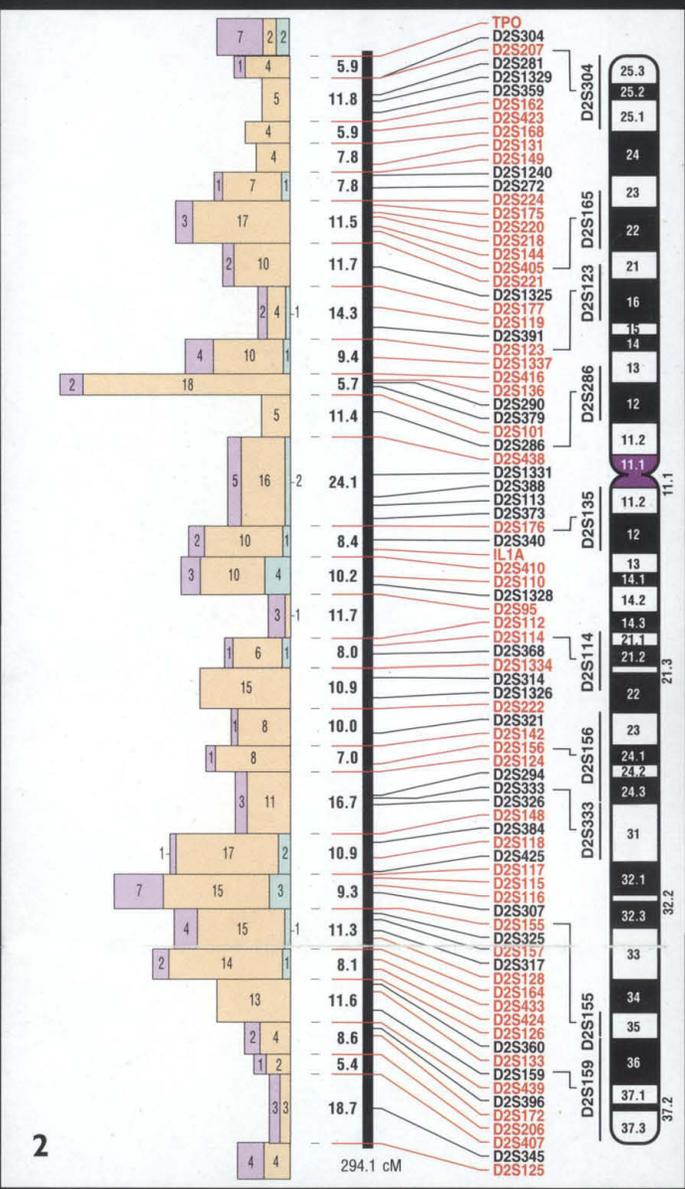
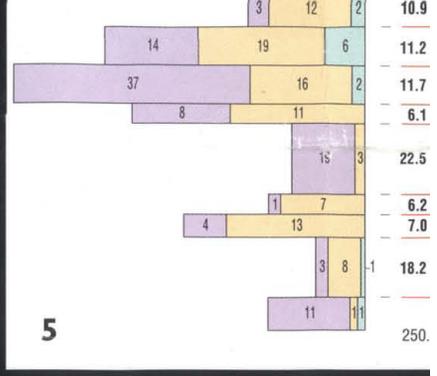
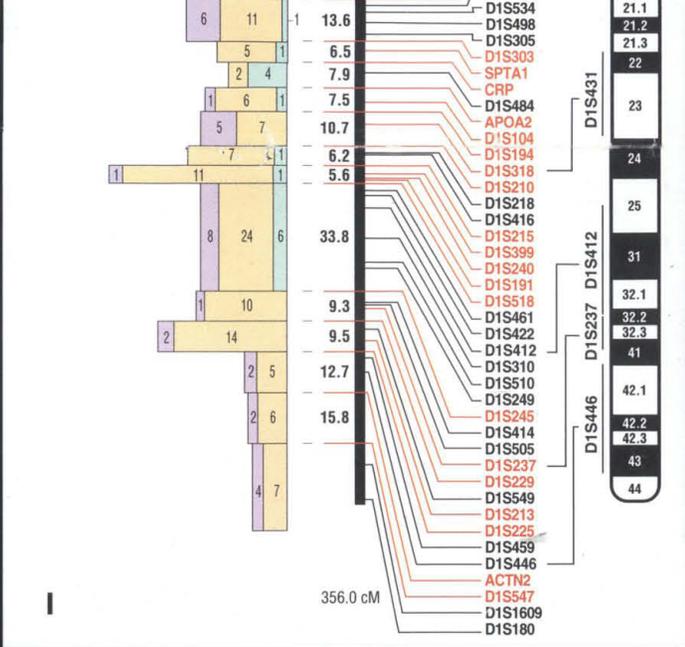


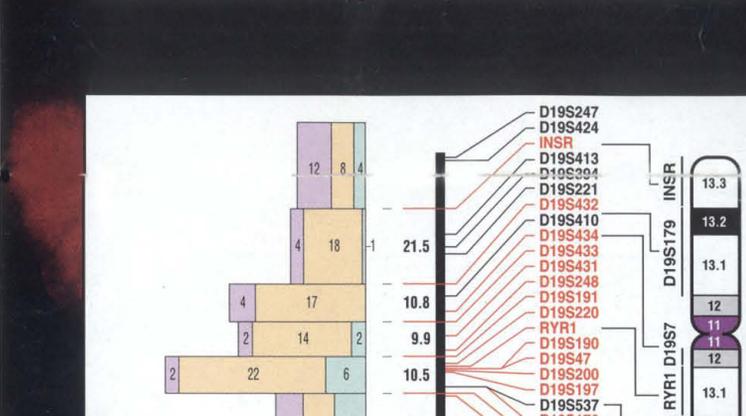
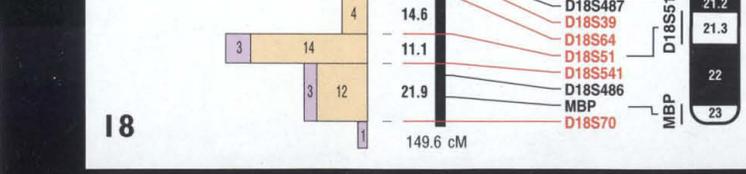
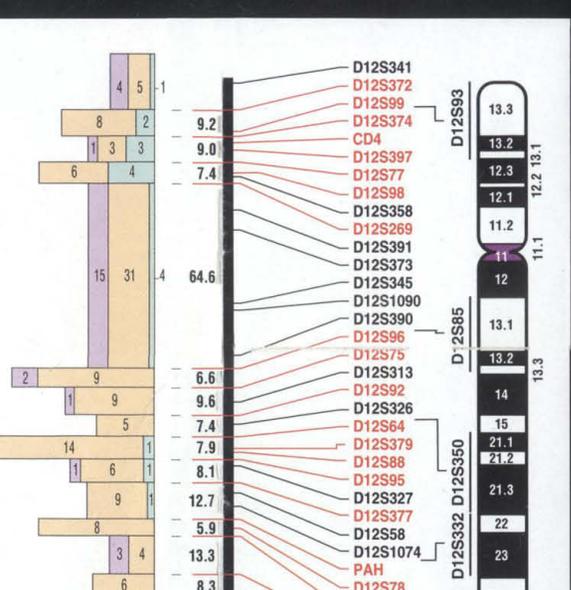
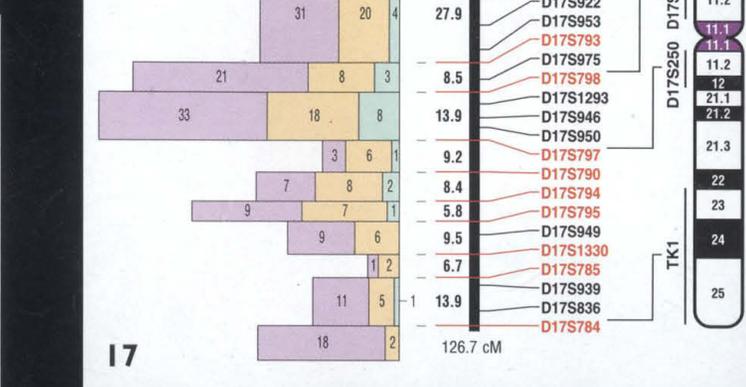
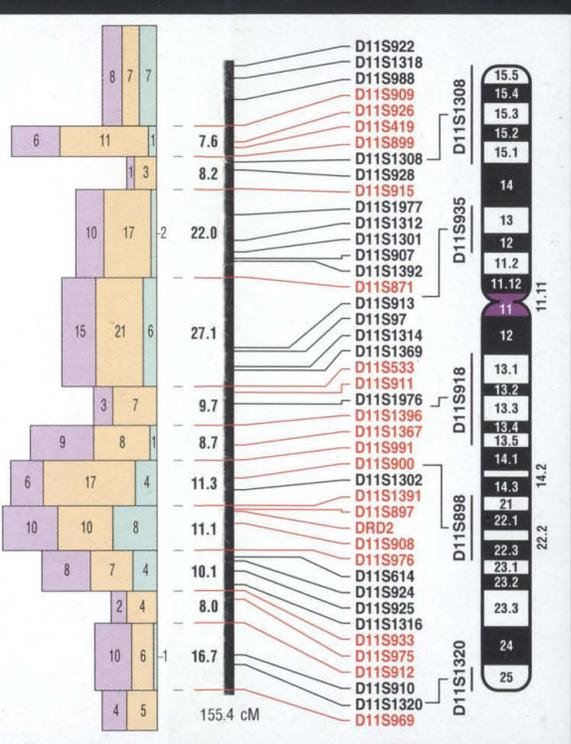
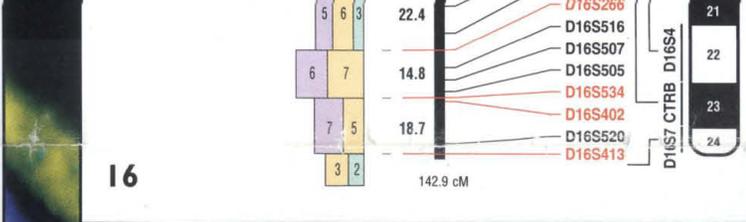
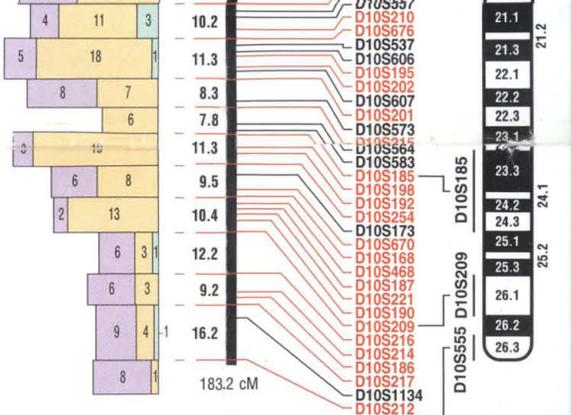
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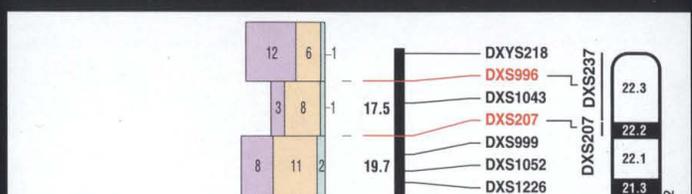
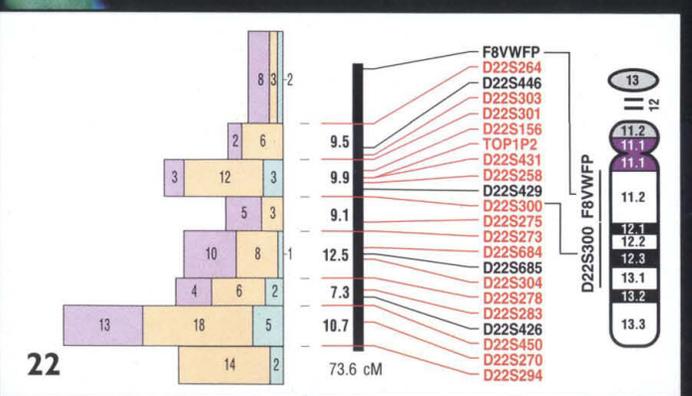
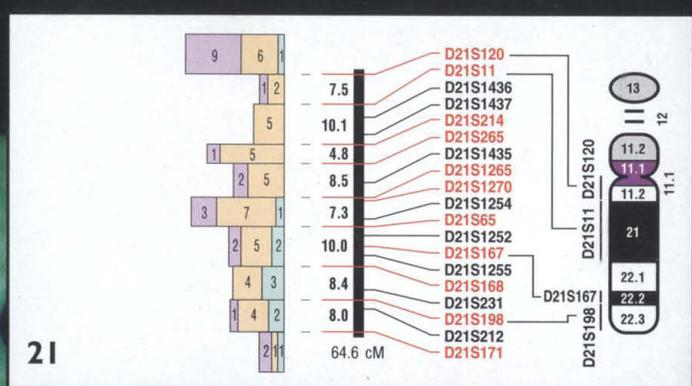
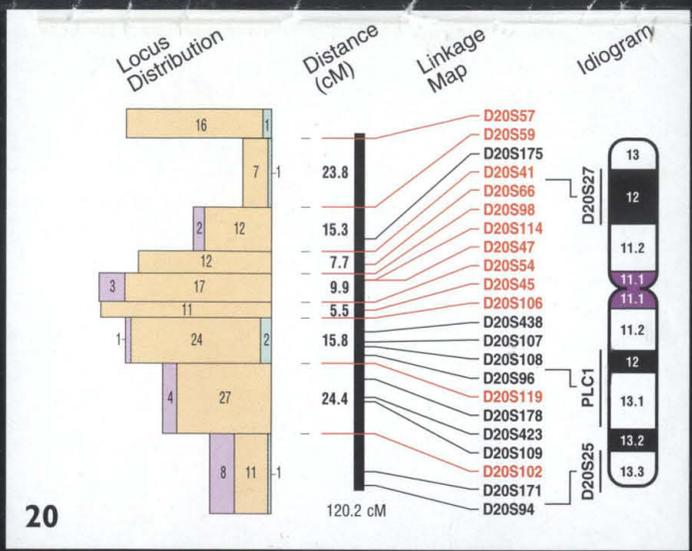
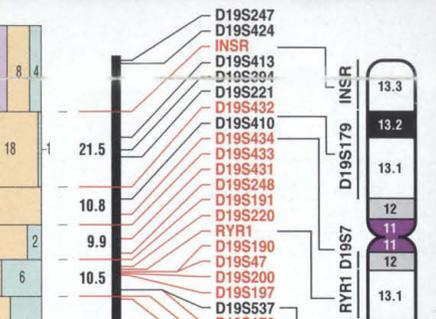
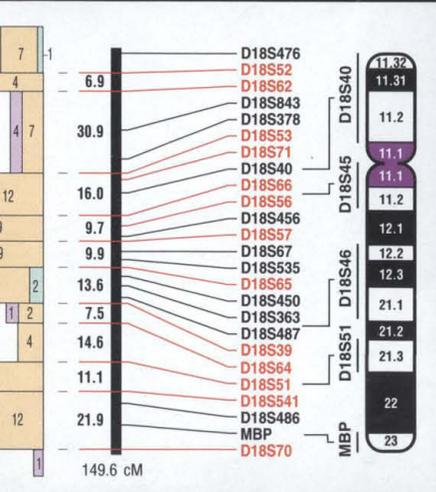
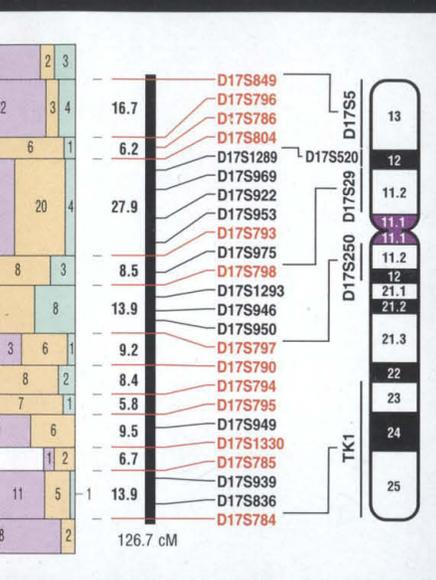
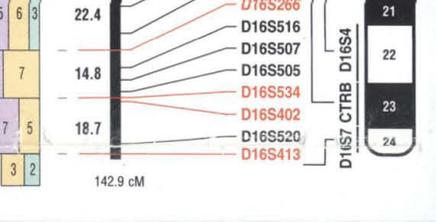
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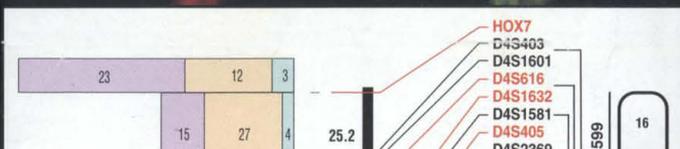
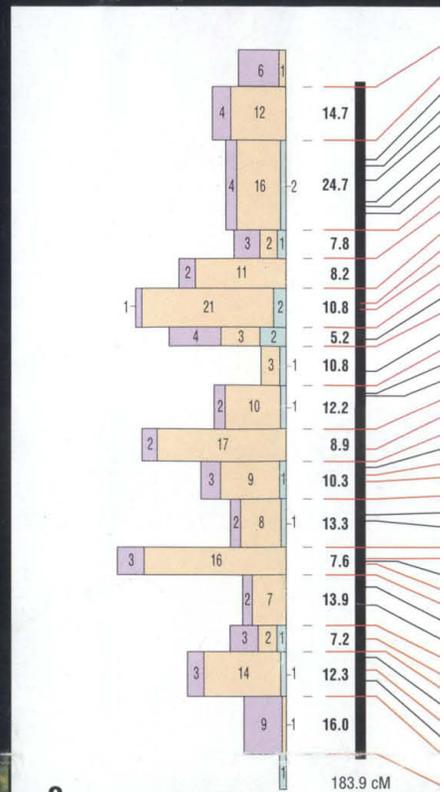
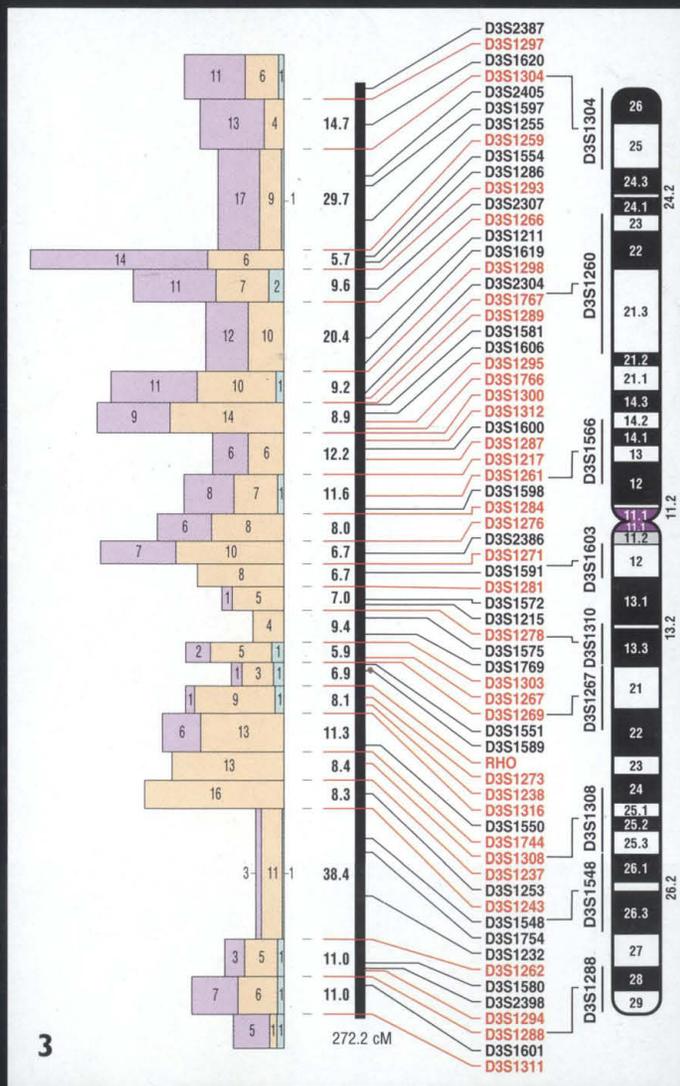
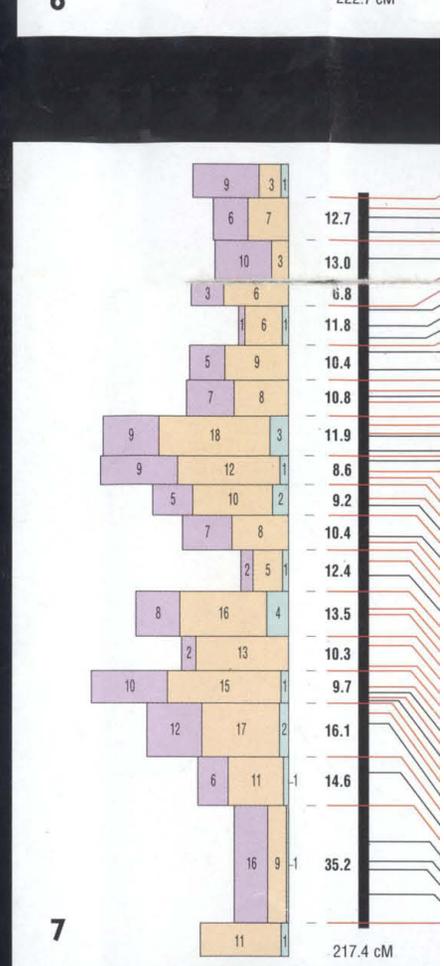
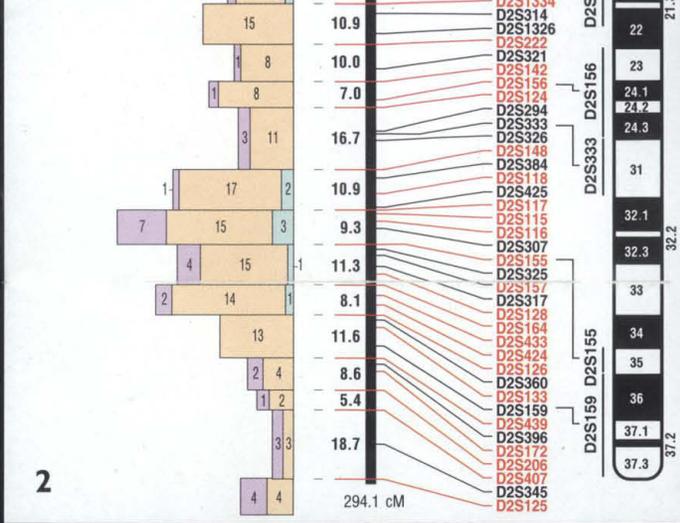
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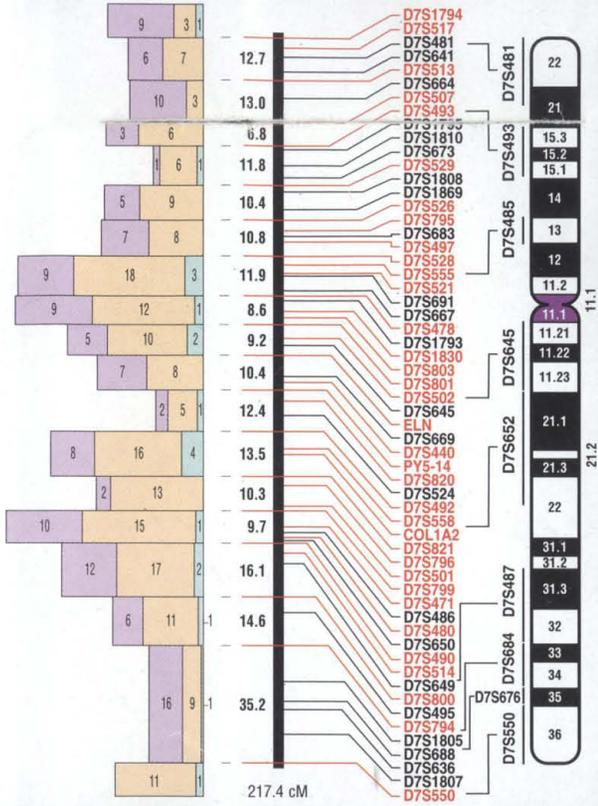




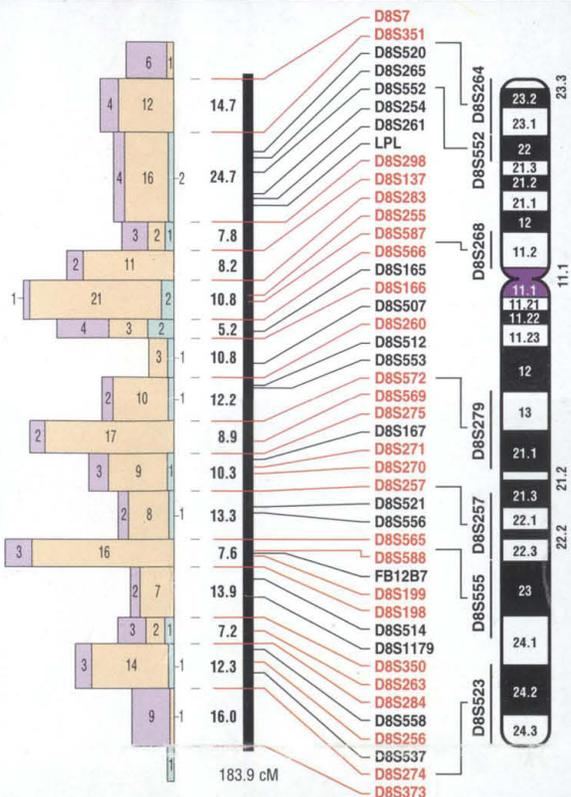




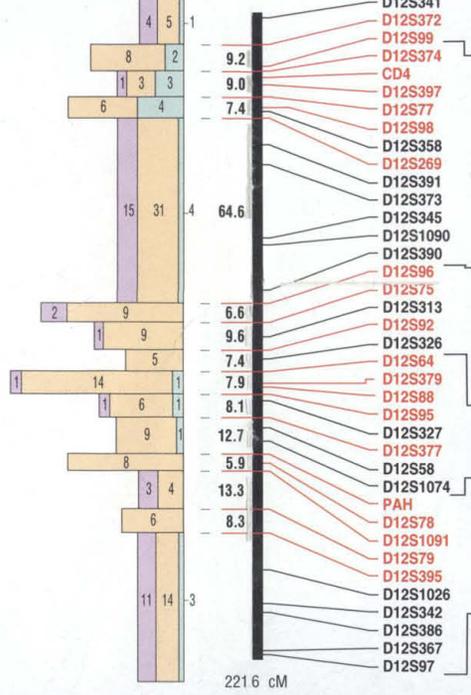
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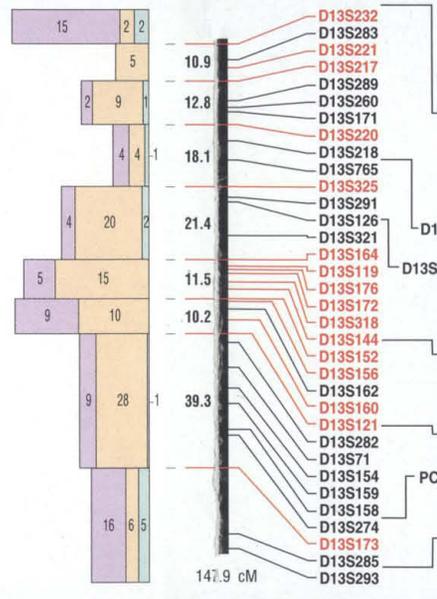
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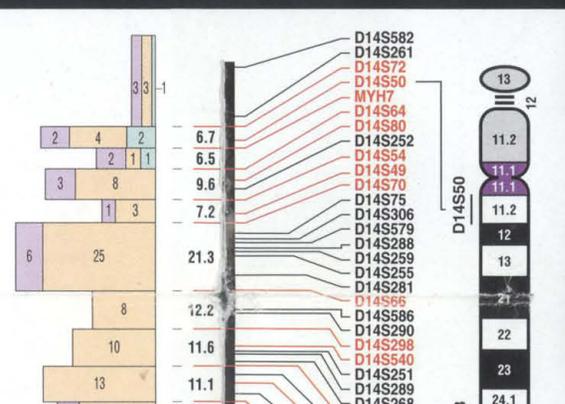
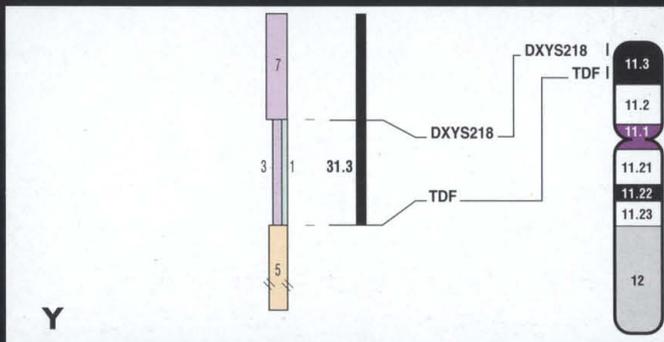
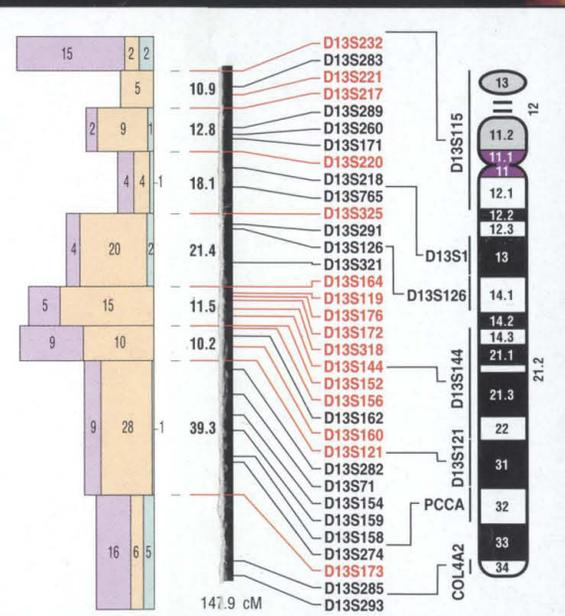
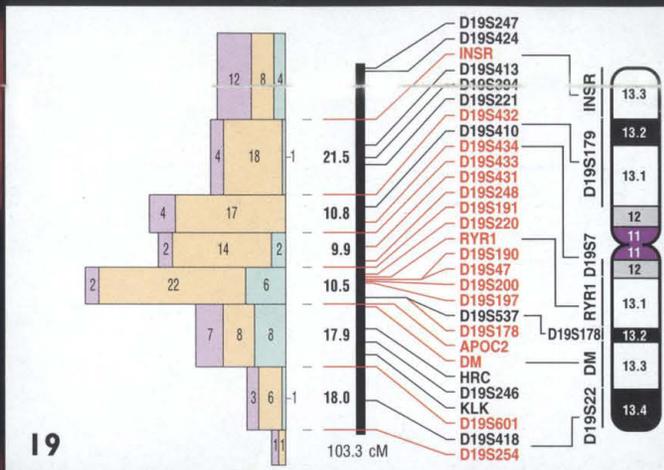
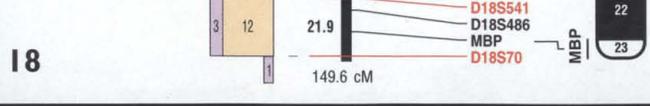
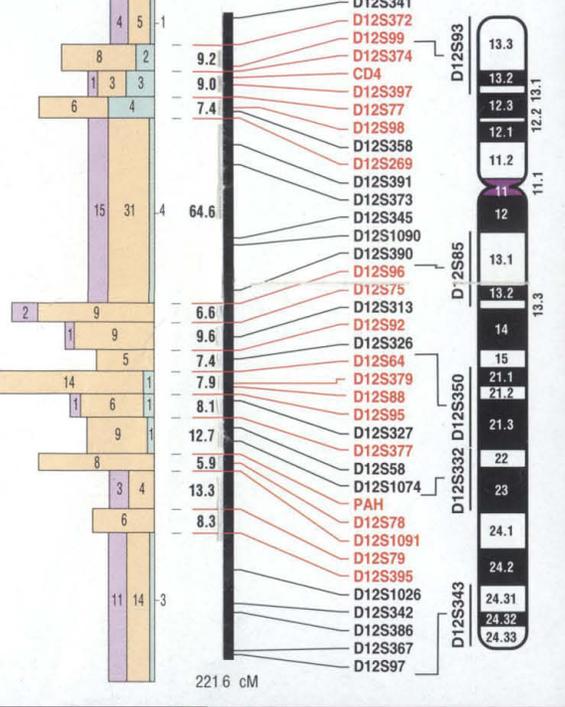


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LEGEND

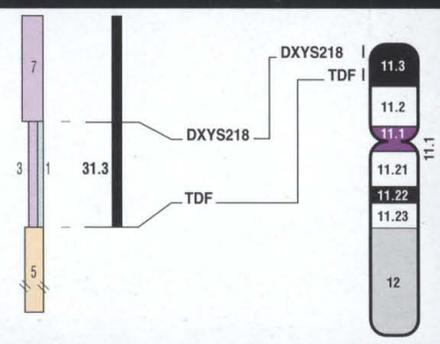
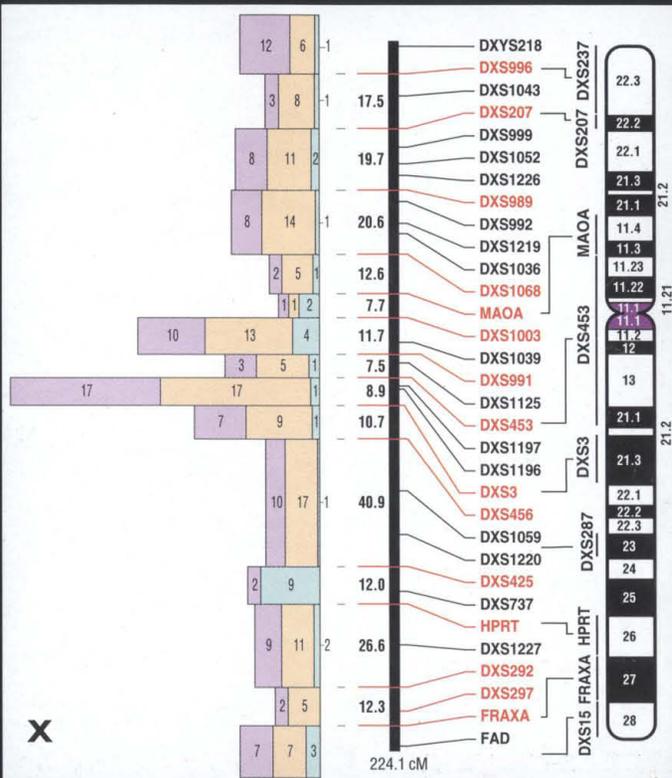
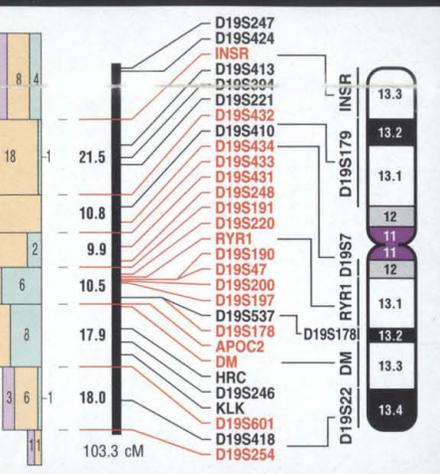
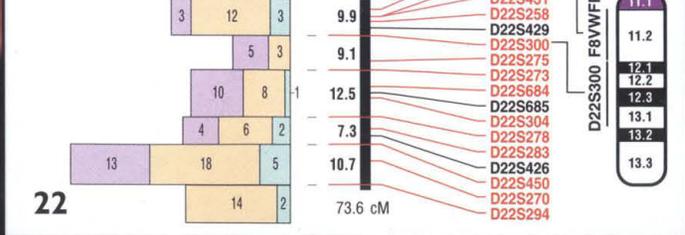
The chart presents a genetic map of the human genome that integrates published data and recombination rates, indicated in centimorgans (cM). The map shows the positions of markers and the distances between them. The markers are labeled with their names and their positions on the chromosome. The recombination rates are indicated by the length of the lines connecting the markers.

The map is composed solely of loci that contain short tandem repeats (STRs) within the map. Data contained in the map were obtained from the International System of Human Cytogenetic Nomenclature (ISCN) and the National Institute of Standards and Technology (NIST) Human Genome Center (UTAH). The data from Génétique Humaine (Génétique Humaine) consist of markers with tri-, tetra-, and pentanucleotide repeats. All data used were generated from the CEPH reference panel.

The map presented on the chart was constructed by first building a map that exceeded all others by odds of 1000:1. The map was constrained by the families used and did not inflate the map with their addition by the inclusion of markers that were not in the primary data. The loci that are contained in this map are those that are not excluded by any other interval.

This map was used to define reference intervals for the construction of reference intervals if they could be excluded from all other intervals by the minimum number of recombination events is shown. A single interval is shown for each marker. The average and sex-specific recombination rates for these maps are shown in the legend.

The banding patterns for each chromosome shown on the ideograms correspond to the cytogenetic and the genetic maps. The correspondences between the cytogenetic and the genetic maps are shown in the legend.



END

The chart presents a genetic linkage map of the human genome (current as of July, 1994) that integrates published data sets with those provided by recent genome-wide mapping efforts. Genetic distances, indicated in centimorgans (cM), are an additive measure of the proportion of cross-over

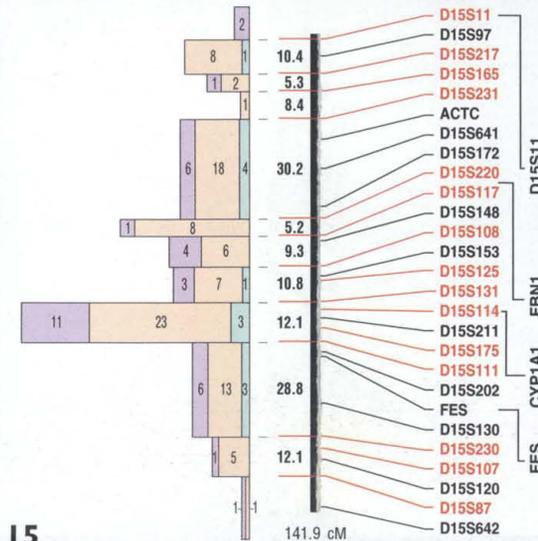
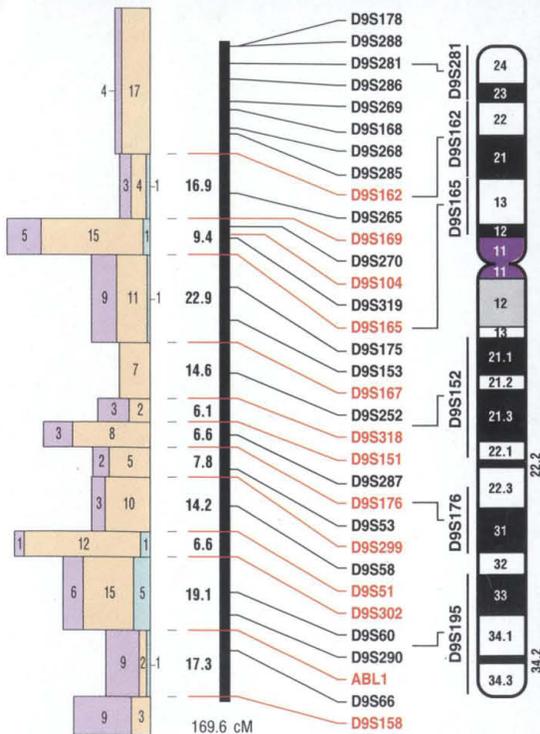
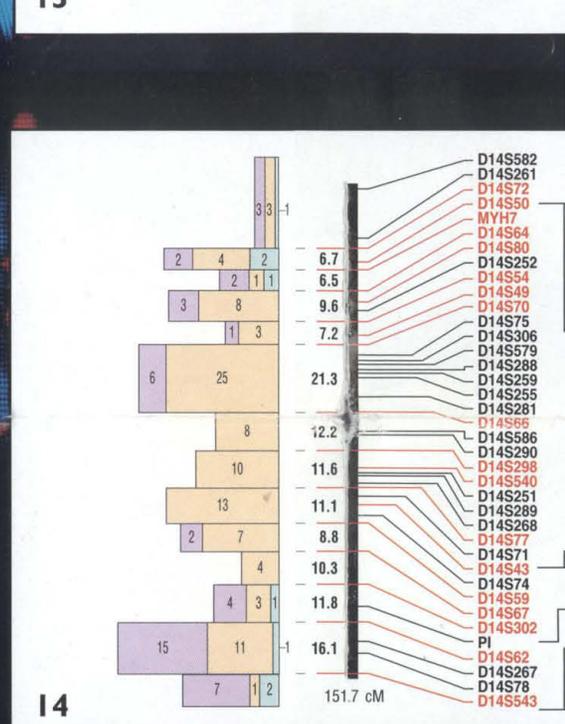
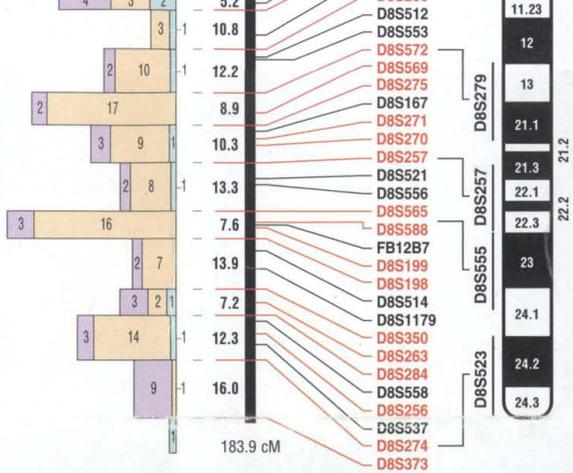
between defined points (loci) on the map that occurred during meiosis.

Proposed solely of loci that contain short tandem repeat polymorphisms (STRPs). A total of 971 loci are uniquely ordered. Loci contained in the map were obtained from four primary sources: Généthon, the mapping consortium of the Centre National de la Recherche Humaine (CEPH) and the National Institutes of Health (NIH), the Cooperative Human Linkage Center (CHLC), and the University of Utah Genome Center (UTAH). The data from Généthon are based on markers containing CA repeat polymorphisms. The markers with tri-, tetra-, and pentanucleotide repeat polymorphisms. The UTAH markers are primarily tetranucleotide repeat polymorphisms were generated from the CEPH reference panel of families.

The map was constructed by first building a reference map. The support for the order of loci in the reference map was by odds of 1000:1. The map was constrained to contain only loci that showed homogeneity of recombination among chromosomes. It did not inflate the map with their addition by more than 2 cM. The degree of map inflation is an estimate of the frequency of recombination events. The loci that are contained in this map are shown in red.

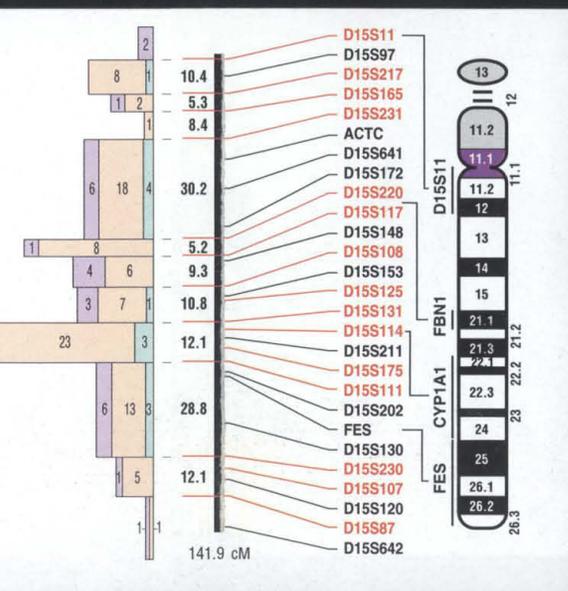
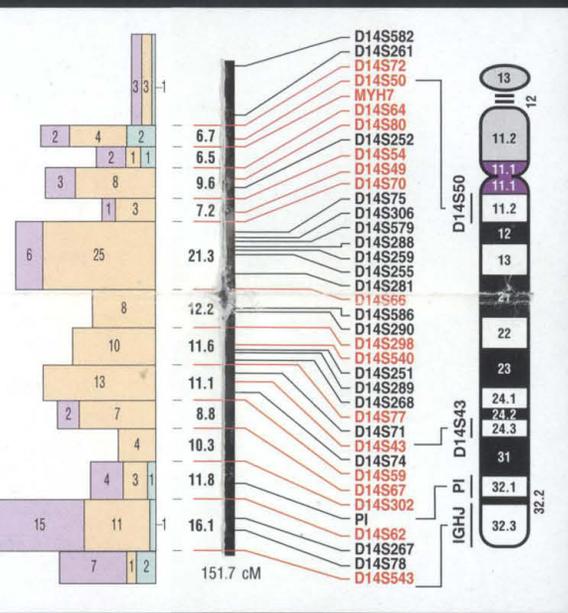
The map was used to define reference intervals for the construction of a more detailed map. Additional loci were assigned to the reference map. Loci could be excluded from all other intervals by odds of 1000:1. The relative order of loci within an interval that resulted in the most frequent recombination events is shown. A single recombination event was sufficient to establish order between loci. Sex-specific recombination rates for these maps are described in the accompanying article.

Genetic patterns for each chromosome shown on the idiograms are still a fundamental part of genetic diagnosis and gene localization. Differences between the cytogenetic and the genetic linkage maps are based on fluorescence in situ hybridization analyses of



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The map is composed solely of loci that contain short tandem repeats (STRPs). All data used were generated from the CEPH reference panel.

The map presented on the chart was constructed by first building a map that exceeded all others by odds of 1000:1. The map was constrained by the families used and did not inflate the map with their addition by the possibility of errors in the primary data. The loci that are contained in this map were selected so that the minimum number of recombination events is shown. A single interval is averaged and sex-specific recombination rates for these maps are shown.

This map was used to define reference intervals for the construction of reference intervals if they could be excluded from all other intervals by the minimum number of recombination events is shown. A single interval is averaged and sex-specific recombination rates for these maps are shown.

The banding patterns for each chromosome shown on the ideogram correspond to the cytogenetic and the genetic maps. The correspondences between the cytogenetic and the genetic maps, yeast artificial chromosomes (YACs) and data from the Genome Data Bank are shown so that marker density could be evaluated.

Accompanying the map is a histogram that includes the composition of additional markers available through the public version of the map. The map distances between these pairs of markers are shown (with the exception of the X and Y chromosomes). Each of the loci within an interval is presented for a total of 5840 polymorphic loci. The histogram is shown for each interval. As each interval varies in size, the width of the bar indicates the density of markers (loci per centimorgan).

Further information on the construction of the chart and the article (CHLC *et al.*) in the 30 September, 1994 issue of *Science*, which they were constructed can be obtained electronically through the Internet.

— Reference marker ■ Genes ■ STRPs

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• **Art:** DIRECTOR, Amy Decker Henry; DESIGN, Coblyn Designs; ILLUSTRATIONS, Coblyn Designs; ILLUSTRATIONS, Coblyn Designs; ILLUSTRATIONS, Coblyn Designs.

• **Fluorescent chromosomes:** Paul Meltzer, Jeffrey M. Trent, National Center for Human Genome Research, Bethesda, MD, USA. • We would like to acknowledge the staff at CEPH, whose work was essential to the construction of the human genome map.

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...the map is a histogram that includes the complete set of loci from the data sets described above, as well as the place-... rkers available through the public version of the CEPH database. A subset of reference loci was used to define larger... gram. The map distances between these points are shown on the left side of the genetic map and are sex-averaged (with... and Y chromosomes). Each of the loci within the data set was assigned to its most likely interval. Genetic locations are... of 5840 polymorphic loci. The histogram is subdivided to indicate genes, STRPs, and other polymorphisms assigned to... h interval varies in size, the width of the bar was scaled by dividing the number of markers by the interval's map dis-... bar width indicates the density of markers (per centimorgan) in that interval.

...ation on the construction of the chart and the state of human genetic linkage mapping can be found in the accompanying... n the 30 September, 1994 issue of *Science*. Access to more detailed presentations of the maps and the data sets from... ructured can be obtained electronically through [ftp.chlc.org](ftp:chlc.org), <gopher.chlc.org>, or <http://www.chlc.org/>.

marker ■ Genes ■ STRPs ■ Other polymorphisms // Distance uncertain

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