HIGHLIGHTS

GENETIC MAPPING

Recharting the human genome

Like maps of old, genetic maps are not always perfect, and so require constant updating with findings from new expeditions. Such an expedition has recently been carried out by scientists from deCODE genetics, who now report a new human genetic map. This map adds substantially to earlier ones, corrects previous errors and provides a detailed picture of recombination patterns across male and female genomes, revealing some intriguing differences.

Kong *et al.* began their expedition by genotyping a sample of 869 individuals, from 146 nuclear Icelandic families and containing 1,275 meioses, for 5,136 microsatellite markers. By comparison, the previous benchmark map — the Marshfield map — was derived from 188 meioses from eight large, three-generation families from the CEPH collection, which were genotyped for 8,000 microsatellite markers. Although lacking a third generation, the deCODE map achieves five times the resolution of the Marshfield map.

This increased marker resolution has several key benefits, such as allowing clustered markers to be resolved with greater reliability. For example, of the 5,012 markers shared by this and the Marshfield map, 3,690 were given distinct positions on this map compared with 2,866 positions on the Marshfield map. Increased marker resolution also highlighted assembly errors in the physical maps of the draft human genome — Kong *et al.* made 104 modifications to a 2001 release of the goldenPath assembly. Increased marker resolution also allowed the authors to better estimate genetic distances between markers and to survey genome-wide recombination patterns in detail.

Several findings emerged from this survey. Among them was that recombination rates, which vary locally along each chromosome, strongly correlate with sequence content — such as with G+C, CpG and poly(A) content — and with cytogenetic features, such as G bands. However, there are hints that other, sequence-independent, factors also influence recombination activity — for example, recombination occurs 1.65 times more frequently in female, than in male, autosomes, and recombination hot spots occur in different places in male and female genomes. No doubt, future studies will look for the stochastic, environmental and genetic factors that influence female recombination rates, which varied between the meioses of each woman studied.

There is much to be gained from this map in addition to that mentioned above, such as the availablity of sex-specific maps, which will provide better estimates of genetic distances for linkage studies. Although produced by a private company, these maps are available at the *Nature Genetics* web site and the primary data on application to deCODE. However, as James Weber remarks in an accompanying News & Views, future mapping expeditions are still much needed to fill in the details for cross-population studies of linkage disequilibrium.

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References and links

ORIGINAL RESEARCH PAPER Kong, A. *et al.* A high-resolution recombination map of the human genome. *Nature Genet.* **31**, 241–247 (2002)
FURTHER READING Weber, J. L. The Iceland map. *Nature Genet.* **31**, 225–226 (2002)
WEB SITE
Supplementary information: http://www.nature.com/ng/journal/v31/n3/suppinfo/ng917_S1.html