Genome Mapping

A PRACTICAL APPROACH

Edited by
PAUL H. DEAR

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Preface

No area of biology has undergone as explosive a growth as that witnessed in genome analysis over the last few years. The 1987 human linkage map, an impressive achievement of its time, gave the locations of 403 markers throughout the genome. More than that number of markers have now been placed on the smallest human autosome.

Several factors have contributed to this rate of advance. Automation has been an increasing factor in making large-scale projects feasible, as has the trend toward larger research groups and well co-ordinated collaborative projects: hundred-author papers are no longer the sole province of particle physics. A second factor has been the development and implementation of new techniques and strategies, each yielding a new generation of human genome maps. A further catalyst has been the sharing of materials and data within the genome community, and the use of shared resources such as clone libraries and mapping panels. The value of these resources increases as their use becomes more widespread, multiplying the benefits to the researchers who use them.

The mapping of non-human genomes (with the notable exceptions of a few model organisms) has so far lagged behind that of the human, but is surely set to undergo a similar revolution over the next few years. Not only are genome maps of agriculturally important species invaluable in their own right, but the need for maps of many plant and animal genomes will become increasingly apparent as the human genome project moves towards its ultimate goal of a complete sequence. Much of the information content of the human genome will be irrelevant or uninterpretable except in the context of comparative molecular genetics.

The methods of genome analysis do not fall into any natural sequence. I have tried as far as possible to arrange them in order of increasing resolution, from linkage analysis to restriction mapping. Two comprehensive appendices give details of resources—both biological and computational—available to the genome community.

Finally, I would like to thank the contributors for producing uniformly excellent chapters, my colleagues for their help and advice, and my wife Denise for her patience as much as for her proof-reading.

Cambridge P. H. D.
June 1997

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Abbreviations

^{5m}C	5-methylcytidine
^{6m}A	6-methyladenosine
AFLP	amplified fragment length polymorphism
ATCC	American Type Culture Collection
azaC	5-azacytidine
BAC	bacterial artificial chromosome
BLAST	Basic Local Alignment Search Tool
BrdU	5-bromodeoxyuridine
BSA	bovine serum albumin
CAD	carbamoyl phosphate synthase/aspartate transcarbamoylase/dihydroorotase
CCD	charge-coupled device
cDNA	complementary deoxyribonucleic acid
CDP-Star	disodium 2-chloro-5-(4-methoxyspiro[1,2-dioxetane-3,2′-(5′-chloro)-tricyclo[3.3.1.1^{3,7}]decan]-4-yl)phenyl phosphate
CEPH	Centre d'Etudes du Polymorphisme Humain
CHEF	contour-clamped homogeneous electric field
CHLC	Co-operative Human Linkage Centre
CHO	Chinese hamster ovary
CIAP	calf intestinal alkaline phosphatase
CISS	chromosomal *in situ* suppression (hybridization)
cM	centiMorgan
cR	centiRay
Cot	concentration × time (DNA reassociation kinetics)
DAPI	4,6-diamidino-2-phenylindole
ddGTP	dideoxy guanosine triphosphate
ddTTP	dideoxy thymidine triphosphate
DIG	digoxigenin
DNA	deoxyribonucleic acid
dNTP	any of dATP, dCTP, dGTP, or dTTP; or a mixture of all four
DOP	degenerate oligonucleotide primer (or primed)
DTE	dithioerythritol
DTT	dithiothreitol
EBV	Epstein–Barr virus
ECACC	European Collection of Animal Cell Cultures
EDTA	ethylenediamine tetraacetic acid (disodium salt unless specified)
EGTA	ethylene glycol *bis*(β-aminoethyl ether) *N,N,N,′N′*-tetraacetic acid
EMBL	European Molecular Biology Laboratory

EST	expressed sequence tag
F_1, F_2	first/ second filial generation
FAM	5-carboxyfluorescein
FITC	fluorescein isothiocyanate
F(B)CS	foetal (bovine) calf serum
FISH	fluorescence *in situ* hybridization
FSC	flow-sorted chromosome
FTP	file transfer protocol
GART	glycinamide ribonucleotide formyltransferase
Gb	billion (10^9) bytes (computing)
GCG	Genetics Computing Group
GDB	Genome Data base
HAT	hypoxanthine/aminopterin/thymidine
Hepes	*N*-2-hydroxyethylpiperazine-*N'*-2-ethanesulfonic acid
HGMP (RC)	Human Genome Mapping Project (Resource Centre)
hnRNA	heterogeneous nuclear RNA
HPRT	hypoxanthine phosphoribosyl transferase
HTML	hypertext markup language
IMAGE	Integrated Molecular Analysis of Genomes and their Expression
IRS	interspersed repeated sequence
JOE	6-carboxy-2',7'-dimethoxy-4',5'-dichlorofluorescein
LINE	long interspersed repeat element
LMP	low-melting point (agarose)
Lod	logarithm (base 10) of odds
Mb	million base pairs (DNA) *or* million bytes (computing)
MAS	marker-assisted selection
MES	2-(*N*-morpholino)-ethanesulfonic acid
MMCT	microcell-mediated chromosome transfer
OMIM	Online Mendelian Inheritance in Man
PAC	P1-derived artificial chromosome
PACE	programmable autonomously-controlled electrophoresis
PBS	phosphate-buffered saline
PCR	polymerase chain reaction
PEG	polyethylene glycol
PFG(E)	pulsed-field gel (electrophoresis)
PHA	phytohaemagglutinin
PI	propidium iodide
PIC	polymorphism information content
PMSF	phenylmethylsulfonyl fluoride
QFH	quinacrine/ fluorescence/ Hoechst 33258
QTL	quantitative trait locus
RAPD	randomly amplified polymorphic DNA
rDNA	ribosomal DNA

REM PCR	repeat element-mediated PCR
RFLP	restriction fragment length polymorphism
RH	radiation hybrid
ROX	6-carboxy-X-rhodamine
SDS	sodium dodecyl sulfate
SHMT	serine hydroxymethyl transferase
SINE	short interspersed repeat element
SSC	standard saline citrate
SSCP	single-strand conformational polymorphism
STR(P)	short tandem repeat (polymorphism)
STS	sequence-tagged site
TAFE	transverse alternating field electrophoresis
TAMRA	*N,N,N′N′*—tetramethyl-6-carboxyrhodamine
T_{ann}	annealing temperature (of DNA probe or oligonucleotide)
TEMED	*N,N,N′,N′*-tetramethylethylenediamine
TIFF	tagged information file format
TK	thymidine kinase
T_m	melting temperature (of DNA probe or oligonucleotide)
tRNA	transfer ribonucleic acid
UMP	universal mapping probe
UMPS	uridine monophosphate synthase
UV	ultraviolet
WWW	World Wide Web
YAC	yeast artificial chromosome

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