

Contents

<i>Preface</i>	vii
1 Introduction; Aims of Applied Genetics; Revision of Basic Genetic Concepts and Terminology	1
1.1 Introduction	1
1.2 The aims of applied genetics in humans, animals, plants and fungi; the use of genetic variation; improving the harvest index	8
1.3 Revision of basic genetic concepts, definitions and symbols	14
1.3.1 Alleles, genes, loci, wild-types and mutants	14
1.3.2 Ploidy	17
1.3.3 Genotype and phenotype; homozygotes and heterozygotes; hemizygotes; the time of gene expression	18
1.3.4 Dominance and recessiveness; incomplete dominance and additive action; primes; overdominance; co-dominance; pure breeding	19
1.3.5 Additive and multiplicative gene action	22
1.3.6 Pleiotropy	22
1.3.7 Mutation	23
1.3.8 Recombination; linkage; syntenic and non-syntenic loci; coupling and repulsion arrangements	24
1.3.9 Allelism and the cis/trans test	26

1.3.10	Heritability	29
1.3.11	Selection: natural, sexual, artificial and commercial	30
1.3.12	Populations	31
1.3.13	Polymorphism	33
1.3.14	Random mating	34
	Suggested Reading	35
2	The Inheritance and Analysis of Qualitative and Quantitative Characters	37
2.1	Single-locus qualitative characters: autosomal loci with complete dominance, partial dominance, additive action, overdominance; X-linked and holandric loci	37
2.1.1	Qualitative characters	37
2.1.2	Autosomal loci: complete dominance	38
2.1.3	Autosomal loci with partial dominance, additive action or overdominance	38
2.1.4	X-linked and holandric loci	40
2.2	Multiple-loci qualitative characters: dihybrid ratios and gene interactions such as epistasis causing modified ratios	42
2.2.1	Standard dihybrid ratios	42
2.2.2	Gene interactions such as epistasis causing modified dihybrid ratios	45
2.3	Quantitative characters; quantitative trait loci and polygenes; modifiers; threshold characters	49
2.4	Threshold characters	62
	Suggested Reading	64
3	Regression, Transgression, Environmental Effects and Heritability; Correlations Between Characters; Genotype, Phenotype and Breeding Values	65
3.1	Genetic and environmental causes of regression and transgression	65
3.2	Environmental effects on phenotypes	70
3.2.1	Sex-limited characters	70

3.2.2	Phenocopies, conditional mutants and environmental effects	70
3.2.3	Phenotype plasticity	71
3.2.4	Variable expressivity	72
3.2.5	Incomplete penetrance	72
3.3	Narrow and broad sense heritabilities; equations, estimation and use; use of twins; realised heritabilities; correlations between characters	74
3.4	Genotype value, phenotype value and breeding value	80
3.5	Estimated Breeding Values (EBV) and Best Linear Unbiased Predictions (BLUP)	83
	Suggested Reading	84
4	Population Genetics: Allele Frequencies, Genetic Equilibria, Population Mixing, Genetic Drift and Gene Flow	86
4.1	Introduction	86
4.2	The Hardy-Weinberg equilibrium for one locus and two loci; linkage disequilibrium, and population mixing	87
4.2.1	One locus, two alleles	87
4.2.2	One locus, more than two alleles	89
4.2.3	Allele segregation at two loci and the importance of recombination frequencies	89
4.2.4	What happens when two pure-breeding but different populations mix?	91
4.3	Genetic drift, fixation and effects of population size	97
4.4	Gene flow and population structure	100
4.5	Effects on allele frequencies of selection, mutation, migration and gene conversion; frequency-dependent selection; equilibria between forces in populations	105
4.5.1	Selection	105
4.5.2	Migration	109
4.5.3	Mutation	110
	Suggested Reading	112

5	Types and Uses of Selection	114
5.1	Natural, artificial and sexual selection	114
5.2	Stabilising selection, towards uniformity	116
5.3	Directional selection, favouring one extreme	118
5.4	Cyclic selection, alternatively favouring different extremes	119
5.5	Disruptive selection, selecting against the average type	120
5.6	Pedigree selection; breed records, prepotent stud males and grading-up	121
5.7	Progeny testing	123
5.8	Half-sib and family selection	125
5.9	Selecting for correlated characters	126
5.10	Selection for more than one character: tandem selection, independent culling levels and index selection	126
5.10.1	Tandem selection	127
5.10.2	Independent culling levels	127
5.10.3	Index selection, including PTA, PIN and £PLI	128
5.11	Selection intensities and rates of response to selection; a key equation for selection responses	132
5.12	<i>In vitro</i> selection	134
5.13	Selection at the haploid stage	135
5.14	Selection for meat characteristics	135
	Suggested Reading	138
6	Departures from Random Mating	140
6.1	Positive and negative assortative mating	140
6.2	Inbreeding and outbreeding and their consequences; inbreeding depression and outbreeding depression . . .	143
6.3	Why breeders often use inbreeding	149
6.4	Wright's inbreeding coefficient, F ; Wright's equilibrium for genotype frequencies under inbreeding; calculation of F from pedigrees	151
	Suggested Reading	160

7	Mutation and its Uses	161
7.1	Molecular types of mutation and their revertibility; mutation frequencies	161
7.1.1	Revertibility	161
7.1.2	Germ-line and somatic mutations	162
7.1.3	Base substitutions	163
7.1.4	Frame shifts	164
7.1.5	Large deletions	164
7.1.6	Unstable length mutations	165
7.2	Spontaneous and induced mutation; mutagenic agents	166
7.2.1	Spontaneous mutations; mutation mechanisms; varieties or breeds they have produced	167
7.2.2	Induced mutations	170
7.3	Mutation control and repair systems	174
7.3.1	Mutagen access	174
7.3.2	Repair systems	174
7.3.3	Suppressor mutations	175
7.3.4	Optimum mutation rates	176
7.4	How different types of mutation can complicate population genetics calculations	176
7.5	Using induced mutations	177
	Suggested Reading	180
8	Recombination, Mapping and Genomics	181
8.1	Recombination, genetic distances and the numbers of progeny needed to get particular recombinants	181
8.2	Types of recombination and their effects; meiotic and mitotic crossovers; interference and map functions	183
8.3	Numbers of types of gamete, and of offspring genotypes and phenotypes, for different numbers of segregating loci	187
8.4	Calculation of the frequencies of particular genotypes and phenotypes	188
8.5	Mapping, including physical mapping	188

8.6	Locating genes on particular chromosomes, including the use of pseudodominance, parasexual methods and hybridisation probes	192
8.6.1	Pseudodominance	192
8.6.2	Parasexual methods	194
8.6.3	Hybridisation probes	195
8.7	Practical uses of molecular markers in agriculture	196
8.8	Genomics	198
8.9	Human gene sequencing, including the HapMap Project	201
	Suggested Reading	202
9	Structural Chromosome Aberrations: Their Origins, Properties and Uses	205
9.1	Introduction	205
9.2	Deletions	207
9.3	Inversions, paracentric and pericentric; their effects on fertility	210
9.4	Duplications and the origin of new genes	213
9.5	Translocations, single and multiple	215
	Suggested Reading	219
10	Changes in Chromosome Number: Their Effects and Uses	220
10.1	Background	220
10.2	Changes in ploidy	222
10.3	Monoploids and anther culture	224
10.4	Diploids	228
10.5	Triploids	228
10.6	Tetraploids	228
10.7	Higher polyploids	232
10.8	Loss or gain of single chromosomes: aneuploids, monosomics and trisomics	232
10.9	Chromosome manipulations and substitutions	233
	Suggested Reading	236

11	Supernumerary ("B") Chromosomes	238
11.1	Definition, origins, numbers, size and functions	238
11.1.1	Origins	238
11.1.2	Numbers, occurrence and transmission	238
11.1.3	Size, sequences and function	240
11.2	Effects on phenotype and fertility	240
	Suggested Reading	242
12	Human and Medical Genetics	244
12.1	Introduction	244
12.2	Finding out how or whether characters are inherited: pedigree studies; twin studies; Hardy-Weinberg analysis; familial incidence	246
12.2.1	Pedigree studies	246
12.2.2	Twin studies	248
12.2.3	Hardy-Weinberg analysis of populations	253
12.2.4	Familial incidence	253
12.3	Single gene characters and disorders, and their treatments	255
12.3.1	Autosomal genes	256
12.3.2	X-linked genes	265
12.4	Polygenic and multifactorial disorders	266
12.5	X-inactivation and Barr bodies	268
12.6	Sex differences in disease susceptibility	271
12.6.1	X-linked diseases	271
12.6.2	Non-X-linked diseases	273
12.7	Causes of human mutation; cancer	274
12.7.1	UV light	277
12.7.2	Chemicals	277
12.7.3	Ionising radiations	280
12.7.4	Temperature	281
12.7.5	Infection	282
12.7.6	Cancer genes	282
12.8	Chromosome number abnormalities and chromosome aberrations	283
12.8.1	Human chromosome methods	283

12.8.2	Autosomal abnormalities	289
12.8.3	Sex chromosome abnormalities	296
12.9	Selection before and after birth	297
12.10	Blood groups, especially ABO and Rhesus	299
12.10.1	Blood groups and transfusions	299
12.10.2	The ABO blood group	302
12.10.3	The Rhesus blood group	303
12.10.4	Blood groups in farm animals	305
12.11	The major histocompatibility complex	305
12.12	Prenatal, neonatal and adult screening	307
12.12.1	Amniocentesis	307
12.12.2	Early amniocentesis	308
12.12.3	Chorionic villus sampling	308
12.12.4	Ultrasound screening	309
12.12.5	Maternal blood sampling	309
12.12.6	Pre-implantation genetic diagnosis	310
12.12.7	Foetal DNA screening	311
12.12.8	Neonatal screening (birth to one month of age)	312
12.12.9	Adult or adolescent screening	313
12.13	Effects of human inbreeding	315
12.14	Genetic counselling	319
12.15	Twins and other multiple births	327
12.16	Mosaics, chimeras and hermaphrodites	334
12.17	Gene therapy	338
12.17.1	Introduction	338
12.17.2	Methods for somatic gene therapy	339
12.17.3	Progress	342
12.18	Stem cell therapy, including the use of cord blood	345
	Suggested Reading	348
13	Plant and Animal Breeding Methods and Examples	353
13.1	Using hybrid vigour	353
13.1.1	Definition of hybrid vigour (heterosis)	353
13.1.2	Explanations of hybrid vigour	354
13.1.3	Typical F1 hybrid breeding programmes	355
13.1.4	Hybrid maize production	358

13.1.5	Hybrid sprout production	361
13.1.6	Hybrid rice production	362
13.1.7	Hybrid animals	362
13.2	Selection methods for inbreeders	363
13.2.1	Single line selection	363
13.2.2	Using a mixture of lines, with agricultural mass selection	364
13.2.3	Bulk population breeding	364
13.2.4	Pedigree breeding	365
13.3	Selection methods in outbreeders or random-maters	365
13.4	Recurrent backcrossing for gene transfer	368
13.5	Interspecific and intergeneric hybrids	369
13.5.1	Interspecific hybrids	369
13.5.2	Intergeneric hybrids	374
13.6	Making polyploids	376
13.7	Examples of plant and animal breeding programmes	377
13.7.1	Semi-dwarf wheats and rice — the “Green Revolution”	377
13.7.2	Broad beans	379
13.7.3	Semi-leafless combining pea	379
13.7.4	Coca plants for cocaine	380
13.7.5	Potato breeding and disease-resistance	380
13.7.6	Lime-tolerant Rhododendrons	384
13.7.7	Sugar cane	385
13.7.8	Hops in Britain — dwarf “hedgerow” hops	386
13.7.9	Sheep, including cross-breds and Border Leicesters	387
13.7.10	Cattle, including Ayrshires (dairy) and Aberdeen-Angus (beef)	389
13.7.11	Peruvian guinea pigs	396
13.7.12	Chinese crocodiles	397
13.7.13	Pig breeding to meet the buyer’s specifica- tions; meat quality factors	397
13.8	Breeding for shows; breeds and varieties	399

13.9	Breeding programmes from crosses to selection, to local and national trials, possible commercial release, and approved lists	402
13.10	Selection in feral animals; feral and farmed animals	405
13.11	DNA fingerprinting	407
	Suggested Reading	408
14	Genetic Engineering in Plants, Animals and Micro-Organisms	411
14.1	Introduction	411
14.2	Restriction endonucleases and ligases	412
14.3	Vectors	414
14.4	Getting a particular piece of DNA into a vector, and recognising a clone containing it	416
14.5	Site-directed mutagenesis	418
14.6	Gene targeting; cosuppression; RNA interference in humans, animals and plants	419
14.7	Genetic engineering in plants	423
14.8	Genetic engineering in animals	428
14.9	Genetic engineering in micro-organisms	433
14.10	Some dangers of genetic engineering; the amount of genetically engineered crops grown	434
	Suggested Reading	439
15	Genetic Variation in Wild and Agricultural Populations; Genetic Conservation	442
15.1	The forces controlling the amounts of variation in a population	442
15.1.1	The forces or processes which increase or maintain genetic variation within a population	442
15.1.2	The forces or processes which reduce genetic variation within a population	445
15.1.3	The interactions of forces or processes affecting the amount of variation within a population	445

15.2	Using a knowledge of the origins of genetic variation to solve a practical problem	446
15.3	The maintenance of polymorphism in populations . . .	447
15.4	The need for genetic conservation; methods of conservation	450
15.4.1	The need for genetic conservation and the value of some old varieties	450
15.4.2	Conservation programmes and methods of genetic conservation	452
	Suggested Reading	460
16	Genetic Methods of Insect Pest Control	462
16.1	Introduction	462
16.2	The release of sterile insects, or of fertile insects giving inviable progeny	463
16.3	The breeding of insect-resistant varieties	466
	Suggested Reading	469
17	Reproductive Physiology in Plants, Animals and Humans; Crossing Methods	470
17.1	Plants	470
17.1.1	Plant sexual reproduction	470
17.1.2	Incompatibility in higher plants and ways of overcoming it	477
17.1.3	Crossing methods	481
17.1.4	Getting uniform fruit, seed or bud ripening	483
17.1.5	Somaclonal variation and vegetative propagation; grafting and rootstocks	484
17.1.6	Plant protoplast fusion	494
17.1.7	Gene expression and natural and artificial selection at the haploid stage	495
17.2	Animals	496
17.2.1	Sex ratios	496
17.2.2	Flow cytometry for sexing sperm	497
17.2.3	Anatomy, progeny per pregnancy, and temperature effects	499

17.2.4	Breeding seasons and oestrous cycles	501
17.2.5	Sperm; natural and artificial insemination	505
17.2.6	Egg transplantation and embryo freezing	510
17.2.7	Animal cloning	511
17.3	Humans	513
	Suggested Reading	516
18	Applied Fungal Genetics	519
18.1	General fungal genetics: life cycles; wild-types and mutants; spore types; control of sexual and vegetative fusions; genomics	519
18.1.1	Life cycles	519
18.1.2	Wild-types and mutants	522
18.1.3	Spores	523
18.1.4	The control of vegetative and sexual fusions	524
18.1.5	Fungal genomics: nuclear, mitochondrial and plasmid	525
18.2	The commercial importance of fungi	527
18.3	Recombination and sexual mapping	528
18.4	The parasexual cycle and parasexual mapping	534
18.5	The induction and isolation of mutants, including auxotrophs	541
18.6	Obtaining improved strains for industry	544
18.6.1	Aims and methods	544
18.6.2	Different aims in different yeasts	547
18.6.3	Improving baker's yeast	550
18.6.4	Improving enzymes in industrial fungi	554
18.6.5	Penicillin production	557
	Suggested Reading	559
19	The Economics of Agricultural Products and Breeding Programmes	562
19.1	Basic economics: economic systems; price theory; factors affecting supply and demand; perfect and imperfect competition; monopolies; inflation	562

19.1.1	Factors of production; types of economic system	562
19.1.2	Price theory	564
19.1.3	Types of competition	570
19.1.4	Inflation	574
19.2	Economics applied to agriculture	575
19.2.1	Gluts and shortages; how governments intervene in agriculture; European Union policies	575
19.2.2	Seasonal and perishable produce	581
19.2.3	The value of rarities	582
19.2.4	"Health foods" and "organic" products	585
19.2.5	Discounted cashflow assessment of breeding programmes	588
19.2.6	Breeders' rights	590
19.2.7	Breeding or using for niche markets	592
19.2.8	Who does the breeding?	593
	Suggested Reading	594
	Index	595