CHAPTER 5

Human Karyotypes and Chromosome Behavior

5-1. The orange and black patches are due to alleles in the X chromosome. In heterozygous females, because of X inactivation, some cell lineages express the allele for orange fur and other lineages express the allele for black fur.

5-2. For 47 chromosomes: 47, +21 (trisomy 21 or Down syndrome), 47,XXX or 47,XXY; For 45 chromosomes; 45, X.

5-3. The mosaic females are heterozygous; in some skin cell lineages the X chromosome with the wildtype allele is expressed, in other skin cell lineages the X chromosome with the mutant allele is expressed.

5-4. It is true but unexpected, because 47,XXX would be expected to produce many XX eggs and 27,XXY many XY or XX sperm. Apparently the extra X chromosome is eliminated from the nucleus prior to meiosis.

5-5. (a) 1; (b) 0; (c) 0; (d) 0; (e) 2.

5-6. The chromosomes underwent endoreduplication, resulting in an autotetraploid.

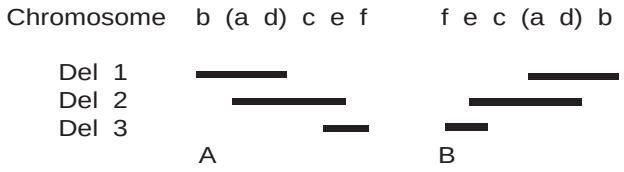
5-7. It is not literally true. It means that the products of single crossing-over are not recovered in the progeny. Single crossovers within the inverted segment give rise to aneuploid and usually nonviable progeny. In case of paracentric inversion, a single crossover would result in two abnormal chromatids (one with two centromeres and one with none). In case of a pericentric inversion, all the chromatids would be monocentric, but one chromatid would have a deletion and the other a duplication of genetic material. Thus an inversion does not prevent crossing-over but merely renders chromatids with one crossover unrecoverable in the progeny. On the other hand, although they are rarely formed, some chromatids resulting from multiple crossovers within an inversion can result in viable recombinant progeny.

5-8. The inversion has the sequence *A B E D C F G*, the deletion *A B F G*. The possible translocated chromosomes are (1) *A B C D E T U V* and *M N O P Q R S F G* or (2) *A B C D E S R Q P O N M* and *V U T F G*. One of these possibilities includes two monocentric chromosomes, and the other includes a dicentric and an acentric. Only the translocation with two monocentrics is genetically stable.

5-9. The calico male is XXY and results from sex-chromosome nondisjunction in the father.

5-10. The gene order is a e d f c b or the reverse.

5-11. Problems of this sort are worked by noting that genes uncovered by a single deletion must be contiguous. The gene order is deduced from the overlaps between the deletions. The overlaps are *a* and *d* between the first and second deletions and *e* between the second and third deletions. The gene order (as far as can be determined from these data) is diagrammed in part A of the accompanying illustration. The deletions are shown by the heavy lines. Gene *b* is at the far left, then *a* and *d* (in unknown order), then *c*, *e*, and *f*. (Gene *c* must be to the left of *e*, because otherwise *c* would be uncovered by deletion 3.) Part B is a completely equivalent map with gene *b* at the right. The ordering can be completed with a three-point cross among *b*, *a*, and *d* or among *a*, *d*, and *c* or by examining additional deletions. Any deletion that uncovers either *a* or *d* (but not both) plus at least one other marker on either side would provide the information we need to complete the ordering.



5-12. Gene *a* is present in band 2, *b* in band 1, *c* in band 3, *d* in band 5, *e* in band 4, and *f* in band 6.

5-13. The mother has a Robertsonian translocation that joins the long arm of chromosome 21 with the long arm of another acrocentric chromosome. The affected child has 46 chromosomes. This karyotype differs from the usual karyotype for trisomy 21 because the extra chromosome 21 is no a free chromosome.

5-14. The *Cy* gene is now linked with the Y chromosome, the most likely reason being because of a reciprocal translocation between 2 and the Y chromosome.

5-15. One possibility is that the metacentric chromosome in *D. texana* originally consisted of two acrocentrics that underwent a Robertsonian translocation (centromeric fusion). Another is that the metacentric in *D. texana* is the ancestral state and that two acrocentrics in *D. virilis* were formed by the splitting of the centromere (and the addition of telomeres). Since the ancestral karyotype of *Drosophila* is like that of *D. virilis*, the first possibility is almost certainly the correct one.

5-16. **(a)** In homozygotes, there is no impediment to crossing-over. Hence, for a map distance of 12 map units, one should expect to observe 12 percent recombination, because over this length of genetic interval, multiple crossing-over can be neglected. **(b)** In heterozygotes, the products of recombination would not be recovered, and if the whole region were involved in the inversion the frequency of recombination would be 0. Because only 1/3 of the interval is inverted in this case, the recombination frequency is expected to be $(2/3) \times 12 = 8$ %.

5-17. (a) 71/595 = 11.9 %; **(b)** 25 % for each of the four classes of progeny.

5-18. Species A ($n = 6$) hybridizes with species B ($n = 6$). The F_1 progeny will have 12 chromosomes and be sterile. The sterility can be overcome by endoreduplication in an F_1 organism, which creates a fertile new species with a chromosome number of 24. This new species ($n = 12$) hybridizes with a third species, C ($n = 6$), yielding another sterile F_1 progeny with 18 chromosomes. Endoreduplication in one of these sterile F_1 organisms gives rise to a fully fertile new species with 36 chromosomes. Note that this scenario is very similar to that which produced hexaploid wheat.

5-19. (a) The process is polyploidy, possibly hybridization between species followed by doubling of the chromosome number in the hybrids to produce fertile individuals. **(b)** The basic number in a single monoploid set of chromosomes is 13. **(c)** The 156-chromosome species has 12 sets of chromosomes (a dodecaploid).