recognized by the presence of inversion loops in meiotic pachytene chromosomes. These structures occur because of the affinity of the two homologues. The only was the two homogues can pair is if one twists on itself and makes a loop, while the other makes a loop without a twist. These loops can best be seen in the polytene chromosomes of organisms such as Drosophila pseudoobscura.

4. **Translocations:** A **translocation** is the movement (by breaking and rejoining) of a chromosomal segment from one chromosome to another, non-homologous chromosome. There are two types of translocations, an **interstitial translocation**, involving the one-way movement of a segment, and the more common **reciprocal translocation**, involving a two-way exchange of chromosomal segments. If two of the segments that join in a reciprocal translocation are large and the other two are small, the smaller translocated chromosomes are often lost. In this case, the number of chromosomes is reduced by the chromosomal exchange. Obviously, translocations can change both the size of chromosomes and the position of the centromere. Even though chromosomal segments have been exchanged between chromosomes in a reciprocal translocation, the affinity of the homologous regions results in pairing during meiosis I. If nearly equal parts of chromosomes are exchanged or not exchanged, the paired chromosomes in a translocation heterozygote have a cross appearance in metaphase I. During anaphase I, two major types of segregation occur: one in which adjacent centromeres goes to the same pole (adjacent I.) and two, the alternate centromere goes to the same poles. When alternate centromeres go to the same pole, the chromosomes often form a figure eight shape in early anaphase I. The products of this event, which is known as **alternate segregation**, are balanced so that each gamete has a full complement of chromosomes; either two untranslocated or two balanced translocated. On the other hand, when adjacent centromeres segregate together, **adjacent segregation**, the chromosomes appear as a ring at metaphase I. When this occurs, the products are unbalanced, resulting in duplications and deletions in the gametes. Some plants, and also a few animals, have a series of reciprocal translocations, so that chromosomal heterozygotes also have nearly

all the chromosomes associated in a large ring (or rings) in meiosis. However, at anaphase these chromosomes may undergo an orderly alternate segregation, producing only zygotes with a balance chromosomal complement. Although translocations can resulting in normal chromosomes, they can also cause several human diseases. For example, about 5% of individuals with **Down syndrome** have one parent who is heterozygous for a translocation. In this instance, chromosome 14 is translocation onto chromosome 21. Half of the time, the heterozygote produces either the normal set or a balanced translocated set of chromosomes, making the progeny either normal or translocation heterokaryotypes, respectively. The other half of the time, unbalanced chromosomes are produced, either a 14 without the translocated 21 segment or a translocated 14 without the translocated 21 segment or a translocated 14 with the attached 21 plus a normal 21. In the first case, offspring get only one 21 chromosome, a lethal chromosomal component. In the second instance, three 21 chromosomes are received, resulting in Down syndrome. Overall then, approximately one-third of the live births from some a translocation heterokaryotype can be expected to have Down syndrome. In actual fact the proportion is less than this, primarily because some Down individuals do not survive gestation. Note that this cause of Down syndrome has implications for genetic counseling. First, Down syndrome could recur in children of a transolocation heterokaryotye, whereas normally Down syndrome does not recur in sibs Second, half of the phenotypically normal sibs of Down individuals are themselves translocation heterokaryotypes, and therefore could produced Down progeny.

Changes in chromosomal number

The numbers of chromosomes may vary in two basic ways: **euploid** variants, in which the number of chromosomal sets differ, and **aneuplid** variants, in which the number of a particular chromosome is not diploid. As one might expect, changes in chromosome number, either euplid or aneuploid, generally have a greater effect on survival than do changes in chromosome structure. In fact, in humans, more than half of

homologous, just as they are in a diploid. But in allopolyploids, the different chromosomal sets generally vary somewhat and are called **homeologous** or partially homologous.

Triploid organisms are usually **autopolyploids** (AAA) that result from fertilization involving a haploid and a diploid gamete. They are normally sterile because the probability of producing balanced gametes is quite low. For example, most bananas are triploids; they produced unbalanced gametes, and as a result, are seedless (they are propagated by cuttings). **Allopolyploids:** Most naturally occurring polyploids are allopolyplods, and they may result in a new species. For example, the bread wheat Tritium aestipum is an allohexaploid with 42 chromosomes. By examining wild related species, it appears that bread wheat is descended from three different diploid ancestors, each of which contributed two sets of chromosomes (in this case designated as AABBDD). Pairing occurs only between the homologous sets, so that meiosis is normal and results in balanced gametes of n = 21.

Aneuploidy: The cause of aneuploidy is non-disjuction; that is, two homogolous chromosomes fail to separate properly during meiosis or mitosis. Non-disjuction in meiosis itself is thought to result from improper pairing of homologous chromosomes on opposite sides of the metaphase plane, or from failure of chiasma formation. As a result, both chromosomes may go to the same pole, leaving one daughter cell with an extra chromosome and the other daughter cell with no chromosome. When these gametes are fertilized by a normal gamete, they either have an extra chromosome, 2n + 1, termed **trisomy,** or are missing a chromosome, 2n – 1, termed **monosomy.** Non-disjuction is most common in meiosis 1, but it can occur in meiosis II as well. Non-disjuction can also take place in mitosis, resulting in mosaics for normal and aneuploid cells. Other combinations of extra chromosomes are possible, the most important being a tetrasomic with 2n + 2 chromosomes and a nullisomic with 2n – 2 chromosomes, in which no copies of a particular homologue exist. Trisomics are known in many different species. They are viable in many plants, but are less frequently viable in animals. For example, among the aneuploids that have been most thoroughly studies are those in the Jimson weed, or thorn apple. A series of Datura mutants with strange

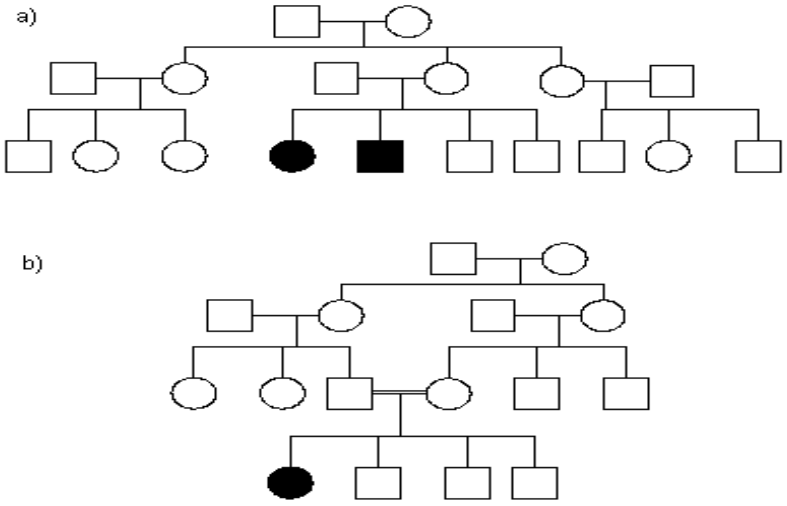
2. Replace the herd sires culled with animals whose pedigrees indicate there should be only minimal probabilities of the new sires being heterozygous for the defects.
3. Remove all females which have produced defective offspring from the seed-stock herd itself. They may be placed in an auxiliary herd and used to progeny – test future herd sires to determine whether they are heterozygous for the gene(s) responsible for the defect.
4. Cull other close relatives of affected individuals including normal offspring of sires and dams which have produced defective individuals.
5. If the affected individuals are viable and fertile, retain them for progeny – testing prospective breeding animals.
6. Progeny – test prospective herd sires before using them extensively in the herd.

Penetrance and Expressivity

A gene does *not* determine a phenotype by acting alone, it does so only in conjunction with other genes and with the environment. In the examples of gene interactions, the genetic basis of the dependence of one gene on another has been worked out from clear genetic ratios. However, in other situations, where the phenotype ascribed to a gene is known to be dependent on other factors but the precise inheritance of those factors has not been established, the terms *penetrance* and *expressivity* may be useful in describing the situation.

Penetrance is defined as the percentage of individuals, with a given genotype which exhibit the phenotype associated with that genotype. For example, an organism may have a particular genotype but may not express the phenotype normally associated with that genotype because of modifiers, epistatic genes or suppressors in the rest of the genome or because of modifying effect of the environment. Penetrance can b used to measure such an effect when it is not known which of these types of modification underlies the effect. On the other hand, expressivity describes the extent

a) A 'typical' autosomal recessive pedigree, and b) an autosomal pedigree with inbreeding:



If the parents are related to each other, perhaps by being cousins, there is an increased risk that any gene present in a child may have two alleles identical by descent. The degree of risk that both alleles of a pair in a person are descended from the same recent common ancestor is the degree of inbreeding of the person. Let us examine b) in the figure above. Considering any child of a first cousin mating, we can trace through the pedigree the chance that the other allele is the same by common descent. Let us consider any child of generation IV, any gene which came from the father, III_3 had a half chance of having come from grandmother II_2, a further half chance of being also present in her sister, grandmother II_4 a further half a chance of having been passed to mother III_4 and finally a half chance of being transmitted into the same child we started from. A total risk of½ x ½ x ½ x ½ = 1/16

This figure, which can be thought of as either the chance that both maternal and paternal alleles at one locus are identical by descent or the proportion of all the individual's genes that are homozygous because of identity by common descent, is known as the coefficient of inbreeding and is usually given the symbol F.