CHAPTER XII

HETEROPLOIDS

IRREGULARITIES in the division or the separation of the chromosomes occasionally cause a single chromosome to be added to the group. Conversely, one may be lost from the group. In so far as the addition of one or more chromosomes to, or loss from, a given group produces a new number, the word heteroploid has been used. Another word, trisomic, has also been used for cases where three of one kind are present (in contrast to triploid, where there are three of each kind present) and the word triplo combined with the name of the particular chromosome in triplicate has also been used, as triplo-IV in Drosophila. Still earlier, an extra chromosome was called a supernumerary or m-chromosome, etc. The loss of one member of a pair is designated by the term haplo- combined with the name of the particular chromosome, as in the haplo-IV type in Drosophila.

Certain mutant types of Oenothera have been found to be associated with the addition of a fifteenth chromosome.

Normally Lamarck's evening primrose has 14 chromosomes. Certain mutant types, known as lata and semi-lata, have 15 chromosomes, *i.e.,* one additional chromosome (Fig. 100). The lata plants differ from Lamarckiana in many small details, although most of the differences are so slight that only an expert would notice them. According to Gates, one of the lata mutants is almost completely male-sterile, and its production of seed is also greatly reduced. In one of the semi-lata types some good pollen is produced.

The frequency of occurrence of lata types varies in different progenies from 0.1 to 1.8 per cent, according to Gates.

At the maturation of the pollen of the 15 chromosome types, 8 chromosomes are present. Seven are in pairs

FIG. 100.
Oenothera lata. (After Anne Lutz.)

and 1 is unpaired. The conjugants separate and pass to opposite poles at the first maturation division. The unpaired chromosome does not divide at this time, but passes intact to one or to the other pole. Other irregularities in the maturation divisions occur in some cases, but whether or not they are caused by the extra chromosome is unknown, although Gates states that these irregu-

larities are much more frequent in triplo-typic individuals than in normals.

From the 15 chromosome types two kinds of germ-cells are expected, one with 8, one with 7 chromosomes. It has been shown that these two kinds are produced. From a genetic standpoint the lata type, crossed to a normal type, should produce equal numbers of lata (8+7) and normal (7+7) offspring. This is approximately what happens.

The most interesting question concerning these triplotypes relates to the particular chromosome that becomes the supernumerary. Since there are seven kinds of chromosomes, we may anticipate that any one may appear in triplicate. De Vries has recently suggested that there are seven trisomic types in Oenothera, corresponding to the seven possible supernumeraries.

It is also important to bear in mind that types with two supernumeraries (either like or unlike), the tetrasomic types, may not be as viable as trisomic types. It is known that such types occur. For instance, amongst the offspring of a triplo-type there seems to be a good chance for the formation of an individual with two like supernumeraries when an 8-chromosome pollen grain fertilizes an 8-chromosome egg. This would give a tetra-type or tetrasomic group for one particular chromosome. It would be a stable type to the extent that 8 paired chromosomes are present in each germ-cell, but it might be even more unbalanced than a triplo-type with only one extra chromosome. Sixteen-chromosome types have been recorded, some of which are probably multiples of the same chromosome when they are derived from a 15 triplotype, but their relative viability is not recorded.

It seems, *a priori*, possible that duplication of any pair of chromosomes may be brought about through a triplotype giving rise to a tetratypic individual. But even if

stability should be attained, the more important factor of gene balancing may make it improbable that a permanent increase in the chromosome pairs could be established in this way. When a large chromosome number is present the initial stages of unbalancing might be slight as compared with forms having fewer chromosomes, because in the former the ratio would be less disturbed.

In Drosophila Bridges found a triplo-type for the small IV-chromosome, and since three genetic factors are present in this small chromosome it has also been possible to study not only the characters that are affected by the presence of an additional IV-chromosome, but the bearing of this condition on genetic questions in general. On the other hand, it has been found that an individual with three X-chromosomes usually dies, and that individuals with either chromosome-II or -III in triplicate do not live.

The triplo-IV Drosophila is not strikingly different from the normal, and the two can be distinguished only with difficulty. The general color of the body is a little darker and the trident marking on the thorax is absent (Fig. 32); the eyes are somewhat smaller and have a smooth surface; the wings are narrower and more pointed than those of the wild type. That these slight effects are due to the presence of an additional small chromosome was shown both by a cytological demonstration of its presence (Fig. 32) and by genetic tests. When a triplo-IV is crossed to eyeless (eyeless is a IV-chromosome recessive mutant type) some of the offspring (F_1) can be distinguished by the characters given above as triplo-IV flies. If these are back-crossed to eyeless (Fig. 33), flies with full eyes and flies with "eyeless eyes" are produced approximately in the ratio of 5 to 1. As shown in Fig. 33 this result agrees with expectation provided that one normal gene is dominant to two eyeless genes.

When two triplo-IV flies (obtained in the way described above) that have two ordinary IV-chromosomes and another IV-chromosome carrying eyeless, are mated, they give approximately 26 full-eyed flies to one eyeless.

From this cross some flies might be expected that contained four chromosome-IV's, since half of the eggs and half of the sperm-cells carry two of these chromosomes. If such tetra-typic flies developed, the expected ratio would be 35 full-eyed to one eyeless. The ratio found (26 to 1) instead of the expected ratio (on the assumption that the tetra-typic flies come through) is due to the death of the tetra-types. In fact, no flies of this composition have been detected, which means that, despite the smallness of these chromosomes, the presence of four of them upsets the balance of the genes to such an extent that such an individual does not develop into an adult.

In contrast to these triplo-types of Drosophila there is another heteroploid type, the haplo-IV type (Fig. 29), in which one of the small chromosomes is absent. This type has appeared very often, which is interpreted to mean that one of these small chromosomes is sometimes lost in the germ track—possibly as a result of two passing to one pole at the reduction division. The haplo-IV has a paler body color but a more marked trident on the thorax, rather large eyes with a rough surface, slender bristles, and somewhat shortened wings, and the aristae are reduced or even absent. In all these respects its characters are the opposite of those of the triplo-type. This is not at all surprising if the IV-chromosome contains genes that affect many parts of the body in conjunction with other genes. These effects are increased by the presence of an additional chromosome and diminished when one is absent. The haplo-IV's emerge four or five days later than the normals; they are often sterile and generally poor producers; their mortality is very high. There is abun-

dant cytological and genetic evidence that these flies owe their peculiarities to the absence of one chromosome.

Flies lacking both IV-chromosomes have not been found and the ratio obtained when two haplo-IV's are bred together (giving 130 haplo-IV's to 100 normals) shows that the nullo-IV's die.

If a diploid fly that is eyeless is mated to a haplo-IV fly carrying wild type genes in its single chromosome-IV, some of the F_1 offspring will be eyeless and these will be haplo-IV. Theoretically, half of the offspring should be eyeless, but the presence of the eyeless gene in the single fourth chromosome lowers the viability of the haploid 98 per cent of expectation, and this relation holds when the other recessive mutant types (bent and shaven) are present in the single IV-chromosome. According to Bridges, bent lowers survival by 95 per cent and shaven, 100 per cent, *i.e.*, haplo-shaven does not develop.

The Jimson weed, Datura stramonium, has 24 chromosomes. A number of types under cultivation have been detected by Blakeslee and Belling with 25 chromosomes (2n+1). It is probable that there are 12 such types, each of which has a different extra chromosome. The slight but constant differences shown by these 12 triplo-types (2n+1) involve all parts of the plant. These differences are well shown in the capsules (Fig. 101). In two of these, at least (triplo-globe and triplo-poinsettia), in which Mendelian factors are present in the extra-chromosome group, it has been shown by Blakeslee, Avery, Farnham, and Belling, that the twenty-fifth chromosome involved is a different one in the two cases. In one of these in particular, namely, the trisomic type poinsettia, involving a chromosome that carries the gene for purple stem pigment and white flower color, the effects on the inheritance due to one extra chromosome have given the clearest results. These show that those germ-cells carrying the extra chro-

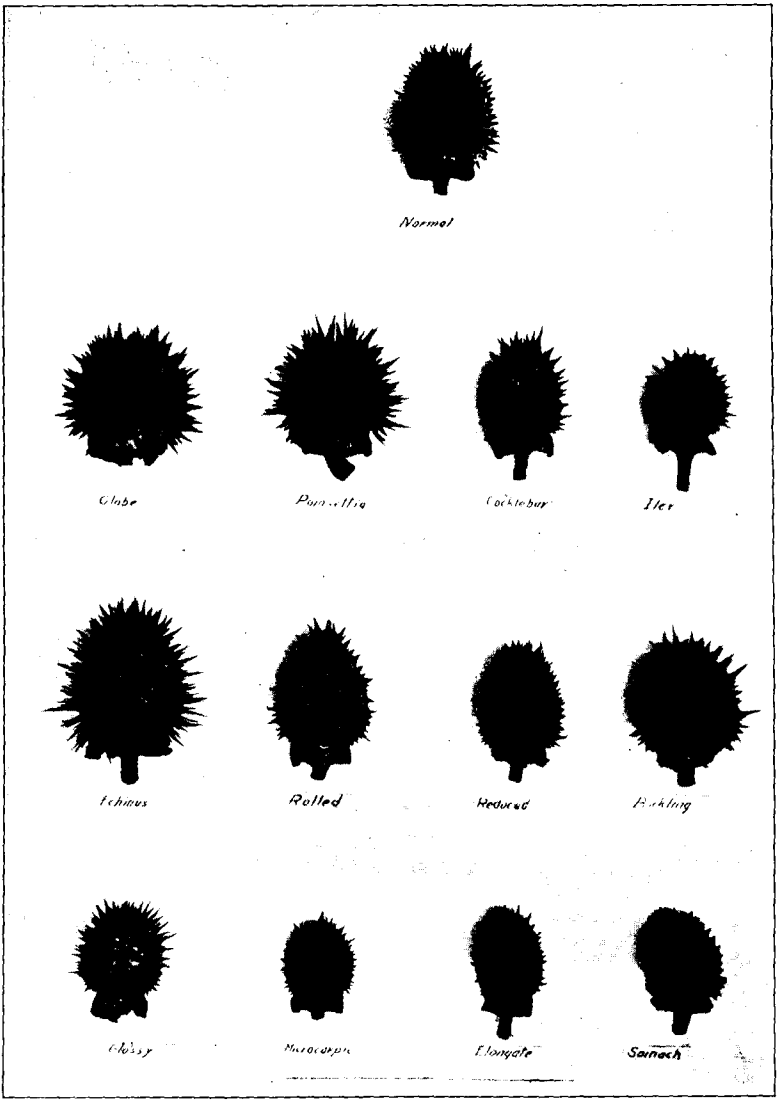

FIG. 101.

The original type of seed-capsule of Datura stramonium, and the twelve probable trisomic types. (After Blakeslee, in Journal of Heredity.)

mosome are less viable than the normal, hence deficiencies in certain expected classes occur; in fact, these germ-cells (n+1) are not transmitted at all through the pollen

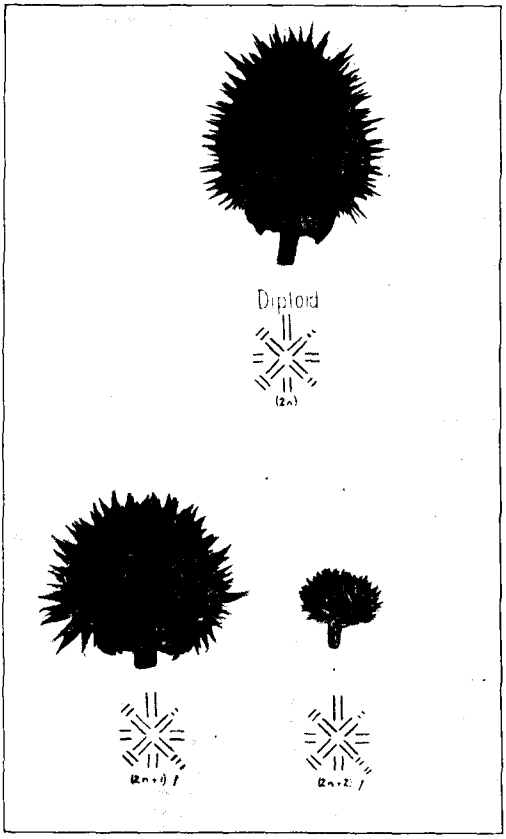

FIG. 102.

Diploid type of capsule of Datura (2n) as contrasted with 2n+1 and 2n+2 types of capsule. (After Blakeslee, in Journal of Heredity.)

(or only to a slight extent), and through only about 30 per cent of the eggs. When these relations are allowed for, the genetic results agree with expectation.

In their study of the trisomic types of Datura, Blakeslee and Belling have found about 12 distinct types belonging to the 2n+1 or trisomic series. Since there are just 12 pairs of chromosomes, only 12 simple trisomic types are expected, and, in fact, evidence has been found that there are only 12 such primary types. The rest, called secondaries, appear to belong to one or another of the 12 primary types (Fig. 102). The evidence for this comes from several sources, from similarities in external appearance, from internal structures (as shown by Sinnott), from their similar mode of inheritance (giving the same trisomic inheritance for marked chromosomes), *from the reciprocal throwing of one member of the group* by the other, and from the sizes of the extra chromosomes (Belling).

In the following table a list of the primaries and their secondaries is given. These have been derived from triploids.

PRIMARY AND SECONDARY (2n+1) TYPES IN OFFSPRING FROM TRIPLOIDS
(Primaries are printed in capitals, secondaries in lower case type.)

	3n × SELF	3n × 2n	TOTAL		3n × SELF	3n × 2n	TOTAL
1. GLOBE	5	46	51	8. BUCKLING	9	48	57
2. POIN-SETTIA	5	34	39	Strawberry	..	...	...
Wiry	..	..	..	Maple	..	...	...
3. COCKLE-BUR	6	32	38	9. GLOSSY	2	30	32
Wedge	..	1	1	10. MICRO-CARPIC	4	46	50
4. ILEX	4	33	37	11. ELONGATE	2	30	32
5. ECHINUS	3	15	18	Undulate	..	...	...
Mutilated	..	(2?)	(?)	12. SPINACH(?)	..	2	2
Nubbin (?)	..	..	..	Totals (2n + 1)	43	381	424
6. ROLLED	..	24	24	(2n + 1 + 1)	11	101	112
Sugarloaf	..	..	..	2n	30	215	248
Polycarpic	..	..	..	4n	3	...	3
7. REDUCED	3	38	41	Grand Totals	87	697	784

The spontaneous occurrence of primaries and secondaries is given in the next table. The primaries arise in this way more frequently than the secondaries. Breeding experiments have shown that whereas primaries may occasionally throw secondaries, the secondaries regularly throw their primaries more frequently than they throw new mutants belonging to the other groups. Thus of 31,000 offspring from poinsettias about 28 per cent were poinsettia and about 0.25 per cent were the secondary wiry. Conversely, when wirys were the parents about 0.75 per cent of the offspring were the primary poinsettia.

SPONTANEOUS OCCURRENCE OF PRIMARY AND SECONDARY (2n+1) MUTANTS
(Primaries are printed in capitals, secondaries in lower-case type.)

	FROM 2n PARENTS	FROM UNRELATED (2n + 1) PARENTS	TOTALS		FROM 2n PARENTS	FROM UNRELATED (2n + 1) PARENTS	TOTALS
1. GLOBE	41	107	148	8. BUCKLING	27	71	98
				Strawberry	1	1	2
2. POINSETTIA	28	47	75	Maple	..	2	2
Wiry	..	1	1	9. GLOSSY	8	11	19
3. COCKLEBUR	7	17	24	10. MICROCARPIC	64	100	164
Wedge	..	..	..	11. ELONGATE	..	2	2
4. ILEX	19	27	46	Undulate	..	1	1
5. ECHINUS	10	11	21	12. SPINACH(?)	6	4	10
Mutilated	2	4	6				
Nubbin(?)	1	..	1	Totals (2n + 1)	269	506	775
6. ROLLED	24	47	71	Related (2n + 1)			
Sugarloaf	3	9	12	types	....	22,123	22,123
Polycarpic	3	..	3	2n	32,523	70,281	102,804
7. REDUCED	25	44	69	Grand totals	32,792	92,910	125,027

The breeding experiments of Wedge—a Secondary of the Cocklebur group—furnishes the following evidence as to the relation of secondaries to primaries. "Both Poinsettia and its Secondary Wiry give trisomic ratios for the color factors P, p, but give disomic ratios for

spine factors A, a, indicating that both Poinsettia and Wiry have their extra chromosomes in the set carrying the factors P, p, but not in the set with the factors A, a. Similarly, the ratios for Cocklebur indicate that this Primary has its extra chromosome in the set carrying the factors A, a, but not in the set with factors P, p. Its Secondary Wedge, however, fails to give trisomic ratios for A, a. The ratios actually found resemble those in disomic rather than in trisomic inheritance and seem to indicate a deficiency in the extra chromosome of Wedge for the locus A, a, since the evidence strongly indicates that it is a Secondary of Cocklebur. If A′ indicates the modified chromosome and A and a go to opposite poles at reduction division in a Wedge plant with the formula AA′a, the gametes would be A+a+AA′+aA′. Such behavior would account for the ratios [in table 5]. If A′ is deficient for the factor A, the gamete aA′ would carry no factor for A ; hence the disomic ratios between armed and *inermis* Wedges found but not shown in the table. If A and a, occasionally should go to the same pole, the gametes would be A′ (which would probably die) and Aa, which would go to form a Primary Cocklebur occasionally thrown by Wedges.

"The hypothesis of a deficiency in the extra chromosome of Secondaries has been strengthened by Dr. Belling's cytological findings. His hypothesis of reversed crossing-over, however, completes the picture by indicating a doubling of a part of the chromosome along with a deficiency of the remaining portion."

Tetraploids of Datura with an additional chromosome have also been reported (Fig. 103). In one of these shown in the figure there are five like chromosomes in one group, and in the other there are six like chromosomes.

Belling and Blakeslee have studied the modes of union of the three chromosomes in the primary and in the sec-

ondary trisomic types of Datura, and have found certain differences that offer a suggestion as to the relation of these two types. In the upper row of Fig. 104 the differ-

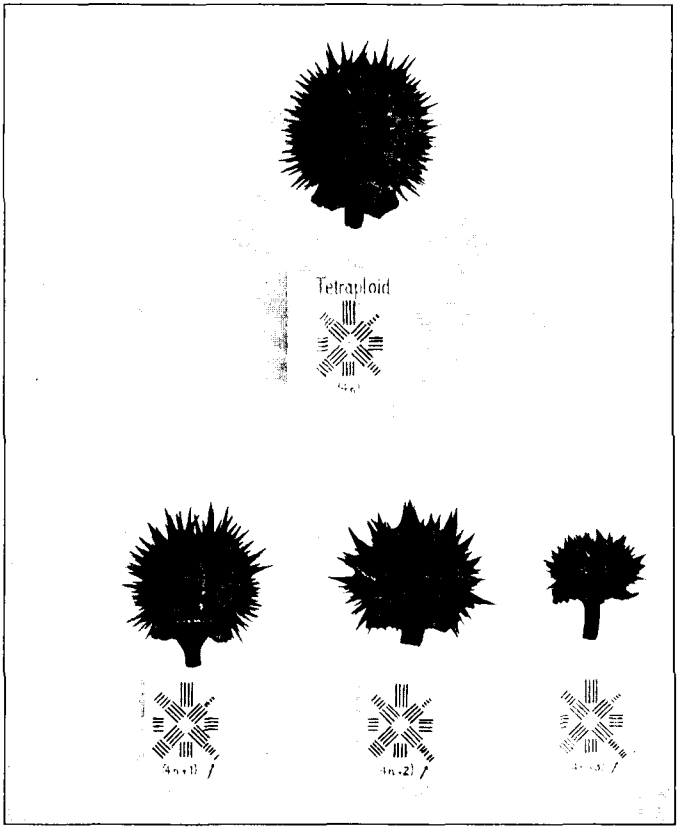

FIG. 103.

Tetraploid capsule above, and below 4n+1, 4n+2, and 4n+3 capsules. (After Blakeslee, in Journal of Heredity.)

ent ways in which the three chromosomes of the primary type are united are shown. The numbers below each show the frequency of the type. Of these types the triva-

lent V is the most common form of union (48); next in frequency is the ring-and-rod type (33); then the Y (17); the straight chain (9); the ring (1); the double ring (1); the ring of two with the third member left over (9+).

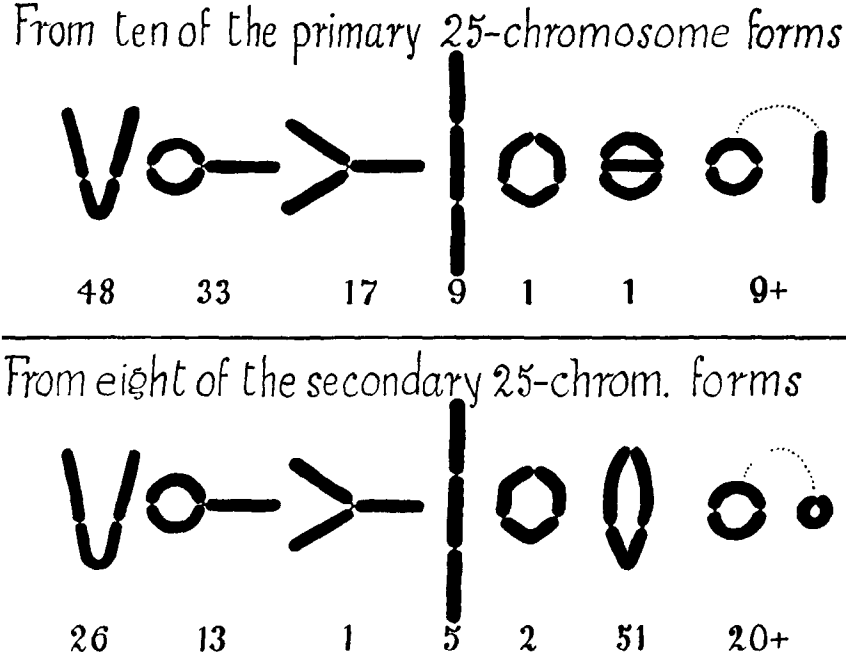

FIG. 104.

Methods of union of the three chromosomes of a *trisomic* type of Datura. (After Belling and Blakeslee.)

Since chromosomes are supposed to conjugate by like ends coming together it is reasonable to assume that, in these types, like ends (a and a, Z and Z) are still in contact (see Fig. 104, upper row).

In the lower row of Fig. 104 the different ways in which the three chromosomes of the secondary types are united are shown. In general the types are the same as those of

the primaries, but the frequencies are different. The most noticeable features are seen in the last two types (to the right). One of these is an elongated ring of three chromosomes, the other is a ring of two chromosomes and a small single-ring chromosome. These two types suggest

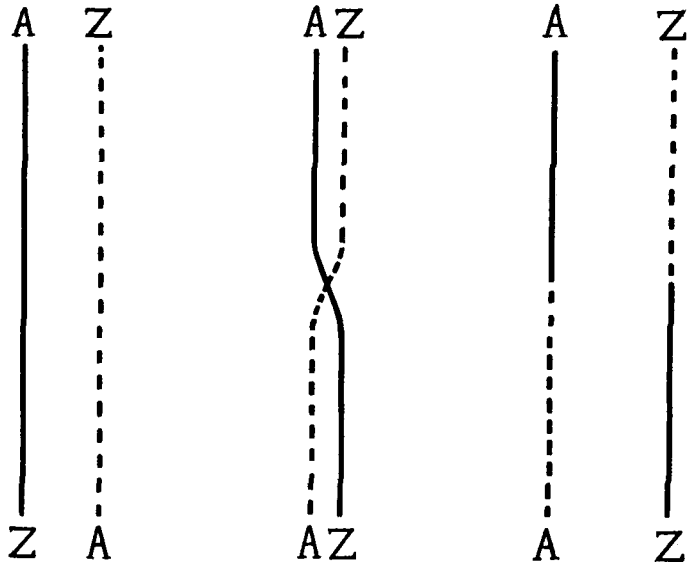

FIG. 105.

Diagram illustrating possible conjugation of two chromosomes, turned in opposite directions.

that, in some way, the end of one chromosome has been changed. Belling and Blakeslee offer the following provisional suggestions as to how such a change may have been brought about at a preceding stage in the triploid parent or in a trivalent of a primary type. Suppose, for example, two chromosomes should come to lie side by side in reversed position as shown in Fig. 105, and suppose they should cross over in the middle, which is the only level

at which like genes come together. The result will give two chromosomes each having its two ends alike, *i.e.*, one has A and A at its ends, the other Z and Z. If now such a chromosome becomes in the next generation a member

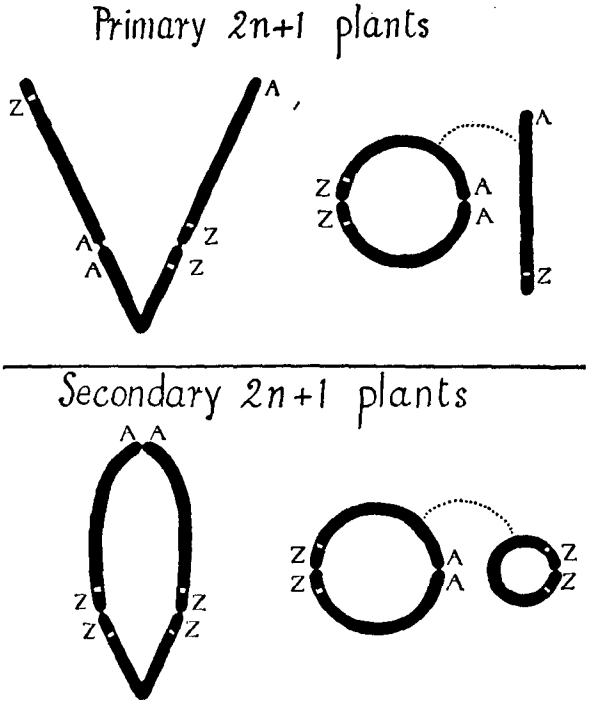

FIG. 106.

Diagram illustrating possible types of conjugation of three chromosomes of trisomic types. (After Belling and Blakeslee.)

of a trivalent group, it is possible to construct such modes of union as indicated in Fig. 106, where in a Z-Z chromosome, combined with two normal partners, like ends meet each other.

If these rings, peculiar to the secondaries, can be accounted for in the way suggested, it follows that one of

the trivalent chromosomes differs from the other two by a duplicated half. Hence the secondary has a different gene combination from the primary.

Kuwada reports 20 chromosomes (n=10) for corn (Zea mays), but certain sugar corns were found to have 21, 22, and even 23 or 24 chromosomes. Kuwada suggests that corn is a hybrid, one of whose parents was the Mexican teosinte (Euchlaena). One of the corn chromosomes that is longer than its mate was derived from teosinte, he thinks, and its mate from some unknown species. The longer one sometimes breaks into two pieces, which accounts for the additional chromosomes found in sugar corns. If this interpretation is verified (it has recently been questioned), these 21, 22, and 23 chromosome types are not strictly trisomic.

De Vries' conclusions relating to the extra chromosome types of Oenothera Lamarckiana had an important bearing on his interpretation of the origin of progressive mutation, hence on his interpretation of the relation of mutation to evolution. The numerous small changes in the characters of the individual frequently observed in trisomic types fulfill de Vries' early definition as to what constitutes an elementary species, causing at a stroke, as it were, the appearance of two elementary species.

It should be observed that when a mutational effect is produced by the addition of a whole chromosome the result involves, so far as the germ material is concerned, an enormous alteration in the actual number of the hereditary units. This change is scarcely compatible with the comparison to a change in a single chemical molecule. Only by treating the chromosomes as a unit could such a comparison have any weight. The constitution of the chromosomes, from the viewpoint of their genes, is hardly consistent with such a comparison.

The chief interest in these heteroploids, as I interpret

them, lies in the explanation they offer of a peculiar and interesting genetic situation arising from the occasional erratic behavior of the mechanism that is involved in the processes of cell division and maturation. Unstable forms are produced, that, in so far as they maintain themselves, do so by remaining unstable, *i.e.*, with an extra chromosome. In this respect they differ obviously from normal

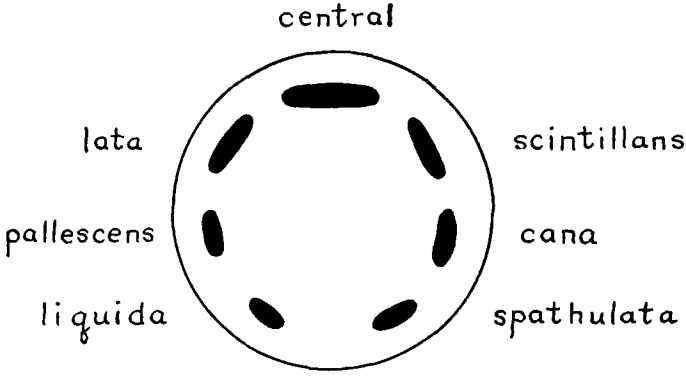

FIG. 107.

Diagram illustrating de Vries' idea of the relation between the seven chromosomes of O. Lamarckiana and types of trisomic mutants. (After de Vries and Boedijn.)

types and species. Furthermore, most of the evidence indicates that these heteroploids are not so viable as the balanced types from which they arise, hence would rarely be able to replace them or act as substitutes in a different environment.

Nevertheless, the occurrence of heteroploidy must be regarded as a significant genetic event whose explanation promises to clear up many situations that would be very puzzling without the information which a study of their chromosomes has revealed.

De Vries identifies six trisomic mutant types, and a seventh one, also, that differs genetically more strikingly from the other six than they do from each other. These seven trisomic types may, he suggests, correspond to the seven chromosomes of the evening primrose. A list of six of them is given below. A diagram of the corresponding chromosome groups is given in Fig. 107.

15-chromosome mutants.

1. Lata group.
    a. Semi-lata.
    b. Sesquiplex mutants: albida, flava, delata.
    c. Subovata, sublinearis.
2. Scintillans group.
    a. Sesquiplex mutants: oblonga, aurita, auricula, nitens, distans.
    b. Diluta, militaris, venusta.
3. Cana group: candicans.
4. Pallescens group: lactuca.
5. Liquida.
6. Spathulata.

This list of six 15-chromosome primary mutants includes some secondary mutant types arranged under their primaries. Their interrelations are shown not only by similarities in characters, but also by the frequency with which one throws the other. Two of them, albida and oblonga, have two kinds of eggs but only one kind of pollen, and are called one-and-one-half or sesquiplex mutants. Another secondary, candicans, is also a sesquiplex type. The central or largest "chromosome" of the group (Fig 107) carries the "factors" for velutina or for those of lacta. De Vries assigns to them also, from evidence found by Shull, the new mutants funifolia and pervirens. It may seem probable, therefore, following Shull, that

the factors for five other mutant types[1] of Lamarckiana belong in this group, as well as the lethal factors that put these factors in a balanced lethal condition. According to Shull the appearance of these recessive characters is due to crossing-over between the members of a pair of chromosomes here identified provisionally as the large central chromosome.

[1] Rubricalyx buds, and its allelomorph red stem (intensifier), nanella (dwarf), pink-coned buds, sulfur colored flowers.