CHAPTER 6 ANEUPLOIDY

Mutants with atypical behavior

Soon after the genetic analysis of Drosophila by Morgan and his group began, a different type of mutation was noted in Datura stramonium, the Jimson weed (1910). These mutants were characterized by phenotypic changes in many characters of the individual. The first one of this type had globose capsules and changes in several other characters. It appeared in Blakeslee's cultures being grown for class demonstrations at Storrs, Connecticut (cf. Blakeslee, Belling and Farnham 1920, Avery et al. 1959). When these "globe" plants were selfed or crossed as ♀ with normal plants, about 25% of the progeny had the globe-shaped capsules, as contrasted with the reciprocal cross of normal x globe ♂ in which few or none had globose capsules. The character appeared to be dominant, but did not breed true, was transmitted mainly through the ♀ and did not occur in the expected numbers. After Blakeslee moved in 1915 to the Station for Experimental Evolution at Cold Spring Harbor, more plants of Datura were grown. Other "mutants" with different morphologies, but with similar breeding behavior were found (Blakeslee and Avery 1919). There were differences in capsule size and shape as shown in Fig. 43. They also differed in leaf size and shape, growth habit, and vigor. In 1920, Belling, a young cytologist, was working in the laboratory and provided the clue to their behavior. He found that these off-type plants had one extra chromosome, i.e. 24 + 1 or 25, whereas the normal number was 24. At meiosis, the plant showed 11 bivalents plus a trivalent or had 12 bivalents plus a univalent. It was said to have a trisome and to be trisomic; also written as 2n+1, i.e. 11 bivalents plus a trivalent, often written as 11 II-III. The genes in one chromosome were present in triplicate, those in the other 11 in duplicate. The change in phenotype of these plants was assumed to be a result of the change

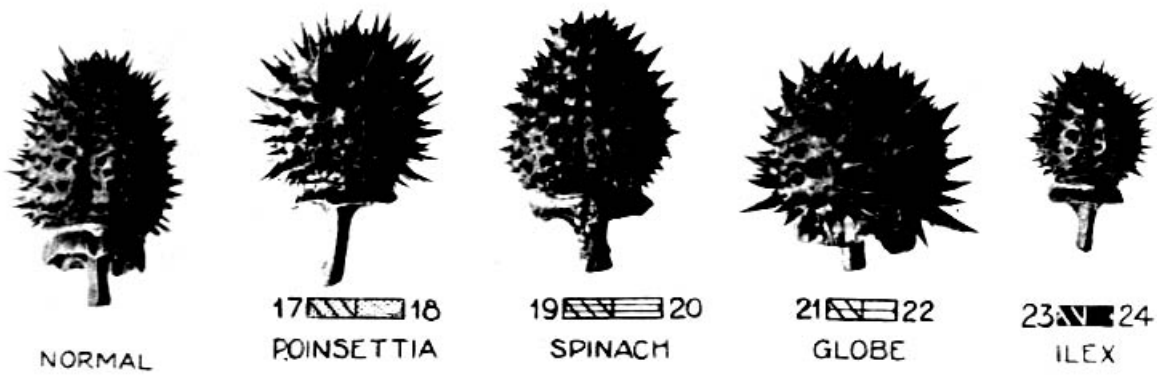


FIG. 43. Capsules of a normal 2n Jimson weed (Datura stramonium) and of four of the 12 primary trisomic, 2n + 1, types (Blakeslee 1934, from Fig. 6, p. 87, Jour. of Heredity 25 which also appears in Blakeslee: The Genus Datura by Amos G. Avery, Sophie Satina and Jacob Rietsema. Copyright 1959. The Ronald Press Company).

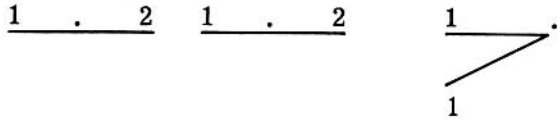
in chromosome number, and hence of the change in dosage (Blakeslee, Belling and Farnham 1920, Blakeslee 1921, 1931, 1934). The differences between the different mutants might be expected if in each type the extra chromosome were a different one. This led them to the conclusion that a change in the relative proportions of a group of genes had an effect on the phenotype, the particular changes being dependent on what genes were increased or decreased. Or, in other words gene balance is important. Blakeslee (1921) recognized the importance of this principle in evolution and focused his attention for many years on studies of the genetic contributions of whole chromosomes or large segments of them. Lines with changed chromosome numbers that were relatively true breeding for the phenotypic changes were established (Blakeslee et al. 1936). Here was evolution being duplicated in his cultures. Those who have heard his talks will remember him holding up a magnetic board on which he arranged models of the chromosomes of the new types three or four of a kind, or four or more in circles. Examples of these diagrams may be found in many of his publications, e.g. Blakeslee (1929, 1934). These also have photographs of the capsules and growth habits of the various trisomics.

This accounted for the new phenotypes, but what about the non-mendelian genetic behavior? The extra chromosome tended to be lost at the meiotic divisions; also the seeds that were $2\underline{n} + 1$ often did not germinate as well as those that were 2n. Both of these behaviors lowered the frequency of $2n + 1$ from the 50% expected if the behavior had been normal. Crosses made on normal plants gave few or no trisomics because of the inability of the $\underline{n} + 1$ pollen to compete with the $\underline{n}$ pollen. For further details, see page 148. Thus the presence of extra chromosomes also explained the breeding behavior.

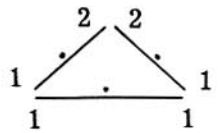
As the studies progressed size differences in the chromosomes were noted by Belling who designated part of them as long, medium and short, L, M, and S; and the remainder that were only a little shorter than these as l, m, and s, (Belling and Blakeslee 1922), as shown in the first column of Table 51. It became advantageous also to number the two ends of each chromosome. The longest chromosome was numbered 1. 2, the next one 3. 4 etc., diagrammed as $\underline{1 \ . \ 2}$, $\underline{3 \ . \ 4}$, where the . designated the centromere. The primary trisomic, globe, had three normal $\underline{21 \ . \ 22}$ chromosomes, and at meiosis they formed a chain of three or a pair plus a rod-shaped univalent. Here the entire chromosome was present in three doses.

As more and more trisomics were established in Datura the first expectation was that there would be only 12, since 12 is the haploid number. When the 13th appeared and still more were added, explanations had to be found. Careful study revealed two additional types. In one of these certain of the phenotypic characteristics of the primary trisomic were accentuated, in the other certain characteristics of two different primary trisomics were combined. The former were called secondary, the latter tertiary trisomics. The basis for the different types became obvious when Belling and Blakeslee (1924) reported the results of studies at meiosis. The aberrant plants had an extra whole chromosome, but in contrast to the primary trisomics in which the univalent was always rod-shaped, in the secondaries it often was ring-shaped or there was a circle or ring of 3 chromosomes (written as ⊙3). In the tertiary trisomics, the extra chromosome often was included in a chain of five chromosomes. It was the connecting link between two pairs of chromosomes. When present as a univalent, it was always rod-shaped.

To explain these configurations, they assumed that the extra chromosome in the secondary trisomic was a double half chromosome, i. e. that one arm was duplicated. To this type of chromosome Darlington (1939) applied the term isochromosome. The chromosomes associated at meiosis in the secondary trisomic might be illustrated as:



and the diagnostic configurations at meiosis should be a ring of 3 chromosomes (⊙3), illustrated as

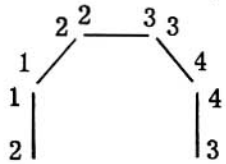


or a u-shaped ring formed by the univalent 

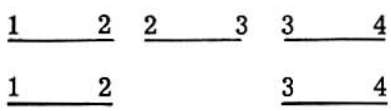
Such a plant has four doses of end number 1. Two kinds of secondaries for each primary are possible if an entire arm is duplicated.

They assumed that the extra chromosome in the tertiary trisomic was an interchange chromosome, i.e. a chromosome in which a segment of one chromosome had been replaced by a segment from a non-homologue. For example if in chromosome 1 . 2, the 1 end had been replaced by 3 from chromosome 3 . 4, the new chromosome would be 3 . 2. The tertiary trisomic would have 12 normal pairs plus 3 . 2 as the extra chromosome. The diagnostic configurations would be:

a chain of five;



or two pairs linked by a single chromosome.

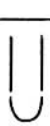
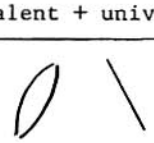
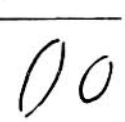


It has three doses of end number 2 and of end number 3.

It was a tertiary trisomic in which the extra chromosome was smaller than the others in the chain that led Belling and Blakeslee (1926) to conclude that non-homologous chromosomes could exchange end segments, the "segmental interchange hypothesis", and that this could explain semisterility and its mode of inheritance (Belling 1925); as discussed in Chapter 4.

Since the extra chromosome is probably the result of an exchange of end segments, following breaks that might occur at any position, the potential number of different tertiaries is almost infinite.

Table 50. Frequencies of the various configurations observed at late diakinesis in primary and secondary trisomics in Datura. (Belling and Blakeslee, 1924, p. 117, Proc. Nat. Acad. Sci. 10).

	Trivalent						bivalent + univalent	
								
primary trisomics	48	33	17	9	1	1	9+	0
secondary trisomics	26	13	1	5	53	0	0	20 +

Another type, that in which the extra chromosome was a fragment, was found also by Belling and Blakeslee; but not studied to any great extent. It is probable that in certain of them the fragment was one chromosome arm including the centromere, i.e. a telocentric chromosome; the plant being 2n + telocentric, which might be referred to as a telosomic trisomic.

The various kinds of configurations and their frequencies observed by Belling at late diakinesis in primary and secondary trisomics are summarized in Table 50.

Although counts of the univalent frequency were not complete for the secondary trisomics, the presence of the ⊙3 or the doughnut-shaped univalent and their absence in the primaries, agree with the assumptions as to the nature of the extra chromosome. All except the one ring of 3 in the primary trisomic plants are expected if only homologous ends are associated.

No counts were reported for the tertiary trisomics, but a chain of five, 11 II + III or 12 II + rod-shaped univalent were observed.

At this point we will consider the kinds of changes in number that may occur, and the terminology which is applied to the various types (Sharp 1934, Darlington 1937).

Terminology

Whenever populations of plants of a given species are observed in detail, occasional individuals may be found with a chromosome number that differs from the somatic number characteristic of the species. These individuals are said to be heteroploids. They may be classified into two types: (1) euploids, in which there is an exact multiple of a basic, monoploid number and (2) aneuploids, in which the number is not an exact multiple. These terms are also applied to cells, tissues, individuals, races or species whose nuclei have these numbers (Sharp 1934).

The somatic chromosome number is referred to as 2n, the gametic number as n; whether it be a diploid or a polyploid.

Individuals with 2n+1, 2n+2, 2n+1+1, 2n-1 etc. chromosomes are aneuploids, and are referred to as being trisomic, tetrasomic, double trisomic, and monosomic, respectively. One with a pair missing is nullisomic. For example in a species with 24 chromosome pairs, e.g. Nicotiana tabacum, a trisomic or 2n+1 has 49 chromosomes, it has 23 II + 1 III; a tetrasomic has 23 II + 1 IV, a double trisomic has 22 II + 2 III, and a monosomic has 23 II + I. A nullisomic has 23 II.

A chromosome that has one arm duplicated is an isochromosome. A fragment chromosome that has one entire arm with a terminal centromere is a telocentric chromosome. In Lycopersicon esculentum - (tomato) and in Nicotiana tabacum - (tobacco), the chromosomes are designated as A, B, C etc.; a plant that is trisomic for the A chromosome is triplo-A, etc. (Lesley 1928). In Drosophila melanogaster, aneuploids for chromosome 4 have been described by Bridges (1925), namely 2n+1 (triplo-4) and 2n-1 (haplo-4). Aneuploids that occur in diploid species are discussed in this chapter; those that occur in polyploids are discussed in Chapter 8.

An individual with a 2n or diploid count may be 2n-1+1, a quasi-diploid or individual cells or sectors may have this constituion. This condition was recognized in somatic cells of the Chinese hamster (work of Yerganian, cited by Hsu 1959), owing to favorable morphological differences in the chromosomes.

Phenotypes of trisomics in *Datura*

As mentioned earlier, 12 different cultures of Datura were established, each segregating for one of the 12 different chromosomes. These, referred to as the primary trisomics, and the secondary trisomics are listed with their laboratory names in Table 51.

Table 51. A list of the primary and secondary trisomics in Datura stramonium, their relative sizes, laboratory names and number designations of the ends.

Relative size*	Chrom. No.	Primary trisomics Desig. for 2 arms	Name	Corresponding secondary trisomics Desig.	Name	Desig.	Name
L	1	1.2	Rolled	1.1	Polycarpic	2.2	Sugar Loaf
l	2	3.4	Glossy	3.3	Smooth		
l	3	5.6	Buckling	5.5	Strawberry	6.6	Areolate
M	4	7.8	Elongate	7.7	Undulate		
M	5	9.10	Echinus	9.9	Mutilated	10.10	Thistle
M	6	11.12	Cocklebur	11.11	Wedge		
M	7	13.14	Microcarpic	13.13	Marbled	14.14	Mealy
M	8	15.16	Reduced	15.15	Scalloped		
m	9	17.18	Poinsettia	17.17	Dwarf		
m	10	19.20	Spinach	19.19	Divergent		
S	11	21.22	Globe				
s	12	23.24	Ilex				

* L, M, S = long, medium and short, with a shorter class for each, designated l, m, s, respectively.

As shown in Table 51, only 14 of the 24 possible secondary trisomics were established. Only four of the primaries are represented by both secondaries.

The primary trisomics in Datura differ from normal and from each other in several characters. These include differences in capsule size and shape; size and length of spines; size of plant; growth habit; and leaf, flower and stigma size, shape and form. These differences result from the change in genic balance brought about by the added chromosomal material. The following descriptions of a few primary trisomics and secondaries are based on the photographs and descriptions published by Blakeslee and Belling (1924), and Blakeslee (1927, 1931, 1934), and Avery et al. (1959).

1. 2 Rolled - narrow, in-rolled leaves, capsule smaller than normal, short spines.
1. 1 Polycarpic - very small, weak plants; narrow, lanceolate leaves; capsule very much smaller than in rolled, perfect oval in shape. A comparison of the primary and its two secondaries shows that extra doses of the 1. arm make the plant more erect; extra doses of the 2. arm make it more spreading.
2. 2 Sugar loaf - plant shorter, leaves larger, capsule larger than in rolled, but conical in shape, short spines.
5. 6 Buckling - large leaves with irregular surfaces. The . 5 half contains factors for erect habit, dark stem, small short capsules and narrow leaves. The . 6 half has factors for spreading habit, light stem, long capsules and broad leaves with irregular pale areas.

17. 18 Poinsettia - plant less branched; leaves long, drooping.
17. 17 Dwarf - short plant, narrower leaves which droop.
21. 22 Globe - globe-shaped capsule.

The primaries are usually intermediate between the secondaries. Using a knowledge of the characters of the primary and one of the secondaries, they were able to predict the characters of the unknown secondary from the peculiarities of the primary not shared by the known secondary (Blakeslee 1924, 1927).

The effect on capsule size and shape of adding extra 21. 22 chromosomes to the 2n and 4n, i. e. 2n+1, 2n+2; 4n+1, 4n+2 and 4n+3 (Blakeslee 1934) is shown in Fig. 44.

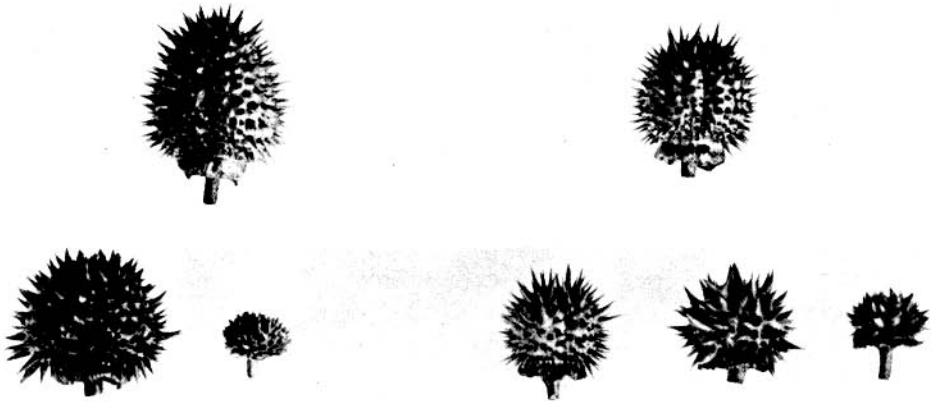


FIG. 44. Capsules of five "Globe" types showing the effect of adding one or more of the same chromosome to the 2n and to the 4n types. From left to right:
Upper row: diploid(2n) and tetraploid (4n) capsules
Lower row: 2n + 1, 2n + 2; 4n + 1, 4n +2, 4n + 3.
(Blakeslee, 1934, from Fig. 8, p. 89, Jour. of Heredity 25).

The 2n+2 and 4n+3 had very small capsules. Sinnott et al. (1934) have studied the various types histologically.

An example of a tertiary trisomic is one Blakeslee called Pinched. It was similar to Sugarloaf, a secondary (2. 2) for chromosome 1 (1. 2) and to Strawberry, a secondary (5. 5) for chromosome 3 (5. 6); but less extreme in appearance for leaf shape, capsule form, growth habit and other characters. Pinched is 2n+2. 5.

Another example is Hedge (probably 2n + 1. 9) which has similarities to Polycarpic, the other secondary (1. 1) for chromosome 1 and Mutilated, a secondary (9. 9) for chromosome 5. Since the extra chromosome is an interchange chromosome, the pieces exchanged might be less than entire arms. Hence the extra chromosome probably has less than one arm of one chromosome, more than one arm for the other.

As mentioned earlier, changes in phenotype that accompany the change in chromosome number must be the result of a change in genic balance brought about by the addition of the extra chromosome. The kind of change would depend on the genes present in the chromosome. This change in phenotype dependent on a change in gene dosage is a very important phenomenon in genetics and evolution. Blakeslee recognized its importance and emphasized it in his work on Datura.