interchanges if compared with O. hookeri, a race with 7 pairs selected as a standard normal race (see Chapter 5). Many different trisomics are possible in the progeny of multiple interchange heterozygotes such as Oenothera.

Trisomics in cotton, wheat and tobacco which are polyploids will be discussed in Chapter 8.

Drosophila melanogaster

Triplo-IV and haplo-IV individuals have been reported in Drosophila (Bridges, 1921). Non-random segregation was reported in triplo-IV females (Sturtevant 1936). In the stock having the X chromosomes attached at the centromere end (attached-X, actually an isochromosome), the females are XXY (Morgan, 1938a). They may be considered as a modified trisomic. There are also stocks in which the females are XXY but the X chromosomes are not attached. Only rarely do triplo-X flies survive (Bridges and Brehme, 1944).

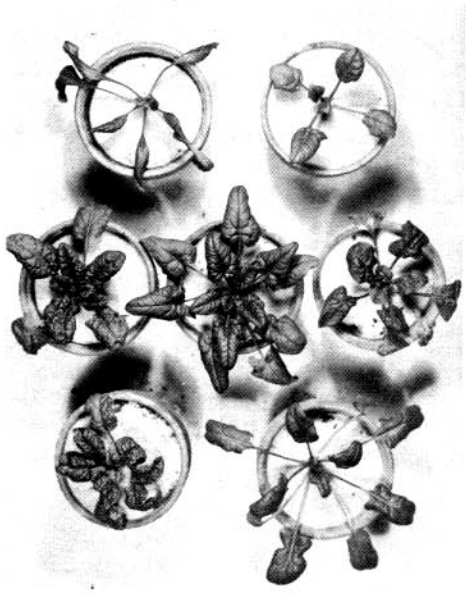


FIG. 50. The six primary trisomics of spinach, Spinacia oleracea. The normal 2n (Long Standing Bloomsdale variety) is in the center.
Top row: curled, reflex (sexchromosome)
Middle row: wild, normal diploid, oxtongue
Lower row: savoy, star.
(modified from Ellis and Janick, 1960, Fig. 3, p. 213, American Journal of Botany 47).

Man

Trisomics have been reported in man. Mongoloid individuals are trisomic for a particular autosome (Lejeune et al. 1959, and Jacobs et al. 1959). An individual that was mongoloid and also had the Klinefelter's syndrome had the extra autosome and was also XXY (Ford et al. 1959). Other subjects with genetic defects have been shown to be trisomic for a D_1 chromosome (Patau et al. 1961). A ♀ with gonadal dysgenesis had only one X chromosome (Tjio et al. 1959).

Genetic ratios

Genetic ratios in trisomic individuals are a means of locating genes and of establishing the independence of the linkage groups.

Primary trisomics

Since there is one trivalent (a group of three homologues) in a primary trisomic, the genetic ratios for factors that are located on these chromosomes will be very different from the ratios for genes on any of the other bivalent chromosomes. In a 2n + telocentric, the ratios for genes located in the arm that is trisomic will be modified. The ratios are modified still further by the low transmission of n + 1 through the pollen. The primary trisomic stocks, therefore, have been used in several species to determine the linkage group to which a genetic marker belongs, to identify the specific chromosome that carries it, and the 2n + telocentric or isochromosome has been used to determine the arm in which it is located. A trisomic may have the following genotypes for a particular locus, Aa as follows: AAA, AAa, Aaa, and aaa, referred to as triplex, duplex,

simplex and nulliplex. An example is furnished by the data for trisomic 10 in corn tested for segregation for markers in different linkage groups (McClintock and Hill 1931). The data from tests for the R factor for aleurone color in selfs and reciprocal backcrosses are summarized in Table 66. The trisomic plants could not be separated phenotypically from their diploid sibs.

Table 66. Data from various tests of plants trisomic for chromosome 10 and heterozygous for R vs. r (McClintock and Hill 1931, p. 178-183, Genetics 16).

Cross or Self	Colored aleurone	Colorless	Total	Observed % colorless	Observed ratio
RRr					
RRr selfed	396	41	437	9.4	10:1
RRr x rr	819	213	1032	20.6	4:1
rr x RRr	941	486	1427	34.1	2:1
Rrr					
Rrr x rr	679	836	1515	55.2	1:1
rr x Rrr	1392	2685	4077	65.9	1:2
Checks (2n)					
Rr selfed	608	204	812	25.1	3:1
Rr x rr	1161	1196	2357	50.7	1:1
rr x Rr	132	135	267	50.6	1:1

As shown in Table 66, for duplex (RRr) plants selfed the ratio of colored:colorless was about 10:1; and for the backcrosses the ratio was 4:1 through the ♀, 2:1 through the ♂. The deviations from the observed ratios in disomic plants, 3:1 in F_2 and 1:1 in the backcrosses, are all highly significant.

The data from tests of simplex (Rrr) plants, also in Table 66, show about a 1:1 ratio from Rrr x rr and 2:1 from rr x Rrr. For the simplex trisomic genotype only the backcross using the trisomic as the pollen parent and also selfs give ratios that are markedly different from those for disomic plants.

Plants trisomic for chromosome 10 were crossed also with markers in seven of the other linkage groups. The data from selfing or backcrossing of trisomic F_1 plants from those crosses are summarized in Table 67.

Table 67. Data from tests of plants trisomic for chromosome 10 in maize and heterozygous for genetic markers in other linkage groups (from McClintock and Hill, 1931, Table 12, p. 157-158, Genetics 16).

Linkage group	Character	Kind of test	Dominant	Recessive	Total	Percent recessive
2	B b	F_2	54	19	73	26.0
3	A_1 a_1	bkc.	144	173	317	54.6
4	Su su	F_2	1758	586	2344	25.0
5	Pr pr	F_2	1030	356	1386	25.7
6	Y y	bkc.	323	298	621	48.0
7	Gl_1 gl_1	bkc.	113	105	218	48.2
9	C c	bkc.	172	184	356	51.7

The data in Table 67 are all disomic segregations, about 25% recessives for F_2 or about 50% for backcrosses; very different from the trisomic ratios. Hence plants trisomic for chromosome 10 do not carry factors in the seven linkage groups represented in the tests summarized in Table 67. Since the same trisomic gave trisomic ratios for R, the R linkage group was shown to be independent of the seven other linkage groups tested. Until these tests were made, independence of the linkage groups in maize was based only on linkage data. Independence of the other linkage groups in maize and their identification with particular chromosomes was accomplished in large part by McClintock by the use of trisomics (Cf. Rhoades and McClintock 1935). A few were identified in connection with the studies of interchanges (Burnham 1934).

Data on genetic segregations in species in which the trisomics could be classified phenotypically were reported by Blakeslee and Farnham (1923) in Datura stramonium, by Rick and Barton (1954) in tomato, Lycopersicum esculentum, and by Tsuchiya (1956) in barley, Hordeum vulgare. The data from the barley trisomics will be summarized and discussed here as an example. Each of the seven trisomics was crossed with stocks carrying gene markers for the seven linkage groups as known at that time. The data showing trisomic segregations are summarized in Table 68.

Table 68. Summary of F_2 data from those crosses in barley showing trisomic ratios; all from duplex trisomic plants; VVv, BBb, etc.*

Name of trisomic	Chromosome	Gene pair	Observed numbers in 2n		2n+1		Total			%	%
			Dom.	Rec.	Dom.	Rec.	Dom.	Rec.	Total	Rec.	2n+1
Bush	1	Nn	281	24	89	0	370	24	394	6.1	22.6
"	1	Br br	280	25	90	0	370	25	395	6.3	22.8
"	1	Fc fc	258	16	138	0	396	16	412	3.9	33.5
Slender	2	Vv	235	29	61	0	296	29	325	8.9	18.8
Pale	3	Uz uz	357	17	136	0	493	17	510	3.3	26.7
Robust	4	Kk	53	11	24	0	77	11	88	12.5	27.3
"	4	Bl bl	201	25	128	0	329	25	354	7.1	36.2
Pseudo-normal	5	Bb	76	3	39	0	115	3	118	2.5	33.1
Purple	6	**									
Semi-erect	7	Ss	100	9	45	2	145	11	156	7.1	30.1
Total (avg. %)			1841	159	750	2	2591	161		6.4	27.9

* From Tsuchiya, 1959. Table 3, p. 21, Jap. Journ. Bot. 17. The other data were from Tsuchiya et al., 1960, Tables 2, 3, 4, pp. 155, 156 and 157, Jap. Jour. Genetics 35.

** Only disomic ratios were observed with markers for the linkage groups then known.

The ratios for the totals in Table 68 as well as those within the 2n and 2n + 1 classes deviate widely from disomic ratios, and indicate that the particular marker is located on the chromosome that was trisomic in that stock. Also note that the trisomic having the "bush" phenotype (trisomic for chromosome 1) produced trisomic segregations for characters previously thought to be in separate linkage groups; naked (n) in what had been linkage group 3 and brachytic (br) and chlorina (fc) in 7. This confirmed a conclusion arrived at earlier from linkage tests with interchanges (Kramer, et al. 1954). For

each trisomic, the segregations for the markers in the other linkage groups were disomic. For trisomic 6, all the markers tested showed disomic segregations, since no gene marker for that chromosome was known at that time. The two recessive trisomic plants from chromosome 7 are the result of double reduction which is discussed on pages 185-192.

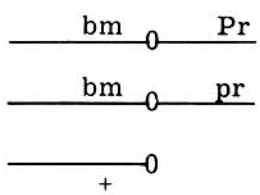
Data were reported by Rick and Barton (1954) for segregation of marker genes in the 12 trisomics in tomatoes. In two instances, two linkage groups previously supposed to be independent were found to be carried by one chromosome. One new linkage group was found.

The presence of lethals linked with the genetic marker in the trisome modifies the ratios, e.g. in Matthiola (Frost 1931). Modified ratios might be expected also in many of the Oenothera trisomics derived from interchange heterozygotes which carry lethals. In a tertiary trisomic if the two normal chromosomes carry a recessive marker that is lethal and closely linked with the breakpoint, the surviving offspring will be the tertiary trisomics (Ramage 1955).

Data illustrating the use of aneuploids to determine the arm in which genetic markers are located will be considered next.

Telosomic trisomic

The segregation for several markers in chromosome 5 in maize was studied in a stock 2n + telocentric for the short arm of 5 (Rhoades 1933b, 1936, 1940). The 2n + telocentric plants had a distinctive phenotype, very short, broad leaves. The genetic constitution of the F_1 and the backcross parents in one cross (Rhoades 1936) were:



x bm bm pr pr

The results from one backcross, segregating for an independent colorless aleurone factor, were:

	2n	2n + telo	Totals	Ratio
Pr vs. pr	63:64	31:35	94:99	1:1
Bm vs. bm	1:171	85:0	86:171	1:2

Note the ratio of 1 Pr:1 pr in the 2n and 2n + telo classes of offspring, whereas all but one of the 2n plants was bm, and all the 2n + telo plants were Bm. These results indicate that the bm locus is in the arm represented by the telocentric, i.e. the short arm, very close to the centromere; and pr in the long arm. The one 2n plant with Bm phenotype must have arisen by crossing over between the centromere and the bm locus. Based on the total offspring, the ratio of Bm:bm was 1:2. This suggests a trisomic ratio, but in a primary trisomic, this type of backcross of a simplex genotype is expected to yield a 1:1 ratio. The clue to a probable explanation is the absence of any bm trisomic offspring which would be expected if the telocentric and a normal chromosome paired and disjoined, leaving the other normal chromosome free to pass to either pole. If the two normal chromosomes always paired and the telocentric lagged in one-third of the meiocytes the observed ratio of 1 Bm:2 bm is expected. A similar low frequency of primary trisomics was found in Datura in the progeny of a 2n + telocentric (see Table 57).

Table 69. Location of 72 genes in Datura stramonium (Blakeslee, et al. Report Carnegie Inst. of Wash., Yearbook No. 39:201-211. 1940. Also Abstr., Genetics 26:138-139. 1941).

Chromosome	No. of genes	Centromere		
		in odd-numbered half	in even-numbered half	not located to arm
1.2	5	pale-7, pale-16 spotted leaves, rough-4	-----	albino-11
3.4	8	-----	bronze, ferox white, tufted	Q spot, glaucous-1, albino-10, albino-12, non-flowering
5.6	6	-----	-----	sickly-3; pale-19; mottled; pa-2; pa-12*; slow pollen tube-2
7.8	3	-----	-----	pale-25; fused; puckered
9.10	4	-----	-----	red seed; dyad; male-sterile 1; pa-24
11.12	9	pa-22	inermis; early; short flowers; lobed-pollen; chromosome doubling; sickly-1	triforked; wilt-3
13.14	7	albino-2	-----	pa-8; pa-11; peach, wilt-4; short pollen tube-2; pregermination pollen
15.16	8	pale-8; glaucous-3	compact; pale-5; tricarpel; white-2	rough-3; short burst pollen tube-3
17.18	6	curled-1; wilt-1	pa-5; short burst pollen-tube-2; white-1	pale-6
19.20	5	pa-1; pa-16; short	curled-2	short pollen tube-1
21.22	6	-----	-----	pale-1; pale-3; slender capsule; asynapsis; bunchy-2, pa-17
23.24	5	-----	-----	swollen; broad leaves-1; zigzag; ragged leaves; pa-23

* pa = pollen abortion.

In Datura, many of the genes were located as to arm as shown in the list of genes, Table 69. The interchanges were apparently regarded as half-chromosome exchanges. Location to arm was accomplished by using secondary and tertiary trisomics. Location to chromosome was determined by the use of trisomics, linkage with other genes of known position, linkage with pollen abortion genes (using the heterozygote as the ♂ crossed on the recessive), linkage with certain interchanges that showed sterility when heterozygous, and linkage with compensating trisomics.

The effect of crossing over between a gene locus and the centromere on the expected genotypes from trisomics and tetrasomics, and on the expected ratios will be considered in the next chapter.

Special uses of aneuploids

1. To study the effects of duplication for blocks of genes, e.g. whole chromosomes, one arm etc. The possible phenotypic effects are illustrated by the work of Blakeslee and his group on Datura stramonium. The physiological effects or potentialities are probably of even greater importance, and should be explored.

2. To determine the linkage group carried by each chromosome. This may be done by establishing the different primary $2n + 1$ types and determining for each the one group of factors for which trisomic ratios result. This also serves to establish the independence of the linkage groups. If the chromosome can be distinguished, it is possible to state which linkage group is carried by each morphologically distinct chromosome. Trisomics have been used in this manner for the chromosomes in maize (McClintock 1929 and unpub.); barley (Ramage 1955, 1960; Tsuchiya 1958, 1959, Tsuchiya, et al. 1960); and tomato (Rick and Barton 1954). In barley, the final identification was made possible by utilizing information from the studies of interchanges (Hagberg and Tjio 1950, Tjio and Hagberg 1951, Burnham and Hagberg 1956).

3. The primary trisomics may be used to determine the linkage group to which a new gene belongs. This is useful in cases where long regions of a chromosome are as yet unmarked with any known gene.

4. The secondary trisomics, and the $2n$ + telocentric types may be used to determine which genetic factors are carried in each arm of a particular chromosome. For example, in maize plants that are $2n$ + telocentric for the short arm of chromosome 5 segregation for bm is trisomic but for bt is disomic, thus placing them in opposite arms, although the cross-over frequency between them is less than 1% (Rhoades 1933a).

5. Tertiary trisomics (an interchange chromosome is the extra one) may be used to localize still further the genetic factors.

6. The tetrasomic and also the trisomic types may be used to determine relative distances of genes from the centromere as measured by the frequency of double reduction. The most reliable results are obtained from data in which the trisomic plants in the segregating progeny of trisomics can be identified. If they are not recognizable by their phenotype, the progeny of the first backcross may be selfed or backcrossed again and their progeny grown to determine which plants were trisomic and the frequencies of the various kinds of gametes contributed to the first backcross progeny by the F_1. These frequencies may be used in maximum likelihood equations to calculate α, the frequency of double reduction (see Chapter 7).

7. Any of these types might be used to study the effect of extra chromosomes on crossing over. For example, the $2\underline{n} + 1$ type would be much more usable than a triploid which is highly sterile. The tetrasomic could be compared with the autotetraploid.

8. The primary trisomics are of use in identifying the chromosomes involved in chromosomal interchanges, i.e. observations at diakinesis or metaphase in crosses of $2\underline{n} + 1$ x interchange will determine if the extra chromosome is homologous with one in the ring. Trisomics might be useful in identifying the chromosomes involved in other abnormalities, e.g., duplications, deficiencies, or inversions.

9. The $2\underline{n}$ + telocentric and $2\underline{n}$ + isochromosome stocks are useful in determining which arm of a given chromosome is involved in the abnormality, again using diakinesis or metaphase configurations. Conceivably they may be useful for this purpose for species in which pachytene analysis is not feasible, as for example in barley.

10. Polysomics offer the possibility of recovering certain crossover products or segregation products which otherwise are deficient.

11. A $2\underline{n}$+ isochromosome stock might be used to produce a ring chromosome by introducing an inversion, as was done in attached-X Drosophila.

12. All of these results further strengthen our ideas on the relation between chromosomes and heredity; and add additional evidence regarding chromosome homology and pairing.

13. They furnish evidence that character expression is a matter of balance between a large number of genes acting in different ways. Hence added chromosomal material may be of great importance in evolution. It may be important also in future breeding operations.

Monosomics in diploids (also see P. 226). In heterozygotes for the r-X1, X-ray induced, female transmissible deficiency for the r locus (Stadler, 1933), female gametes carrying the deficiency are also often n-1 or n+1 for different chromosomes (Satyanarayana, unpublished, University of Wisconsin). Progeny* from crosses with normal include about 11% monosomics, 11% triploids, and 78% diploids with an occasional double or triple monosomic (Plewa and Weber, 1973). The event is postmeiotic. It probably results from non-disjunction, and leads to non-correspondence of embryo and endosperm.

*r r-x_1 embryos from R r-x_1 x r r.

Monosomics for each of the 10 chromosomes have been isolated. Most, but not all of them can be used either as pollen or as female parents, or both, but do not transmit the monosomic condition (Weber, 1974). Examples of their use are: Plewa and Weber (1973) a study of genes for lipid content, Phillips et al. (1974) for setting up one of the stocks in a series for comparison of ribosomal gene number for plants with multiples of the nucleolus organizer region. For use in gene mapping, as described by Weber (1974), the r-X1 deficiency is introduced into Mangelsdorf's multiple chromosome tester, one recessive marker in each chromosome (numbers 1 to 10, respectively): bm_2; lg_1; a_1; su_1; pr; y_1; gl_1; j_1; wx; g_1. In crosses with a mutant, e.g. m (if on 7), the female parent would be glM/glM, the male parent would be Glm/Glm. The monosomic for that chromosome from the cross would be Glm/ ---, the glM chromosome having been lost in the female. When crossed on the multiple tester, all the progeny would be Gl; whereas all the other genes would be segregating 1:1.

In the tomato, monosomics for chromsome 5 (Ecochard and Merkx, 1972), chromosome 11 (Rick and Khush, 1961), and chromosome 12 (Khush and Rick, 1966) are viable.

Also in the tomato, segmental monosomics of two types have occurred: 1. translocated or tertiary monosomics which have a translocated chromosome and only one each of the two in the normal set whose arms are represented in the interchange (Khush and Rick, 1966; Ramanna, 1969). 2. haplo-triplo-disomics which have a normal chromosome replaced by one of its 2 isochromosomes. Neither of the above can be reproduced sexually (Khush and Rick, 1967). The same is true of the primary monosomics.