

## Trisomics

An organism containing a normal chromosome complement and one extra chromosome is known as a trisomic.

The genetics of trisomic individuals were first described by Belling, Blakeslee and Farnham in 1920 while studying *Datura* (Jimson weed). Once the change in chromosome number was described, Belling recognized the importance of gene dosage and gene balance in determination of phenotype and viability.

# Trisomics

## Types

### 1. Primary trisomics ( $2n=2x+1$ )

An individual with a normal chromosome complement plus an extra chromosome.

### 2. Secondary trisomic ( $2n=2x+iso$ (i.e. $2n=2x+1L.1L$ ))

An individual with a normal chromosome complement plus an extra isochromosome (both arms of the isochromosome are homologous).

### 3. Tertiary trisomic ( $2n=2x+\#.\#$ (i.e. $2n=2x+1L.3L$ ))

An individual with a normal chromosome complement plus an interchange chromosome involving two nonhomologous chromosomes in the interchange.

### 4. Telotrisomic ( $2n=2x+telo$ (i.e. $2n=2x+1S$ ))

An individual with a normal chromosome complement plus an extra telosomic chromosome. The telocentric chromosome contains a terminal kinetochore. A plant with an extra telocentric chromosome is designated a monotelotrisomic. A plant with two extra telocentric chromosomes that are derived from opposite arms of the same chromosome is designated a ditelotrisomic.

# Trisomics

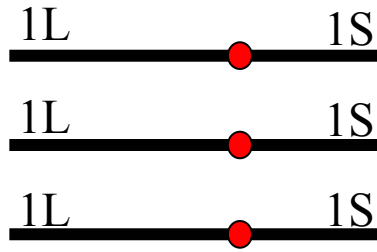
## Types

### 5. Acrotrisomic ( $2n=2x+acro$ (i.e. $2n=2x+acro\ 1S$ ))

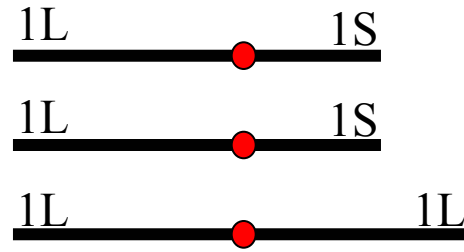
An individual with a normal chromosome complement plus an acrocentric chromosome. An acrocentric chromosome has a portion of one chromosome arm deleted. Acrotrisomic stocks have been useful in locating genes to physical chromosome segments in barley.

### 6. Compensating trisomic

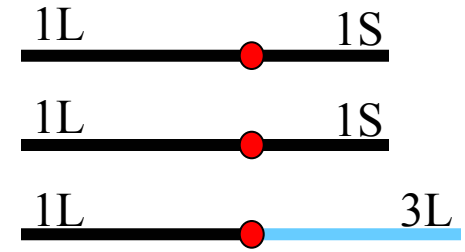
An individual with a missing chromosome that is replaced by two tertiary chromosomes or by a secondary and a tertiary chromosome where the missing chromosome 1 is compensated by tertiary chromosomes 1L.3L and 1S.9L in the following illustration. The chromosome formula would be written as  $2n-1+1L.3L+1S.9L$ .



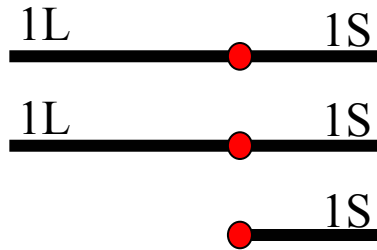
Primary trisomic



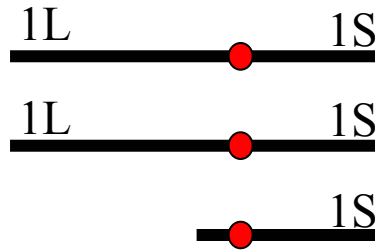
Secondary trisomic



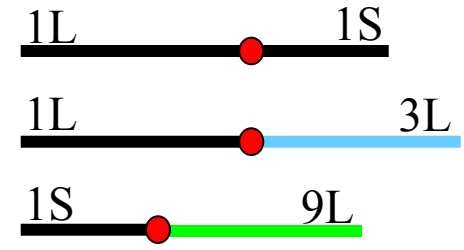
Tertiary trisomic



Telotrisomic



Acrotrisomic



Compensating trisomic

# Primary Trisomics

## 1. Primary trisomics ( $2n=2x+1$ )

An individual with a normal chromosome complement plus an extra chromosome.

### Sources:

#### 1. Normal disomics

- a. Nondisjunction in the germline of somatic tissue or during meiosis can produce a cell or gamete with an extra chromosome
- b. Failure of a bivalent to reach the meiotic metaphase plate may result in its inclusion in the telophase nucleus
- c. Physical and chemical agents may enhance the probability of producing trisomics usually by affecting the microtubules of the spindle apparatus:
  - Colchicine
  - High or low temperatures
  - Radiation

#### 2. Asynaptic or desynaptic mutants (univalent behavior)

# Primary Trisomics

## Sources:

### 3. Polyploids

- a. Haploids (gametes with a chromosome number that approaches genetic balance will be most likely viable)
- b. Triploids (the best and most dependable source to experimentally produce trisomics)
- c. Tetraploids

### 4. Chromosome interchange heterozygote numerical nondisjunction

### 5. Other aneuploids (i.e. primary trisomics, tetrasomics, multiple trisomics, secondary trisomics, tertiary trisomics, compensating trisomics)

### 6. Monosomics

### 7. Other chromosome abnormalities

Any event that effects normal chromosome pairing and disjunction may lead to the production of somatic cells or gametes with an extra chromosome. Interspecific hybrids often produce trisomic plants in their segregating progeny.



# Cytology of Primary Trisomics

## Pairing

### a. Pachytene configurations:

Two homologous chromosomes usually pair at any given point along the length of the chromosome, but the third member may pair with the other two at different sites. Only two by two pairing occurs along the synaptonemal complex.

### b. Metaphase I configurations will be determined by homolog pairing and chiasma position

### c. Anaphase I segregation:

Two members of the trivalent usually go to one pole and the third to the other pole. The orientation of homologous centromeres to opposite poles is conferred by the pairing of homologs.

All three members will rarely migrate to the same pole and result in the production of  $n-2$  and  $n+2$  spores.



# Secondary Trisomics

## 2. Secondary trisomic ( $2n=2x+iso$ )

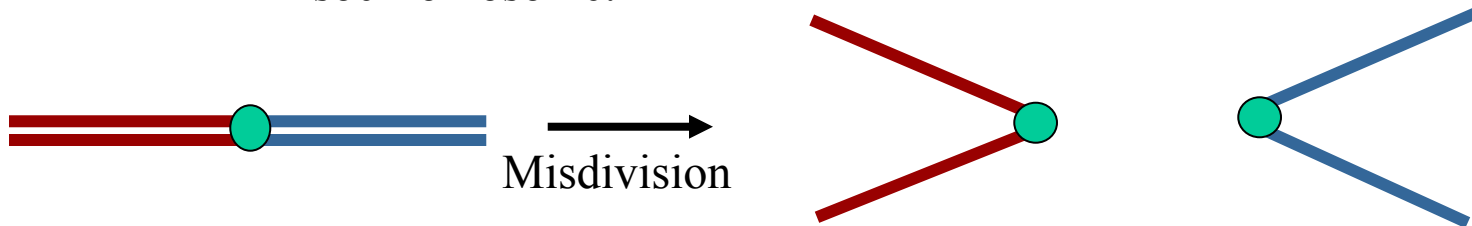
An individual with a normal chromosome complement plus an extra isochromosome (both arms of the isochromosome are homologous).

### Sources:

The origin of secondary trisomics is dependent on four events:

#### 1. Synthesis of the isochromosome

- a. Misdivision of a univalent at the centromere to produce an isochromosome:



# Secondary Trisomics

## Sources:

### 1. Synthesis of the isochromosome

- a. Misdivision of a univalent at the centromere to produce an isochromosome:

Any genotype that produces a univalent potentially may be a source of secondary trisomic:

Monosomics

Primary trisomics

Asynaptic and desynaptic mutants

Triploids

Haploids

### 2. Production of $n+1$ spores

- 3. Viability of the gametophyte with the extra chromosome (the degree of genetic imbalance is greater with an extra isochromosome than with an extra normal chromosome)

### 4. Viability of the secondary trisomic zygote

# Cytology of Secondary Trisomics

## Meiotic Pairing

### a. Univalent

The two arms of the extra chromosome are completely homologous and may pair internally resulting in  $n$  bivalents and a small ring univalent. A univalent of a secondary trisomic is nearly always the isochromosome.

### b. Trivalent

Trivalents formed in secondary trisomic individuals may assume **eight different** types of configurations, resulting from a variable number and position of chiasmata



# Cytology of Secondary Trisomics

## c. Anaphase I segregation:

An  $n$  (II+I) will produce  $n$  and  $n$ +isochromosome spores

Segregation of 3 chromosomes from the trivalent produces 3 types of functional spores.

- i.  $n$
- ii.  $n$ +secondary chromosome
- iii.  $n$ +related primary chromosome

The frequency of  $n+1$  gametes in which the extra chromosome is primary instead of secondary depends on the frequency of trivalent formation and the type of segregation it undergoes at anaphase I.

## Tertiary Trisomics

### 3. Tertiary trisomic ( $2n=2x+\#. \#$ )

An individual with a normal chromosome complement plus an interchange chromosome involving two nonhomologous chromosomes in the interchange.

#### Sources:

1. Spontaneous occurrence in the progenies of normal disomic (rare).
2. Asynaptic or desynaptic mutants produce univalents that may misdivide at the centromere and nonhomologous arms may rejoin to form tertiary chromosomes.
3. Polyploids may produce tertiary trisomics depending on two events:
  - a. Chromosome rearrangement
  - b. Production of  $n+1$  spores having the extra tertiary chromosome
4. Numerical non-disjunction from a multivalent of an interchange heterozygote may produce  $n+1$  spores with the interchange chromosome as the extra chromosome.

# Cytology of Tertiary Trisomics

## Meiotic Pairing

### a. Pachytene pairing

The extra chromosome may pair with either of the two chromosome pairs (III) from which it was derived or with both (V).

Rarely it may be left out as a univalent at pachytene.

A pentavalent (V) configuration is likely to be produced.

### b. Metaphase I

The type and frequency of the configurations formed depends on:

- i. Length of the duplicated (interchange) segment
- ii. Chiasma frequency

# Cytology of Tertiary Trisomics

Tertiary trisomic pairing configurations:

1.  $(n-2) \text{ II} + \text{V}$
2.  $(n-2) \text{ II} + 1 \text{ IV} + 1$
3.  $(n-1) \text{ II} + 1 \text{ III}$
4.  $(n-2) \text{ II} + 1 \text{ III} + 2 \text{ I}$
5.  $n \text{ II} + 1 \text{ I}$
6.  $(n-1) \text{ II} + 3 \text{ I}$

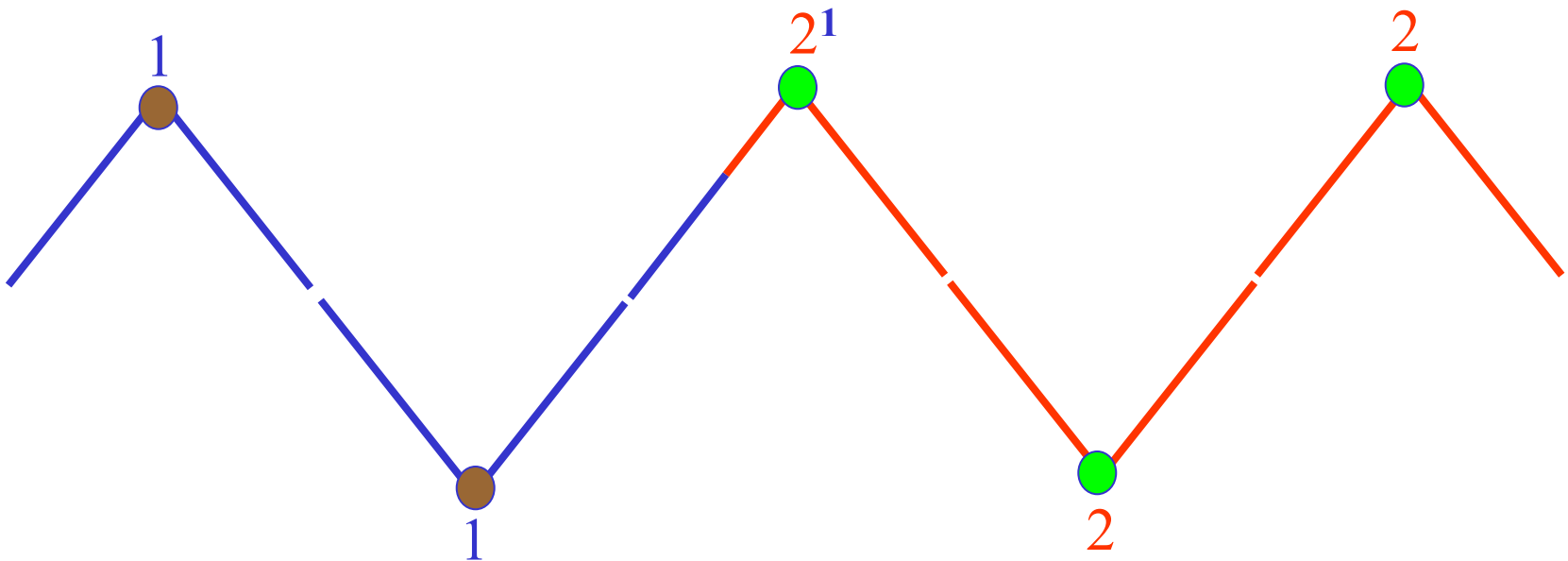
An extra chromosome with two long arms would tend to form a V at a higher frequency than those with two short arms.

Chromatids from univalents will often be lost during meiosis.

# Cytology of Tertiary Trisomics

## Gametes:

- ✓ The kinds of gametes formed will be determined by the frequency of different kinds of associations at metaphase I and the type of disjunction at anaphase I.
- ✓ Orientation of the trivalents or the multivalents of the tertiary trisomic is usually such that the two fully homologous chromosomes separate to opposite poles while the tertiary chromosome goes to either pole.



## Transmission of the extra chromosome from trisomic plants

A  $2n+1$  plant would be expected to produce  $n$  and  $n+1$  gametes with equal frequency, but 50%  $n+1$  gametes is rarely achieved.

Factors responsible for reduced transmission of  $n+1$  gametes through the female include:

1. Elimination of the extra chromosome during meiosis due to lagging or misdivision of a univalent
2. Megaspore replacement
3. Reduced viability of  $n+1$  spores or gametes relative to  $n$  gametes
4. Subnormal development of  $2n+1$  zygotes
5. Poor and delayed germination of  $2n+1$  seeds
6. Reduced vigor of  $2n+1$  seedlings
7. The effect of genetic background (degree of tolerance of genetic imbalance)

## Transmission of the extra chromosome from trisomic plants

Male transmission of the extra chromosome is usually extremely low relative to female transmission.

Factors responsible for reduced transmission of  $n+1$  gametes through the male include:

1. All the factors that reduce transmission of the extra chromosome through the female probably affect male transmission in a like manner
2. The ability of  $n+1$  pollen to compete with  $n$  pollen may be reduced since  $n+1$  pollen may:
  - a. Mature later than  $n$  pollen
  - b. Produce slow growing pollen tubes
  - c. Produce defective pollen tubes
  - d. Fail to germinate
  - e. Fail to complete post-meiotic stages of microsporogenesis due to genetic imbalance of the nucleus



## Genetic ratios expected from trisomic segregation

- ✓ Three genotypes are possibly in disomic individuals considering two alleles at a locus:
  - $AA$  homozygous dominant
  - $Aa$  heterozygous
  - $aa$  homozygous recessive
- ✓ Four genotypes are possible in trisomic individuals considering two alleles at a locus:
  - $AAA$  triplex
  - $AAa$  duplex
  - $Aaa$  simplex
  - $aaa$  nulliplex
- ✓ Genetic ratios of factors located on trisomic chromosomes will be different from the ratios of genes on disomic chromosomes.
- ✓ Ratios will be further modified by low transmission of  $n+1$  gametes through the pollen in addition to the trisomic condition.
- ✓ Genetic segregation ratios of trisomic individuals are a means of locating genes to chromosomes and establishing independence of linkage groups.



## Genetic ratios expected from trisomic segregation

### Trisomic segregation from a selfed duplex ( $AAa$ )

Assume:

1. No double reduction (requires that after crossing over between the centromere and gene marker in adjacent chromosomes, these chromosomes go to the same pole at anaphase I).
2. 50% transmission of  $n+1$  gametes through both the pollen and ovule

Ovule	Pollen			
	1AA	2Aa	2A	1a
1AA	1AAAA	2AAAa	2AAA	1AAa
2Aa	2AAAa	4AAaa	4AAa	2Aaa
2A	2AAA	4AAa	4AA	2Aa
1a	1AAa	2Aaa	2Aa	1aa

## Genetic ratios expected from trisomic segregation

Trisomic segregation from a selfed duplex (AAa)

1	AAAA	10	AAa
4	AAAa	4	AA
4	AAA	4	AAaa
4	Aaa	4	Aa
1	aa		

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35 A\_ : 1 aa

## Genetic ratios expected from trisomic segregation

Trisomic segregation from a selfed duplex ( $AAa$ )

Assume:

1. No double reduction
2. 50% transmission of  $n+1$  gametes through the female
3. 0% transmission of  $n+1$  gametes through the male

		Pollen	
		2A	1a
50% n+1	1AA	2AAA	1AAa
	2Aa	4AAa	2Aaa
50% n	2A	4AA	2Aa
	1a	2Aa	1aa

2 AAA	
5 AAa	
2 Aaa	
4 AA	
4 Aa	
1 aa	
<hr style="width: 100%;"/>	
17 A_ : 1 aa	

## Genetic ratios expected from trisomic segregation

Trisomic segregation from a selfed duplex ( $AAa$ )

Assume:

1. No double reduction
2. 25% transmission of  $n+1$  gametes through the female
3. 0% transmission of  $n+1$  gametes through the male

		Pollen	
		2A	1a
25% n+1	1AA	2AAA	1AAa
	2Aa	4AAa	2Aaa
75% n	3 x 2A	12AA	6Aa
	3 x 1a	6Aa	3aa

2 AAA  
 5 AAa  
 2 Aaa  
 12 AA  
 12 Aa  
 3 aa

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33  $A\_ :$  3 aa  
 or  
 11  $A\_ :$  1 aa

## Double reduction will affect genetic expectations in the progeny of trisomic

Double reduction gametes are aa gametes

### Requirements:

1. Multivalent pairing
2. Crossover between the gene and the centromere
3. Crossover chromatids pass to the same pole at division I
4. Crossover chromatids pass to the same pole at division II

