

Pericentric Inversions (around the centromere)

Kinds of chromatids produced by various crossovers within the inversion in
a pericentric inversion heterozygote

C.O. event	Position of C.O.	<u>Constitution of 4 chromatids after C.O.</u>		
		Normal	Inversion	dp + df
Single C.O	Any point	1	1	2
<u>Double</u>				
2-Strand	1,2	2	2	0
3-Strand	1,3 or 1,4	1	1	2
4-Strand	1,5	0	0	4

The numbers in column 2 refer to those in inversion figure. C.O.=Crossover

Paracentric Inversions

(beside the centromere)

In plants

- ✓ A homozygous inversion will produce normal pollen and seed set
- ✓ A heterozygous inversion will produce partial ovule and pollen abortion
- ✓ The degree of pollen abortion is dependent upon the amount of crossing-over within the inversion loop
- ✓ To distinguish a homozygous inversion from a homozygous normal individual, the unknown can be crossed with a homozygous normal individual

If the unknown is a homozygous inversion, the F_1 will be heterozygous for the inversion and be partially sterile

Paracentric Inversions (besides the centromere)

Kinds of chromatids produced by various crossovers in a paracentric inversion heterozygote

C.O. event	Position of C.O.	<u>Constitution of 4 chromatids after C.O.</u>		
		Normal	Inversion	dp + df
Single C.O	Any point	1	1	2 (dicentric+acentric)
<u>Double</u>				
2-Strand	1,2	2	2	0
3-Strand	1,3 or 1,4	1	1	2 (dicentric+acentric)
4-Strand	1,5	0	0	4 (dicentric+acentric)
IN/OUT	1 in loop + 1 in interstitial	1	1	2 (dicentric+acentric)
IN/OUT	4-strand double + 1 in interstitial	0	0	4 (dicentric+acentric)

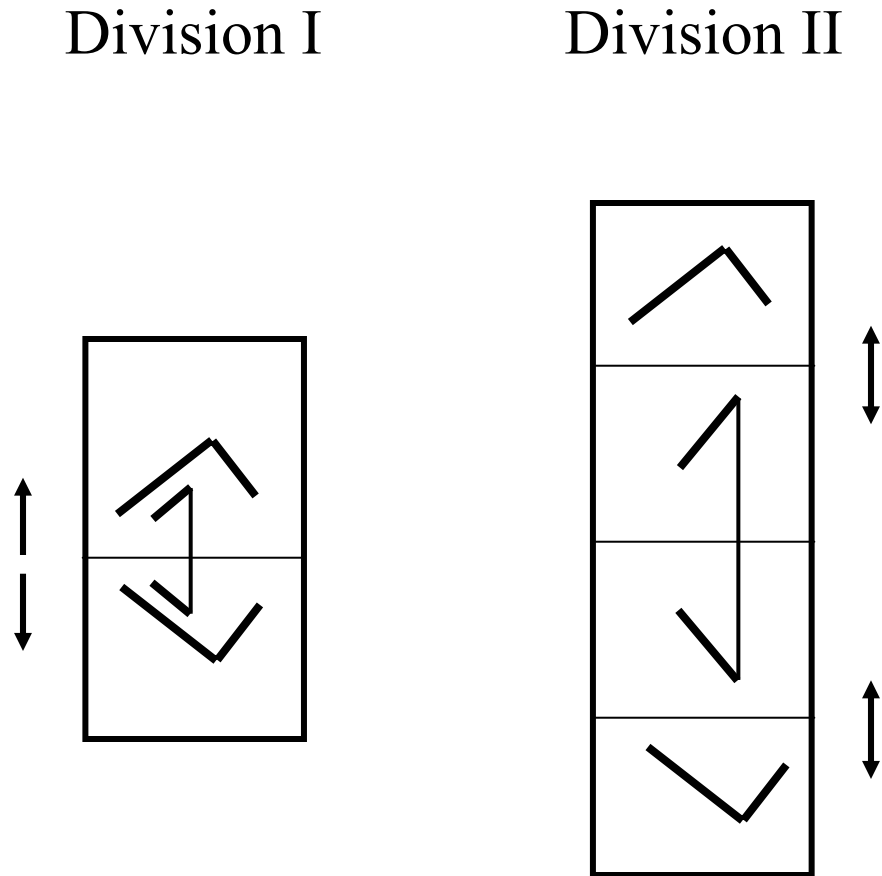
Paracentric Inversions (beside the centromere)

- ✓ Crossing-over in paracentric inversions result in bridges (dicentric chromosomes) and fragments (acentric chromosomes)
- ✓ The size of the acentric fragment represents the length of the inverted region plus twice the length of the distal segment
- ✓ If deficiencies for the segment distal to the inversion, resulting from the loss of the acentric fragment, cause gamete spore abortion, the pollen abortion percentage can be predicted by cytological observation of meiosis

Chromatid tie in *Dorsophila* female Oogenesis

A dicentric bridge orients crossover chromatids away from the poles at division I

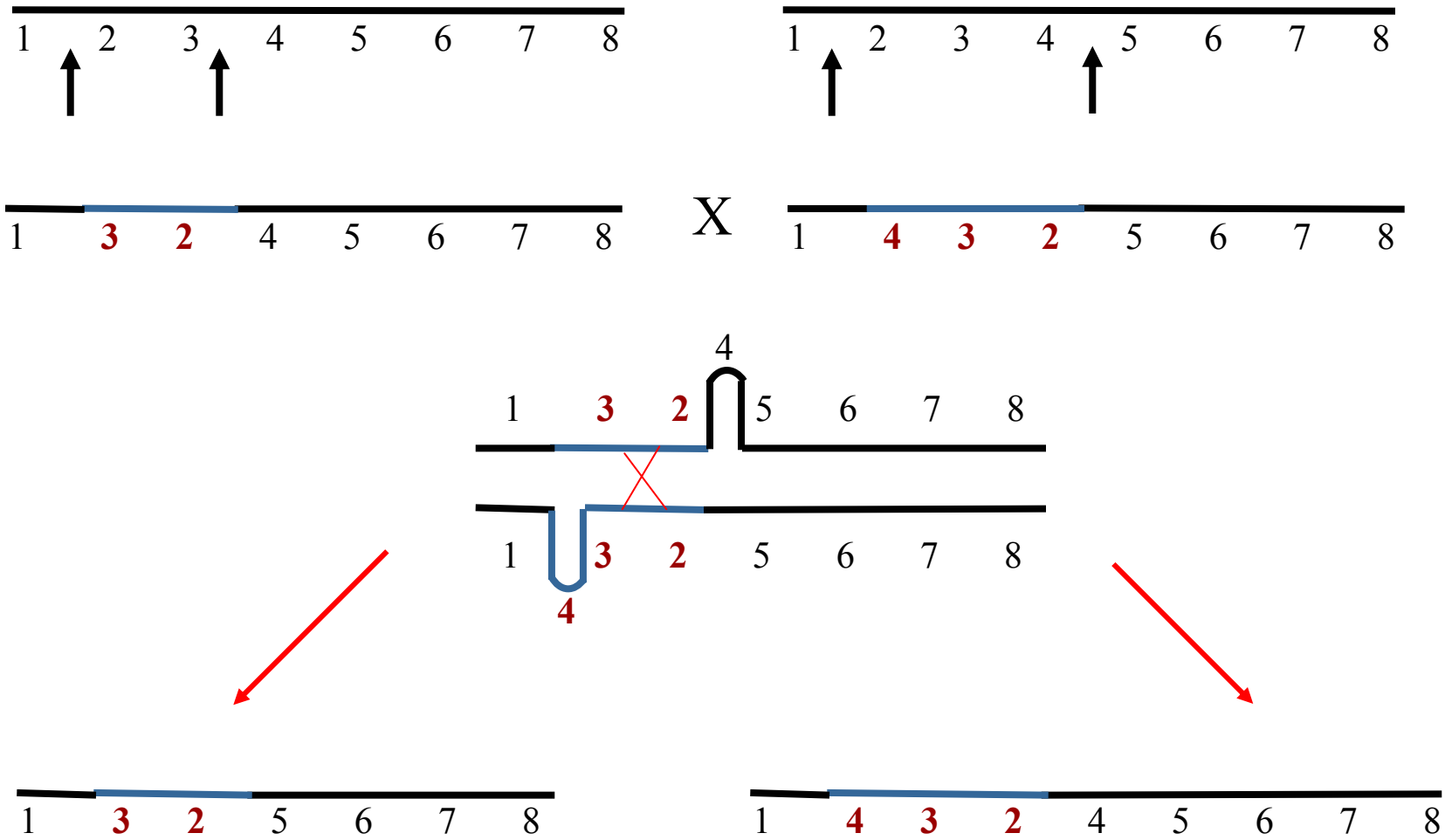
Deficiency-duplication chromatids will occur in intercalary cell and polar cells will produce a fertile ovum with intact chromosomes



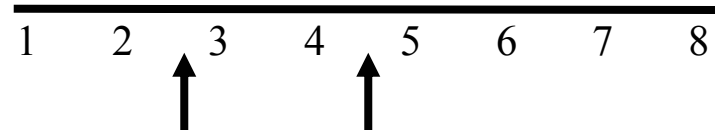
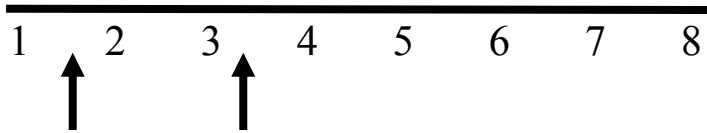
- ✓ During female gamete formation in plants and animals, only one polar megaspore will function in production of the ovule
- ✓ The duplication-deficiency chromosomes are oriented to the intercalary cells by the chromatid tie and the normal chromosomes will be included in the nuclei of the polar cells more often than the dp-df chromosomes
- ✓ The result will be nearly normal female fertility of inversion heterozygotes, but recombination will be substantially reduced in regions involved in the inversion

Use of inversions to produce duplications without deficiencies

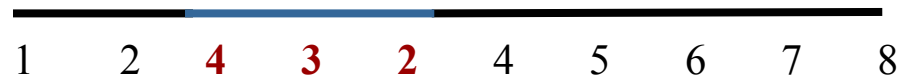
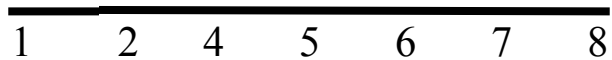
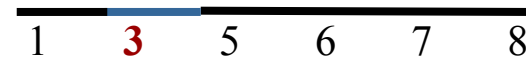
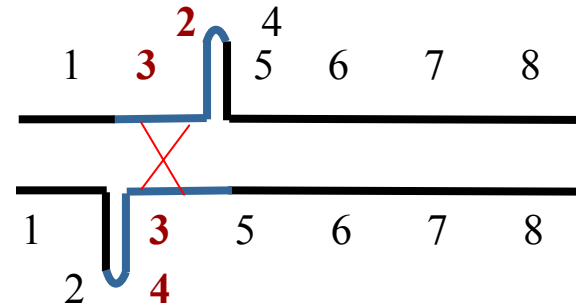
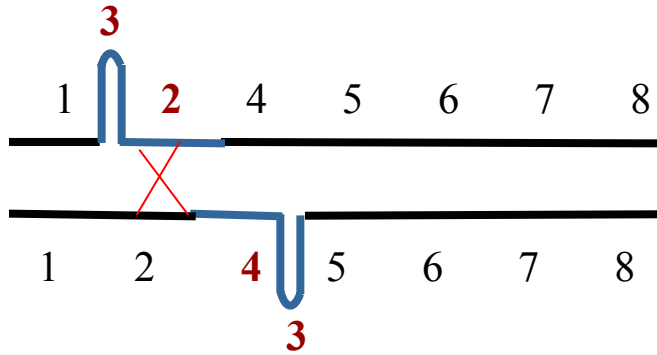
Intercross two inversions with one breakpoint in common:

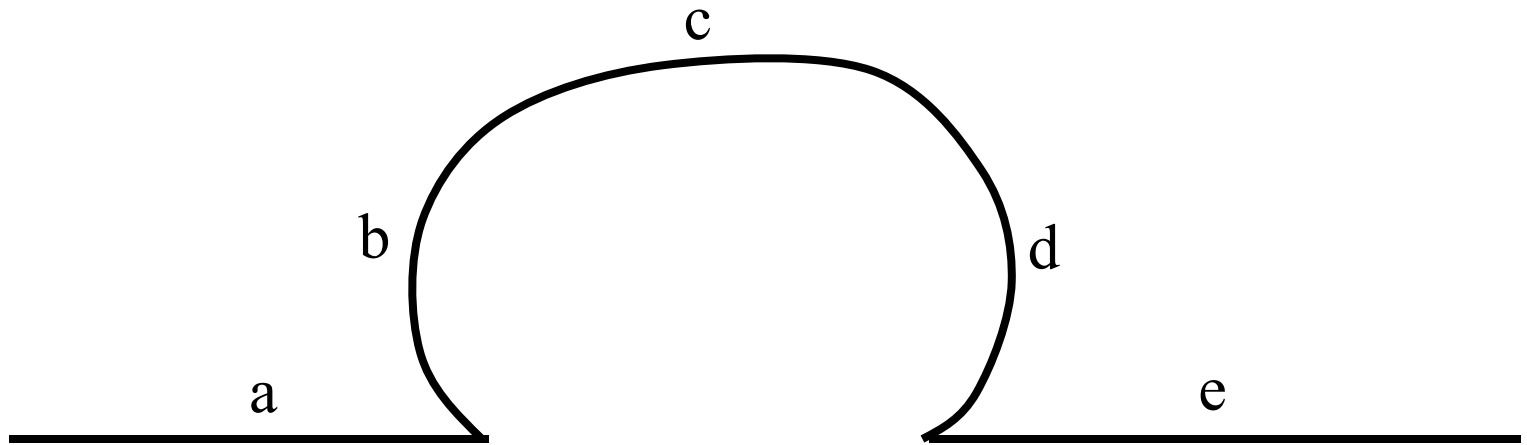


Intercross overlapping inversions:



X

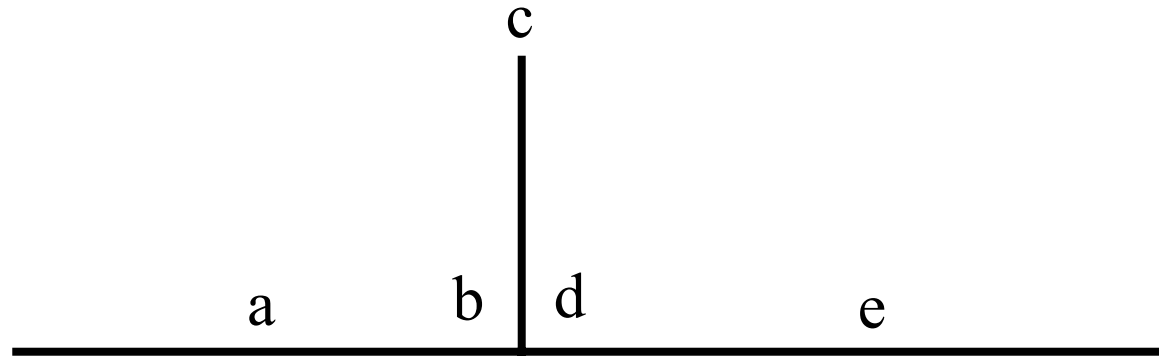




A double crossover is required for recovery of chromatid with recombination within the inversion loop

c will have greatest amount of recombination with a

b and d will have equal amount of recombination with a, but less than c



Interchromosomal Translocation

(part of one chromosome is attached to another)

Types

1. Interstitial translocations (intercalary)

A segment from one chromosome is transferred to a position in another chromosome. Requires three breaks.

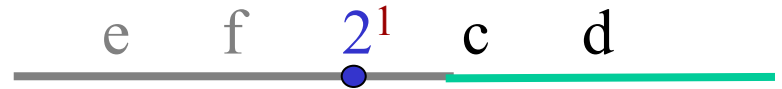
2. Reciprocal translocation (interchange)

Two non-homologous chromosomes have symmetrically exchanged segments. One break in each chromosome is sufficient. Nearly always involves terminal end segments.

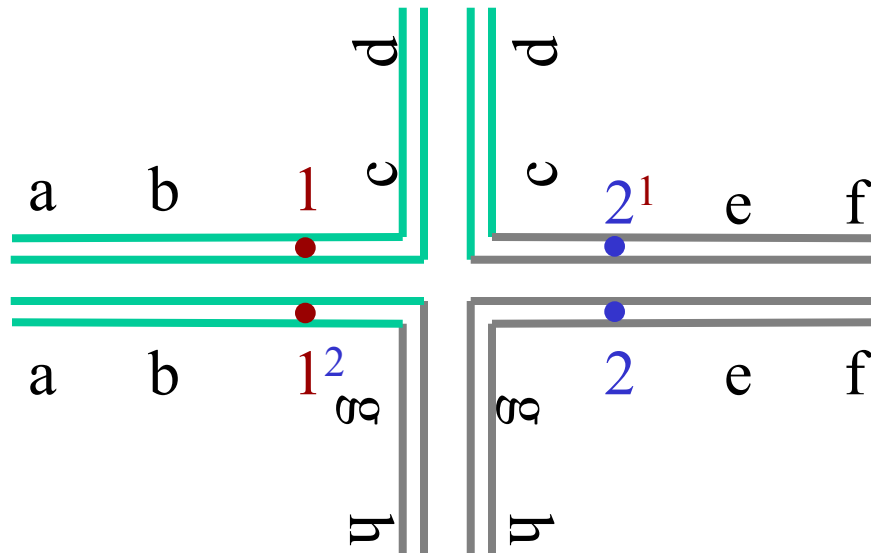
Interstitial segment = segment of an interchange chromosome between the breakpoint and the centromere.

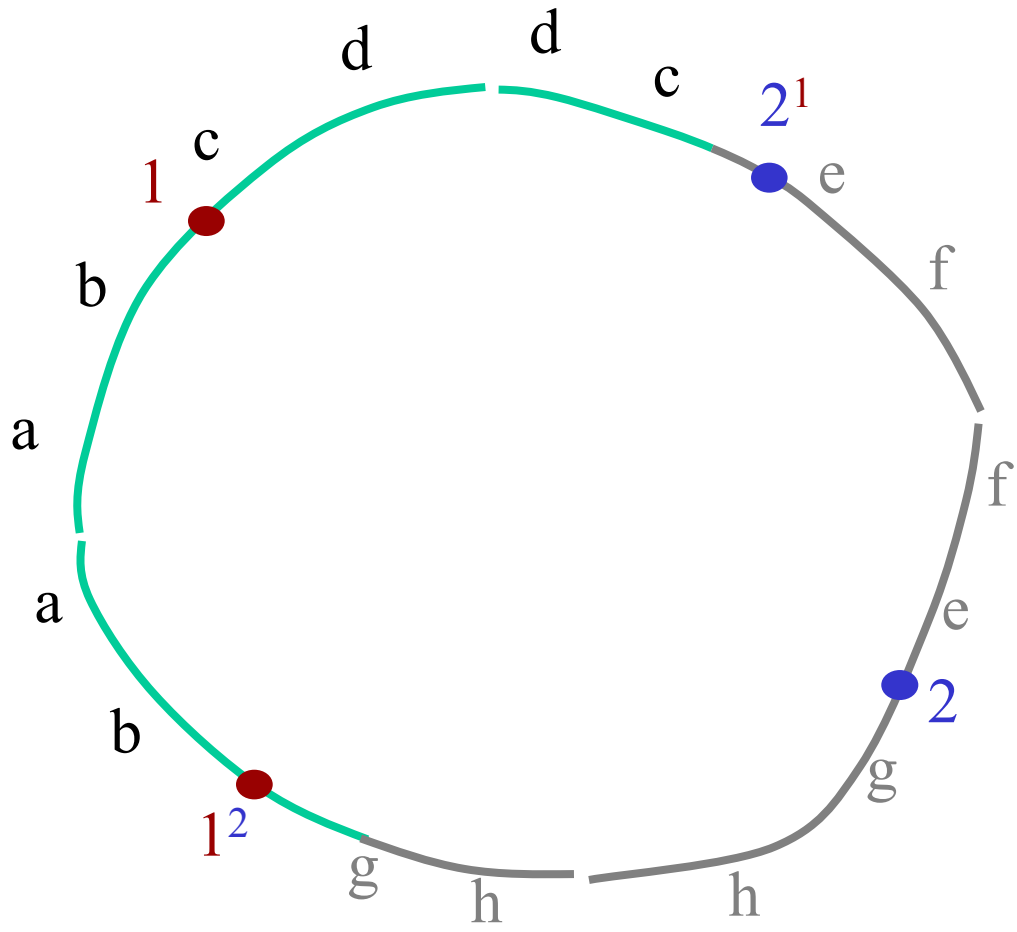
Interchromosomal Translocation

- ✓ A cross configuration is formed at pachytene of interchange heterozygotes. The position of the cross is a reflection of where the breakpoint has occurred.
- ✓ During diplotene and diakinesis, the chromosomes shorten, the chiasma terminalize, and the cross configuration opens up to form a ring of 4 if chiasma are present.



Pachytene pairing of interchange heterozygote





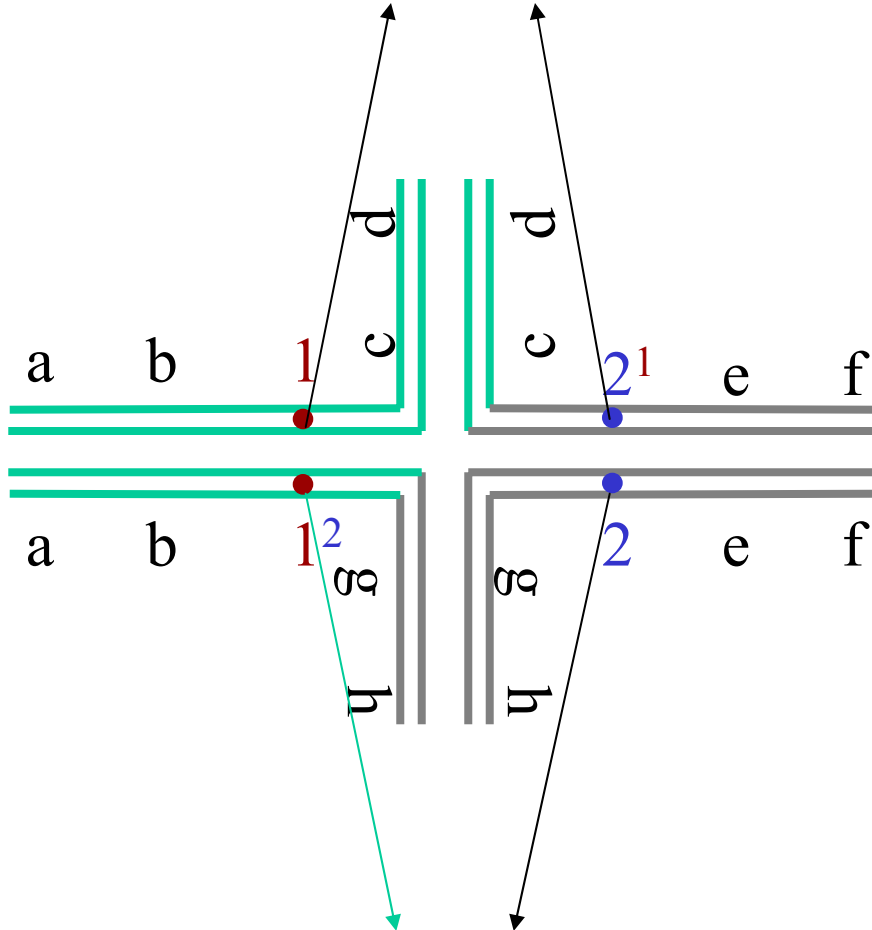
Interchromosomal Translocation

Observed meiotic configurations depend on the occurrence of chiasmata

No. of Arms with Chiasma	Diakinesis Configuration
4	Ring of 4 ($\odot 4$)
3	Chain of 4 (IV, 4 types)
2 adjacent arms	Chain of 3 + univalent (III+I, 4 types)
2 alternate arms	2 pairs (2II, 2 types)

Orientation of interchange heterozygote quadrivalent at Metaphase I

Adjacent I

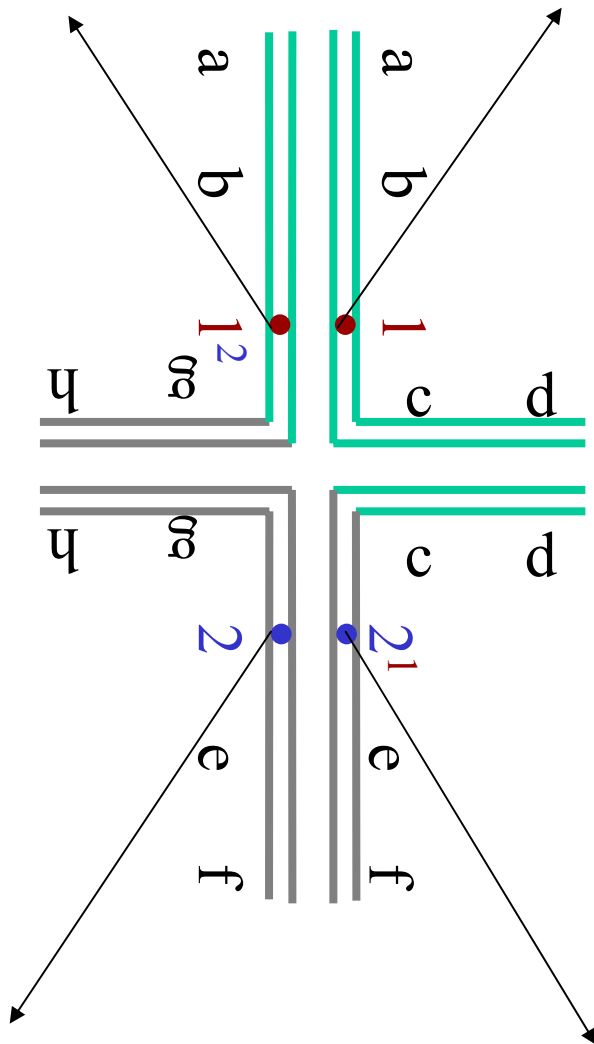


Adjacent non-homologous centromeres pass to the same pole

$1 + 2^1$ dp cd + df gh
 $1^2 + 2$ dp gh + df cd

Orientation of interchange heterozygote quadrivalent at Metaphase I

Adjacent II



Adjacent homologous centromeres
pass to the same pole

1 + 1²

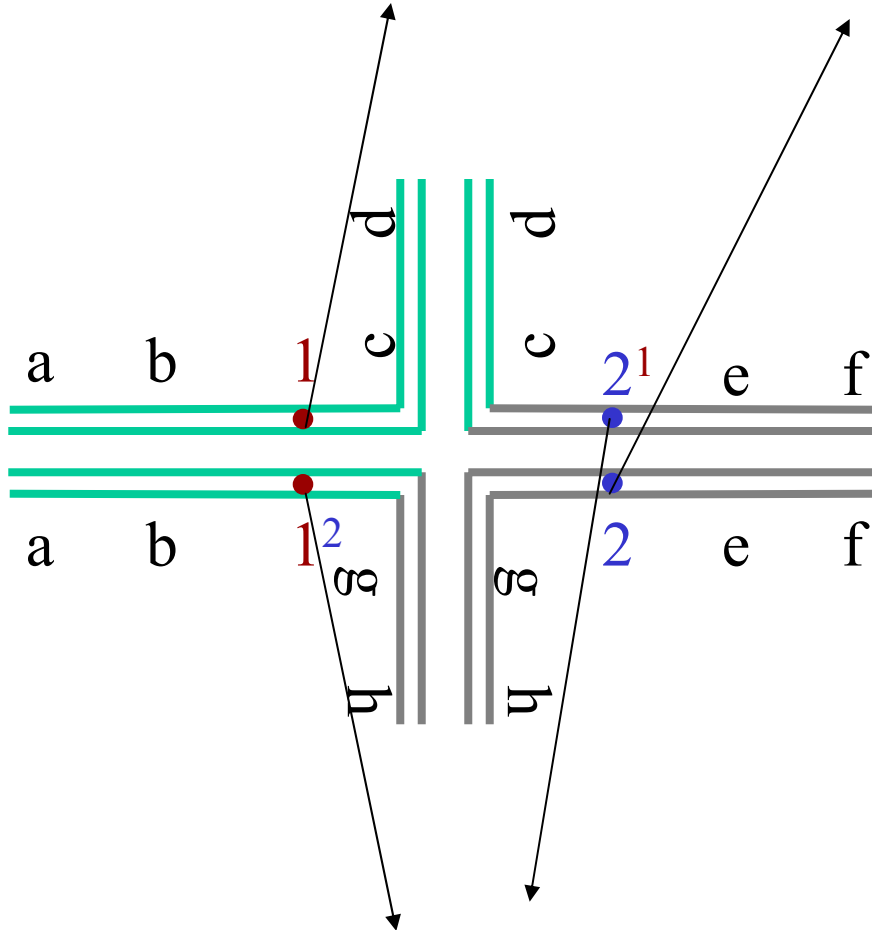
dp ab + df ef

2¹ + 2

dp ef + df ab

Orientation of interchange heterozygote quadrivalent at Metaphase I

Alternate



Alternate disjunction of non-homologous centromeres

$1 + 2$	Normal
$1^2 + 2^1$	Balanced translocation

Disjunction from a ring quadrivalent

Orientation of chromosomes of a ring of 4 may be either an open or a zig-zag configuration leading to either adjacent or alternate chromosome disjunction.

Adjacent I disjunction

Adjacent but non-homologous centromeres migrate to the same pole.

$1+2^1$	Dp fe +Df jk	Dp=duplication
1^2+2	Dp jk +Df fe	Df=deficiency

Gametes usually abort.

Adjacent II disjunction

Occurs rarely if ever. Adjacent but homologous centromeres migrate to the same pole.

$1+1^2$	Dp abcd +Df ghi
2^1+2	Dp ghi+Df abcd

Gametes abort.

Disjunction from a ring quadrivalent

Alternate disjunction

Alternate centromeres migrate to the same pole at anaphase I.

$1+2$	Normal chromosome complement
1^2+2^1	Interchange chromosome complement

Both combinations produce viable gametes.

Factors influencing orientation of a ring quadrivalent

- ✓ Considering 2 normal bivalents, there is complete independence and adjacent I and alternate disjunction will occur with equal frequency.
- ✓ Adjacent II should be impossible since there is no opportunity for co-orientation between non-homologous centromeres.
- ✓ With production of quadrivalent co-orientation of non-homologous centromeres becomes possible.
- ✓ With random co-orientation:
alternate disjunction frequency = adjacent disjunction frequency
- ✓ Even within species there is considerable genetic variation affecting the ratio of alternate and adjacent disjunction.
- ✓ In most cases either alternate or adjacent predominates so that co-orientation is not a reality.
- ✓ Random orientation may occur in early prophase but soon forces act on quadrivalent, changing the orientation of the quadrivalent.

Factors influencing orientation of a ring quadrivalent

- ✓ Forces acting on the quadrivalent:
 1. Contraction of chromosomes resulting in stiffness and torsion.
Short stiff chromosomes or those with little tendency for chiasma terminalization do not have sufficient flexibility for alternate disjunction.
 2. Centromere activity
Centromere orientation is maintained by the presence of counter-force exerted on the centromere
- ✓ Alternate orientation provided more stable counter forces and will not readily revert to adjacent orientation.
- ✓ With adjacent orientation if the pull from a single opposite centromere lapses, both co-orienting centromeres become unstable and resume equal probabilities to orient to either pole.
- ✓ With time the alternate orientation often accumulates.
- ✓ In rye interchange heterozygotes, alternate rings may occur in up to 95% of PMCs in late metaphase.

Factors influencing orientation of a ring quadrivalent

1. Forces acting on the quadrivalent.
2. Length of interchange and interstitial segment.
3. Localization and terminalization of chiasmata.

Genetic consequence of interchange

- ✓ An interchange behaves like a single genetic factor.
- ✓ Two reciprocal translocations that do not have a chromosome in common segregate independently.
- ✓ In the translocation homozygote, the linkage relationship will be changed.
- ✓ Genes in the translocated segment fail to show linkage with genes in the chromosome where they originally occurred.