Chromosome Deficiencies and Duplications

- ✓ Deficiencies (df) usually cause abortion and are expressed in the form of gametic abortion in plants and zygotic abortion in animals. However, a small deficiency and/or one for a nonessential region frequently may be transmitted through the female parent in plants or the zygote may survive in animals.

- ✓ In plants duplications (dp) usually do not cause abortion, but are not transmitted through the male gamete due to the lack of competitive ability of duplication-bearing pollen with normal pollen. Since there is no competition between ovules, duplications may be transmitted through the female parent. Sparse pollination may reduce competition and allow transmission of duplications through the male parent.

Chromosome Deficiencies and Duplications

- ✓ In plants, deficiency heterozygotes are semi-sterile.
- ✓ Duplication heterozygotes may be fertile, but the duplication-bearing pollen grains may be abnormal in appearance.
- ✓ A deficiency heterozygote would not be expected to produce recombinants in the deficient region.
- ✓ Adjacent chromosomal regions in a deficiency or duplication heterozygote may also exhibit reduced recombination due to asynapsis or nonhomologous pairing.

Chromosome Inversions

- ✓ Muller (1916) observed crossover reducers that were noticeable in *Dorsophila* only in heterozygous stocks.
  - ➢ Genetic crossing over was practically eliminated in the portion of the genetic map.
  - ➢ Maps of the region indicated a reverse order of the genes in the region of reduced crossing over.
  - ➢ Muller determined a section of the chromosome had been inverted.

Chromosome Inversions

- ✓ Chromosome inversions are a change in the linear sequence of the genes in a chromosome which results in the reverse order of genes in a chromosome.

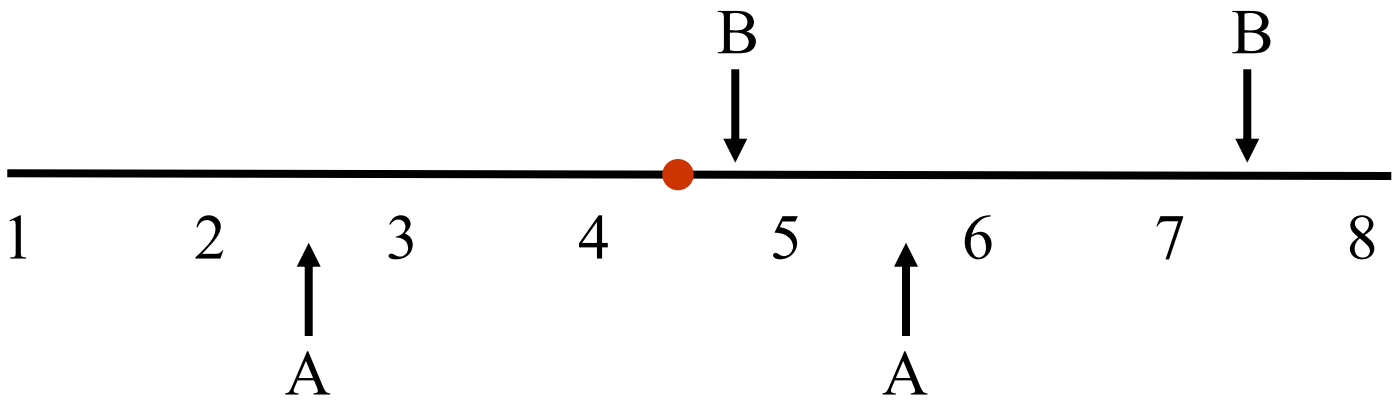
- ✓ Sources:

    1. Occur in natural populations
    2. Induced by radiation and chemical mutagens

- ✓ Result from chromosome breakage followed by healing to produce a new chromosome structure.

Chromosome Inversions

Types of inversion:



A. _____ inversion (around the centromere)

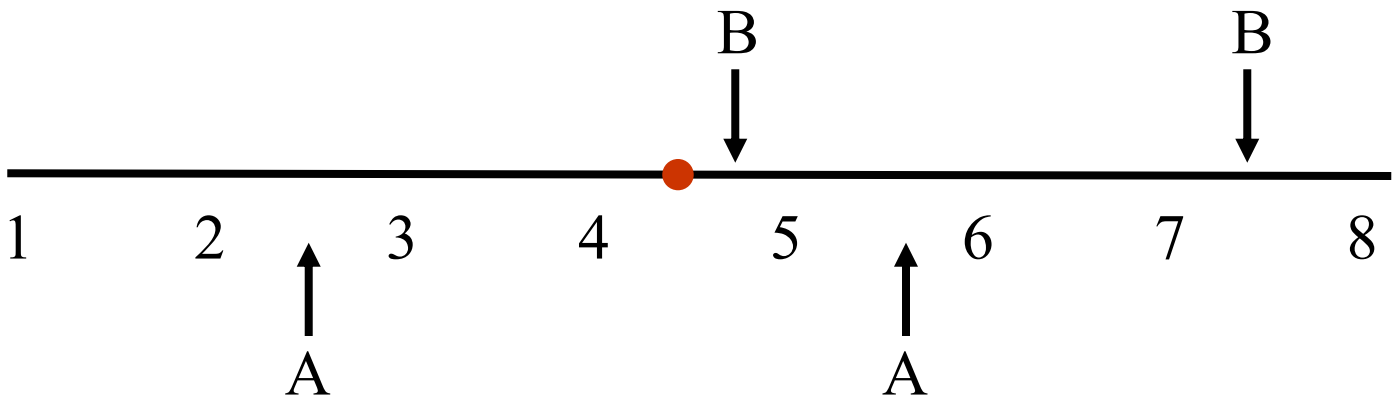
The arm ratio may change

✓ _____ inversion (beside the centromere)

The arm ratio does not change

Chromosome Inversions

Types of inversion:



A. Pericentric inversion (around the centromere)

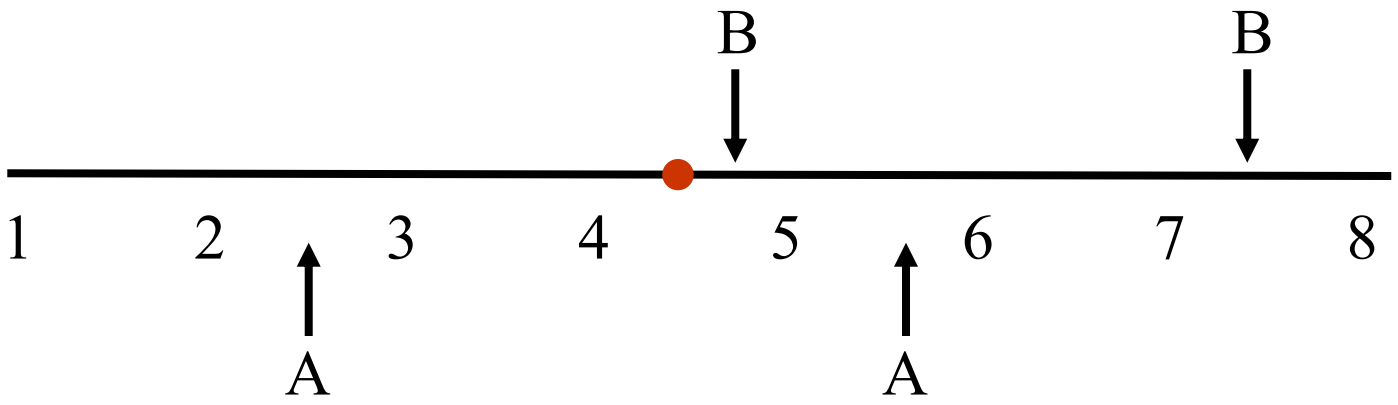
The arm ratio may change

✓ _____ inversion (beside the centromere)

The arm ratio does not change

Chromosome Inversions

Types of inversion:



A. Pericentric inversion (around the centromere)

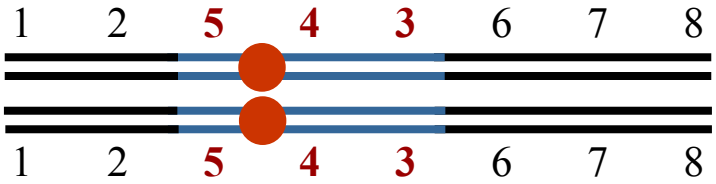
The arm ratio may change

B. Paracentric inversion (beside the centromere)

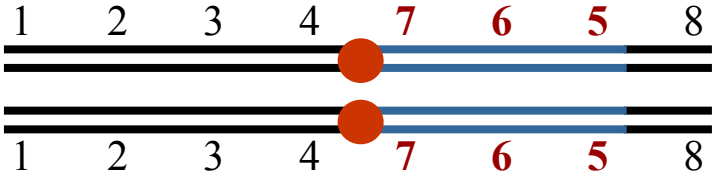
The arm ratio does not change

Chromosome Inversions

✓ Chromosome Pairing of homozygous inversions:

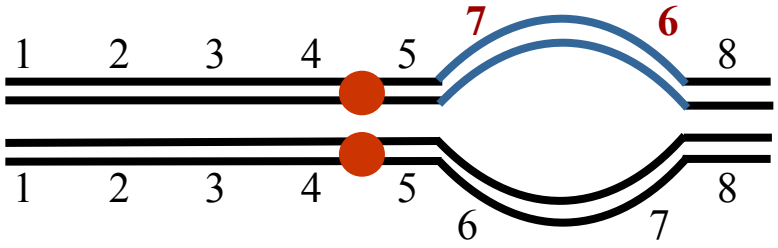


A. Pairing is normal
B. Meiosis is normal
C. Fertility is normal



✓ Chromosome Pairing of heterozygous inversions:

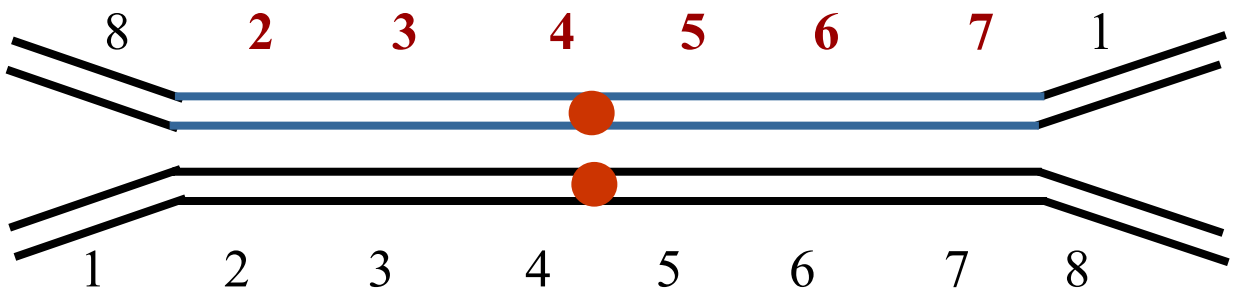
Small inverted segments



Chromosome Inversions

✓ Chromosome Pairing of heterozygous inversions:

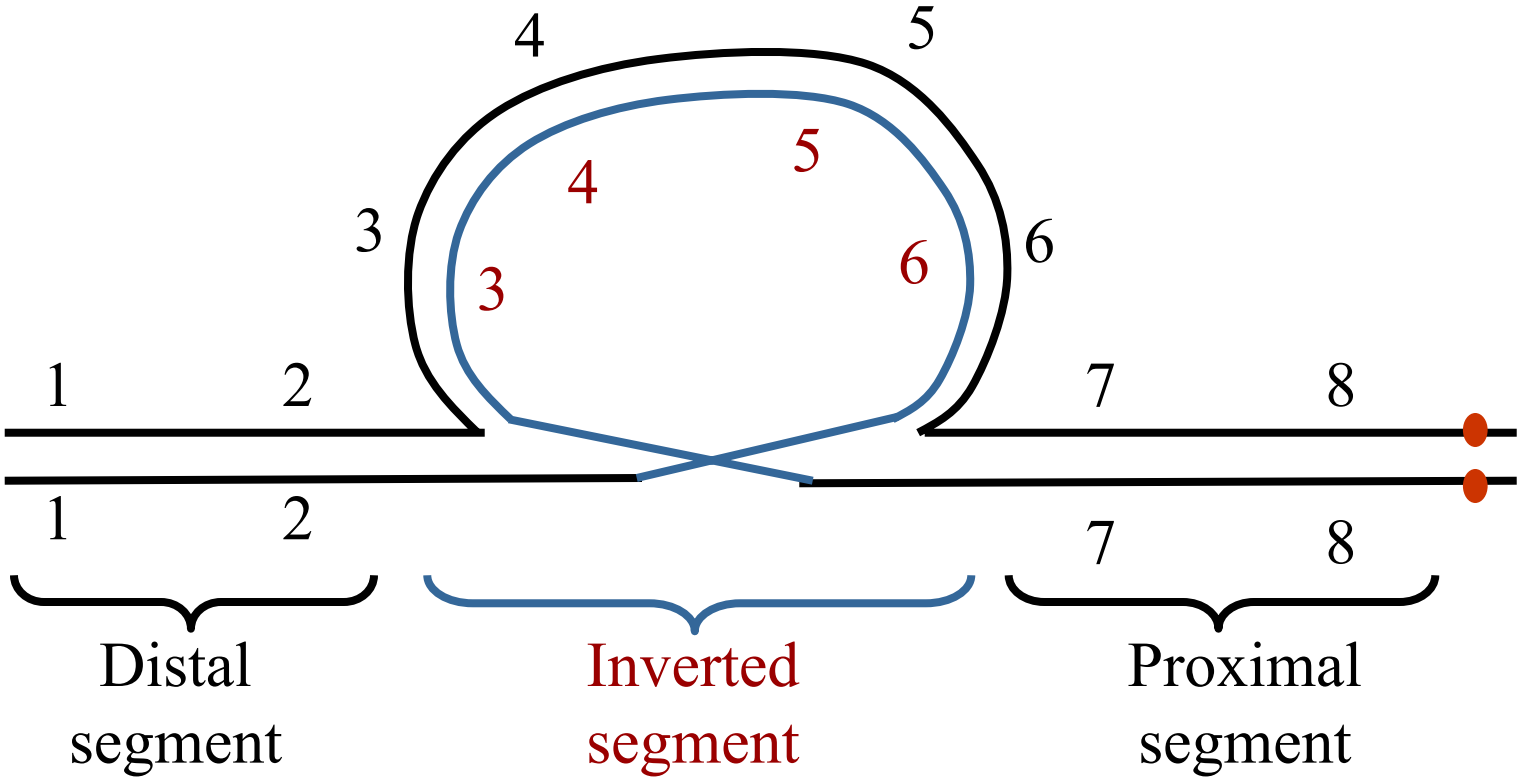
Large inverted segments that includes the majority of the chromosome



Chromosome Inversions

✓ Chromosome Pairing of heterozygous inversions:

Inverted segments long enough to be flexible



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.	Constitution of 4 chromatids after C.O.		
		Normal	Inversion	dp + df
Single C.O	Any point	1	1	2
Double				
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

Pericentric Inversions
(around the centromere)

- ✓ The two types of inversions (para- and peri-centric) result in different cytological events
- ✓ Chromosome inversions have no effect on mitotic divisions, but do effect meiosis
- ✓ Meiosis is normal in individuals with homozygous inversions
- ✓ If the inverted regions of the inversion heterozygote is large enough for crossing-over to occur within the inversion loop, a portion of the resulting gametes will be abnormal

Pericentric Inversions
(around the centromere)

- ✓ Pericentric inversions result in dp + df gametes which is related to the chromosome segments distal to the breakpoints of the inverted segment

- ✓ Products of crossover event within the loop are lost

    1. Recombinant types are not recovered
    2. Crossing over may not be suppressed cytologically
    3. Only 2 strand double crossover types are recovered

- ✓ No bridges or fragments produced at anaphase I or II

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

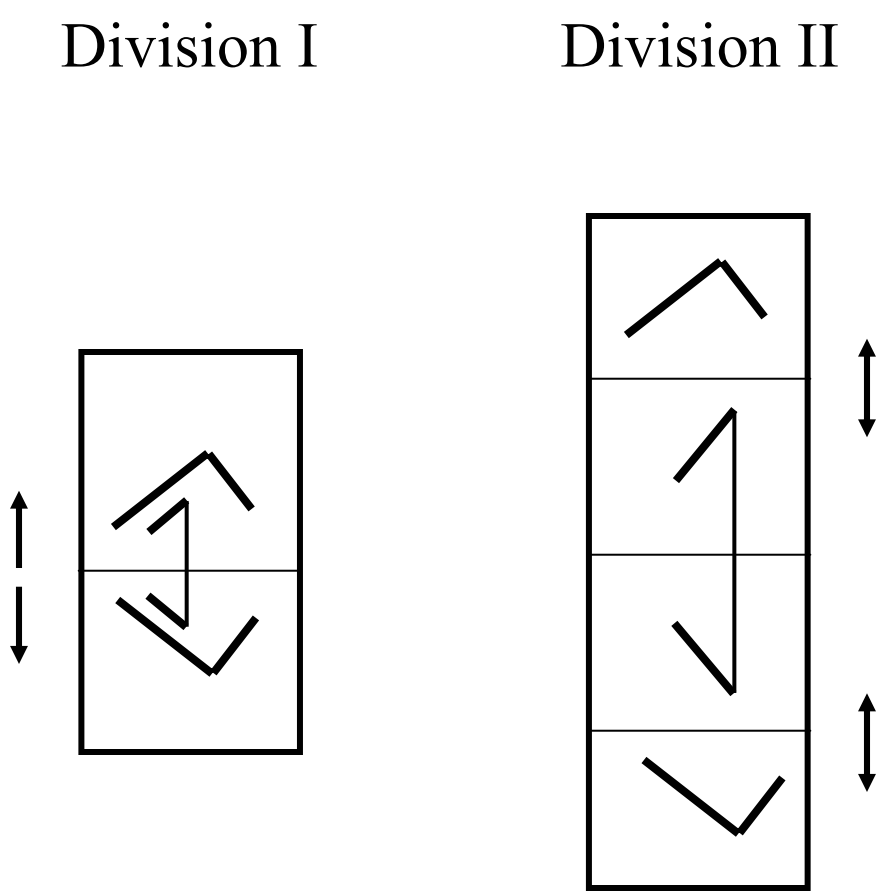
Paracentric Inversions
(beside the centromere)

- ✓ 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



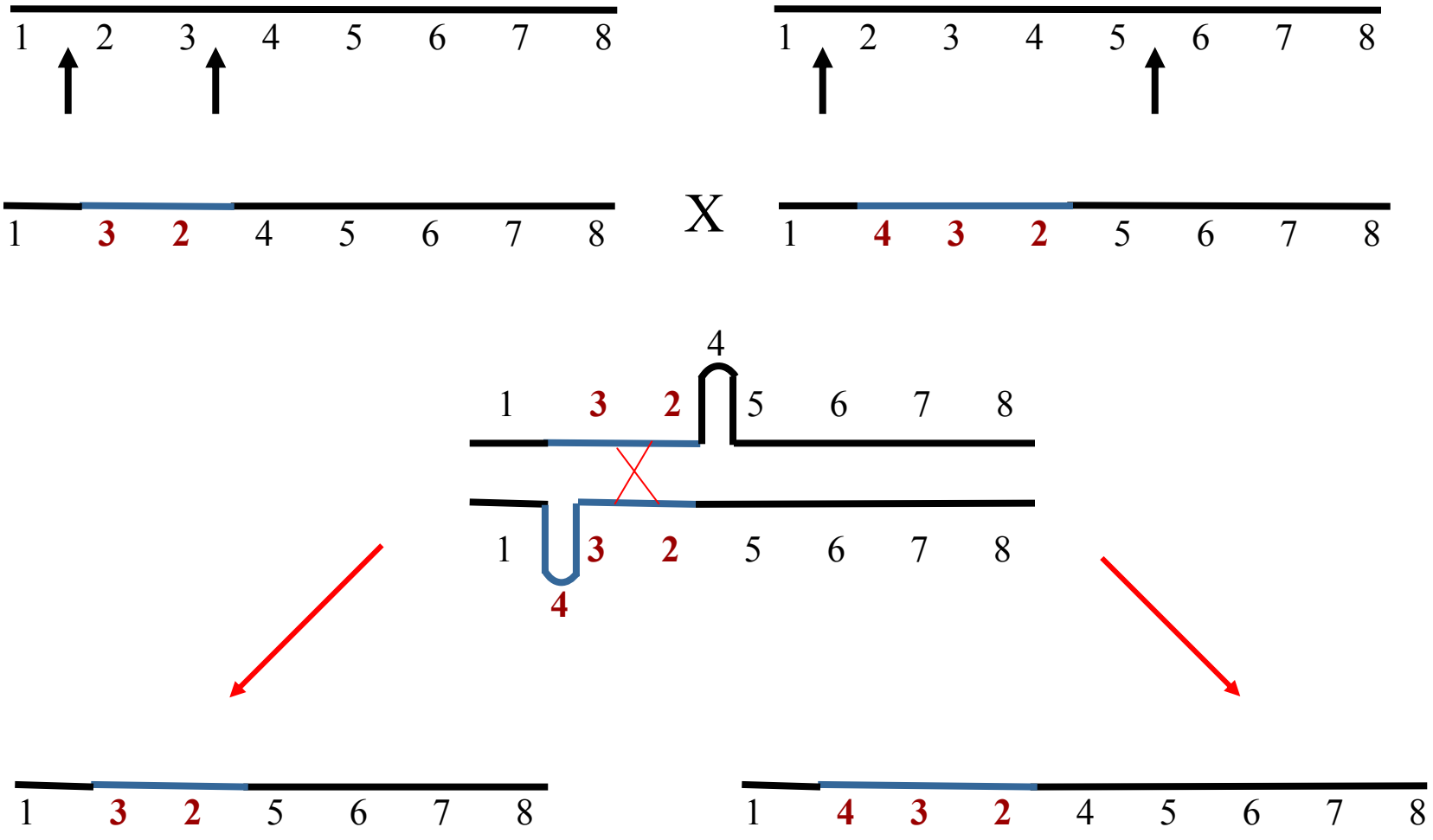
- ✓ During female gamete formation in plants and animals, only one polar megaspore will function in production of the ovule

- ✓ 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

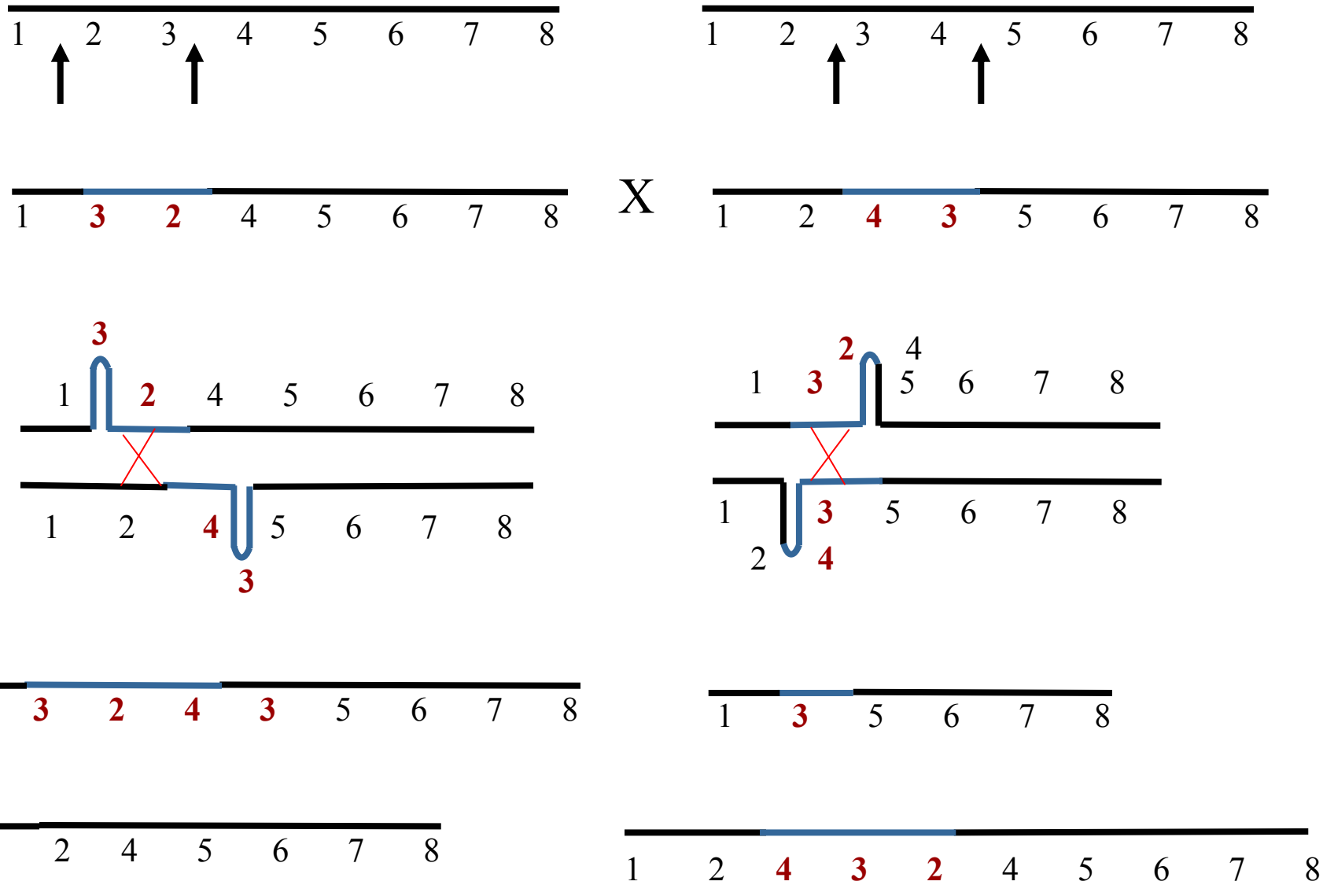
- ✓ The result will be nearly normal female fertility of inversion heterozygotes, but recombination will be substantially reduced in regions involved in the inversion

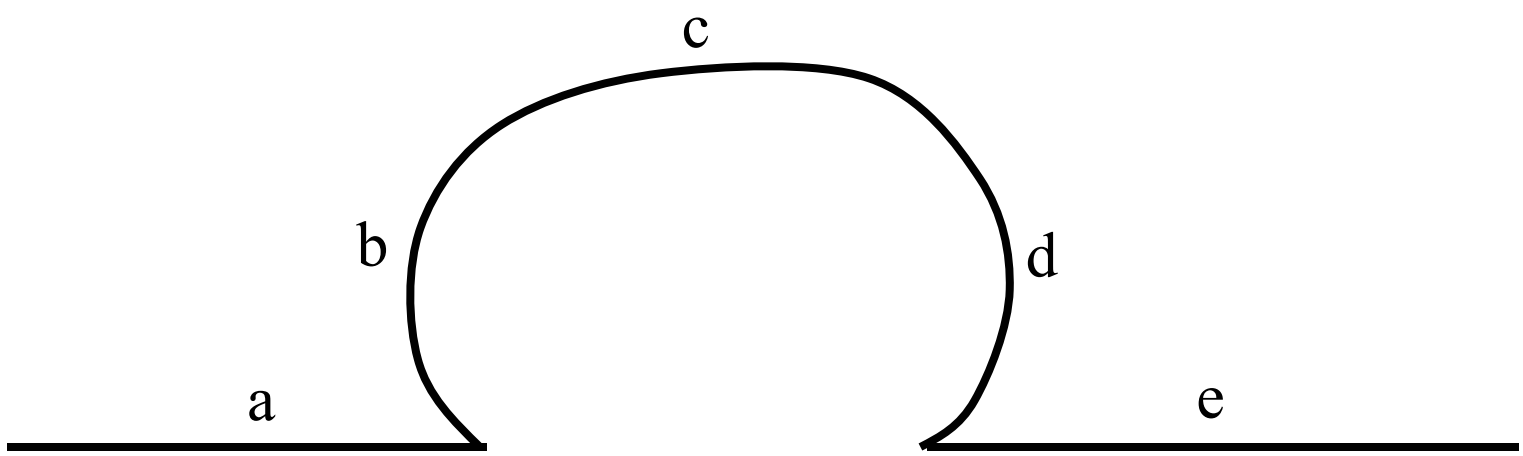
Use of inversions to produce duplications without deficiencies

Intercross two inversions with one breakpoint in common:



Intercross overlapping inversions:





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