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.

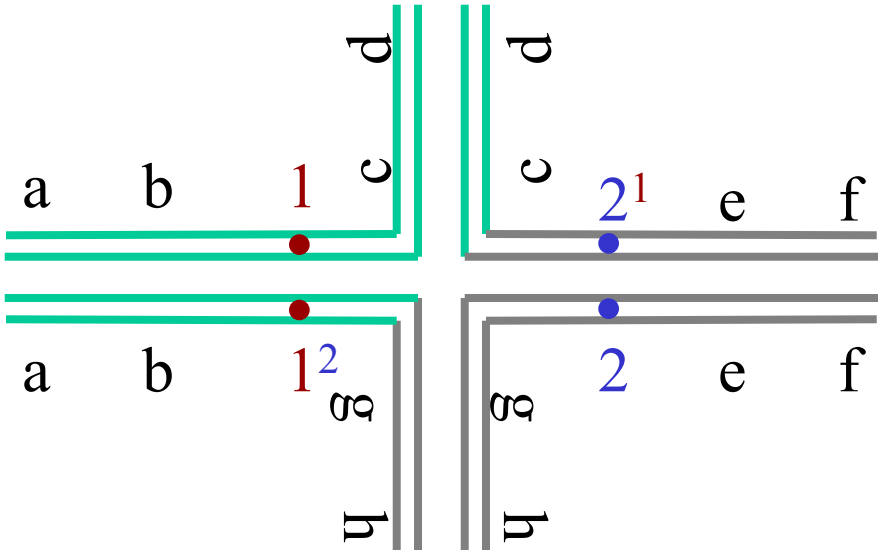
Interstitial segment = segment of an interchange chromosome between the breakpoint and the centromere.

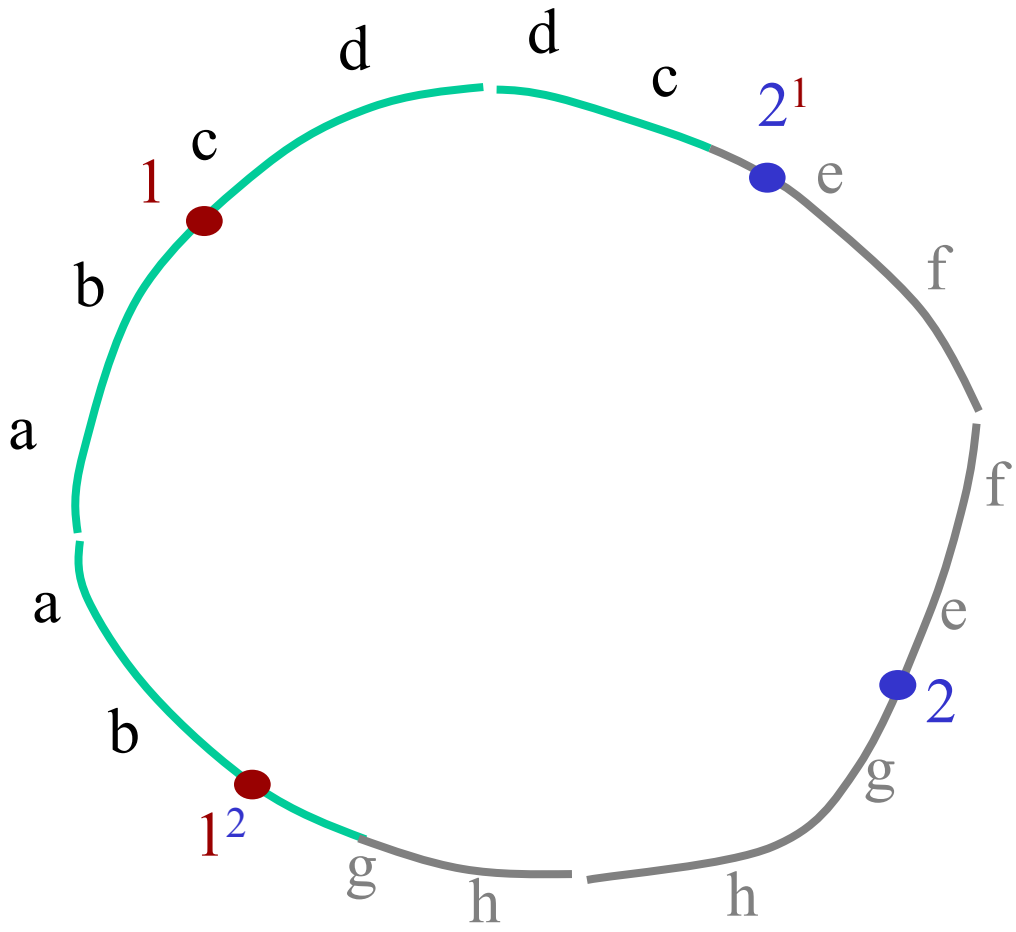
Interchromosomal Translocation

- ✓ 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



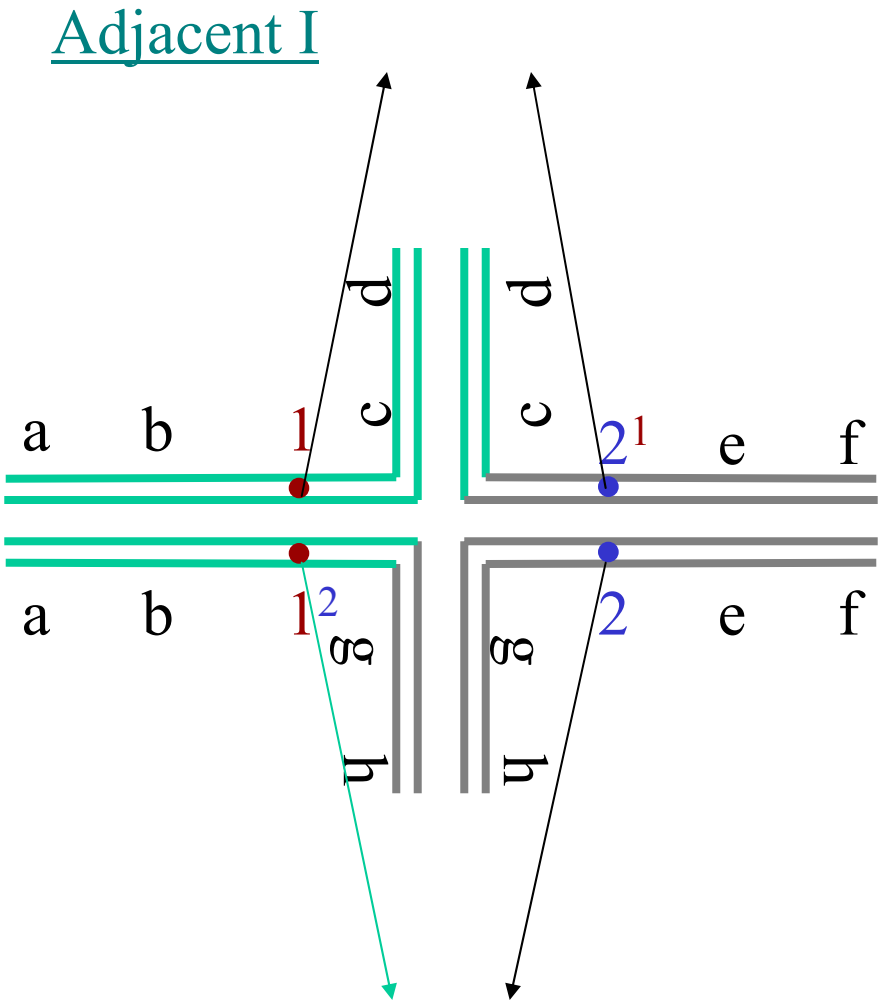

d
d
c
2^1
e
f
f
e
2
g
h
h
g
1^2
b
a
a
b
1
c

Interchromosomal Translocation

Observed meiotic configurations depend on the occurrence of chiasmata

No. of Arms with Chiasma	Diakinesis Configuration
4	Ring of 4 (⊙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

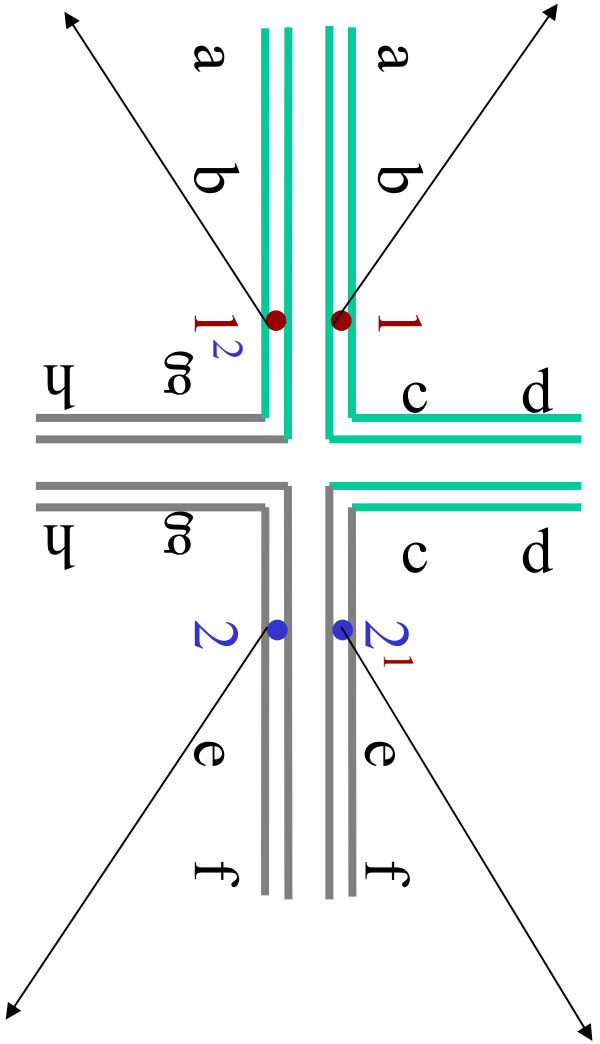


Adjacent non-homologous centromeres pass to the same pole

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

Orientation of interchange heterozygote quadrivalent at Metaphase I

Adjacent II



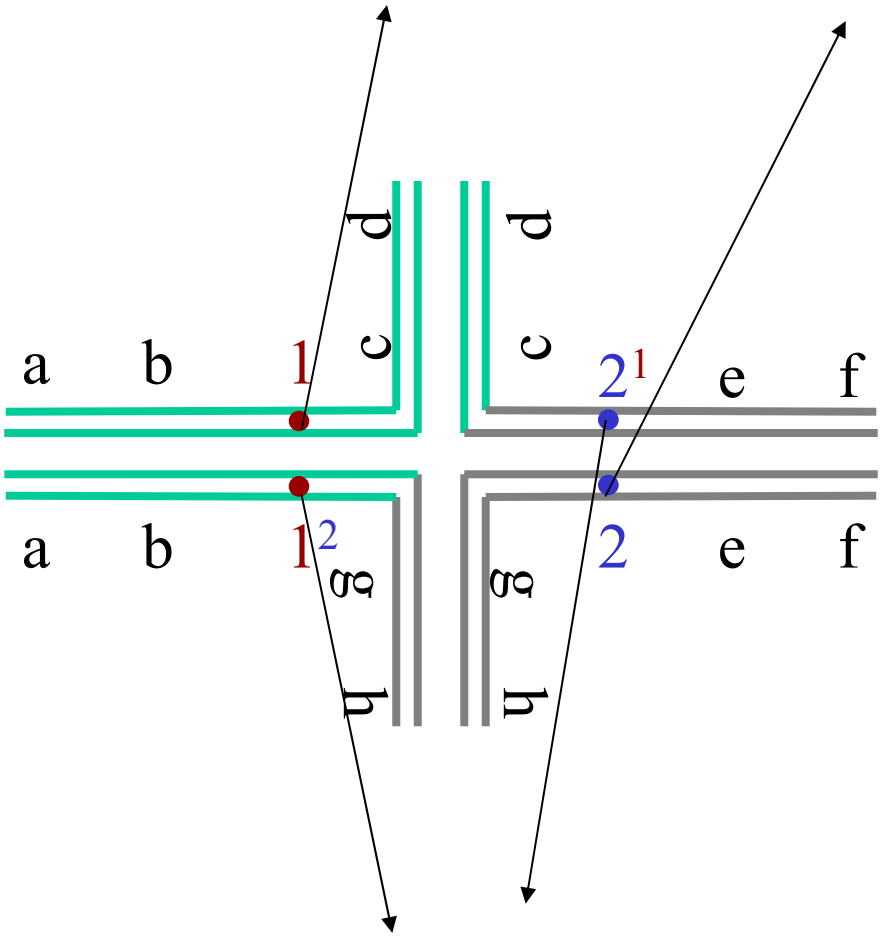
Adjacent homologous centromeres pass to the same pole

$1 + 1^2$ dp ab + df ef

$2^1 + 2$ 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.

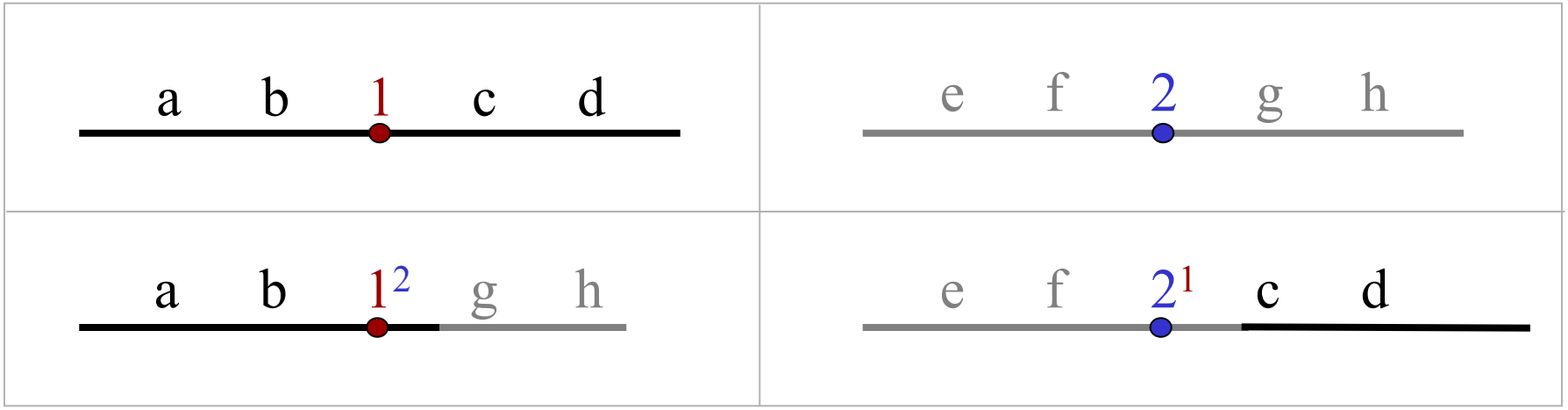
Factors influencing orientation of a ring quadrivalent

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

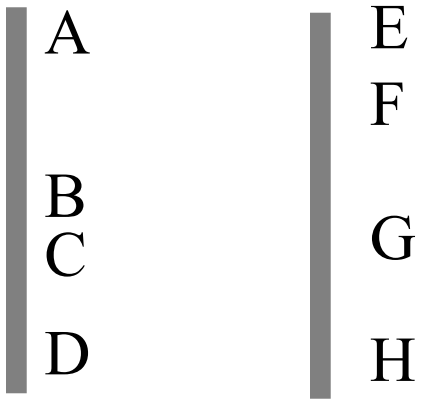
Genetic consequence of interchange

- ✓ 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.

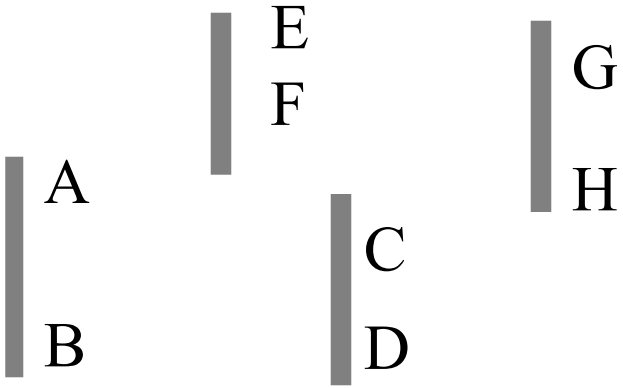
Products of Alternate disjunction



Normal linkage map



Linkage map from progeny of translocation heterozygote



Identification of chromosomes involved in interchanges

1. Cytology

   - Pachytene analysis of chromosomes involved in the cross configuration
   - Karyotype analysis of somatic cells
     - → Unequal size of exchange segment allows identification of change in chromosome lengths
     - → Banding pattern of chromosomes
     - → Direct observation of Dorsophila salivary gland chromosome bands

2. Genetic Linkage

   - Genes on one chromosome become linked to those on another
   - Genes known to be linked or independent suddenly change relationship

Identification of chromosomes involved in interchanges

1. Use of trisomic tester

   → 2n+1 known trisomic tester lines are crossed with unknown interchange stocks
   → If one of the chromosomes involved in the translocation is the trisomic chromosome, a chain of 5 is expected
   → If the trisomic does not involve one of the interchange chromosomes, a ring of 4 plus a trivalent are expected

2. Chromosome identification set

   ➢ Cross a series of known interchange stocks with the unknown interchange stock and examine the F1 at meiotic metaphase I
      ✓ Two rings of 4 indicate the interchanges are independent
      ✓ A ring of 6 indicates one chromosome of the interchange is in common with one of the tester interchange chromosomes
      ✓ An F1 from a cross between interchange stocks involving the same two chromosomes will not produce an association larger than a ring of 4 or may produce mostly/only pairs