Mendel

- What was Mendel's contribution to our understanding of Heredity?

- What is the Law of Segregation?

- What is the Law of Independent Assortment?

- What is a punnett square and how is it used to illustrate the principles of inheritance?

1

Human Mendelian Traits

- OMIM estimates that about 20,000 genes are inherited in a simple Mendelian way.

- Many blood charateristics and diseases and disorders - some simple physical traits

- Examples: Sickle cell, tented eyebrows, hitchhiker's thumb

- Most human "traits" are more complicated, but particles of inheritance - genes - are passed on in a simple Mendelian way

2

Terminology

genotype
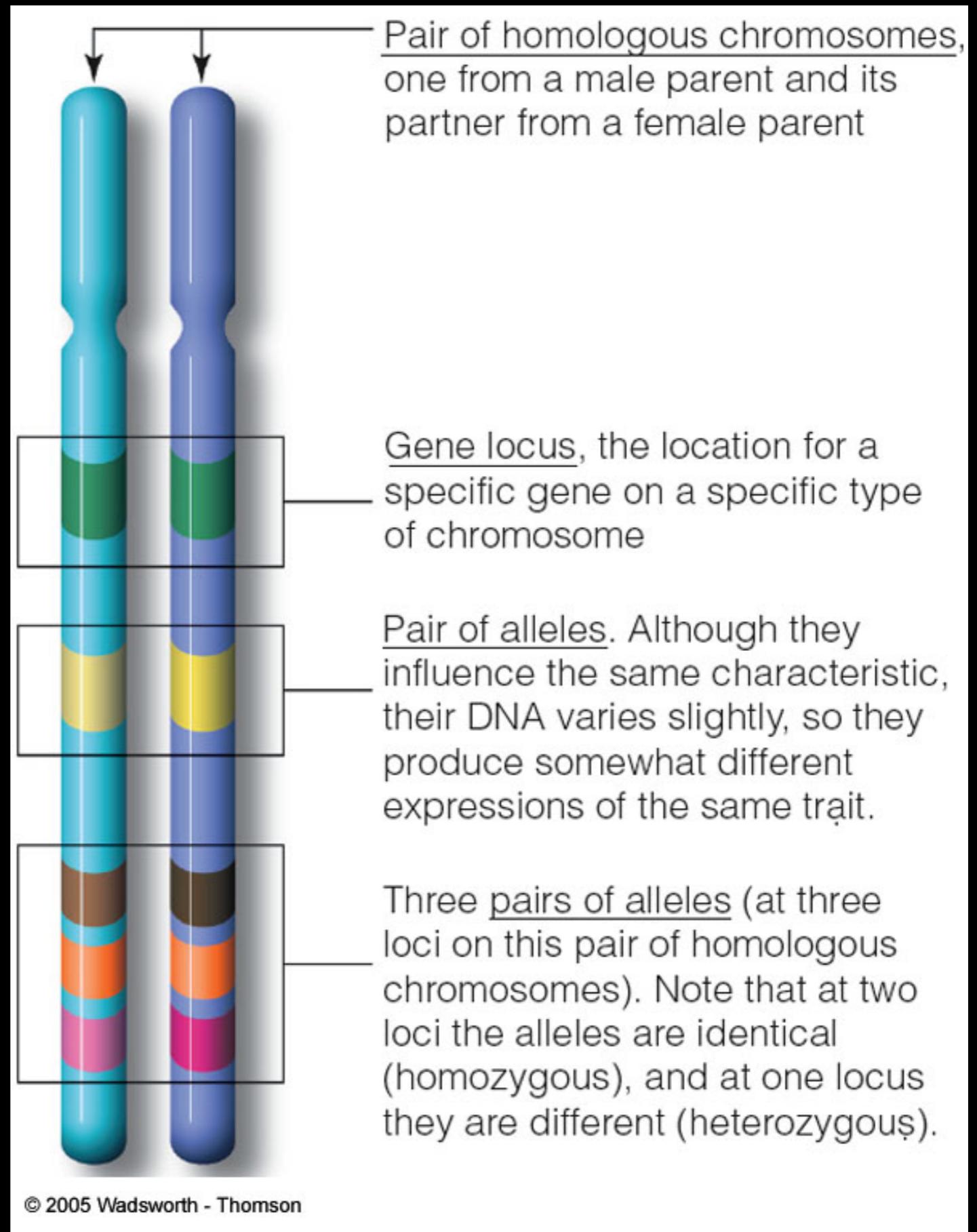phenotype
gene
allele
locus

DNA
chromosomes
bases

3

locus= the location of a gene on a chromosome

Allele= alternative form of a locus

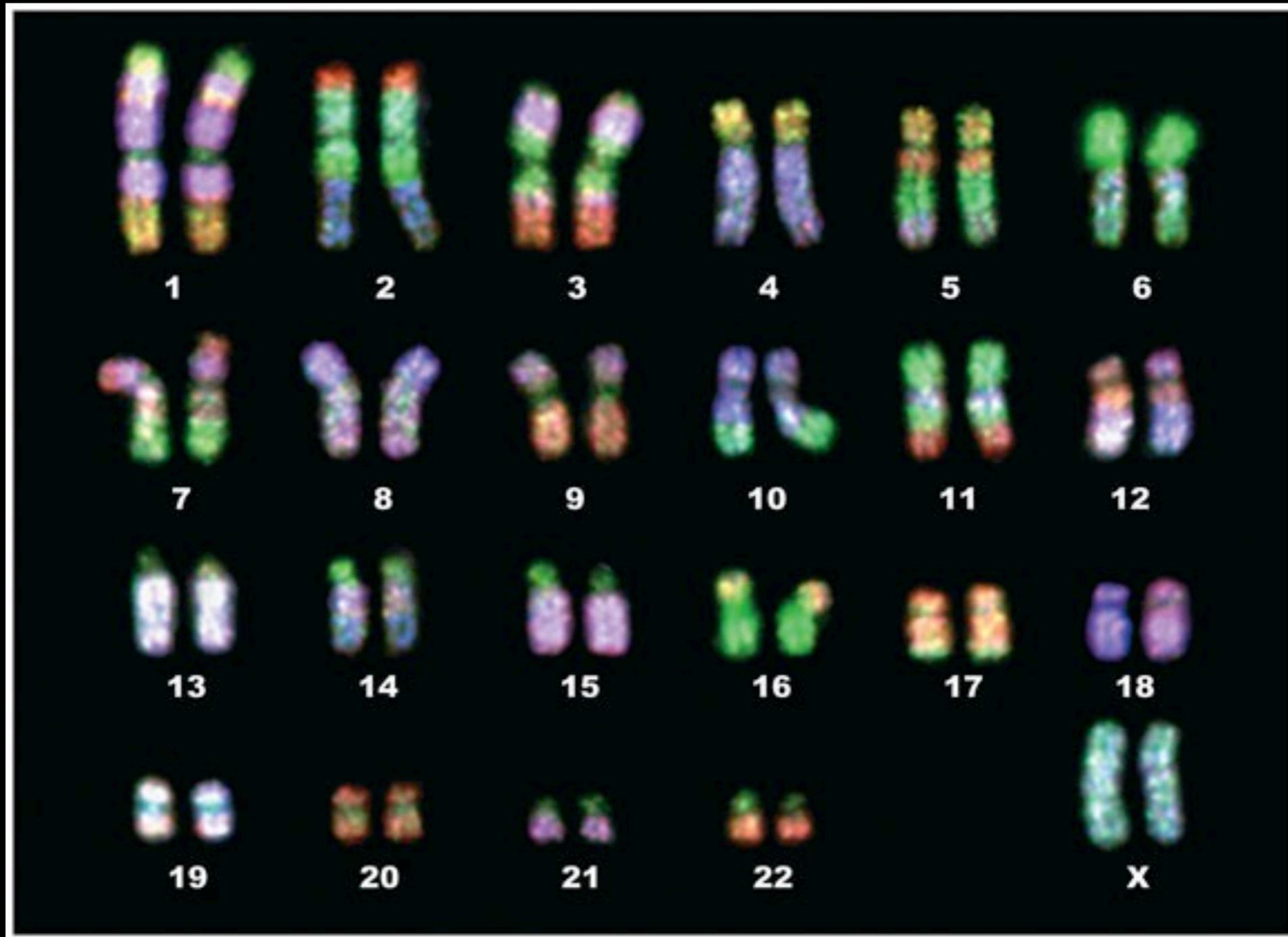homozygous= having the same allele at the locus on both chromosomes

heterozygous= having different alleles at the locus on both chromosomes

Pair of homologous chromosomes, one from a male parent and its partner from a female parent

Gene locus, the location for a specific gene on a specific type of chromosome

Pair of alleles. Although they influence the same characteristic, their DNA varies slightly, so they produce somewhat different expressions of the same trait.

Three pairs of alleles (at three loci on this pair of homologous chromosomes). Note that at two loci the alleles are identical (homozygous), and at one locus they are different (heterozygous).

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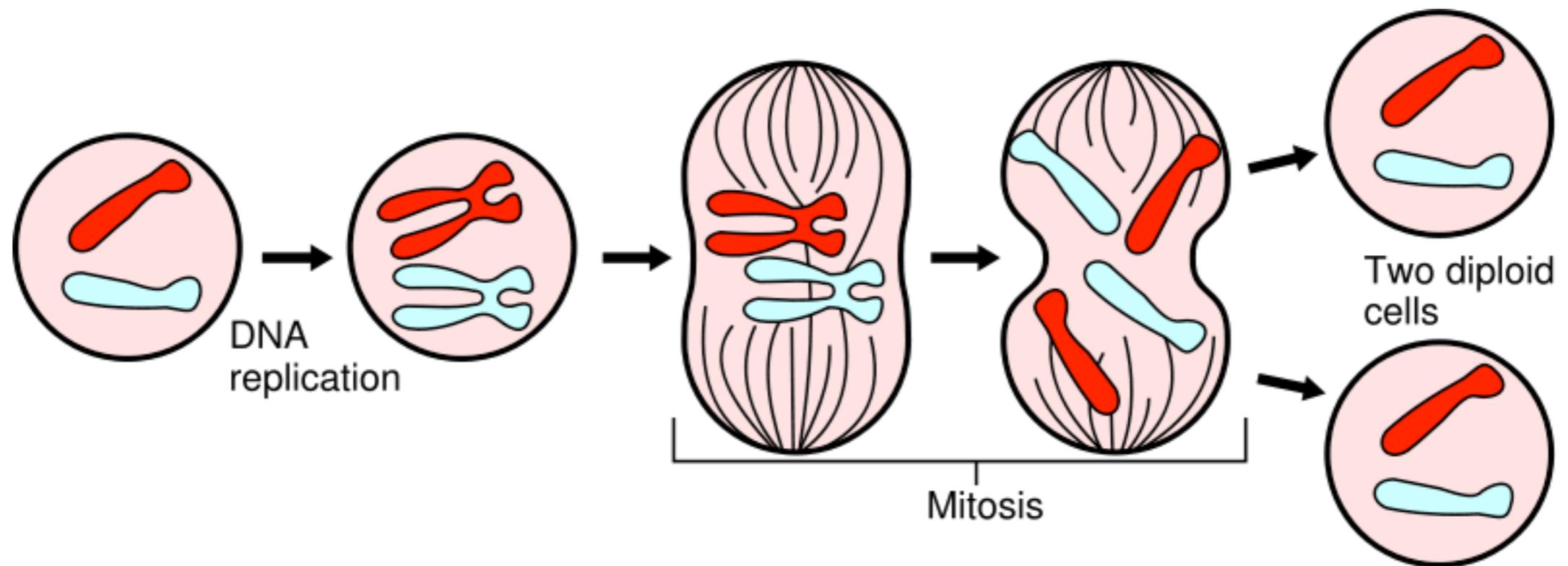
4

Heritability - a 3 Part question

- How come we resemble our parents? That is, **how is our heritable information passed from generation to generation?**

- **How does the genetic code create a characteristic?**

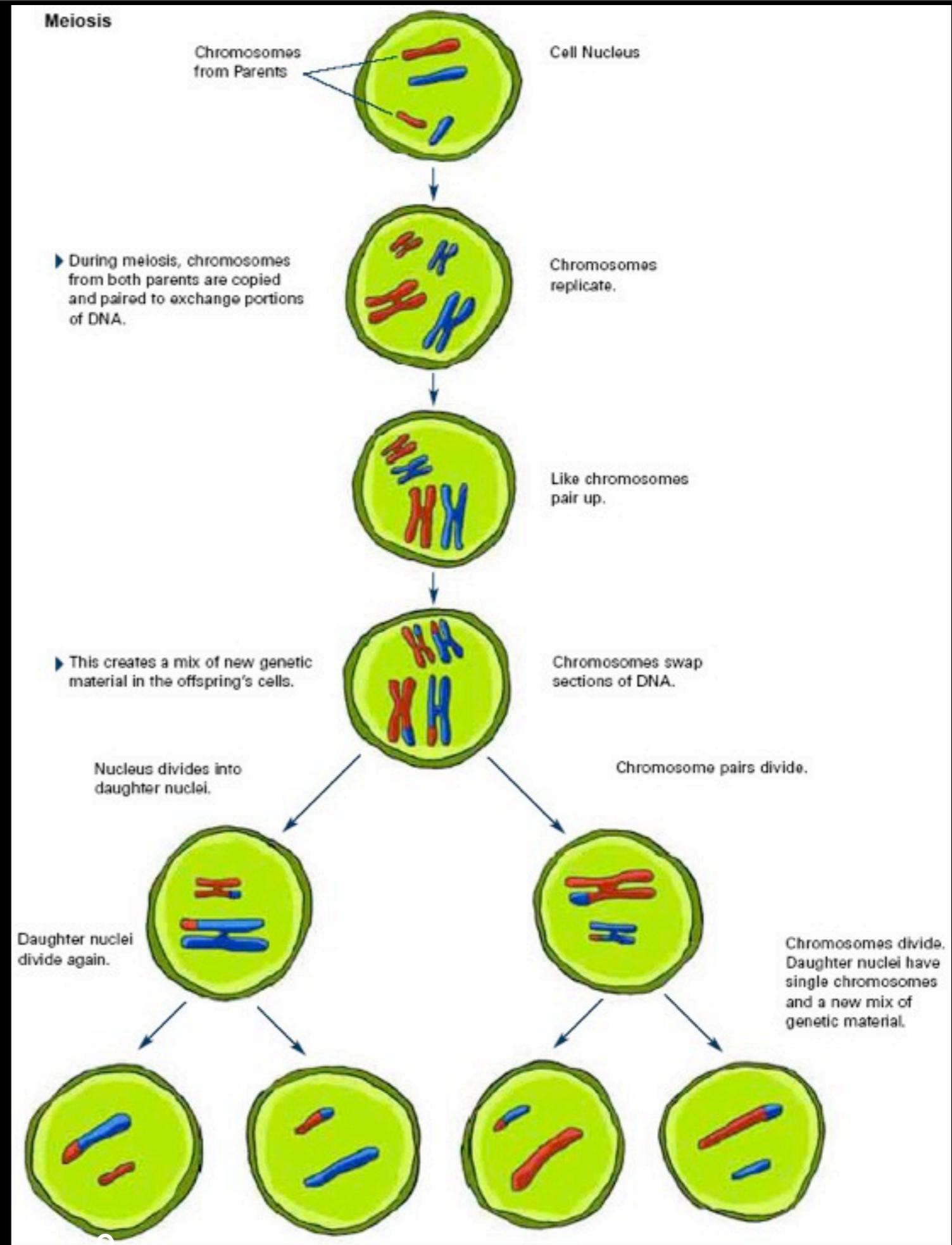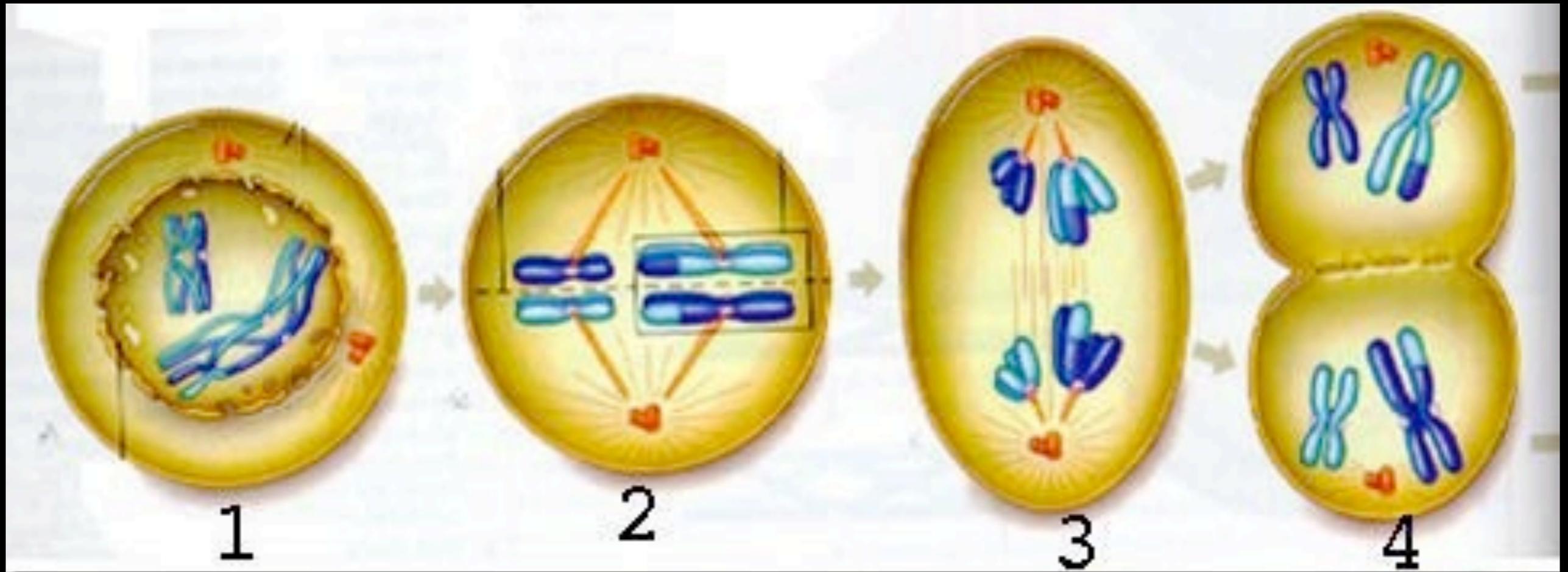- Where does variation in the code come from?

5

Human Karyotype

- **MITOSIS** - somatic cell division
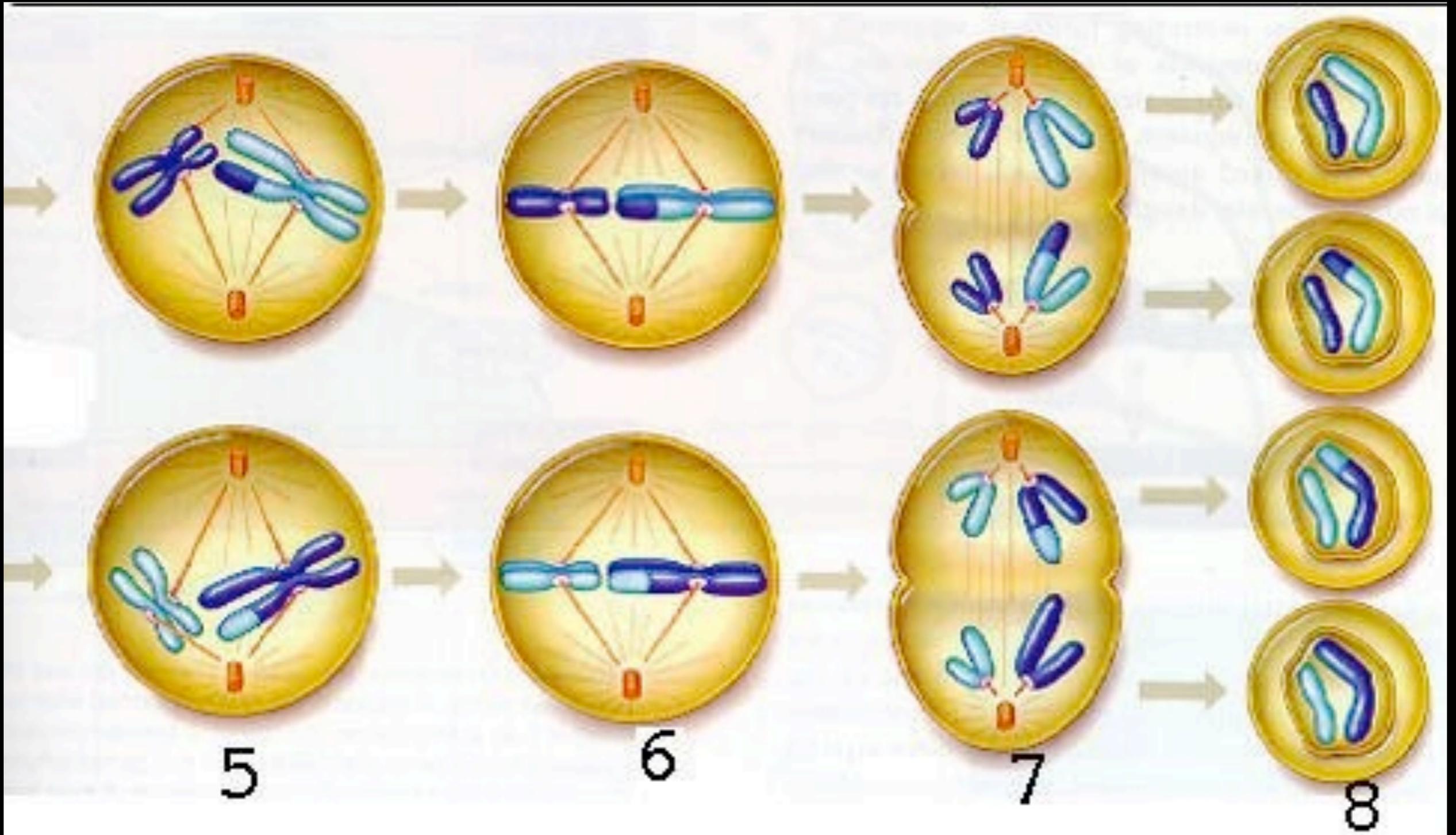
- **MEIOSIS** - gametic cell division

7

Mitosis

Meiosis

Meiosis

Chromosomes from Parents · Cell Nucleus

▶ During meiosis, chromosomes from both parents are copied and paired to exchange portions of DNA.

Chromosomes replicate.

Like chromosomes pair up.

▶ This creates a mix of new genetic material in the offspring's cells.

Chromosomes swap sections of DNA.

Nucleus divides into daughter nuclei.

Chromosome pairs divide.

Daughter nuclei divide again.

Chromosomes divide. Daughter nuclei have single chromosomes and a new mix of genetic material.

9

1   2   3   4

10

5

6

7

8

II

Heritability - a 3 Part question

- How come we resemble our parents? That is, **how is our heritable information passed from generation to generation?**

- How does the genetic code create a characteristic?

- Where does variation in the code come from?

12

Variation comes from

- Recombination
- Crossing Over
- Mutation

13

Recombination

2 chromosomes
x
2 possibilities for each
=
4 possible combinations

14

Recombination

23 chromosomes

x

2 possibilities for each

=

2 to the 23rd power

=

15

Recombination

23 chromosomes

x

2 possibilities for each

=

2 to the 23rd power
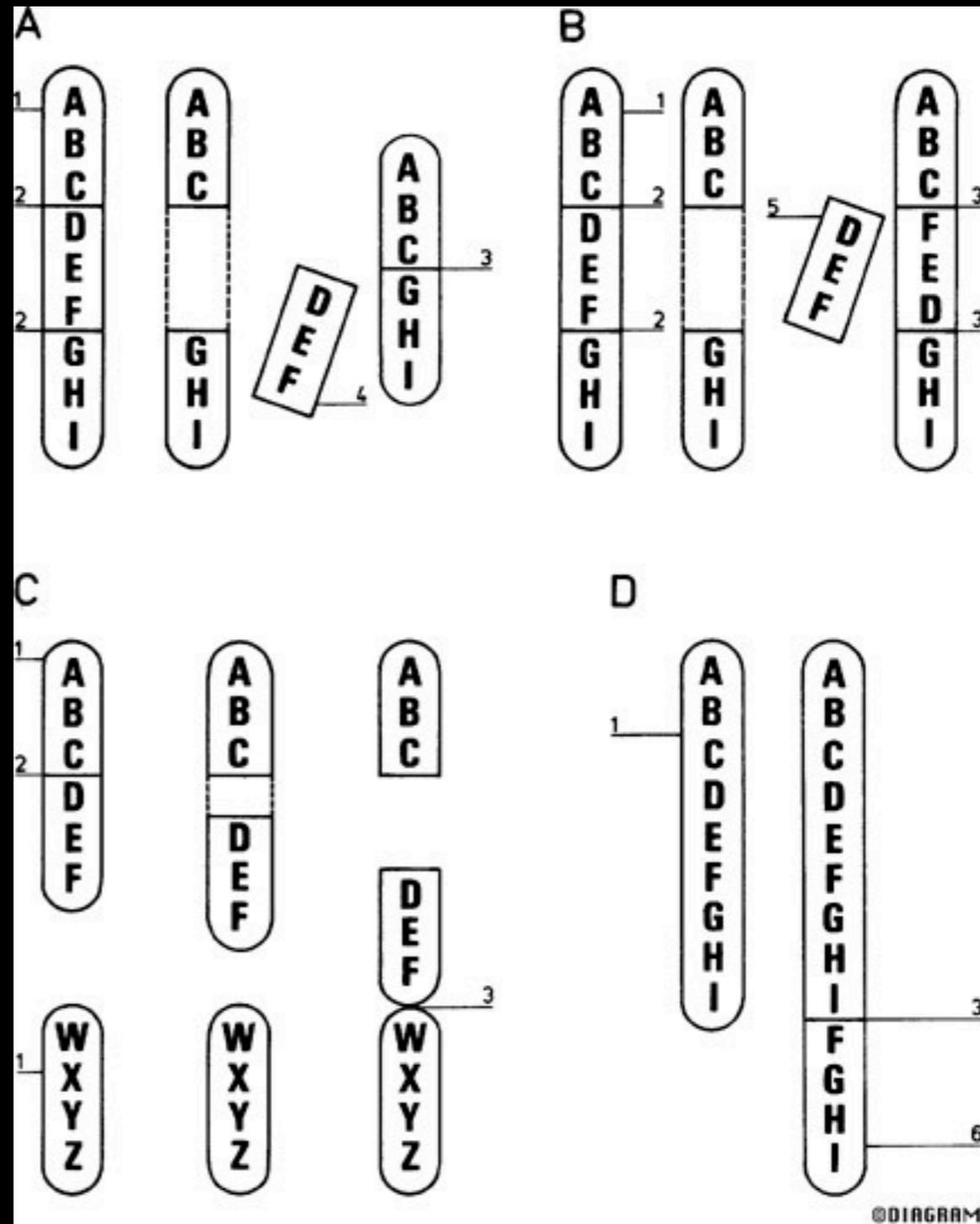
=

8,388,608

16

Variation comes from

- Recombination
- Crossing Over
- Mutation
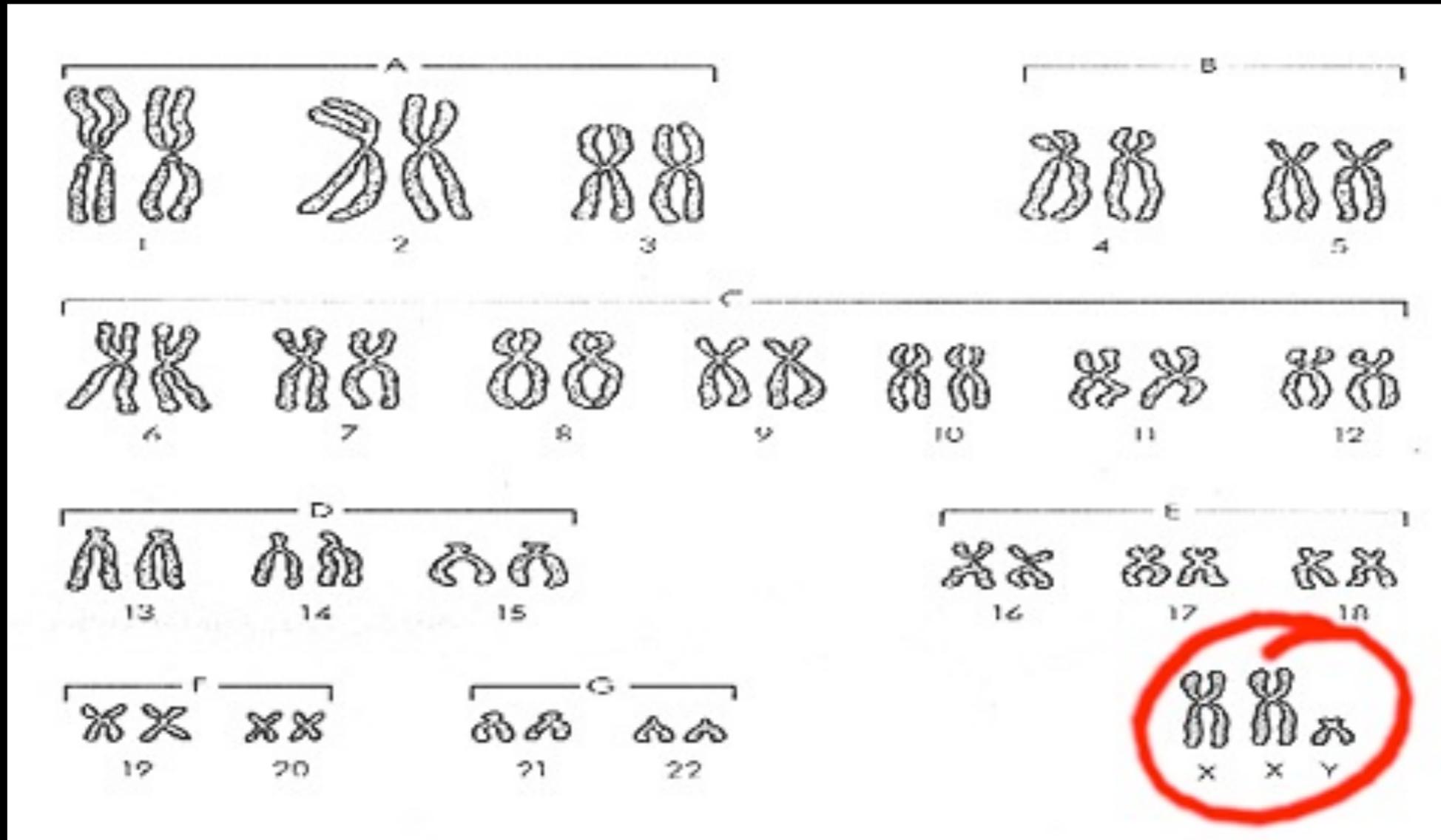
17

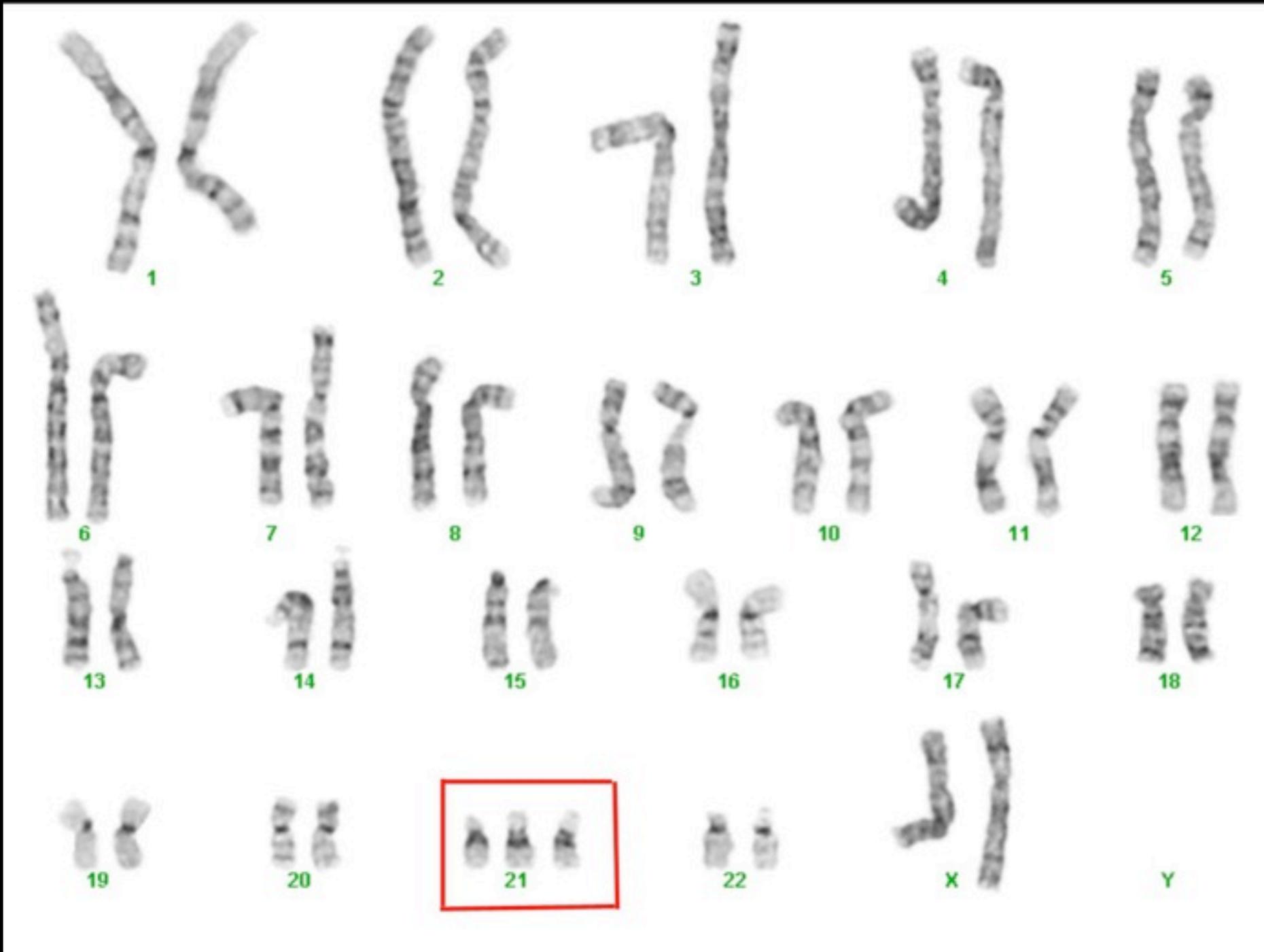Crossing over during meiosis

18
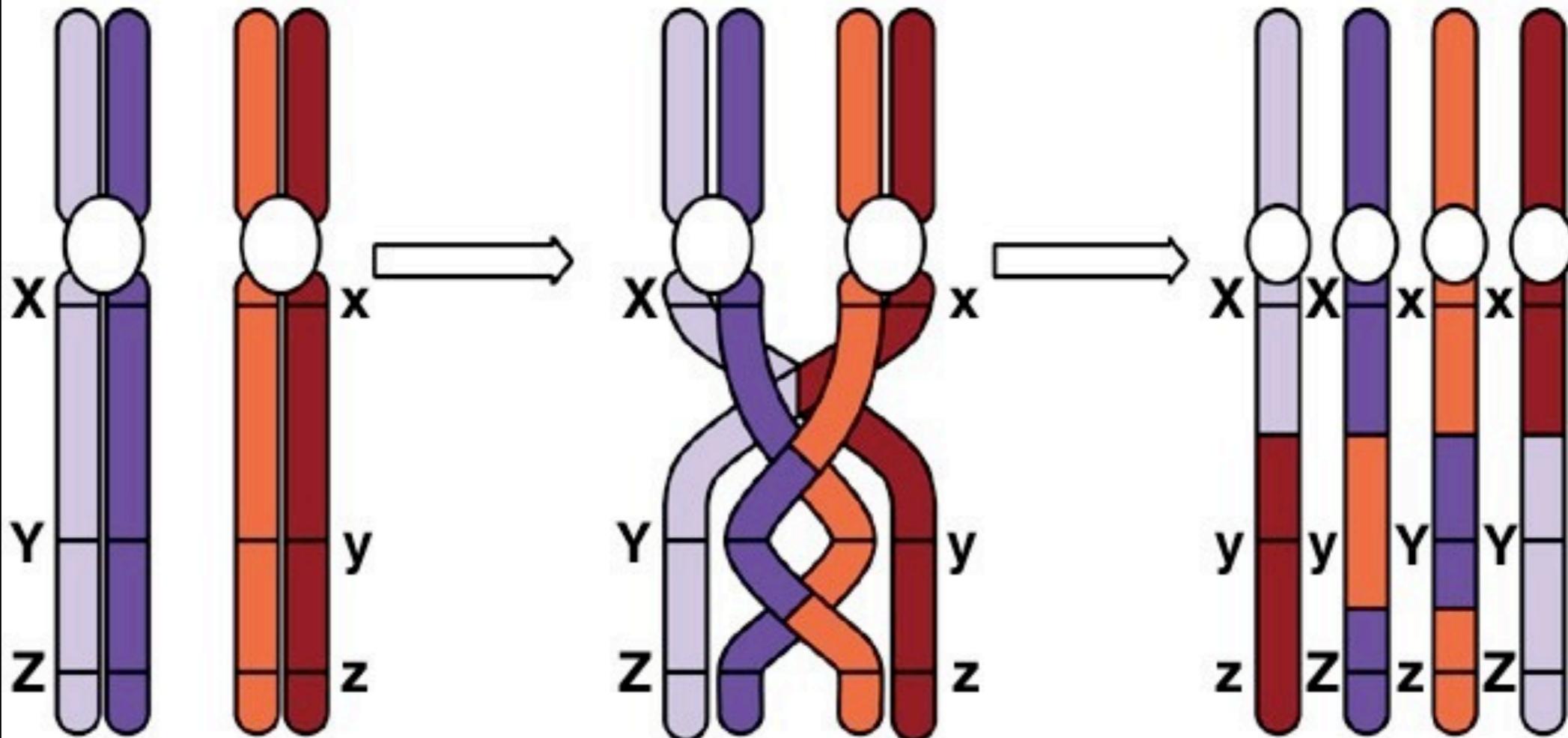
Chromosomal mutations

- Down's syndrome -21

- Klinefelter's syndrome -Sex
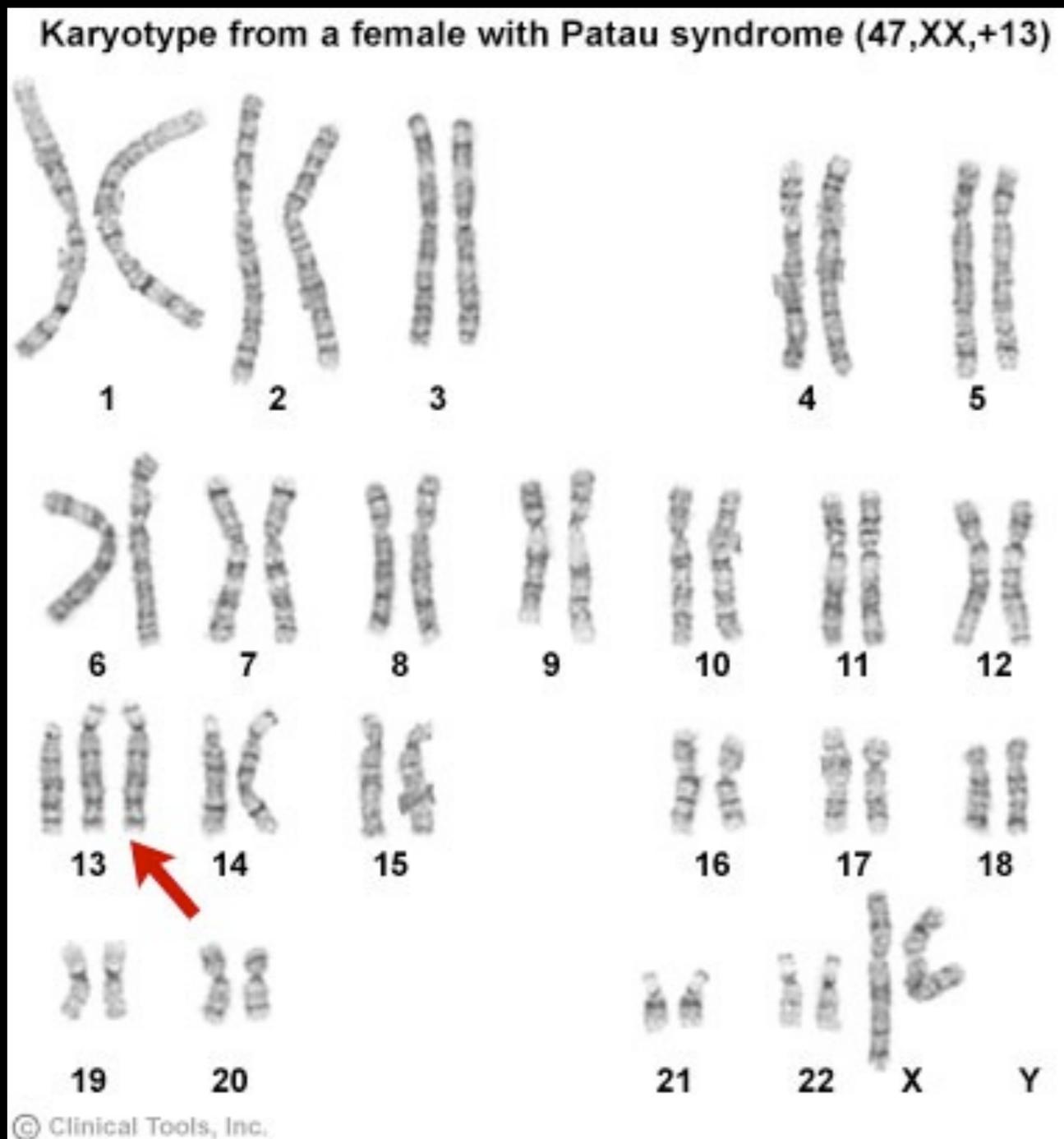
- Turner's syndrome - sex

- William's Syndrome - 7

Klinefelter's Karyotype

Down's Karyotype

Sunday, February 20, 2011

Trisomy 13

Karyotype from a female with Patau syndrome (47,XX,+13)

1 2 3 4 5
6 7 8 9 10 11 12
13 14 15 16 17 18
19 20 21 22 X Y
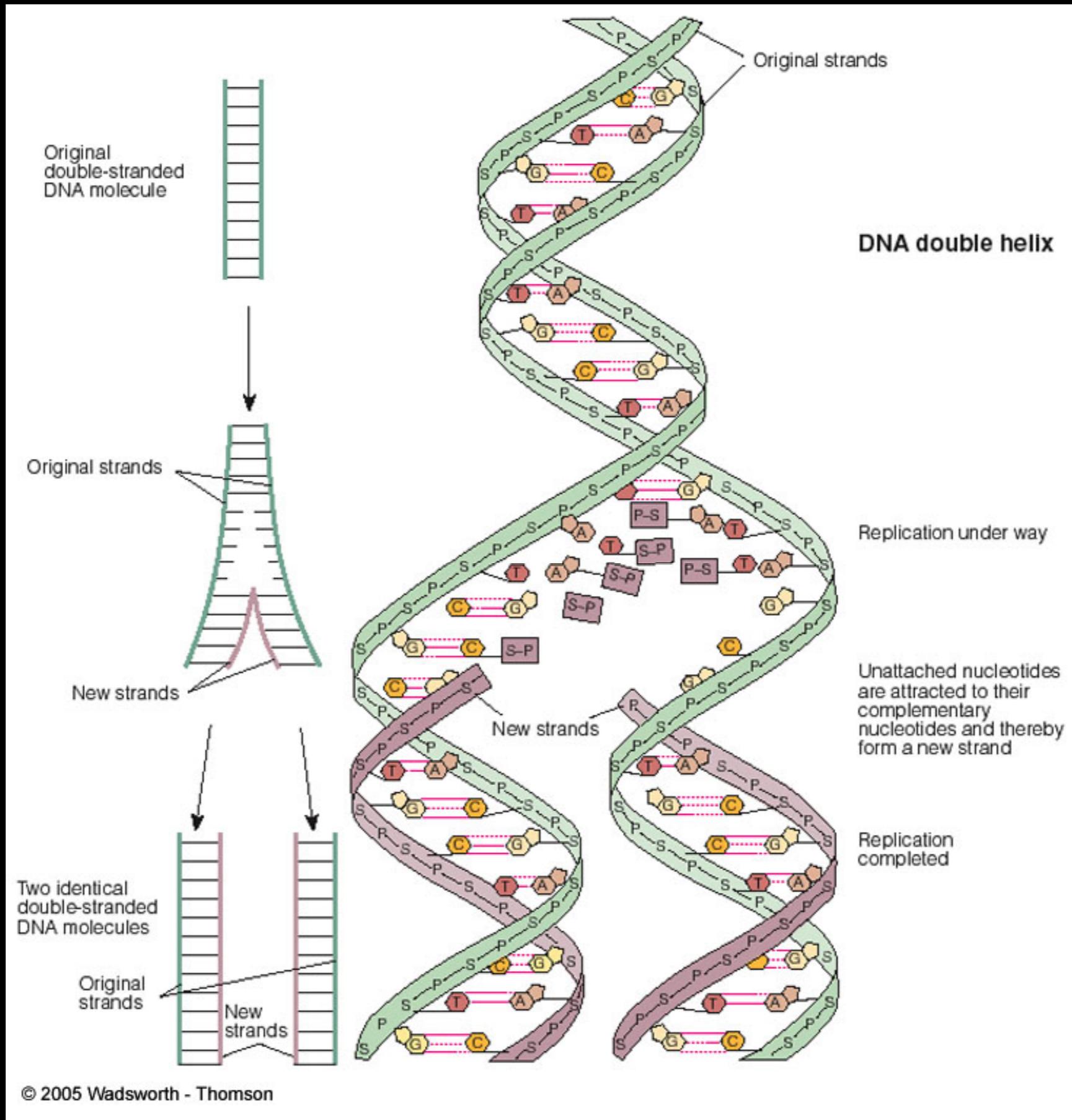
© Clinical Tools, Inc.

-small head
small eyes
cleft lip
ear shape
palm differences
extra fingers/toes
heart defects
kidney defects
etc.

Variation comes from

- Recombination
- Crossing Over
- Mutation

23

Mutation

- Change in base sequence of DNA

- Occurs during replication stage of meiosis (or mitosis)

- MAY change the amino acid change and therefore the protein

24

Original double-stranded DNA molecule

Original strands

New strands

Two identical double-stranded DNA molecules

Original strands

New strands

DNA double helix

Original strands

Replication under way

Unattached nucleotides are attracted to their complementary nucleotides and thereby form a new strand

New strands

Replication completed

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25

Kinds of Mutations

- Substitution - replace one base with another

- Frame Shift -

  - Insertion- an extra base gets pulled in

  - Deletion- a base gets omitted

How common is mutation?

- happens all the time

- assume a rate of one in a million per locus per gamete

- assume approximately 50,000 loci

- $(1 \times 10^{-6}) \times (5 \times 10^{4}) = 0.05$
  5% of gametes have a mutation

- an individual is combination of two gametes
  $2 \times 0.05 = 0.1 - 10\%$

27