

# Overview: Hereditary Similarity and Variation

- Living organisms
  - are distinguished by their ability to reproduce their own kind

# • Heredity

is the transmission of traits from one generation to the next

## Variation

 shows that offspring differ somewhat in appearance from parents and siblings



### Genetics

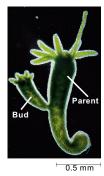
- is the scientific study of heredity and hereditary variation
- Offspring acquire genes from parents by inheriting chromosomes

### **Inheritance of Genes**

- Genes
  - are the units of heredity
  - are segments of DNA
- Each gene in an organism's DNA has a specific locus on a certain chromosome
- We inherit one set of chromosomes from our mother and one set from our father

# **Comparison of Asexual and Sexual Reproduction**

- In asexual reproduction
  - one parent produces genetically identical offspring by mitosis



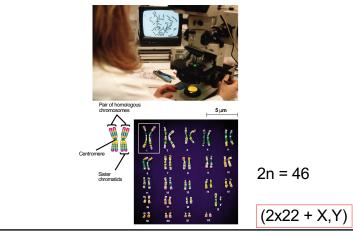
# In sexual reproduction Two parents give rise to offspring that have unique combinations of genes inherited from the two parents Fertilization and meiosis alternate in sexual life cycles A life cycle is the generation-to-generation sequence of stages in the reproductive history of an organism Sets of Chromosomes in Human Cells in humans each somatic (body) cell has 46 chromosomes, made up of two sets one set of chromosomes comes from each parent

# **Homologous Chromosomes**

- The two chromosomes, one from each of your parents, which carry the information for similar genes
- We get 23 from mom, 23 from dad

### A Karyotype

 is an ordered, visual representation of the chromosomes in a cell



# What is an Allele?

- Alleles are alternative forms of a gene
- One allele for a gene carried on one chromosome
  - Chromosome can have many, many genes
- Example:
  - If you have type AB blood, you got the A allele from one parent, the B allele from another

# **Diploid vs. Haploid**

- Most human somatic (body) cells have 46 chromosomes
  - This is called the diploid condition

-2n = 46 (2x22 + X,Y)

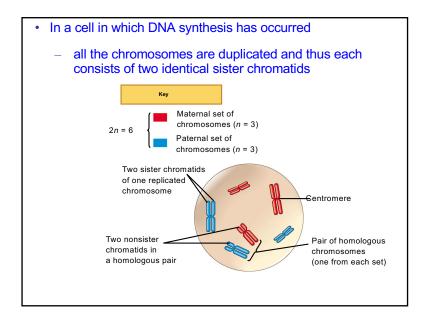
- Gametes (sperms and eggs) are an exception: they only have 23 chromosomes
  - This is the haploid condition
  - n = 23

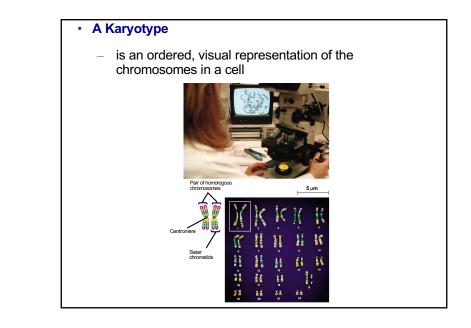
# **Trick Question**

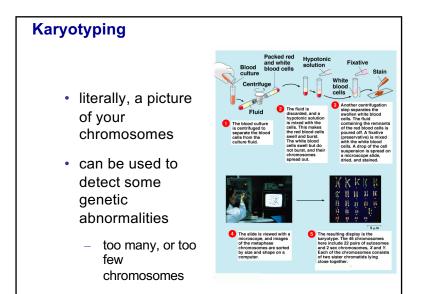
• We get 23 chromosomes from our mom, and 23 from our dad. Which 23 does each of us pass on to our children?

### Answer

- The 23 that we pass on to our offspring, are each a medley of the 23 pairs of chromosomes that we have.
- Hence, we pass on parts of both!
- Meiosis allows us to do this







- Homologous chromosomes
  - are the two chromosomes composing a pair
  - have the same characteristics
  - may also be called autosomes
- Sex chromosomes
  - are distinct from each other in their characteristics
  - are represented as X and Y
  - determine the sex of the individual, XX being female, XY being male
- A diploid cell
  - has two sets of each of its chromosomes
  - in a human has 46 chromosomes (2n = 46)

# Trisomy 21: Down's syndrome

- 3 copies of chromosome #21 present
- Many symptoms
  - Round face
  - Flattened nose
  - Small, irregular teeth
  - Short stature
  - Heart defects
  - Respiratory infection
  - Leukemia
  - Alzheimer's disease (APP on chr21) / similar symptoms

# XYY

- Extra Y chromosome
- Often taller
- Produces an otherwise normal male

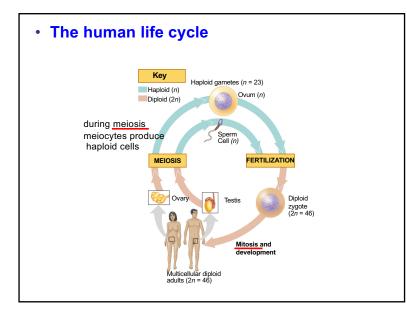
# Metafemale

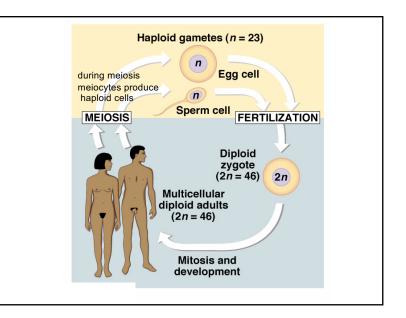
- XXX
- · Reduced fertility
- Otherwise normal female

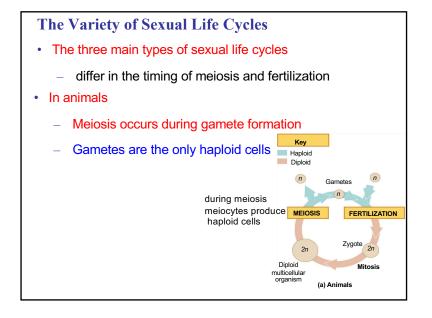
- Unlike somatic cells
  - gametes, sperm and egg cells are haploid cells, containing only one set of chromosomes

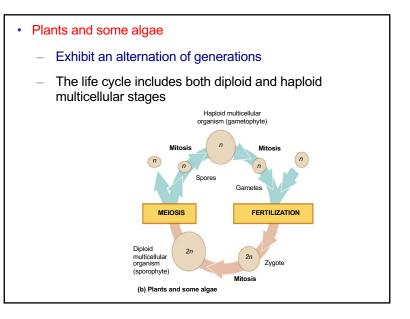
Behavior of Chromosome Sets in the Human Life Cycle

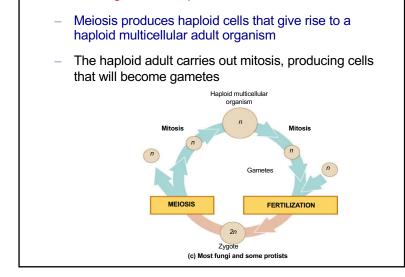
- At sexual maturity
  - the ovaries and testes produce haploid gametes by meiosis
- During fertilization
  - these gametes, sperm and ovum, fuse, forming a diploid zygote
- the zygote
  - develops into an adult organism











# Meiosis Basic Concepts

- Two things achieved:
  - Recombines the chromosomes your parents gave you
  - Produces haploid gamete cells, having only 23 chromosomes

# **Two Parts of Meiosis**

• In most fungi and some protists

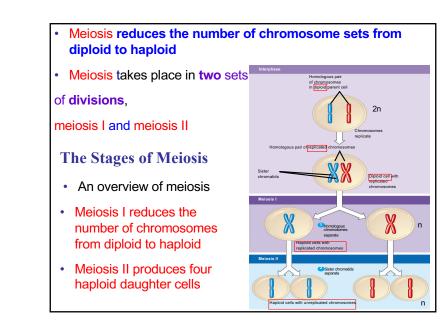
- Meiosis I
- Meiosis II
- Both are similar to, but also different from mitosis (---> see cell cycle, cell proliferation)

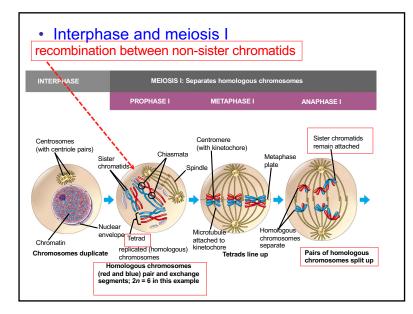
# **Meiosis** I

- This step reduces chromosomes
  - Starts with diploid (2n = 46) cell
  - Produces two haploid (n = 23) cells
- · Also is where recombination occurs

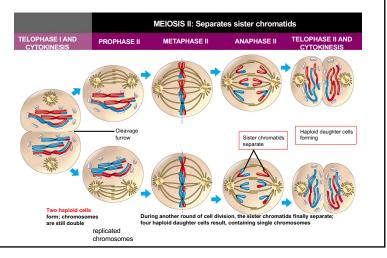
# **Meiosis II**

- · Starts with two haploid cells
- Produces four haploid cells
  - These are 4 sperms or 4 eggs







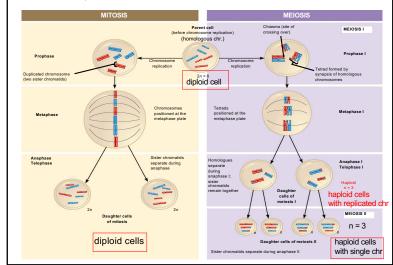


# A Comparison of Mitosis and Meiosis

- Meiosis and mitosis can be distinguished from mitosis
  - by three events in Meiosis I
- Synapsis and crossing over
  - Homologous chromosomes physically connect and exchange genetic information
- Tetrads on the metaphase plate
  - at metaphase I of meiosis, paired homologous chromosomes (tetrads) are positioned on the metaphase plates
- Separation of homologues: at anaphase I of meiosis, homologous pairs move toward opposite poles of the cell; in anaphase II of meiosis, the sister chromatids separate

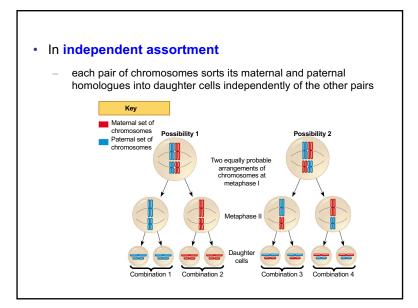
Roy	w Mitosis	Meiosis
	In somatic cells	In cells in the sexual cycle
۱	One cell division, resulting in two daughter cells	Two cell divisions, resulting in four Meiocyte $\longrightarrow \longrightarrow \longrightarrow$
2	$\begin{array}{c} \text{Chromosome number} \\ \text{per nucleus maintained} \\ (e.g., \text{for a diploid cell}) \end{array} \qquad \begin{array}{c} 2n \\ \hline 2n \\ \hline 2n \end{array}$	$\begin{array}{c} \text{Chromosome number} \\ \text{halved in the products} \\ \text{of meiosis} \end{array} \xrightarrow{(n)} \begin{array}{c} 0 \\ 0 \end{array} \xrightarrow{(n)} \begin{array}{c} 0 \\ 0 \end{array} \xrightarrow{(n)} \begin{array}{c} 0 \\ 0 \end{array}$
3	One premitotic Sphase per cell division	Ons premeiotic Sphase for both cell divisions
4	Normally, no pairing of homologous chromosomes in prophase Chromosome No Chromosome tetrads	Full synapsis of of homologous chromosomes in prophase
5	Normally, norecombination in prophase	At least one recombination between nonsister chromatids
6	Bi-oriented sister kinetochores	Co-orientation of sister kinetochores
7	Loss of cohesion between sister chromatid arms during metaphase	Maintenance of cohesion between sister chromatid arms during metaphase of meioses I
8	Centromeres divide at anaphase	Centromeres do not divide at anaphase I but do at anaphase II
	Conservative process: daughter cells' genotypes identical with parental genotype	Promotes variation among the products of meiosis
	Cell undergoing mitosis can be diploid or haploid	Cell undergoing meiosis is diploid

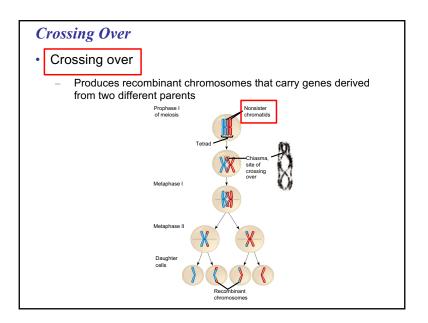
• A comparison of mitosis and meiosis



# **Independent Assortment of Chromosomes**

- Homologous pairs of chromosomes
  - orient randomly at metaphase I of meiosis





### **Random Fertilization**

• the fusion of gametes will produce a zygote with any of about 64 trillion diploid combinations

### **Evolutionary Significance of Genetic Variation Within Populations**

- · Genetic variation
  - is the raw material for evolution by natural selection
- Mutations
  - are the original source of genetic variation
- Sexual reproduction
  - produces new combinations of variant genes, adding more genetic diversity

- Genetic variation produced in sexual life cycles contributes to 'evolution'
- Reshuffling of genetic material in meiosis
  - produces genetic variation

# Origins of Genetic Variation Among Offspring

- In species that produce sexually
  - the behavior of chromosomes during meiosis and fertilization is responsible for most of the variation that arises each generation

# Taking home message: Epigenetics

In biology, and specifically genetics, epigenetics is the study of inherited changes in phenotype (appearance) or gene expression caused by mechanisms other than changes in the underlying DNA sequence, hence the name epi- (Greek:  $\epsilon \pi forer, above$ ) -genetics. These changes may remain through cell divisions for the remainder of the cell's life and may also last for multiple generations. However, there is no change in the underlying DNA sequence of the organism; instead, non-genetic factors cause the organism's genes to behave or express themselves differently.

The hypermethylation of CpG islands in the promoter regions of tumor suppressor genes is a common hallmark of human cancer, and it is associated with inactivation of these genes.

p60TRP is particularly interesting due to its location on the X chromosome (Xq23), one copy of which is randomly inactivated by DNA methylation in females.