

PowerLecture:
Chapter 12

Chromosomes and Human Inheritance
Section 12.0: Weblinks and InfoTrac

See the **latest Weblinks and InfoTrac articles** for this chapter online or click **highlighted articles below (articles subject to change)**

- Section 12.0: Nobel Prize, Economics, 1994—John F. Nash, Jr. et al
- Section 12.0: Equilibrium, Mental and Mathematical. Jeremy Bernstein. *Commentary*, Aug. 1998.

How Would You Vote?

The following is the question for this chapter. See national results below.

Would you support regulation of diagnostic genetic tests in order to protect individual privacy?

Impacts, Issues: Strange Genes, Richly Tortured Minds

- Schizophrenia is a neurobiological disorder
- Symptoms: delusions, hallucinations, disorganized speech, abnormal social behavior

Impacts, Issues: Strange Genes, Richly Tortured Minds

- Emotionally healthy people who show creative brilliance tend to have personality traits in common with the mentally impaired
- Altered gene expression in certain brain regions may boost creativity as well as invite illness

Section 12.1: Weblinks and InfoTrac

See the **latest Weblinks and InfoTrac articles** for this chapter online or click **highlighted articles below (articles subject to change)**

- Section 12.1: Human Genome Landmarks
Chromosomes & Cancer
- Some genes on chromosomes control cell growth and division
- If something affects chromosome structure at or near these loci, cell division may spiral out of control

- This can lead to cancer

Philadelphia Chromosome

- First abnormal chromosome to be associated with a cancer
- Reciprocal translocation
- Causes chronic myelogenous leukemia (CML)

Genes

- Units of information about heritable traits
- In eukaryotes, distributed among chromosomes
- Each has a particular locus
 - Location on a chromosome

Genes

Homologous Chromosomes

- Homologous autosomes are identical in length, size, shape, and gene sequence
- Sex chromosomes are nonidentical but still homologous
- Homologous chromosomes interact, then segregate from one another during meiosis

Alleles

- Different molecular forms of a gene
- Arise through mutation
- Diploid cell has a pair of alleles at each locus
- Alleles on homologous chromosomes may be same or different

Sex Chromosomes

- Discovered in late 1800s
- Mammals, fruit flies
 - XX is female, XY is male
- In other groups XX is male, XY female
- Human X and Y chromosomes function as homologues during meiosis

Section 12.2: Weblinks and InfoTrac

See the latest Weblinks and InfoTrac articles for this chapter online or click highlighted articles below (articles subject to change)

- Section 12.2: Cytogenetics Gallery
- Section 12.2: Photographing Chromosomes—The Colored Bodies. *PSA Journal*, Dec. 1998.

Karyotype Preparation - Stopping the Cycle

- Cultured cells are arrested at metaphase by adding colchicine
- This is when cells are most condensed and easiest to identify

Karyotype Preparation

- Arrested cells are broken open
- Metaphase chromosomes are fixed and stained

- Chromosomes are photographed through microscope
- Photograph of chromosomes is cut up and arranged to form karyotype diagram

Section 12.3: Weblinks and InfoTrac

See the **latest Weblinks** and **InfoTrac articles** for this chapter online or click **highlighted articles below (articles subject to change)**

- Section 12.3: Your Genes, Your Health
- Section 12.3: Genetics and Rare Diseases
- Section 12.3: Parents of Galactosemic Children
- Section 12.3: Manic-Depression Findings Spark Polarized Debate. Virginia Morell. *Science*, April 5, 1996.

Autosomal Dominant Inheritance

Trait typically appears in every generation

Huntington Disorder

- Autosomal dominant allele
- Causes involuntary movements, nervous system deterioration, death
- Symptoms don't usually show up until person is past age 30
- People often pass allele on before they know they have it

Achondroplasia

- Autosomal dominant allele
- In homozygous form usually leads to stillbirth
- Heterozygotes display a type of dwarfism
- Have short arms and legs relative to other body parts

Autosomal Recessive Inheritance Patterns

- If parents are both heterozygous, child will have a 25% chance of being affected

Galactosemia

- Caused by autosomal recessive allele
- Gene specifies a mutant enzyme in the pathway that breaks down lactose

Section 12.4: Weblinks and InfoTrac

See the **latest Weblinks** and **InfoTrac articles** for this chapter online or click **highlighted articles below (articles subject to change)**

- Section 12.4: One Week of Joy (photo essay)

- Section 12.4: The Premature Aging Syndromes: Insights into the Aging Process.
Christopher Dyer et al. *Age and Aging*, Jan. 1998.

Hutchinson-Gilford Progeria

- Mutation causes accelerated aging
- No evidence of it running in families
- Appears to be dominant
- Seems to arise as spontaneous mutation
- Usually causes death in early teens

Section 12.5: Weblinks and InfoTrac

See the **latest Weblinks** and **InfoTrac articles** for this chapter online or click **highlighted articles below (articles subject to change)**

- Section 12.5: HHMI BioInteractive: The Meaning of Sex—Genes and Gender
- Section 12.5: Talking about Sex: Biology and the Social Interpretations of Sex in Sport. Sandra Kirby et al. *Canadian Woman Studies*, Winter–Spring 2002.

The Y Chromosome

- Fewer than two dozen genes identified
- One is the master gene for male sex determination
 - SRY gene (sex-determining region of Y)
- SRY present, testes form
- SRY absent, ovaries form

The X Chromosome

- Carries more than 2,300 genes
- Most genes deal with nonsexual traits
- Genes on X chromosome can be expressed in both males and females

Section 12.6: Weblinks and InfoTrac

See the **latest Weblinks** and **InfoTrac articles** for this chapter online or click **highlighted articles below (articles subject to change)**

- Section 12.6: Thomas Hunt Morgan and His Legacy

Thomas Morgan

- Embryologist
- *Drosophila*, fruit fly's X chromosome has the gene for eye color and another gene for body color (*p193*)

Thomas Morgan

- *D. melanogaster*
- Reciprocal crosses

Discovering Linkage

Section 12.7: Weblinks and InfoTrac

See the latest Weblinks and InfoTrac articles for this chapter online or click highlighted articles below (articles subject to change)

- Section 12.7: Case Study—Hemophilia
- Section 12.7: Not Seeing Red (or Blue or Green). Elia T. Ben-Ari. *BioScience*, Aug. 2000.

X-Linked Recessive Inheritance

- Males show disorder more than females
- Son cannot inherit disorder from his father

Examples of X-Linked Traits

- Color blindness
 - Inability to distinguish among some of all colors
- Hemophilia
 - Blood-clotting disorder
 - 1/7,000 males has allele for hemophilia A
 - Was common in European royal families

Color Blindness

Fragile X Syndrome

- An X-linked recessive disorder
- Causes mental retardation
- Mutant allele for gene that specifies a protein required for brain development
- Allele has repeated segments of DNA

Section 12.8: Weblinks and InfoTrac

See the latest Weblinks and InfoTrac articles for this chapter online or click highlighted articles below (articles subject to change)

- Section 12.8: Introduction to Chromosome Abnormalities
- Section 12.8: Which of Our Genes Make Us Human (comparison of primate DNA). Ann Gibbons. *Science*, Sept. 4, 1998.

Duplication

- Gene sequence that is repeated several to hundreds of times

- Duplications occur in normal chromosomes
- May have adaptive advantage
 - Useful mutations may occur in copy
- Karyotype of duplicated chromosomes of the gibbon.

Duplication

Deletion

- Loss of some segment of a chromosome
- Most are lethal or cause serious disorder

Deletion

- Cri-du-chat

Inversion

A linear stretch of DNA is reversed
within the chromosome

Translocation

- A piece of one chromosome becomes attached to another nonhomologous chromosome
- Most are reciprocal
- Philadelphia chromosome arose from a reciprocal translocation between chromosomes 9 and 22

Translocation

Chromosome Structure

- Alterations to chromosome structure are usually bad
- Duplications are adaptive: one gene functions normally - the other is free to mutate
- Chromosome structure evolves
- Human body cells have 23 pairs of chromosomes
- Chimpanzees, gorilla, or orangutan body cells have twenty-four pairs of chromosomes
- During human evolution, two chromosomes in an early ancestor fused to form chromosome 2 (p197)

Chromosome Structure

Section 12.9: Weblinks and InfoTrac

See the latest Weblinks and InfoTrac articles for this chapter online or click highlighted articles below (articles subject to change)

- Section 12.9: National Down's Syndrome Society

- Section 12.9: Turner Syndrome Clinical Studies
- Section 12.9: Chromosomal Mosaicism
- Section 12.9: Life as We Know It: A Father, a Son, and Genetic Destiny. Michael Berube. Harper's Magazine, Dec. 1994.

Aneuploidy

- Individuals have one extra or less chromosome
- ($2n + 1$ or $2n - 1$)
- Major cause of human reproductive failure
- Most human miscarriages are aneuploids

Polyploidy

- Individuals have three or more of each type of chromosome ($3n, 4n$)
- Common in flowering plants
- Lethal for humans
 - 99% die before birth
 - Newborns die soon after birth

Nondisjunction

Down Syndrome

- Trisomy of chromosome 21
- Mental impairment and a variety of additional defects
- Can be detected before birth
- Risk of Down syndrome increases dramatically in mothers over age 35

Turner Syndrome

- Inheritance of only one X (XO)
- 98% spontaneously aborted
- Survivors are short, infertile females
 - No functional ovaries
 - Secondary sexual traits reduced
 - May be treated with hormones, surgery

Klinefelter Syndrome

- XXY condition
- Results mainly from nondisjunction in mother (67%)
- Phenotype is tall males
 - Sterile or nearly so
 - Feminized traits (sparse facial hair, somewhat enlarged breasts)
 - Treated with testosterone injections

XYY Condition

- Taller than average males
- Most otherwise phenotypically normal
- Some mentally impaired
- Once thought to be predisposed to criminal behavior, but studies now discredit

Section 12.10: Weblinks and InfoTrac

See the latest Weblinks and InfoTrac articles for this chapter online or click highlighted articles below (articles subject to change)

- Section 12.10: Almanach de Gotha
- Section 12.10: The USGenWeb Project
- Section 12.10: Must Ask Your Mom Today. Karen Bannan. *Redbook*, May 2003.
- Section 12.10: Charting the Future? Eileen Beal. *American Visions*, Oct.–Nov. 1994.

Pedigree

Symbols

Pedigree for Polydactyly

Genetic Abnormality

- A rare, uncommon version of a trait
- Polydactyly
 - Unusual number of toes or fingers
 - Does not cause any health problems
 - View of trait as disfiguring is subjective

Genetic Disorder

- Inherited conditions that cause mild to severe medical problems
- Why don't they disappear?
 - Mutation introduces new rare alleles
 - In heterozygotes, harmful allele is masked, so it can still be passed on to offspring

Genetic Disorders and Genetic Abnormalities

Section 12.11: Weblinks and InfoTrac

See the latest Weblinks and InfoTrac articles for this chapter online or click highlighted articles below (articles subject to change)

- Section 12.11: PBS—Our Genes, Our Choices
- Section 12.11: The DNA Files
- Section 12.11: National Society of Genetic Counselors
- Section 12.11: Elvis' DNA: The Gene as a Cultural Icon. Dorothy Nelkin et al. *The Humanist*, May–June 1995.

- Section 12.11: The Icelandic Database—Do Modern Times Need Modern Sagas? Ruth Chadwick. *British Medical Journal*, Aug. 14, 1999.

Phenotypic Treatments

- Symptoms of many genetic disorders can be minimized or suppressed by
 - Dietary controls
 - Adjustments to environmental conditions
 - Surgery or hormonal treatments

Genetic Screening

- Large-scale screening programs detect affected persons
- Newborns in United States routinely tested for PKU
 - Early detection allows dietary intervention and prevents brain impairment

Prenatal Diagnosis

- Amniocentesis
- Chorionic villus sampling
- Fetoscopy
- All methods have some risks

Preimplantation Diagnosis

- Used with in-vitro fertilization
- Mitotic divisions produce ball of 8 cells
- All cells have same genes
- One of the cells is removed and its genes analyzed
- If cell has no defects, the embryo is implanted in uterus

Preimplantation Diagnosis