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Chromosome Theory and Human Genetics

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The road to understanding inheritance

- Sexual reproduction was understood for a long time
  (egg + sperm -> zygote)
- Did the egg and sperm have about equal influence?
- The nuclei of the egg and sperm were roughly equal so…
- Could the nucleus hold the genetic material?

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The road to understanding inheritance

- August Weismann-first to state that the genetic material **must be divided** in half whenever the egg and sperm are formed

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- Theodore Boveri – was the first to observe meiosis in Ascaris (roundworm)

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- Walter Sutton-stated that chromosomes held smaller particles called genes and each gene accounted for a single trait

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The road to understanding inheritance

- Thomas Hunt Morgan - discovered sex chromosomes
- He worked with the famous fruit fly known as ***Drosophila melanogaster*** *
- ***We also learned about X linked traits – traits located on the X chromosome*** *

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The road to understanding inheritance

- Crossing over - when a piece of DNA (a gene or more) on one chromosome exchanges with a piece of DNA on another chromosome

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The road to understanding inheritance

- Crossing over

- If two genes are located close together on a chromosome, the likelihood that a recombination event will separate these two genes is less than if they were farther apart
- Farther apart means more crossover events

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The road to understanding inheritance

- A chromosome map can be made by using crossing over frequencies this map shows the position of genes (gene linkage) on the chromosome
- Genetic maps are lines or circles with marks indicating the relative positions of genetic markers.
- Genetic markers are genetically determined traits or characters

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The road to understanding inheritance

- Gene - a series of bases or nucleotides in the DNA molecule that encodes for the amino acid sequence of a particular trait * (1 trait)

- One human compared other humans and other species differ from one another in the sequence of their DNA bases *

11 ▨ **The road to understanding inheritance**

- Every cell in your body (except RBC's and gametes) have 100% of your genes
- The expression of each gene is controlled by:
  - Hormones
  - Enzymes
  - Specific cell environment

12 ▨ **The road to understanding inheritance**

- Temperature can influence the effect of genes
  - Himalayan rabbit - fur turns dark in cold climate for warmth
  - Drosophila - curly wings at a 25ºC temperature

13 ▨ **The road to understanding inheritance**

- Mutation - a sudden change in a genetic trait

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1 - Types of mutations:
  - Somatic mutation - mutation in a non-sex cell

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  - Germ mutation - mutation in the sex-cells

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- Mutation severity
  - minor - relatively inconsequential mutation which really does not limit activity
  - major - drastic changes
  - lethal mutations - cause death

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- Mutagens - agents that cause mutations
  - Radiation
    - Gamma
    - Beta
    - X-rays
    - UV light
  - Chemicals
    - Gasoline
    - Cyclamates
    - Benzene

17 ▨ **Human Genetics**

- Nature of Human Heredity
  - Humans have 46 chromosomes
  - Human reproduce by sexual reproduction using haploid gametes (23 chromosomes)

18 ▨ **Human Genetics**

- Population Genetics
  - Population is a large group of individuals
  - Gene frequencies - how often a particular gene is found in a population
  - The larger the population the better the data

19 ▨ **Human Genetics**

- Population Sampling
  - Hitchhiker's thumb
  - Attached earlobes

- Tongue rolling
- PTC - tasters vs. non-tasters (dominant TT or Tt)

20 Human Genetics

- Gene pool - all of the genes present in a given population *
  - Some gene pools rarely change
    - cultural barriers
    - physical isolation
  - Gene pools change with emigration (out) and immigration (in)

21 Human Genetics

- Fraternal twins - formed by fertilization of two eggs
  - completely different
  - can be different sexes
  - can look alike or different

22 Human Genetics

- Identical twins - formed when one egg is fertilized and later it splits during developments
  - exactly alike
  - always the same sex
  - look very much alike

23 Human Genetics

- Heredity and Blood Types
- Karl Landsteiner developed the ABO blood typing system
- There are over fifty different kinds of blood typing
- Rh is a second type of blood type

24 Human Genetics

- Blood cells can have a protein substance called antigen on the surface (agglutinogen)
- Blood with different antigens will clot when mixed

25 Human Genetics

- Multiple alleles - three genes are needed for blood type *
- **This is also known as <u>codominance</u> in some books – where more than one gene is expressed (A and B traits in blood type AB)**
  - I^A = dominant for type A antigen
  - I^B = dominant for type B antigen
  - i = recessive for O (no antigen)

26 Human Genetics

- Blood typing procedure
  - clean finger with alcohol
  - puncture finger with a sterile lancet
  - place drops of blood on a clean slide (one on each side)
  - mark one side "A" and one side "B"
  - place one drop of anti-A serum on the side marked A
  - place one drop of anti-B serum on the side marked B
  - mix each solution with a separate applicator stick
  - read slide after 1 minute

27 Human Genetics

- Rh Blood Type

- Rh is a surface antigen found on human red blood cells
- 85 - 88% of humans have Rh antigens and are called Rh^+
- 12 -15% of humans do not have Rh antigens and are called RH^-
- Mothers that are RH^- and have RH^+ babies will have babies at risk of hemolytic disease unless they get medical help

28 ▧ **Human Genetics**

- Multiple alleles *
  - Many genes needed for phenotype *
  - Eye and skin color plus blood types are caused by multiple alleles *

29 ▧ **Human Genetics**

- Eye color

30 ▧ **Human Genetics**

- Eye color

31 ▧ **Human Genetics**

- Albinism – greatly reduced or totally absent production of melanin in the skin (causes skin colors darker than totally and unusually white skin)
- aa – causes albinism *
- AA or Aa gives "normal" non-albino skin colors *
- Parents with Aa x Aa (normal skin color) can produce a child with albinism (aa) * HOW? *

32 ▧ **Human Genetics**

- Sex linkage - traits that are carried on the sex chromosome, especially the X chromosome
  - Examples of sex linkage
    - Color blindness
    - Hemophilia 10X as many men as women
  - The reason males show sex linked traits more often is that males only get one X chromosome
  - They get a sex-linked trait from their mother

33 ▧ **Human Genetics**

- Sex-linked Traits in Human Beings
  - Red-green color blindness
    - X^C (big C) dominant trait for normal color vision
    - X^c (little c) recessive trait for color blindness
  - In most cases, the inability to distinguish red from green, or to see red and green in the same way as most people do, because of an abnormality in the red or green photoreceptors. About 7 percent of men are red-green color blind, compared to 0.4 percent of women.
  - Be sure you can do Punnett squares dealing with colorblindness from your lab book! *

34 ▧ **Human Genetics**

- Color blindness tests

35 ▧ **Human Genetics**

- Color blindness tests One more try…

36 ▧ **Human Genetics**

- Hemophilia – a group of hereditary disorders in which affected individuals fail to make enough of certain proteins needed to form blood clots.
  - Prevalance of Hemophilia: 20,000 people in the United States (NHLBI)
  - Prevalance Rate: approx 1 in 13,600 or 0.01% or 20,000 people in USA [about data]
  - Death rate extrapolations for USA for Hemophilia: 1,681 per year

37 ☐ **Human Genetics**

- Hemophilia

38 ☐ **Human Genetics**

- Sex-influenced traits - baldness
  - BB = bald in either sex
  - Bb = bald in males only
  - bb = normal hair in both

39 ☐ **Human Genetics**

- Sex-limited traits - beards
  - sex-hormones need to be present for gene to function
  - excessive and abnormal testosterone in females may cause beard growth

40 ☐ **Human Genetics**

- Non-disjunction - the failure of a pair (homologous) chromosomes to segregate during meiosis *
- The resultant individual has one less or one too many chromosomes

41 ☐ **Human Genetics**

- Non-disjunction
  - Trisomy - one too many chromosomes (3)
  - Monosomy - only one of a homologous pair (1)

42 ☐ **Human Genetics**

- Trisomy 21 - Down's syndrome
  - 1/600 born with trisomy-21
  - rate increases with very young and older mothers (+35)
  - individuals sometimes called Mongoloids (dated term)

43 ☐ **Human Genetics**

- A karyotype is the complete set of all chromosomes of a cell of any living organism.
- The chromosomes are arranged and displayed (often on a photo) in a standard format: in pairs, ordered by size.
- Karyotypes are examined in searches for chromosomal aberrations, and may be used to determine other macroscopically visible aspects of an individual's genotype, such as sex (XX vs. XY pair).

44 ☐ **Human Genetics**

- Trisomy 21 *

45 ☐ **Human Genetics**

- Trisomy 21

46 ☐ **Human Genetics**

- Monosomy X
- AKA Turner's syndrome
- Characteristics

- Almost all individuals with Turner syndrome have short stature resulting in adult heights of 4 feet 8 inches
- Sterile with underdeveloped sexual organs
- Variety of physiological abnormalities

47 ▧ **Human Genetics**

- Monosomy X

48 ▧ **Human Genetics**

- Trisomy X
  - Many girls and women with Trisomy X have no signs or symptoms. Signs and symptoms vary a lot between individuals, but can include:
  - 1 in 1,000 newborn girls
    - Increased space between the eyes
    - Tall stature (height)
    - Small head
    - Speech and language delays and learning disabilities
    - Delayed development of certain motor skills
    - Behavioral problems
    - Seizures
    - Delayed puberty
    - Infertility
    - Rarely, mental retardation

49 ▧ **Human Genetics**

- Trisomy X

50 ▧ **Human Genetics**

- XXY - Klinefelter's syndrome
- Occurs as frequently as 1 in 500 to 1 in 1,000 male births
- Characteristics (males may have)
  - Enlarged breasts
  - Sparse facial and body hair
  - Small testes
  - An inability to produce sperm
  - Many men live out their lives without ever even suspecting that they have an additional chromosome
  - Although they are not mentally retarded, most XXY males have some degree of language impairment

51 ▧ **Human Genetics**

- XXY - Klinefelter's syndrome

52 ▧ **Human Genetics**

- Amniocentesis - removal of fluid (containing cells of fetus) from a pregnant woman
  - The cells removed are grown
  - Chromosomes are counted and analyzed for abnormalities through a karyotype
  - There is a small risk of miscarriage (1 in 200 or less). Some women have cramping, spotting or leaking of amniotic fluid after the procedure. Serious complications are uncommon