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Patterns of Inheritance

Bio 103 Lecture

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2 ## Topics

✓Mendel's Principles

✓Variations on Mendel's Principles

✓Chromosomal Basis of Inheritance

✓Sex Chromosomes and Sex-Linked Genes

3 ## Experimental genetics began in an abbey garden

✓ Mendel's pea plants
- – easy to grow
- – easily distinguishable varieties
- – mating could be controlled
  - petals of a pea flower almost completely enclose male and female parts
    - – pea plants usually **self-fertilize**
      - »
    - – Mendel could conduct **cross-fertilization**

4  Figure 14.0 Painting of Mendel

5  Figure 14.0x Mendel

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7 ## Experimental genetics began in an abbey garden

✓ 7 characteristics, with two distinct forms
- – flower color, flower position, seed color, seed shape, pod shape, pod color, stem length

✓ Mendel produced **true-breeding** varieties
- – varieties for which self-fertilization produced offspring all identical to parent

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9  Figure 14.x1 Sweet pea flowers

10  Figure 14.x2 Round and wrinkled peas

11 ## Experimental genetics began in an abbey garden

✓ **hybridization** or a **cross**
- – cross-fertilization of two different varieties
- – **hybrids**
  - offspring of different varieties are
- – **P generation**
  - parental plants
- – **F_1 generation**
  - hybrid offspring
- – **F_2 generation**
    - – offspring of F_1 generation when its individuals self fertilize

12  Figure 14.1 A genetic cross

13 Principle of segregation describes inheritance of a single characteristic

✓**monohybrid cross**
- cross in which parents differ in only one characteristic

✓Mendel tracked inheritance of a single characteristic
- developed four hypotheses based on his results

14 Principle of segregation describes inheritance of a single characteristic

✓*(1) There are alternative forms of genes, the units that determine heritable traits*
- gene for flower color in peas exists in one form for purple and another form for white
- **alleles**
  - alternative forms of genes
  - represented as upper and lower case letters, such as **A** and **a**

15 Principle of segregation describes inheritance of a single characteristic

✓*(2) For each inherited characteristic, an organism has two genes, one from each parent. These genes may both be the same allele or may be different alleles*
- for a given characteristic, an individual could have
  - two of same alleles, **AA** or **aa**
  - two different alleles **Aa**

16 Principle of segregation describes inheritance of a single characteristic

✓*(3) A sperm or egg carries only one allele for each inherited trait, because allele pairs separate (segregate) from each other during the production of gametes*
- when sperm and egg unite at fertilization
  - each contributes its allele
    - sperm with allele **A** unites with an egg with allele **a**
      - » produces offspring with two alleles **Aa**
  - restores paired condition to offspring

17 Principle of segregation describes inheritance of a single characteristic

✓*(4) When the two genes of a pair are different alleles and one is fully expressed while the other has no noticeable effect on the organism's appearance, the alleles are called the* ***dominant allele*** *and the* ***recessive allele****, respectively*
- **dominant allele**
  - represented by upper case letter, such as **A**
- **recessive allele**
  - represented by lower case letter, such as **a**

18 Principle of segregation describes inheritance of a single characteristic

✓**homozygous**
- organism with pair of identical alleles for a given characteristic
  - "true-breeding"
- Mendel's true-breeding flowers
  - letters P and p represent alleles for flower color
  - true-breeding purple flower was **homozygous dominant** , **PP**
  - true-breeding white flower was **homozygous recessive, pp**

19 Principle of segregation describes inheritance of a single characteristic

- ✓ Mendel crossed two true-breeding plants
  - one purple and one white flowered
    - each parent could produce 1 type of gamete
      - purple homozygous dominant (PP)
        - » only produced gametes that were P
      - white homozygous recessive (pp)
        - » only produced gametes that were p
    - resulting F_1 hybrid offspring
      - all had purple flowers, each received P & p
  - **heterozygous**
    - organism w/ two different alleles for a trait

20 Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

21 Principle of segregation describes inheritance of a single characteristic

- ✓ **Punnett square**
  - used to determine
    - possible combinations of gametes
      - based on genetic make up of parents
    - proportions of F_2 plants predicted for a given cross
      - | P | p
      - P | PP | Pp
      - p | Pp | pp
      - producing 1 PP, 2 Pp, 1 pp

22 Principle of segregation describes inheritance of a single characteristic

- ✓ **phenotype**
  - expressed, or physical, traits
    - example, purple flowers
  - **phenotypic ratio**
    - ratio of expressed traits (3 purple :1 white)
- ✓ **genotype**
  - organism's genetic makeup (AA or Aa)
  - **genotypic ratio**
    - ratio of different genotypes
      - » 1 PP : 2 Pp : 1 pp

23 Principle of segregation describes inheritance of a single characteristic

- ✓ In cross of 2 true breeders, Mendel found that
  - one parental trait disappears in F_1 generation
  - reappear in 1/4 of the F_2 offspring
- ✓ Mechanism underlying this inheritance pattern
  - stated by Mendel's **principle of segregation**
    - *pairs of genes segregate (separate) during gamete formation*
    - *fusion of gametes at fertilization pairs genes once again*
- ✓ principle of segregation applies to all sexually reproducing organisms

24 Figure 14.2 Mendel tracked heritable characters for three generations

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26 Figure 14.4 Mendel's law of segregation (Layer 2)

27

28 Figure 14.4 Mendel's law of segregation (Layer 1)

29 Figure 14.5 Genotype versus phenotype

30 Figure 14.6 A testcross

31 Homologous chromosomes bear the two alleles for each characteristic

✓ **Homologous chromosomes** (HC)

– the two chromosomes that make up a matched pair in a diploid cell

• 1 inherited from each parent

– have genes for same characteristics located at same positions along their lengths

–

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33 Figure 14.3 Alleles, alternative versions of a gene

34 Principle of independent assortment

✓ **dihybrid cross**

– cross between parental varieties differing in two characteristics

– produces **dihybrids**

✓ were the two traits transmitted from parents to offspring as a package or was each characteristic inherited independently of the other?

35 Principle of independent assortment

✓ If genes for the two traits were inherited together

– F_1 hybrids would produce only same two kinds (genotypes) of gametes that they received from parents

• producing a 3:1 phenotypic ratio

✓ If genes were inherited independently of one another

– F_1 generation would produce four gametes genotypes in equal quantities

• producing a phenotypic ratio of 9:3:3:1

36 Principle of independent assortment

✓ Mendel conducted many dihybrid crosses

– always observed a phenotypic ratio of 9:3:3:1

✓ These results supported **principle of independent assortment**

– *each pair of alleles segregates independently during gamete formation*

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39 Genetic traits in humans can be tracked through family pedigrees

✓ inheritance of many human traits follows Mendel's principles and rules of probability

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✓ Family pedigrees are used to determine patterns of inheritance and individual genotypes

41 Many inherited disorders in humans are controlled by a single gene

✓Most such disorders are caused by autosomal recessive alleles

– Examples:
cystic fibrosis,
sickle-cell
disease

42

✓A few are caused by dominant alleles

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44 Fetal testing can spot many inherited disorders early in pregnancy

✓Karyotyping and biochemical tests of fetal cells and molecules can help people make reproductive decisions

– Fetal cells can be obtained through amniocentesis

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✓Chorionic villus sampling is another procedure that obtains fetal cells for karyotyping

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✓Examination of the fetus with ultrasound is another helpful technique

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✓Mendel's principles are valid for all sexually reproducing species

– However, often the genotype does not dictate the phenotype in the simple way his principles describe

48 Incomplete dominance results in intermediate phenotypes

✓When an offspring's phenotype— such as flower color— is in between the phenotypes of its parents, it exhibits incomplete dominance

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✓Incomplete dominance in carnations: red, pink, white

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✓Incomplete dominance in human hypercholesterolemia

51 Many genes have more than two alleles in the population

✓In a population, multiple alleles often exist for a characteristic

– example, three alleles for ABO blood type in humans

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– alleles for A and B blood types are codominant
  • both are expressed in the phenotype

53 A single gene may affect many phenotypic characteristics

✓A single gene may affect phenotype in many ways

– called pleiotropy

- example, allele for sickle-cell disease

54

55 Genetic testing can detect disease-causing alleles

✓Genetic testing

– can be of value to those at risk of developing a genetic disorder or of passing it on to offspring

56 A single characteristic may be influenced by many genes

✓single characteristic may be influenced by many genes

– creates a continuum of phenotypes

- example: skin color in humans

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✓Genes are located on chromosomes

– Their behavior during meiosis accounts for inheritance patterns

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✓The chromosomal basis of Mendel's principles

60 Genes on the same chromosome tend to be inherited together

✓Certain genes are linked

– tend to be inherited together because they reside close together on same chromosome

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62 Crossing over produces new combinations of alleles

✓Crossing over

– produces gametes with recombinant chromosomes

- fruit fly *Drosophila melanogaster* was used in the first experiments to demonstrate effects of crossing over

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65 Geneticists use crossover data to map genes

✓Crossing over is more likely to occur between genes that are farther apart

– Recombination frequencies

- can be used to map relative positions of genes on chromosomes

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✓A partial genetic map of a fruit fly chromosome

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✓human male

– one X chromosome and one Y chromosome

✓human female

– two X chromosomes

✓Whether a sperm cell has an X or Y chromosome determines sex of the offspring

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✓Other systems of sex determination exist in other animals and plants

70 Sex-linked genes exhibit a unique pattern of inheritance

✓All genes on sex chromosomes are said to be **sex-linked**

– In many organisms

- X chromosome carries many genes unrelated to sex
- Fruit fly eye color is a sex-linked characteristic

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– Their inheritance pattern reflects fact that males have one X chromosome and females have two

72 Sex-linked disorders affect mostly males

✓Most sex-linked human disorders are due to recessive alleles

– Examples: hemophilia, red-green color blindness

– mostly seen in males

- male receives a single X-linked allele from his mother, and will have disorder
- female has to receive the allele from both parents to be affected

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✓A high incidence of hemophilia has plagued the royal families of Europe