

GENDER DISORDER

A Lesson Plan to Augment the DVD of *The Meaning of Sex: Genes and Gender*

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Section A. Sex Determination Lecture Questions

Lecture 1 Questions

“Deciphering the language of sex,” by Dr. David Paige

1. What do genes have to do with infertility?
2. How is a human embryo sex determined?
3. When in human development do sexes become distinct?
4. Give a historical perspective on theories of sex determination.
5. Why did early geneticists think that sex was determined by the absence of the second X chromosome?
6. Explain the occurrence of XX males in 1/20,000 individuals within the population.
7. Why have two sexes?
8. Explain the statement “meiosis is the defining feature of sexual reproduction.”
9. What are the advantages of having sexual and clonal options?
10. Why are males considered “evolution’s spare parts”?

Lecture 2 Questions

“Hermaphrodites, the safer sex,” by Dr. Barbara Meyer

1. What is meant by the unity of life, and how does this concept aid in research?
2. What are the advantages of using model organisms?
3. How can you make a male into a hermaphrodite?
4. Explain the function of XOL-1 in sexual development of *C. elegans*?
5. Under what conditions are males produced in *C. elegans*?
6. What are the advantages of being a hermaphrodite?
7. What are the disadvantages of being a hermaphrodite?

8. What came first, self-fertilization or sex? How do we know this?
9. Why do males still occur in *C. elegans*?

Lecture 3 Questions

“Sex and death, too much of a good thing,” by Dr. Barbara Meyer

1. What causes Down syndrome?
2. Explain the process which gene regulates expression between the sexes.
3. What is dosage compensation and how does it influence sex determination?
4. Why is the absence of XOL 1 lethal?
5. If there is a XOL 1 mutation, how can the lethal effect be overcome?
6. If it is a normal condition for human females to inactivate the X chromosome, why does Turner’s syndrome occur?
7. What happens if a nematode has XXX?
8. How do some genes on the inactivated X chromosome escape inactivation?
9. How does MIX 1 carry out different jobs in the same cell?
10. How can SMC help trace zygote development?
11. What does the term “function conserved” mean?

Lecture 4 Questions

“Sexual Evolution from X to Y,” by Dr. David Paige

1. What makes a male, male?
2. How is the no recombination region significant when looking at human history?
3. Why are inversions tolerated on the Y chromosome?
4. How is sex determination different from mammals in some reptiles?
5. What region of the Y chromosome is particularly prone to loss?
6. Why is it dangerous to pile the genes for spermatogenesis on the Y chromosome?
7. Describe the ICSI procedure and how it benefits infertile couples.
8. Many organisms throughout evolutionary history have used the theme of creating sex chromosomes from autosomes. Give three examples of these variations.

National Science Standards Addressed in this Curriculum for 9th through 12th grade:

Science as Inquiry Standards

Science and Technology Standards

Science in Personal and Social Perspectives

History and Nature of Science

Molecular Basis of Heredity

Oregon State Standards Addressed in this Curriculum for 10th Graders:

Scientific Inquiry.

Forming the Question/Hypothesis

Designing the Investigation

Collecting and Presenting the Data

The Laws of Heredity.

Technical Writing.

Speaking.

Section B. Genetic Disorder Project

GENETIC DISORDER PROJECT POWER POINT VERSION

Birth defects occur in one of every 14 births. These defects are the leading cause of infant death, and they also have a profound effect on the daily lives of people of all ages. Despite their special conditions, many children born with birth defects go on to live full, active lives. For those with any of the more than 3000 known conditions, there is both challenge and reward in life. Scientists do not know the causes of most birth defects, but many of them are inherited or caused by gene mistakes. Each year scientists learn more about such diseases and are continually looking for ways to prevent or correct them.

ASSIGNMENT: You will research a genetic disorder of your choosing and present the material in the form of a power point presentation and brochure. You will give a brief presentation to the class about the disorder using the power point presentation as your visual aid. You may work with one or two partners. *You may choose your partners under the assumption that I may add or deduct members from your group. You will have some class time to research. Any diagrams graphs, charts, pictures, ect. Must be clearly labeled and referenced. Use *your own words* and define any words that are unfamiliar to the general public.

Grading:

Project = 150 points

Power Point = 50 points

Brochure = 50 points

Presentation = 20 points

*Presentations will be given May 28th to June 5th

Presentation times and dates will be randomly selected.

Peer Evaluation = 20 points

Group Evaluation = 10 points

Projects will be graded on accuracy, completeness, neatness, spelling/grammar, visual appeal, labeling, references, and use of current information. Presentations will be graded using CIM speaking standards. More specifically, all group members need to participate, voice, eye contact, interesting, completeness, accuracy, enthusiastic, and ability to answer questions. Peer evaluation will also be a component.

Things to Include: Each disorder will vary slightly in the information that is available and what is important to know about the disorder. The following are general guidelines that should be considered and may or may not directly apply to your project. Feel free to include any additional information you feel is important to know about your disorder.

Name of Disorder: Common name and medical name.

Description: Describe the characteristics and historical information about the disease.

Symptoms: List of symptoms of this disease including visual and laboratory test indicators.

Describe How Inherited: Describe the inheritance patterns (dominant/ recessive/ incomplete dominance/ sex-linked/ autosomal). Provide an example of a cross illustrating the inheritance pattern.

Pedigree or Karyotype: Depending on how the disorder is inherited.

Geographic and/or Population Occurrence: Describe if this trait is found more commonly in specific geographical location/ ethnic groups/ or equally in all populations.

Characterize the Typical Recipients: Describe if age, sex, newborn, children, or elderly are likely characteristics when the condition is first discovered or becomes a factor in the life of the person.

Statistical Information: Describe the occurrence of this condition in the population as a probability or percentage of occurrence and/ or normal but carries of the condition.

Treatment: Describe the treatment options, if any, available to the person. Do any organizations provide assistance to the patients?

Prevention: Describe the prevention actions that may be taken, if any, to avoid this condition. Include if genetic counseling is appropriate for this condition.

Current Research: What is currently being done in the research field regarding this condition.

Bibliography: Bibliography must include a minimum of three sources. Sources must include at least one periodical article. One source must be within last five years. Complete information for each source!

Nitty Gritty:

Power Point Tips:

1. Use outline form with titles and subtitles.
2. Typing should be legible from the back of the room.
3. Do not use lengthy paragraphs. Bullets, Lists, subheading, and short segments of text is easier to understand.
4. Pictures are worth a thousand words! But make sure they are informative. **MUST** be accompanied by a caption.
5. Leave some space open in the design. Tightly packed space can tire the eyes and mind.
6. Aim for a “*WOW*” from your viewers!
7. Credit the source of pictures and diagrams on the page presented. Include a references page at the end.

Brochure Tips:

1. Information should be well organized and easy find. Sub-heading help direct the reader.
2. Type should be single spaced and not larger than 12 font.
3. The length and layout of the brochure is up to you, but make sure that the information is adequately and completely covered and looks like a brochure or flyer.
4. Pictures, graphs, etc. **MUST** have captions and sources credited.
5. Should be written at level for general public.
6. Aim for something that looks ‘*professional*’

and could be used at a hospital or clinic!

GENETIC DISORDER PROJECT WEBSITE VERSION

Birth defects occur in one of every 14 births. These defects are the leading cause of infant death, and they also have a profound effect on the daily lives of people of all ages. Despite their special conditions, many children born with birth defects go on to live full, active lives. For those with any of the more than 3000 known conditions, there is both challenge and reward in life. Scientists do not know the causes of most birth defects, but many of them are inherited or caused by gene mistakes. Each year scientists learn more about such diseases and are continually looking for ways to prevent or correct them.

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Project = 150 points

Website = 50 points

Brochure = 50 points

Presentation = 20 points

*Presentations will be given March 10th through March 12th
Presentation times and dates will be randomly selected.

Peer Evaluation = 20 points

Group Evaluation = 10 points

Projects will be graded on accuracy, completeness, neatness, spelling/grammar, visual appeal, labeling, references, and use of current information. Presentations will be graded using CIM speaking standards. More specifically, all group members need to participate, voice, eye contact, interesting, completeness, accuracy, enthusiastic, and ability to answer questions. Peer evaluation will also be a component.

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2. Typing should be legible from the back of the room.
3. Do not use lengthy paragraphs on the homepage. Bullets, Lists, subheading, and short segments of text is easier to understand.
4. Pictures are worth a thousand words! But make sure they are informative. **MUST** be accompanied by a caption.
5. Aim for a “*WOW*” from your viewers!
6. Credit the source of pictures and diagrams on the page presented. Include a references page at the end.
7. Create links only to credible sources.

Brochure Tips:

8. Information should be well organized and easy find. Sub-heading help direct the reader.
9. Type should be single spaced and not larger than 12 font.
10. The length and layout of the brochure is up to you, but make sure that the information is adequately and completely covered and looks like a brochure or flyer.
11. Pictures, graphs, etc. **MUST** have captions and sources credited.
12. Should be written at level for general public.
13. Aim for something that looks ‘*professional*’ and could be used at a hospital or clinic!

Section C. Genetic Disorder Study Guide and List

- Most are generally caused by **MUTATIONS** (a change in the order of the bases on the DNA).
 - Mutations can result in genes that do not function properly or do not function at all.
 - Mutations become part of the genetic material, and can therefore be passed along from one generation to the next.
 - Mutations can result from many causes including radiation, cosmic rays, x-rays, or certain chemicals.
 - Mutations occur at random; they are not predictable; they can contribute considerable diversity to all life as seen today.
 - Most mutations are **homozygous recessive** and **harmful**.
- Genetic disorders are difficult to prevent because of unpredictability of occurrence.
- Genetic disorders cannot be cured without changing the genes.
Current research in genetic engineering is now experimenting with
some of these possibilities.
- Genetic disorders can be divided into five major groups according to
their specific means of inheritance.

Autosomal Recessive

- Most genetic diseases are caused by recessive alleles.
- They are carried on non-sex (autosomes) chromosomes.
- Most are fairly rare because it can only occur in the homozygous condition.

Albinism - Lack of melanin in skin, eyes, or hair; light-sensitive skin and

eyes; white hair; shortened life expectancy.

Cystic Fibrosis - Very salty tears and sweat, abnormal amounts of

mucous congestion in lungs/pancreas, clogged breathing passages.

Galactosemia - Excess galactose in liver, cataracts, mental retardation.

Gaucher's Disease - Enlargement of liver, lymph nodes, and spleen;

severe psychomotor difficulties.

Hereditary Microcephaly - Small head, severe mental retardation.

Maple Syrup Urine Disease - Maple syrup odor of urine, progressive

deterioration.

Phenylketonuria (PKU) - Excess phenylalanine in blood acts as poison

to brain tissue, unusual body odor, mental deterioration, cannot transform phenylalanine into tyrosine.

Sickle-cell Anemia - Abnormal hemoglobin, blocks small blood vessels,

severe pain.

Tay-Sachs Disease - Failure to produce certain blood enzyme which

helps metabolize fat; fats accumulate in brain;

after

6 - 9 months rapid deterioration of vision and motor skills.

Thalassemias - Lack of sufficient hemoglobin.

Autosomal Dominant

- An individual with just one dominant gene for the disorder will be affected.

- Inheritance of the dominant allele produces a characteristic pedigree.
- Every affected child will have at least one affected parent.

Achondroplasia - Form of dwarfism.

Brachydactyly - Hand formation with short fingers.

Congenital Stationary Night Blindness - Dusk or night blindness.

Familial Polycystic Kidneys - Cysts develop in kidneys; hypertension;

uremia; onset about age 40.

Huntington's Disease - Degeneration of nervous system, uncoordinated

movements and progressive mental decline; onset about age 40.

Juvenile Pernicious Anemia - Insufficient red blood cells; onset during

childhood. (This disease has shown both autosomal dominant and recessive transmission.)

Marfan's Syndrome - Long, thin arms and legs; affects bone, muscle,

and connective tissue.

Polydactyly - Extra fingers or toes.

Retinoblastoma - Cancer of one or both eyes.

Treacher-Collins Syndrome - Malformation of the face and jaw;

deafness.

Sex-Linked

- Recessive alleles carried only on the X-chromosome.
- Males exhibit the trait most commonly as there is no allele on the Y-chromosome to affect the allele on the X-chromosome.
- Affected females can only be homozygous recessive.

Brown Tooth Syndrome - Defects in the enamel; conchoidal appearance of

teeth may also occur.

Cerebellar Ataxia - Atrophy of the cerebellum accompanied by clumsy,

explosive speech.

Christmas Disease - Hemophilia B (lack of blood-clotting factor IX), a

milder form of hemophilia.

Congenital Deafness - Profound deafness, possible deafmutism.

Congenital Night Blindness - Poor vision at dusk.

Duchenne Muscular Dystrophy - Trunk weakness and gradual deterioration-

and shrinkage of muscles.

Gout - High uric acid level in the blood, arthritis.

Hemophilia A - Lack of blood-clotting factor VIII.

Lesch-Nyhan Syndrome - Self-mutilation, appears to be linked to a

defect in purine metabolism.

Nystagmus, Hereditary - Constant involuntary motion of the eyeball.

Red/Green Colorblindness - Unable to detect differences between red

and green colors.

Polygenic

- Disorders that result from the interactions of many genes.

Spina Bifida - Failure of the spinal cord to develop properly before

birth.

Cleft Palate - A split along the roof of the mouth caused by the failure of

the two parts of the palate to join in prenatal development.

Cleft Lip - Split along the center of the upper lip caused by the failure of

the two parts of the lip to join during prenatal development.

Chromosomal

- Involves whole chromosomes or whole segments.
- Not a result of specific gene action.
- Most are so severe, there is a tendency to miscarry early in the pregnancy.

WHOLE CHROMOSOMAL ABNORMALITIES

- **Nondisjunction** - Failure of the chromosomes to separate during meiosis resulting in too many or too few chromosomes.
- **Polyplody** - Meiosis may not reduce chromosome number resulting in increased chromosome number of $3N$ or more.

CHROMOSOMAL ABERRATIONS

- **Inversion** - Reversed position of DNA segment.
- **Deletions** - Small segments of DNA become lost.
- **Translocation** - DNA fragment attaches to another chromosome, may cause duplications of genes.

Down's Syndrome - Multiple physical defects including protruding (Trisomy 21) tongue and lower lip; mental retardation, unique

fold in inner corner of eye (mongolism); nondisjunction of 21st pair takes place prior to conception

in 95% of cases; recurrence rate, same family - 1%.

Down's Syndrome - In 1 - 2% of the cases, nondisjunction occurs after

(Mosaicism) conception. All future cell divisions have the extra

chromosome; recurrence rate in same family not significant.

Patau's Syndrome - Multiple birth defects; severe mental retardation.

(Trisomy 13)

Edward's Syndrome - Multiple congenital abnormalities; severe mental

(Trisomy 18) retardation.

Triple X Syndrome (XXX) - Few, if any, problems.

Turner's Syndrome (XO) - Undeveloped female, usually short in stature, usually sterile, may be mentally retarded.

Klinefelter's Syndrome (XXY) - Undeveloped male, may be sterile and/

or mentally retarded.

Double Y Syndrome (XYY) - Taller than average, aggressive, emotionally unstable, low intelligence, may be prone to violence.

Cat Cry Syndrome - Deletion in the 5th chromosome resulting in peculiar cry, mental retardation.

Down's Syndrome - In 4% of cases of this disorder, the extra 21st chromosome is attached (translocated) to another chromosome (usually #14, #15, or #22). About half the time this type of Down's Syndrome is inherited from a parent who is a "carrier." The

risk

of recurrence in future children from the same parents is therefore much higher than with non-disjunction-caused Down's Syndrome.

4. Explain how two individuals with achondroplasia (dwarfism) can produce a normal child. Use a punnett square and pedigree to justify your answer.

5. T = tall plants tt = short plants G = green plants gg = yellow plants

- a. Draw the punnett square for the cross between two purebred plants. Rule: The two plants can not have any traits in common. The plants must have two traits.

- b. Draw the punnett square for an F1 cross. What is the phenotypic ratio of the offspring resulting from the F1 cross.

Section E. Genetics Exam

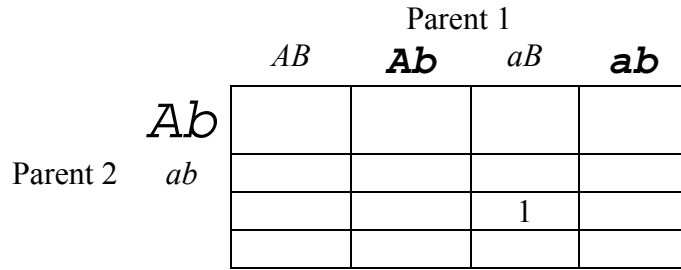
Multiple Choice; Choose the letter of the answer that best completes each statement.

- The genotype of an individual that shows the dominant phenotype can be determined by crossing it with an individual that is
 - heterozygous recessive.
 - heterozygous dominant.
 - homozygous dominant.
 - homozygous recessive.
- A Punnett square is used to determine the
 - probable outcome of a cross.
 - actual outcome of a cross.
 - result of segregation.
 - result of meiosis I.
- When two pea plants that are heterozygous for yellow peas are crossed, the resulting offspring have a genotypic ratio of
 - 9:3:3:1.
 - 1:1.
 - 3:1.
 - 1:2:1.
- Any hybrid organism is
 - recessive.
 - purebred.
 - heterozygous.
 - homozygous.
- The physical appearance of an individual is its
 - phenotype.
 - genotype.
 - heredity.
 - genetics.
- In probability, the larger the number of events, the
 - farther from the expected ratio.
 - closer to the expected ratio.
 - greater the chance of predicting the next event.
 - less the chance of predicting the next event.
- If a family has three daughters, the probability that the next child will be a girl is
 - 1/4.
 - 1/3.
 - 1/2.
 - 3/4.
- Different forms of a gene are
 - diploid.
 - linked.
 - homologs.
 - alleles.
- If a parent that is homozygous dominant is crossed with a parent that is recessive, the offspring are
 - homozygous dominant.
 - homozygous recessive.
 - heterozygous recessive.
 - heterozygous dominant.
- When a purebred pea plant with smooth green pods is crossed with a purebred plant that has constricted yellow pods (this would be the P generation), the phenotypic ratio of the F₂ generation is
 - 3:1:1.
 - 1:2:1.
 - 9:3:3:1.
 - 10:4:2:2.

11. The effect of a recessive allele is not observed while the dominant allele is present, according to Mendel's principle of
- segregation.
 - independent assortment.
 - dominance.
 - blending inheritance.
12. If incomplete dominance is the inheritance pattern, and a homozygous tall pea plant and a homozygous short pea plant are crossed, in the resulting F1 generation
- the recessive trait disappears.
 - the offspring are of medium height.
 - no hybrids are produced.
 - all the offspring are short.
13. Unlike mitosis, meiosis of a single cell results in the formation of
- two identical cells.
 - three identical haploid polar bodies.
 - four identical gamete cells.
 - four unlike gamete cells.
14. In pea plants, tall is dominant over short and yellow seeds are dominant over green seeds. If two pea plants that are heterozygous for plant height and for seed color are crossed, the Punnett square reveals that one of sixteen offspring will probably be
- short with green seeds.
 - short with yellow seeds.
 - tall with yellow seeds.
 - tall with green seeds.
15. If an organism has two identical alleles for a trait, it is
- homozygous.
 - heterozygous.
 - homozygous dominant.
 - heterozygous recessive.
16. When two hybrid organisms are crossed (monohybrid cross), the offspring will probably show a phenotypic ratio of
- 1:1.
 - 1:2:1.
 - 1:3:1.
 - 3:1.
17. If any offspring from a test cross shows a recessive phenotype, the parent with the unknown genotype is
- heterozygous dominant.
 - homozygous dominant.
 - heterozygous recessive.
 - homozygous recessive.
18. The phenotype ratio of the F2 generation of a two-factor cross (dihybrid cross) is
- 3:1.
 - 1:2:1.
 - 2:6:6:2.
 - 9:3:3:1.
19. During meiosis, the production of new gene combinations in the chromosomes results from
- homologs.
 - fertilization.
 - crossing-over.
 - two-factor crosses.
20. A Punnett square does not show the
- genetic makeup of the eggs.
 - probable outcome of a cross.
 - genetic makeup of the sperm.
 - actual outcome of a cross.

21. The sex of a human is determined by the
- presence or absence of a Y chromosome.
 - presence or absence of an X chromosome
 - number of Y chromosomes
 - number of X chromosomes
22. Which human trait is determined by codominant alleles?
- sex
 - skin color
 - height
 - blood type
23. A condition resulting from nondisjunction of autosomes is
- male pattern baldness
 - Down syndrome
 - Duchenne muscular dystrophy
 - albinism
24. Which is not a procedure used to diagnose genetic disorders prenatally?
- tests for absence of RNA sequences
 - tests for biochemical abnormalities in embryonic cells
 - chorionic villus biopsy
 - amniocentesis
25. What is the inheritance pattern for Turner's syndrome?
- autosomal dominant
 - autosomal recessive
 - sex linked
 - chromosomal
26. What is the inheritance pattern for red/green colorblindness?
- autosomal dominant
 - autosomal recessive
 - sex linked
 - chromosomal
27. What is the inheritance pattern for hemophilia a?
- autosomal dominant
 - autosomal recessive
 - sex linked
 - chromosomal
28. What is the inheritance pattern for sickle cell anemia?
- autosomal dominant
 - autosomal recessive
 - sex linked
 - chromosomal
29. Organisms that have two identical alleles for a particular trait are said to be
- haploid
 - homozygous
 - heterozygous
 - diploid
30. What is the total number of chromosomes in a diploid cell of a person with Down syndrome?
- 22
 - 23
 - 44
 - 47

Using Science Skills: Interpreting diagrams



Key:

A = axial flowers

a = terminal flowers

B = red flowers

b = white flowers

1. Name the kind of diagram shown in the figure. _____

What is the purpose of this kind of diagram? _____

2. What are the genotype and phenotype of Parent 1? _____

Of Parent 2? _____

Fill in the boxes in the diagram with the correct information.

3. What biological process is represented by filling in the boxes? Explain. _____

4. Give the genotype and phenotype of the individual symbolized by the box labeled 1.

5. What fraction of the offspring are heterozygous for both traits? _____

What fraction of the offspring have terminal red flowers? _____

Section F. Related Web Sources

www.med.nyu.edu/Sackler/genetics/Cline&Meyer.pdf

A discussion of how sex chromosomes influence gender in flies and worms

<http://www.biol.andrews.edu/gen/17.htm>

Brief descriptions of different ways that gender is determined

<http://www.physci.ucla.edu/html/arnold.htm>

Description of a UCLA professor's research around sex chromosomes and gender determination. Includes lots of references.

[http://www.uvm.edu/~leschnei/develop_html/notes_html/ch26\(3\).pdf](http://www.uvm.edu/~leschnei/develop_html/notes_html/ch26(3).pdf)

Detailed lecture notes on sex determination in different types of animals