MINI-SYMPOSIUM ON GENETIC DISORDERS

Each presentation must be at least three minutes and not more than five minutes in length. A time for questioning will follow each presentation.

Since you are expected to be an expert on the disorder you choose, you must deliver your talk WITHOUT reading from your slides.

What must be included in the presentation?
The presentation should follow the outline below. The questions listed are designed to provoke thought and to help you determine what is significant and what is not. They are not to be answered individually as a question and answer session.

1. **Introduction**
   Give a brief overview and provide pictures of the disorder. Engage the crowd. Ex. Start with picture of famous person.

2. **Mode of inheritance**
   All genetic disorders are inherited. There are several different ways in which they can be inherited. Determine whether your disorder is:
   - autosomal dominant trait
   - autosomal recessive trait
   - X-linked recessive trait
   - chromosomal error - deletion, extra chromosome, etc.
   - polygenic disorder - caused by several genes

   ★ You must include a [Punnett square and/or a pedigree chart in your presentation](#). Be able to explain!

3. **Clinical description of the disorder**
   Identify the following using pictures:
   - Chromosome #
   - Name of gene and location on chromosome
   - Gene sequence of normal gene and sequence of mutated gene (ex. CAG → CTG)
   - Describe the structure/function of normal protein versus mutated protein
   - **Explain the science behind the disorder**! (Ex. if your disease is Muscular Dystrophy then explain to the audience what happens to muscle function because of the lack of the certain protein).
   - What are the features of the disorder?
   - How does it affect the victim?
   - What is it like to have the disorder?
   - What problems are associated with the disorder?
   - Is the disorder physically limiting (makes normal tasks difficult)?
   - Is it life-threatening or fatal?
   - Is it found more commonly in certain groups of people (ethnic or religious group or particular sex)?
4. Treatment

❑ Can anything be done for the disorder?
❑ Can the basic defect be treated?
❑ Can the symptoms or results of the disorder be treated?
❑ Is there a cure for the disorder?
❑ Is there any gene therapy for the disorder?

5. Detection

❑ Can the disorder be detected before its symptoms appear?
❑ Can it be detected before birth (prenatally)? If so, how?
❑ Is there any way to detect whether a parent is a carrier of the disorder? If so, how?

6. Current research - *Must be from 2015-2018

❑ Discuss research from current medical and or scientific journals (sciencedaily.com, library data bases, LibGuide).
❑ Give specific details of recent experiments (gene therapy, genetic engineering, stem cell research, CRSPRs, gene silencing, RNAi therapy).
❑ Include detail of where, when, who, and how the studies were conducted.
❑ Provide summary of article
❑ Include pictures to help explain the research

Important Information

o Due Date: ___________________________
o Will count as a test.
o Before presentation
  1. Share your presentation with your teacher on Google.
  2. Print a copy of your presentation (on your computer or in library) in black and white (6 slides per page).
  3. Hand in the printed copy of the presentation with your rubric attached to the front.

★ To begin research look up “Single Gene Genetic Disorders”
★ If you would like to practice your presentation before class, please schedule a time with me.
YOUR DISORDER WILL BE SELECTED FROM THE LIST BELOW

1. Sickle-Cell Anemia
2. Breast cancer
3. Thalassemia
4. Edward's Syndrome/Patau's Syndrome
5. Turner's Syndrome
6. Polycystic kidney disease
7. Klinefelter's Syndrome
8. G6PD (Glucose 6 phosphate dehydrogenase) Deficiency
9. Cooley's anemia
10. Gaucher's disease
11. Hemophilia
12. albinism
13. Familial Hypercholesterolemia
14. diabetes mellitus
15. Neurofibromatosis
16. diabetes insipidus
17. Marfan Syndrome
18. fragile X syndrome
19. Phenylketonuria
20. Parkinson's disease
21. Cystic Fibrosis
22. Alzheimers
23. Tay Sachs Disease
24. colorblindness
25. Down Syndrome
26. epidermolysis
27. Duchenne Muscular Dystrophy
28. alcoholism
29. Huntington's Disease
30. Burkitt lymphoma
31. Cri-du-chat Syndrome
32. Pituitary dwarfism syndrome
33. Galactosemia
34. Immune deficiency diseases (Boy in the bubble)
35. left Lip/Palate
36. Polydactyl
37. Osteogenesis Imperfecta
38. Sensorineural deafness
39. Spina Bifida/Anencephaly
40. Beta-ketothiolase
41. Pachygyria
42. Ushers syndrome