

## MINI-SYMPOSIUM ON GENETIC DISORDERS

### **WHAT IS IT?**

A symposium is a meeting where participants give oral presentations on subjects revolving around a common theme. Our mini-symposium is based on the theme of genetic disorders. Each student will select one disorder from the list, research information on the disorder, and prepare and deliver a comprehensive, concise, and complete oral presentation to the class.

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 it. You will be given an index card on which you may jot down a few **TOPICS** (not facts). Only one side of the card may be used. You will have it only as a reminder of what you are going to say, **NOT** as a list of statements you are going to make. You may also include any difficult-to-spell terms that you wish to write on the board. The index card will be passed in as soon as your talk is over.

### **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

What is the name of the disorder? Are there any other names by which it is commonly known?

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 an autosomal dominant trait, an autosomal recessive trait, an X-linked recessive trait, a chromosomal error or a multifactorial trait (polygenic disorder). Chromosomal errors may take several types: a particular missing chromosome (e.g. a missing X chromosome), an extra chromosome (e.g. an extra chromosome 21), or a damaged chromosome (e.g. part of a chromosome deleted). A multifactorial disorder is one, which is caused by several genes or by a combination of genetic and environmental factors.

3. Clinical description of the disorder.

What are the features of the disorder? How does it affect the victim? What is it like to have the disorder? How would you describe the disorder to someone else? What is the disorder like externally, internally, biochemically, psychologically, etc. What problems are associated with the disorder? Is the disorder physically limiting? Is it life-threatening? Is it invariably fatal? Is it found more commonly in certain groups of people, such as a particular 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? If so, how? Can it be detected prenatally? If so, how? Is there any way to detect a carrier of the disorder? If so, how?

**HOW WILL THIS ACTIVITY BE GRADED'?**

You will receive a letter grade from A to F. The following factors will be used to determine the grade:

1. Accuracy -How accurate was your presentation? Did you give any misinformation? Did you appear to know the information well? Did you answer questions accurately? Did you make the effort to insure that all terms were pronounced correctly?
2. Following directions -Did you stick to the time limits? Did you make your note card and use it properly? Did you include all the required information?
3. Delivery -Did you speak loudly and clearly? Were you too fast or too slow? Did you appear confident and poised? Did you have proper eye contact with the class? Were you enthusiastic?
4. Use of visual aids -Did you use the board/overhead well? Did you use any other visual aids such as pictures, charts, objects, demonstrations, handouts, etc.?

**YOUR DISORDER WILL BE SELECTED FROM THE LIST BELOW**

1. Sickle-Cell Anemia
1. Breast cancer
2. Thalassemia
3. Turner's Syndrome
4. Klinefelter's Syndrome
5. Hemophilia
6. Albinism
7. Diabetes mellitus
8. Neurofibromatosis
9. Marfan Syndrome
10. Fragile X syndrome
11. Phenylketonuria
12. Parkinson's disease
13. Cystic Fibrosis
14. Alzheimers
15. Tay Sachs Disease
16. colorblindness
17. Down Syndrome
18. alcoholism
19. Huntington's Disease
20. Burkitt lymphoma
21. Cri-du-chat Syndrome
22. Pituitary dwarfism syndrome
23. Galactosemia
24. Immune deficiency diseases (Boy in the bubble)
25. Polydactyl

Biology  
Mr. Addison  
2001

26. Sensorineural deafness
27. Spina Bifida/Anencephaly
28. Achondroplasia
29. Achromatopsia
30. Acid Maltase Deficiency
31. Adrenoleukodystrophy
32. Blue Rubber Bleb Nevus Syndrome
33. Canavan Disease
34. Dercum's Disease
35. Fanconi Anemia
36. Fibrodysplasia
37. Ossificans Progressiva
38. Hurler Syndrome
39. Long QT Syndrome
40. Nephrogenic Diabetes Insipidus
41. Niemann-Pick Disease
42. Osteogenesis Imperfecta
43. Porphyria
44. Prader-Willi Syndrome
45. Proteus Syndrome
46. Shwachman Syndrome
47. Turner's Syndrome
48. Urea Cycle Disorder
49. Waardenburg Syndrome
50. Williams Syndrome
51. Wilson's Disease