

Human Genetic Disorder Project

You have been challenged to incorporate your knowledge about cells, cell division, genetics, and DNA to research and present on a specific genetic disorder. You have already completed your basic study about the ideas of genetics and mutations. Now with the information that you have you are being asked to research a specific genetic disorder.

- **IN YOUR OWN WORDS**, using complete sentences, paragraph style, summarize the information in the spaces provided.
- **DON'T just COPY and PASTE** information from the INTERNET!
Analyse Information from at least 2-3 sites and summarise them in your own words.
- **IMAGES** – at least 3 good and appropriate images need to be included in your report
- You must have the following **sub-headings** in your project.

Introduction

- What is the name of the disorder that you are researching?
- Who discovered this disorder, when and where, etc?
- What is the incidence of this disorder? (eg. One in 10,000 Australia, more in males – males: females = 7:3, is it more often in some ethnic groups)
- Is this disorder more likely to occur in certain populations?

Cause of the Disorder

- What category of genetic disorders does this disorder fit into (single gene defect, multifactorial defect, mitochondrial disorder or chromosome abnormalities)
- Is it a recessive or dominant trait?
- Is it sex-linked
- Which chromosome is the gene for the disorder located on?
- Are there environmental factors or influences?

Genetic Diagram(s) - Make a drawing of how the disorder is inherited (such as a Genetic diagram, Punnett square or pedigree). You need other information if Punnett square is not applicable to your selected disorder.

Signs and symptoms

- How do these problems affect the patient's daily life?
- List the parts of the body that are affected.
- How does the disorder progress? Over time, what happens to the person who has the disorder?

- Is the disorder dangerous?

Treatment of this disorder

- Don't just mention – no cure!
- Since the patient has a lot of associated problems – what sort of medication, physiotherapy etc may help to alleviate his or her problems.
- What is the status of research on the disorder? This information could also include therapy or ways the individual with the disorder adapts.

Prevention of this disorder

- A description of any protocols that may be used to determine carriers and/or individuals who might have the disorder and the probabilities of their passing the disorder to their offspring.
- Genetic Counselling, Genetic Tests

References

- 3 References need to be included – Be specific
Don't just simply site: Google image, Wikipedia etc.
- When you site the internet information, you need to write down the Title and the publisher other than the actual site:

Eg. **Genetic Disorder Information from genomics.energy.gov**

http://www.ornl.gov/sci/techresources/Human_Genome/medicine/assist.shtml

- If it is a book or journal, you need to include the author, year of publication, Title of the book or journal, edition, publisher, page numbers.

Eg. Lofts, Graeme and evergreen, Merrin (2000) “Science Quest 4” 2nd Edition, Jacaranda
p.176-179

TAAB

DNA	You have yet to describe the basic processes by which genetic information is passed from one generations to the next	You have described with some examples, the basic processes by which genetic information is passed from one generations to the next with scientific terms	You have explained, with a variety of examples, the basic processes by which genetic information is passed from one generation to the next with scientific terms	You have explained in depth, the basic processes by which genetic information is passed from one generation to the next with scientific terms
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Punnett squares	You have yet to explore patterns in the inheritance of genetically controlled characteristics	You have described patterns in the inheritance of genetically controlled characteristics	You have explained patterns in the inheritance of genetically controlled characteristics with accuracy and provide a variety of examples	You have coherently explained, patterns in the inheritance of genetically controlled characteristics with accuracy, provide a variety of examples
Variation	You have yet to describe biological ideas relating to genetic variation	You have explained the importance of variation within a changing environment	You have explained the importance of variation within a changing environment, providing various examples	You have coherently explained, the importance of variation within a changing environment, providing various examples
Overall	Working below curriculum level	Working At curriculum level	Working Above curriculum level	Working Beyond curriculum level