



GENETIC DISEASE

Group....

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Group Members

1.
2.
3.
4.
5.
6.
7.

GENETIC DISEASE

A. Introduction

Questions	Expected Answer
Do you know about "Sequence Manipulation Suite (SMS) ?	
What do you know about Genetics?	
What do you know about Gene Mutation?	
What are some examples of genetic diseases that you know?	
Do you know about Cystic Fibrosis?	
Do you know about the CFTR gene?	

Now, let's watch a video on Mutations and Cystic Fibrosis.

a). Genetics

<https://www.youtube.com/watch?v=8rbMQwzmQrk>



b). Mutations

<https://www.youtube.com/watch?v=vl6Vlf2thvl>



c). Cystic Fibrosis

<https://www.youtube.com/watch?v=ppM ZwjcEfxs>



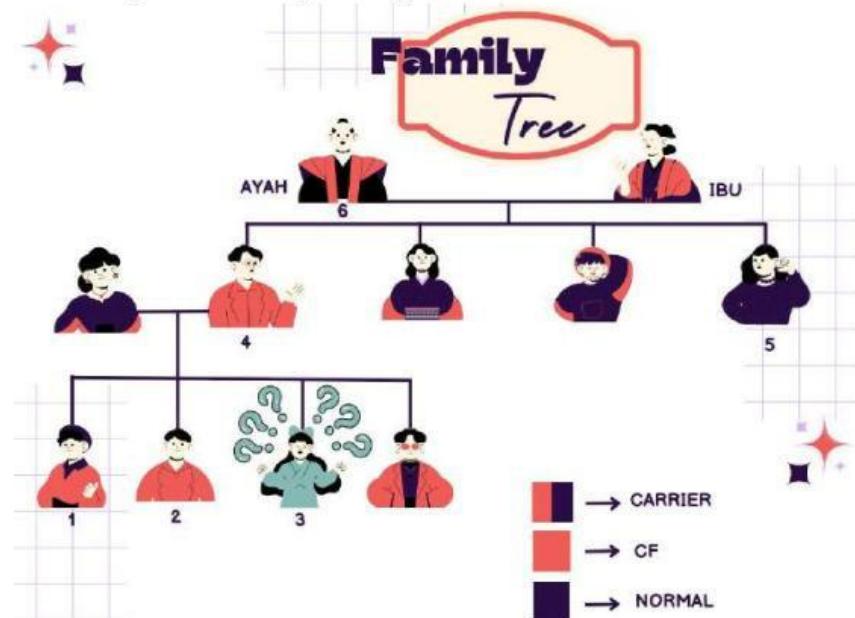
After watching the three videos above, give us your comments and thoughts!

B. Investigations

Cystic fibrosis (CF) affects 1 in 2500 births with symptoms including recurrent lung infections, intractable cough, salty sweat and abnormal stools in Australia. The gene associated with CF produces a protein responsible for transferring salt in and out of cells in our body. This gene is called the CFTR (Cystic Fibrosis Transmembrane Regulator) gene and is located on chromosome seven. Since each person has two copies of chromosome seven, each person also has two copies of the CFTR gene.

Modified from Article: Health Engine. (2008).Genetic Testing for Hereditary Diseases. <https://healthinfo.healthengine.com.au/genetic-testing-for-hereditary-diseases>

Here is the family tree of the phenotype.



The question is, can the child with number 3 get cystic fibrosis symptoms?

To answer this question, researchers extracted DNA and sequenced it from the 6 family members in the family diagram above.

SAMPLE 1

TTCCAAGTCTAGATGTAAAATATTAAGAACAGATCAACTTGGCAGTCAGATGAAAATA
TACTGAAT

SAMPLE 2

TTCCAAGTCTAGATGTAAAAAATTAAGAACAGATCAACCTTGGCAGTCAGATGAAAAT
GCGCTGAAT

SAMPLE 3

TTCCAAGTCTAGATGTAAAAAATTAAGAACAGATCAACCTTGGCAGTCAGATGAAAAT
GCGCTGAAT

SAMPLE 4

TTCCAAGTCTAGATGTAAAAAATTAAGAACAGATCAACCTTGGCAGTCAGATGAAAAT
GCGCTGAAT

SAMPLE 5

TTCCAAGTCTAGATGTAAATATTAAGAACAGATCAACCTTGGCAGTCAGATGAAAAT
ATACTGAAT

SAMPLE 6

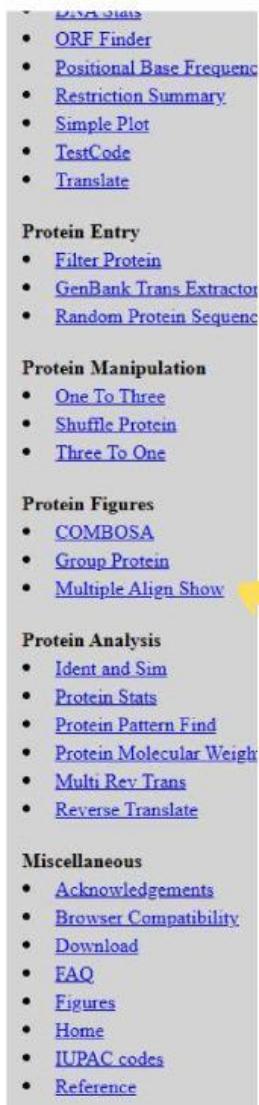
TTCCAAGTCTAGATGTAAAATATTAAGAACAGATCAACTTGGCAGTCAGATGAAAATA
TACTGAAT

1. Using manually

- a. Write back the results of DNA Sequences
- b. Look for differences from DNA Sequences in each sample
- c. Give a mark as a form of distinguishing DNA Sequences in each sample

2. Using *Sequences Manipulation Suite* (SMS)

- a. Access the following link <http://bioinformatics.org/sms/>



The Sequence Manipulation Suite

- The Sequence Manipulation Suite is a collection of web-based programs for analyzing and formatting DNA and protein sequences.
- The output of each program is a set of HTML commands, which is rendered by your web browser as a standard web page. You can print and save the results, and you can edit them using an HTML editor or a text editor.
- Check out some [figures](#) made using the Sequence Manipulation Suite.
- If you are having trouble using a program in the Sequence Manipulation Suite check the [FAQ](#).

[\[home\]](#)

Please make the steps in comparing the DNA Sequences above!

Please answer the following questions again.

Questions	Expected Answer
Do you know about "Sequence Manipulation Suite (SMS) ?	
What do you know about Genetics?	
What do you know about Gene Mutation?	
What are some examples of genetic diseases that you know?	
Do you know about Cystic Fibrosis?	
Do you know about the CFTR gene?	

Let's look at your answers after doing the activities in this lesson. If your answers change, become more detailed and better than before, congratulations! Your understanding is improving. However, if your answers after these activities remain the same, don't worry, please study again and consult your teacher.