SENIOR BIOLOGY

Blueprint of life and *Genetics: the Code Broken?*

NAME

SCHOOL / ORGANISATION

DATE

INSTRUCTIONS

1. Make sure you read the bold text in boxes throughout the worksheet as they contain important information

These boxes contain instructions explaining how to complete the next section of your worksheet

2. To find a particular disease or specimen you may need to refer to it's specimen number. Once you have found the bay, the specimen will have a purple label to help you locate it on the shelf.

Bay 12, 1417

Bay number — Specimen number

3. Detailed information about a specific disease or specimen will either be found on an info sheet, poster or next to the specimen itself. The following symbols are used throughout the worksheet.

Info sheet *A4 displays on table nearest to specimen*

Poster *located throughout museum*

Observation *Find exhibit and describe features*

IMPORTANT

Real people have generously donated their body so that medical scientists can learn about health and disease. Our donors deserve the utmost respect and admiration for their invaluable contribution to medical science.

INTRODUCTORY NOTES

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Blueprint of Life

In this part of the workshop you will address the following syllabus points;

Activity: Punnett Square – Online Punnett square tutorial

- distinguish between homozygous and heterozygous genotypes in monohybrid crosses
- perform an investigation to construct pedigrees or family trees, trace the inheritance of selected characteristics and discuss their current use
- distinguish between the terms allele and gene, using examples
- explain the relationship between dominant and recessive alleles and phenotype using examples
- solve problems involving monohybrid crosses using Punnett squares

Activity: Blood type – online module

- compare the inheritance of the ABO and Rhesus blood groups
- solve problems to predict the inheritance patterns of ABO blood groups

Genetics: The Code Broken?

In this part of the workshop you will address the following syllabus points;

Activity: BLAST

- distinguish between mutations of chromosomes, including;
    - rearrangements
    - changes in chromosome number, including trisomy, and polyploidy
- and mutations of genes, including;
    - base substitution
    - frameshift

Activity: BLAST

- process and analyse information from secondary sources to describe the effect of one named and described genetic mutation on human health
- process information from secondary data to outline the current understanding of gene expression

Activity Station 1: Punnett Square – Online Punnett square tutorial – iPad task

Punnett squares: http://bit.ly/mohdpunnetttute

Activity Station 2: Effect of Environment on cell Genotype – Skin Bay Specimens (Bay 12)
Compare specimens 903 and 2995, and the details about them in the catalogues.

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Activity Station 3: Blood type – online module
compare the inheritance patterns of Codominance instances

Genetics tutorial: http://bit.ly/mohdbiogentute

Activity Station 4: BLAST 1 – Computer Lab

Introduction

Today you are a medical research scientist in a pathology lab who has sequenced part of a patient's DNA. You are not sure what the DNA codes for, but you have 100 nucleotide base pairs of the DNA.

To find out what gene your nucleotide sequence codes for, you will be running an internet-based BLAST search. BLAST, or the Basic Local Alignment Search Tool, compares your sequence with a very large database of known DNA sequences that scientists around the world have compiled.

Find out what your mystery sequence encodes using a BLAST search:

1. Go to the BLAST website: http://blast.ncbi.nlm.nih.gov/Blast.cgi

2. Select "nucleotide" button.

3. Copy and paste the whole of one of the CFTR mystery sequences below into the "Enter accession number" box at the top of the page. **(Figure 1 at end of worksheet)**

>Mystery sequence 1 (in-frame del)

GAATTTCATTCTGTTCTCAGTTTTCCTGGATTATGCCTGGCACCATTAAAGAAAATATCATCGGTGTTTCCTATGATGAAT
ATAGATACAGAAGCGTCA

>Mystery sequence 2 (frameshift del)

TGGACCAGACCAATTTTGAGGAAAGATACAGACAGCGCCTGGAATTGTCAGACATATACCAAATCCCTTCTGTTGATTC
TGCTGACAATCTATCTGAA

>Mystery sequence 3 (frameshift substitution)

TGTGTCTGTAAACTGATGGCTAACAAAACTAGGATTTTGGTCACTTCTAAAATGGGACATTTAAAGAAAGCTGACAAAA
TATTAATTTTGCATGAAGGTAGC

>Mystery sequence 4 (synonymous mutation)

GACTTCATCCAGTTGTTATTAATTGTGATTGGGGCTATAGCAGTTGTCGCAGTTTTACAACCCTACATCTTTGTTGCAACA
GTGCCAGTGATAGTGGCTTTT

4. Under "Choose Search Set" select "Human Genomic + transcript" **(Figure 1 at end of worksheet)**

5. Hit the "BLAST" button at the bottom of the page.. **(Figure 1 at end of worksheet)**

6. Once your BLAST report comes up on the screen, scroll down to the "Alignments" heading. There will be a series of matching sequences that the search found on the database. **(Figure 2 at end of worksheet)**

7. In the top sequence identify the difference and circle it in the sequence above.

8. How many base pairs in the whole gene?_____

9. Which pairs does this short sequence correspond to on the whole gene?_____

10. Repeat for the other sequences then complete the following.

11. Write down the name of the gene your sequence matches. You'll find the name of the gene in the alignment data.

_____

12. Click on the "genbank" link at the top of the description. **(Figure 2 at end of worksheet)**

13. Scroll down and on the right hand side of the page click on "More about gene"

As a pathologist, you need to understand what this gene does in the body.

a) On what chromosome is this gene found?_____

b) What is the function of the protein coded by this gene?

_____

_____

c) What disorders are associated with defective versions of this protein?

_____

The CFTR gene is just one of many on human chromosome 7. Use the chromosome viewer in the banner in the top right hand of the screen to look at how many disorders are caused by genes on chromosome 7. **(Figure 3 at end of worksheet)**
http://web.ornl.gov/sci/techresources/Human_Genome/posters/chromosome/index.shtml

14. *Use this information to answer the following questions:*

g) How many base pairs make up this chromosome? Compare the number of base pairs on chromosomes 1, 7 and 21. _____

h) Browse through the various disorders associated with genes on this chromosome. Name five different parts of the body that could be affected by mutations on this chromosome?

_____

_____

_____

Scientists have found more than 1000 different mutations of the CFTR gene; Some have little or no effect on CTFR function, while others cause cystic fibrosis on a spectrum that varies from mild to severe. Click on this link to view a database of all known mutations in the CFTR gene.
http://www.genet.sickkids.on.ca/cftr/Home.html

The mutations present in your mystery sequences are by far the most common mutation observed in cystic fibrosis patients. The deletion of three base pairs in the CFTR gene causes the loss of an amino acid called phenylalanine at position 508 and the protein does not fold correctly.

Normal CFTR proteins, once translated and synthesised, are transported to the endoplasmic reticulum (ER) and golgi apparatus before they are integrated into the cell membrane. Incorrectly folded CFTR proteins are recognised by the ER and are destroyed before they reach the cell membrane.

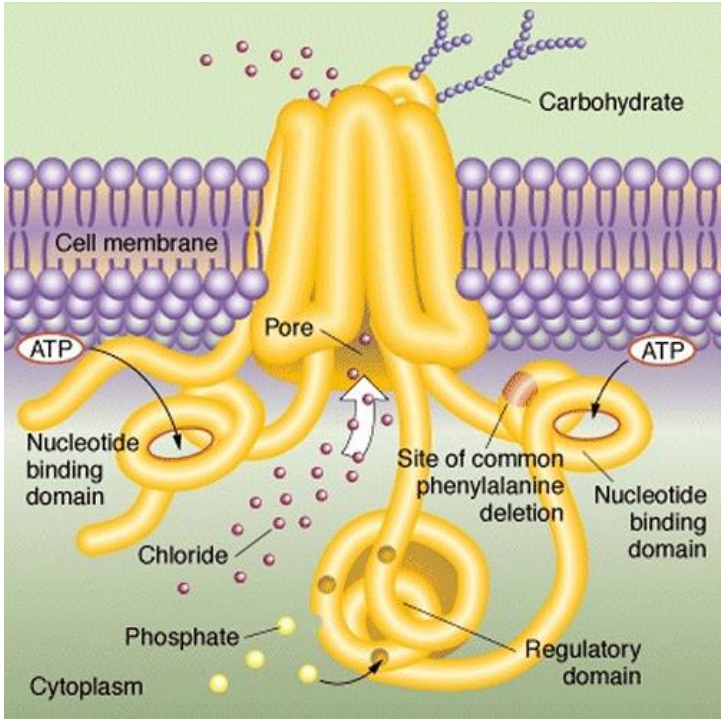

Diagram of how the CFTR protein forms a channel for chloride ions through the membrane of a cell. Image credit: http://massgenomics.org/2011/02/a-promising-new-drug-for-cystic-fibrosis.html

Activity Station 6: Virtual Microscopy

Go to the Microscopy room G05B

Open the link to the annotated CF slide:
https://www.best.edu.au/s/j5ne1cw5/xbfsetze?data=7%40[]!8%401!9%409408!10%40-4986&version=1

What organ is this slide from?

In this slide the stain makes; fibre (scar) Pink, DNA purple and leaves water/air/fat clear.

Using the annotations and zoom function label significant areas on this slide.

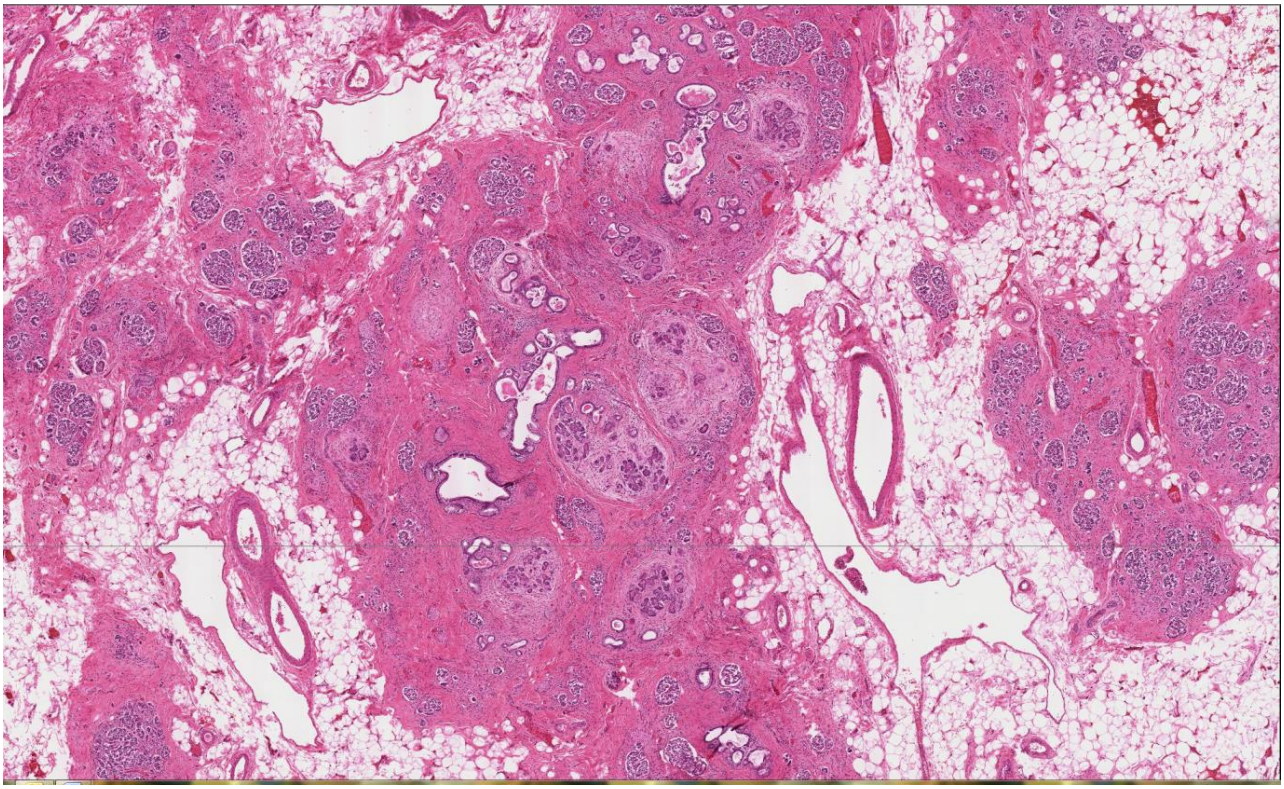

Figure 1 BLAST screen 1

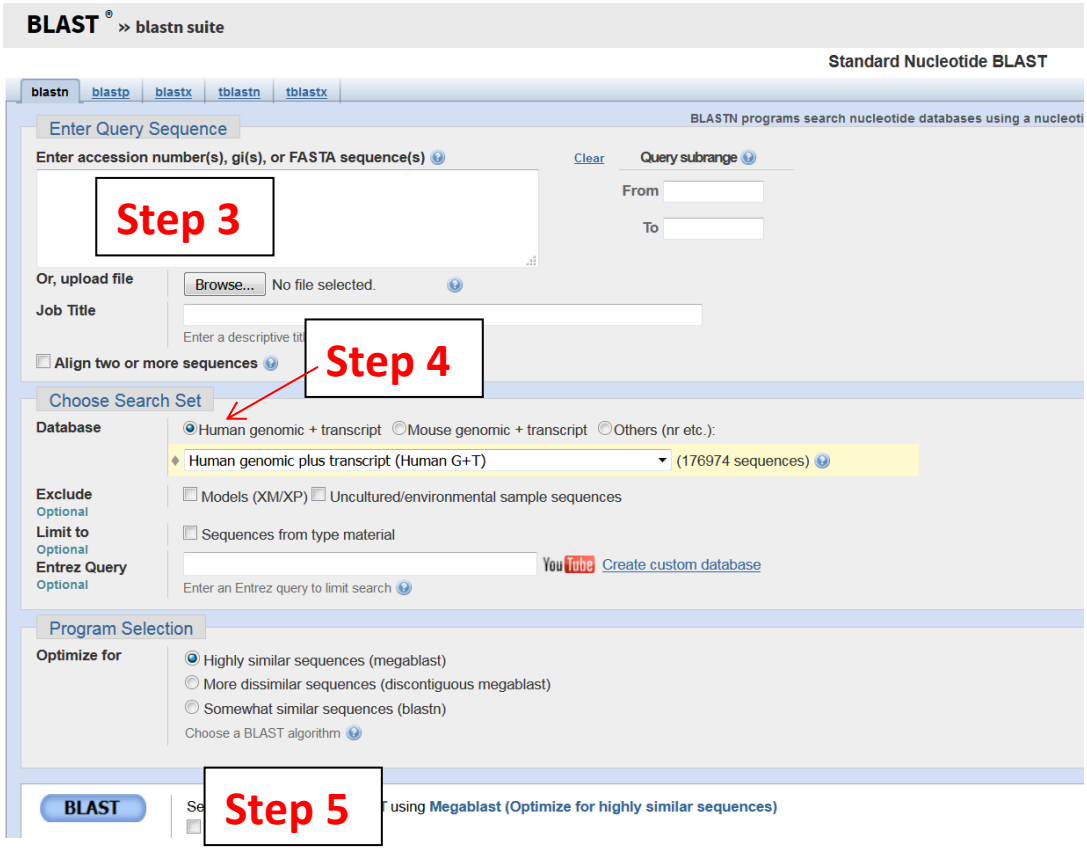

Figure 2 BLAST screen

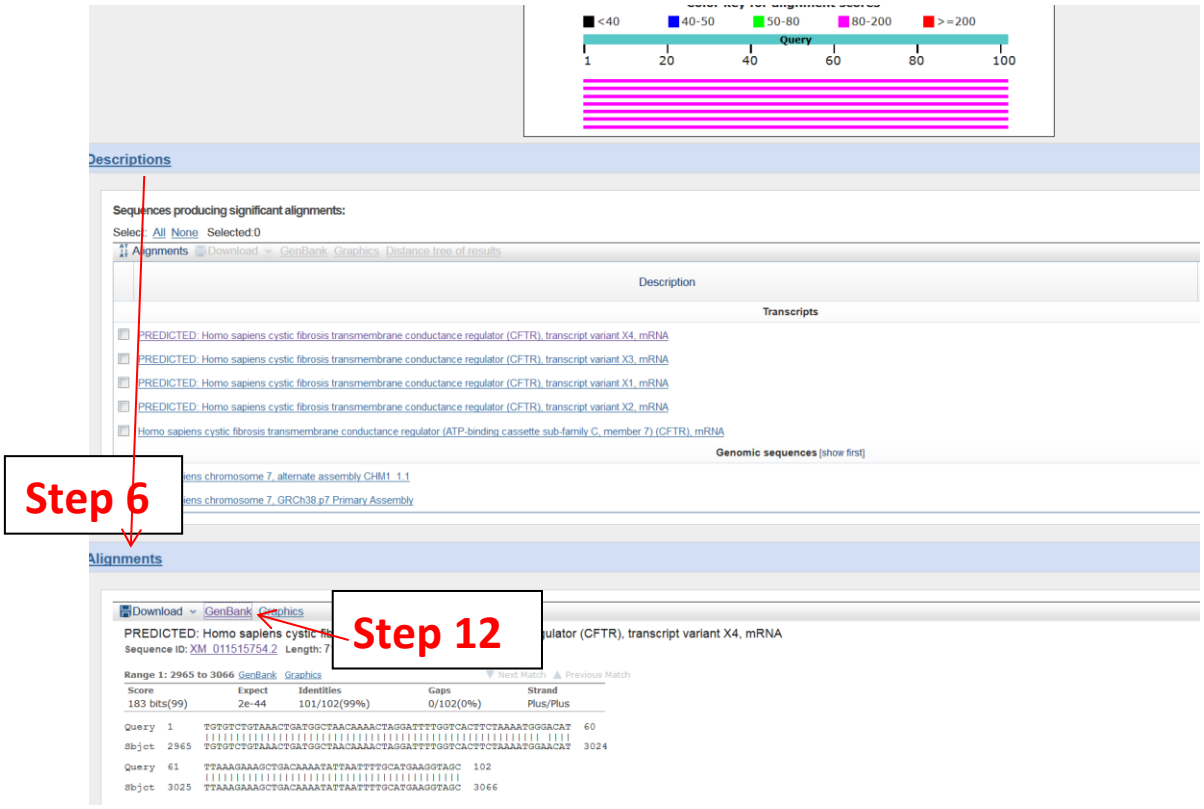

Figure 3 GENE poster screen