Cystic Fibrosis Activity
You Be the Clinician – Part 1

Scenario: You are a doctor in the pediatric intensive care unit (ICU) at a hospital. You have four patients who you suspect might have Cystic Fibrosis, but you can’t make a diagnosis until you run more tests.

The first step in determining whether or not your patients have Cystic Fibrosis is to collect Bronchial Alveolar Lavage Fluid (BALF) samples from their lungs and examine it for the presence of:

- Thick mucus
- Inflammatory cells
- Evidence of infection with the bacteria, *Pseudomonas aeruginosa*

Based on the phenotypes you observe, you will then decide which patients’ DNA needs to be sequenced to determine the genotype of the CFTR allele.

Instructions: DO NOT OPEN THE TUBES!

1.) Examine the 4 Bronchial Alveolar Lavage Fluid (BALF) samples on your desk from patients A, B, C, and D.

2.) Analyze each sample for:
   - mucus consistency (thick or watery/normal)
   - presence of inflammatory cells (white beads)
   - evidence of infection with the bacteria, *Pseudomonas aeruginosa* (green beads)

3.) Record your findings on the next page.

4.) Based on your observations, determine which two patients you think might have Cystic Fibrosis. Write your two choices in the spaces provided. You will vote as a class to determine whose DNA you want to send to the lab for genetic testing.
Record your results below:

**Patient A:**
- BALF sample
- Age: 1 year
- Sex: Female
- Mucus consistency: (Thick/ Normal)
- Inflammatory cells? (Yes/ No)
- *P. aeruginosa* infection? (Yes/ No)

**Patient B:**
- BALF sample
- Age: 2 years
- Sex: Male
- Mucus consistency: (Thick/ Normal)
- Inflammatory cells? (Yes/ No)
- *P. aeruginosa* infection? (Yes/ No)

**Patient C:**
- BALF sample
- Age: 3 months
- Sex: Male
- Mucus consistency: (Thick/ Normal)
- Inflammatory cells? (Yes/ No)
- *P. aeruginosa* infection? (Yes/ No)

**Patient D:**
- BALF sample
- Age: newborn
- Sex: Female
- Mucus consistency: (Thick/ Normal)
- Inflammatory cells? (Yes/ No)
- *P. aeruginosa* infection? (Yes/ No)

Based on what you have found, which two patients are the most likely to have Cystic Fibrosis? _____ and _____

You must now send DNA samples to the lab for further genetic testing...
Cystic Fibrosis Activity
You Be the Clinician - Part 2

Scenario: You sent DNA samples to a genetics lab for sequencing. The lab has sent you the DNA sequence for a small portion of the *cftr* gene from each patient.

Remember how proteins are made?

![DNA to RNA to Protein diagram]

You now have to take the sequences you obtained from the lab and convert them into a protein sequence. You will then be able to compare your patients’ CFTR protein to a normal CFTR protein to make the final diagnosis of Cystic Fibrosis.

Instructions:

1.) First, you must **transcribe** the DNA sequences below to RNA sequences, and write them in the spaces provided.

<table>
<thead>
<tr>
<th>DNA</th>
<th>RNA</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>U</td>
</tr>
<tr>
<td>T</td>
<td>A</td>
</tr>
<tr>
<td>C</td>
<td>G</td>
</tr>
<tr>
<td>G</td>
<td>C</td>
</tr>
</tbody>
</table>

Patient ___ *cftr* DNA sequence:

<table>
<thead>
<tr>
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<th>RNA</th>
</tr>
</thead>
<tbody>
<tr>
<td>T T A</td>
<td>T A G T A G A A A C C A C A A</td>
</tr>
<tr>
<td>RNA:</td>
<td>_ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _</td>
</tr>
</tbody>
</table>

Patient ___ *cftr* DNA sequence:

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</tr>
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<tbody>
<tr>
<td>T T A</td>
<td>T A G T A A A G A C C A C A A</td>
</tr>
<tr>
<td>RNA:</td>
<td>_ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _ _</td>
</tr>
</tbody>
</table>

2.) Now that you have **transcribed** the DNA sequence into an RNA sequence, you need to **translate** the RNA sequence into a protein sequence. Copy your RNA sequences on to the next page in the space provided.
3.) Find the amino acid in the chart below that corresponds with the RNA sequence, and write the protein sequence in the space provided.

Patient ___ RNA sequence:

| RNA: |          |          |          |          |          |          |          |          |          |
|------|----------|----------|----------|----------|----------|----------|----------|----------|
| AAs: |          |          |          |          |          |          |          |          |

Patient ___ RNA sequence:

<table>
<thead>
<tr>
<th>RNA:</th>
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<tbody>
<tr>
<td>AAs:</td>
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</tbody>
</table>

Part of the normal CFTR amino acid sequence

- E
- N
- I
- I
- F
- G
- V
- S

Partial list of Amino Acids:

<table>
<thead>
<tr>
<th>F</th>
<th>N</th>
<th>E</th>
<th>Y</th>
</tr>
</thead>
<tbody>
<tr>
<td>UUU, UUC</td>
<td>AAU, AAC</td>
<td>GAA, GAG</td>
<td>UAU, UAC</td>
</tr>
<tr>
<td>GGU, GGC, GGA, GGG</td>
<td>GUU, GUC, GUA, GUG</td>
<td>AUU, AUC, AUA</td>
<td></td>
</tr>
</tbody>
</table>

4.) Compare the patient’s amino acid sequence to the normal CFTR protein.

Which patient has Cystic Fibrosis?