

## **NeuroNotebook PROMPTS AND GRADING RUBRICS**

### **Prompt #1:**

1. Please provide a 1-page synopsis (double spaced with 0.5 margins) describing the genetic basis of the disease (i.e., which genes are mutated) in Treacher Collins Syndrome (TCS). Based upon the mechanisms that give rise to normal mandible and facial structure and the most common phenotype present in TCS, what cells are likely to be affected in patients diagnosed with TCS? Please use references to support and explain your work.
2. Draw the migratory pathway that a single neural crest cell must take in order to give rise to developing bone and cartilage of the head and neck.
3. Develop a hypothesis to explain how mutations in at least one gene associated with TCS might disrupt the normal migratory pattern of a cranial neural crest cell.

### **Prompt #2:**

1. In your previous notebook assignment, you identified a gene that is important for neural crest cell development. For this assignment, please write a single-page essay describing the function of the gene that you identified in Prompt #1. What cell types express the gene? What is the function of the gene? Does it promote specification, differentiation, proliferation, or cell death? At what point during development does its expression get activated? Are there other disorders associated with mutations in this gene? Your summary should include citations from the literature.
2. Based on what you learned in Question #1 above, refine your hypothesis to predict how your gene of interest affects neural crest cell development. For example "Mutations in Gene X interfere with cellular differentiation." Please focus on the fact that a hypothesis is a testable explanation for an observation. You will need to design an experiment to test your hypothesis for your next prompt.

### **Prompt #3:**

So far, you have developed a hypothesis designed to explain the genetic mutation that causes TCS and how mutations in that gene of interest interfere with neural crest cell development. For this prompt, you will need to design an experiment with which to test your hypothesis. You will have to read the literature to understand techniques/assays that other scientists use to understand gene function. This work requires you to have appropriate citations for your rationale and expectations.

1. The first part of this assignment is to identify an assay to test your hypothesis. In no more than a page, describe the technique you are going to use. What is the goal of the technique? What do you measure? Are you using cells? An animal model? If so, how many individuals do you need? What is statistical significance? How might significance be achieved? What are the advantages and disadvantages of the technique? Are there alternatives? Why is your choice the best choice compared to the alternatives?
2. Draw a depiction of the experimental setup. What materials do you need? How do you use them? Make sure that a person not familiar with your experimental procedure can understand your methodology.

### **Prompt #4:**

In the previous prompt, you developed an experimental procedure to test your hypothesis. You provided alternatives and advantages for your particular choice of experiment. Your experiment included a measurable outcome/variable. The independent variable of an experiment is the variable that an experimenter changes to test an outcome. For example, you might change the temperature of the room (independent variable) and then measure the number of people that leave the room (dependent variable). Now, it is time to determine what potential outcomes you might observe if you performed the experiment. For example, if I change the room temperature there are at three possible outcomes: 1) everyone leaves; 2) people are uncomfortable, but no one leaves; or 3) some people leave and others stay. Based upon this example and your proposed experiment from Prompt #3, answer the following questions.

1. In a single page, state the predicted/anticipated results of the experiment. Provide alternatives as I did in my example above. Remember, you are studying a cellular behavior, so if you are looking

at changes in cell proliferation, you will describe how mutations in your gene affect the number of proliferating cells.

2. Choose one of the predicted results from Question #1. For example, I observe that changes in X increase the number of proliferating cells. In no more than a single page, state a conclusion based on your results. For example, Gene A regulates the proliferation of Y. Now, extend those results to describe the phenotype you are studying (e.g., mutations in Gene A affect the formation of the hypothalamus because the cells that give rise to the hypothalamus fail to proliferate when Gene A is not functioning appropriately). Next, concisely summarize from the literature and provide accurate citations, how does your conclusion relate to what is known about the disease of interest, and how might your findings impact society?

PROMPT #1	Did Not Meet Expectations (10-20pts.)	Met Expectations (30-40pts.)	Exceeded Expectations (40-50pts.)	Score
<b>CONTENT</b>				
<b>Background and Introduction</b>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Does not meet the page limit and spacing requirements</li> <li><input type="checkbox"/> Unclear descriptions of phenotypes, disorders, developmental pathways, and/or brain regions associated with the exercise</li> <li><input type="checkbox"/> Does not use appropriate citations to support writing</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Does not meet the page limit or spacing instructions</li> <li><input type="checkbox"/> Summarizes some phenotypes associated with disease, but neglects to provide sufficient detail</li> <li><input type="checkbox"/> Partially describes developmental processes giving rise to facial development affected in disorder</li> <li><input type="checkbox"/> Summarizes some of the developmental pathways or genes that promote neural crest cells</li> <li><input type="checkbox"/> Uses appropriate citations to support the descriptions above</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Meets the instructions for page limit</li> <li><input type="checkbox"/> Provides clear and concise summary of phenotypes associated with disease</li> <li><input type="checkbox"/> Accurately describes the developmental processes giving rise to craniofacial development</li> <li><input type="checkbox"/> Includes the description of developmental pathways and/or regulatory genes that function in neural crest cells regions described</li> <li><input type="checkbox"/> Use literature review to provide appropriate background statements</li> </ul>	
<b>Connecting Phenotype to Pathways and Hypothesis Formation</b>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Unable to provide a synopsis of syndrome</li> <li><input type="checkbox"/> Lacks a detailed schematic of developmental pathways associated with neural crest cells</li> <li><input type="checkbox"/> Fails to connect disease, brain development, and signaling pathways</li> <li><input type="checkbox"/> No clear hypothesis is stated</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Selects an appropriate gene to study based on the phenotypes associated with disease</li> <li><input type="checkbox"/> Provides a detailed schematic of developmental pathways related to neural crest cells</li> <li><input type="checkbox"/> Unable to accurately connect specific genes to a unique developmental pathway and disease of interest</li> <li><input type="checkbox"/> States hypothesis accurately and clearly</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Selects an appropriate gene to study based on the phenotypes associated with disease</li> <li><input type="checkbox"/> Provides a detailed schematic of a developmental pathway related to neural crest cell migration</li> <li><input type="checkbox"/> Clear connections are made between neural crest cells, pathway analysis, and disease of interest</li> <li><input type="checkbox"/> States hypothesis accurately and clearly</li> </ul>	
<b>Writing Quality and Organization</b>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Unacceptable organization, lacks grammatical expertise, does not use higher order logic to create coherence</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Text is somewhat connected, minor grammatical errors, events are somewhat disjointed</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Expertly organized, with limited grammatical errors and smooth progressions</li> </ul>	

PROMPT #2	Did Not Meet Expectations (10-20pts.)	Met Expectations (30-40pts.)	Exceeded Expectations (40-50pts.)	Score
<b>CONTENT</b>				
<b>Gene Function</b>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Does not meet the page limit and spacing</li> <li><input type="checkbox"/> Unclear descriptions of gene function, development, and mutations</li> <li><input type="checkbox"/> Does not use appropriate citations to support writing</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Does not meet the page limit or spacing instructions</li> <li><input type="checkbox"/> Summarizes some aspects of the target gene, but lacks a comprehensive description of certain developmental aspects</li> <li><input type="checkbox"/> Partially describes the prospect of specific gene mutations in other similar or dissimilar disorders</li> <li><input type="checkbox"/> Uses appropriate citations to support the descriptions above</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Meets the instructions for page limit</li> <li><input type="checkbox"/> Provides a clear and concise summary of the target gene, its developmental function, and its expression</li> <li><input type="checkbox"/> Accurately describes the function of the gene of interest in other disorders or the lack thereof</li> <li><input type="checkbox"/> Use literature review to provide appropriate background statements</li> </ul>	
<b>Connecting Gene and Phenotype with Cellular Function</b>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Unable to describe gene function and identify a gene</li> <li><input type="checkbox"/> Fails to select an appropriate cell behavior to study</li> <li><input type="checkbox"/> Fails to connect NCC development, disease, gene function, and cell type specific expression</li> <li><input type="checkbox"/> No clear hypothesis is stated</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Partially connects gene function to cellular phenotype</li> <li><input type="checkbox"/> Selects at least one cell behavior in the appropriate cell type (i.e., the gene must be expressed in the cell type), but the gene function is not directly related to particular cell behavior</li> <li><input type="checkbox"/> Unable to concisely connect NCC development, disease, gene function, and cell behavior</li> <li><input type="checkbox"/> States hypothesis accurately and clearly</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Makes appropriate connections of specific cell types and phenotype</li> <li><input type="checkbox"/> Selects an appropriate developmental cell behavior for future study, based upon known gene function and cell type specific expression</li> <li><input type="checkbox"/> Clear connections are made between NCCs, disease, brain function, and cell physiology</li> <li><input type="checkbox"/> States hypothesis accurately and clearly</li> </ul>	
<b>Writing Quality and Organization</b>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Unacceptable organization, lacks grammatical expertise, does not use higher order logic to create coherence</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Text is somewhat connected, minor grammatical errors, events are somewhat disjointed</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Expertly organized, with limited grammatical errors and smooth progressions</li> </ul>	

PROMPT #3	Did Not Meet Expectations (10-20pts.)	Met Expectations (30-40pts.)	Exceeded Expectations (40-50pts.)	Score
<b>CONTENT</b>				
<b>Experimental Methodology and Description</b>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Does not meet the page limit and spacing</li> <li><input type="checkbox"/> Fails to select an assay and does not provide descriptions of variable to be measured, the model system, or the methods of the technique</li> <li><input type="checkbox"/> Does not provide alternatives or rationale for selection</li> <li><input type="checkbox"/> Does not use appropriate citations to support writing</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Does not meet the page limit or spacing instructions</li> <li><input type="checkbox"/> Summarizes some aspects of the technique, but does not provide detail about the variable measured, the model system, or the advantages and disadvantages of technique</li> <li><input type="checkbox"/> Partially describes alternative techniques and rationale for decision</li> <li><input type="checkbox"/> Uses appropriate citations to support the descriptions above</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Meets the instructions for page limit</li> <li><input type="checkbox"/> Provides a clear and concise summary of the technique to be used, variable to be measured, the model system and its advantages and disadvantages</li> <li><input type="checkbox"/> Accurately describes alternative techniques available and provides a rationale for the technique chosen</li> <li><input type="checkbox"/> Use literature review to provide appropriate background statements</li> </ul>	
<b>Experimental Set-Up Schematic</b>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Vague experimental schematic</li> <li><input type="checkbox"/> Does not address the materials needed for the assay</li> <li><input type="checkbox"/> Description of the assay is missing essential components for the reader to understand</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Experimental schematic lacks detail sufficient to comprehensively understand technique</li> <li><input type="checkbox"/> Does not describe the materials necessary to complete technique</li> <li><input type="checkbox"/> Some details are missing such that another individual cannot replicate the experiment</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Provides a detailed schematic of the experimental setup based on the literature</li> <li><input type="checkbox"/> Provides a detailed description of the materials needed for the experimental assay</li> <li><input type="checkbox"/> Provides a suitable description so that an individual could replicate their experimental assay</li> </ul>	
<b>Writing Quality and Organization</b>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Unacceptable organization, lacks grammatical expertise, does not use higher order logic to create coherence</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Text is somewhat connected, minor grammatical errors, events are somewhat disjointed</li> </ul>	<ul style="list-style-type: none"> <li><input type="checkbox"/> Expertly organized, with limited grammatical errors and smooth progressions</li> </ul>	

PROMPT #4	Did Not Meet Expectations (10-20pts.)	Met Expectations (30-40pts.)	Exceeded Expectations (40-50pts.)	Score
<b>CONTENT</b>				
<b>Predicted Results</b>	<input type="checkbox"/> Does not meet the page limit and spacing <input type="checkbox"/> Does not provide any alternative outcomes <input type="checkbox"/> Does not use appropriate citations to support writing	<input type="checkbox"/> Does not meet the page limit or spacing instructions <input type="checkbox"/> Provides less than 3 alternative results for the experimental outcome <input type="checkbox"/> Uses appropriate citations to support the descriptions above	<input type="checkbox"/> Meets the instructions for page limit <input type="checkbox"/> Provides at least 3 predictions/anticipated results in sufficient detail to support or refute hypothesis <input type="checkbox"/> Use literature review to provide appropriate background statements	
<b>Conclusions: Connecting Cells to Disease</b>	<input type="checkbox"/> Unable to determine a predicted outcome of any kind <input type="checkbox"/> Does not connect the experimental outcome to the disease or development <input type="checkbox"/> Fails to connect brain development, disease, brain function, cell physiology, and impact <input type="checkbox"/> No clear hypothesis is stated	<input type="checkbox"/> Unable to make accurate conclusions from predicted outcomes <input type="checkbox"/> Does not describe the experimental outcome and its association to disease phenotype or brain development <input type="checkbox"/> Unable to concisely connect brain development, disease, brain function, cell physiology, and impact <input type="checkbox"/> Use literature review to provide appropriate background statements	<input type="checkbox"/> Makes appropriate conclusion from the observed results <input type="checkbox"/> Provides detailed explanation as to how experimental outcome is associated with disease phenotype and brain development <input type="checkbox"/> Clear connections are made between brain development, disease, brain function, cell physiology, and impact <input type="checkbox"/> Use literature review to provide appropriate background statements	
<b>Writing Quality and Organization</b>	<input type="checkbox"/> Unacceptable organization, lacks grammatical expertise, does not use higher order logic to create coherence	<input type="checkbox"/> Text is somewhat connected, minor grammatical errors, events are somewhat disjointed	<input type="checkbox"/> Expertly organized, with limited grammatical errors and smooth progressions	