Bend 3 Remediation/Genetics Unit Review Name: _____KEY_____ Hr: ___

Humans have a variety of genetic traits. For this task we will explore the Human traits below.

Hearing - Normal Hearing or Hearing Loss
Tay-Sachs Disease - Have Tay-Sachs (die young) or Do not have Tay-Sachs
Cheeks - Dimples or No Dimples
Cystic Fibrosis - Have Cystic Fibrosis or Do not have Cystic Fibrosis

Tay Sachs disease progressively destroys nerve cells and children with this disease traditionally die very young. A team of scientists is curious about how Tay-sachs is inherited. Since humans are hard to study they use a mouse model that also has the Tay-Sachs gene. Since mice mature faster than humans they are able to breed before dying from the Tay-Sachs disease. They cross a pure mouse with Tay-Sachs and a pure Mouse without Tays Sachs and count the number of offspring with each trait.

P Generation Cross: Pure male mouse WITH Tay-Sachs X Pure female mouse W/out Tay-Sachs

Generation	Mice WITH Tay-Sachs	Mice W/out Tay-Sachs
F1 (Offspring of the P generation)	0	44

Then the scientists cross mice from the F1 generation with each other and counted the following offspring.
F1 Generation Cross: F1 male mouse W/out Tay-Sachs X F1 female mouse W/out Tay-Sachs

Generation	Mice WITH Tay-Sachs	Mice W/Out Tay-Sachs
F2 (Offspring of the F1 generation)	15	41

Question: How is Tay-Sachs inherited in mice?

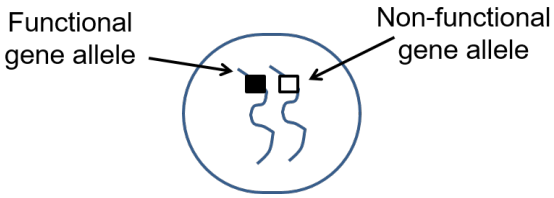
1. **Claim:** **Tay-Sachs is recessive and mice receive one allele from each parent.**

2. **Evidence:** Calculate the % of both Tay-Sachs and Non-Tay-Sachs mice in each generation. Also determine the ratio of mice WITH Tay-Sachs to mice W/Out Tay-Sachs in each generation

	% Mice WITH Tay-Sachs	% Mice W/Out Tay-Sachs	Ratio of WITH : W/OUT
F1 Generation	0%	100%	0:1
F2 Generation	27%	73%	1: 2.7

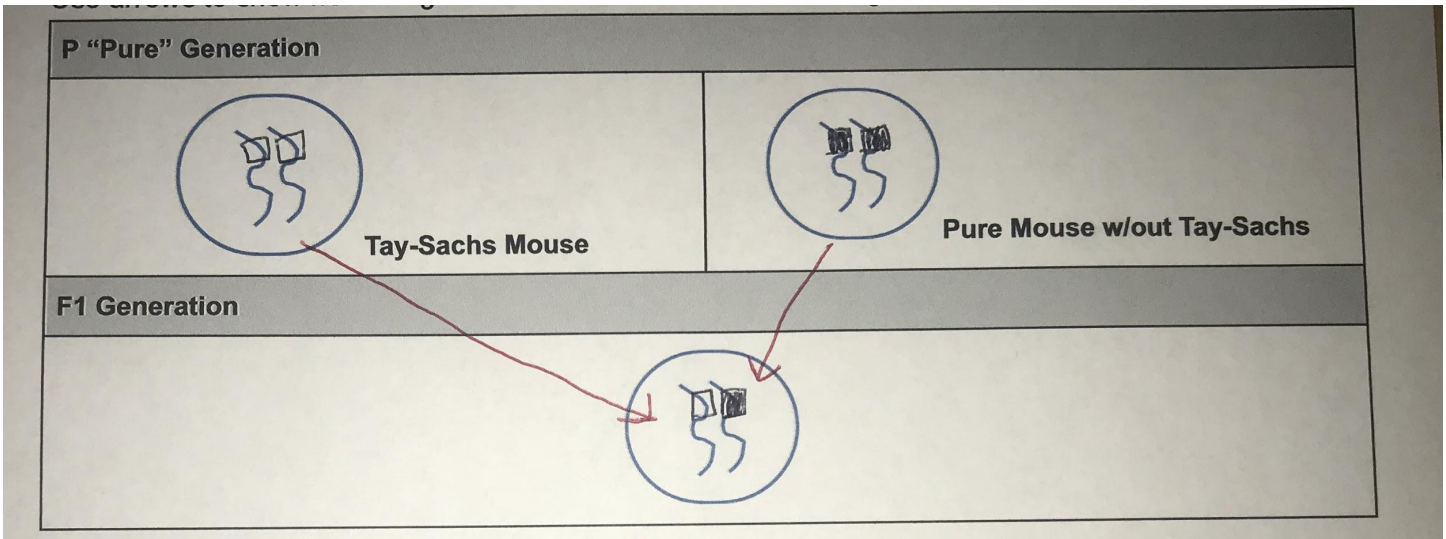
3. **Reasoning -** Use the information below to create a model demonstrating your reasoning on the next page.

The functional version of HEXA gene codes for an enzyme that breaks down toxins that destroys nerve cells. The Non-functional version of the HEXA gene does not break down those toxins.



Model Part 1-

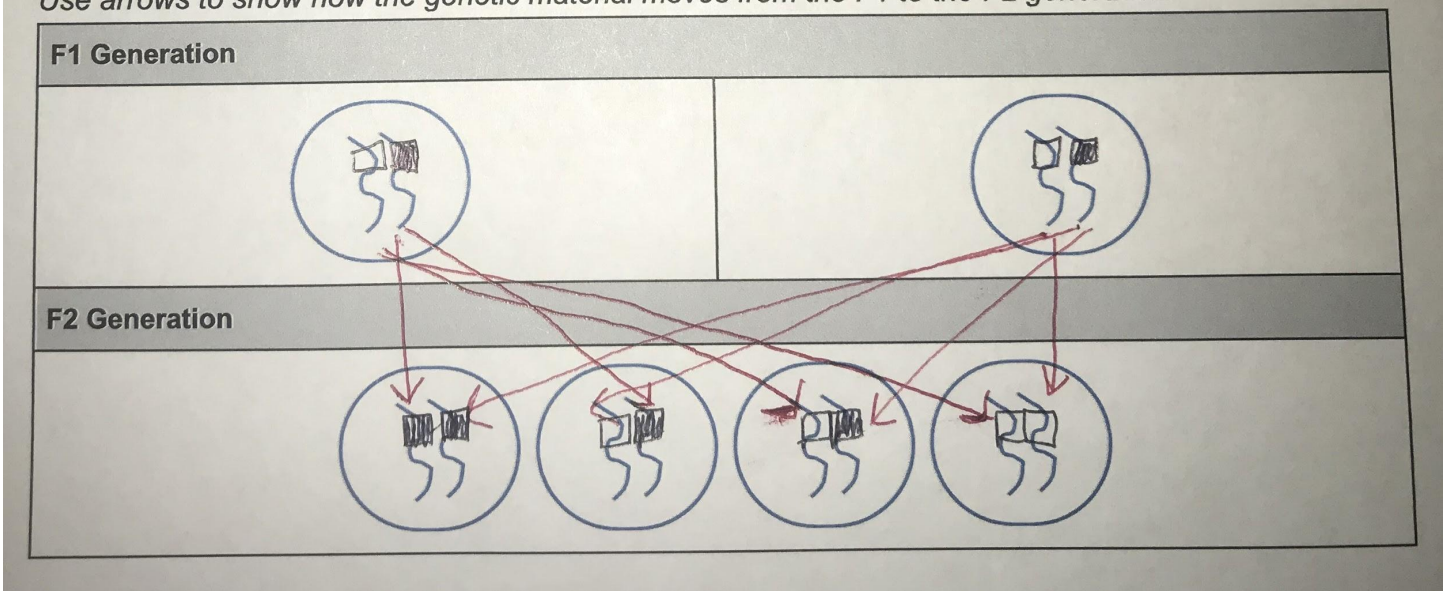
Use arrows to show how the genetic material moves from the P generation to the F1 generation.



Model Part 2-

Complete the model below showing the different offspring that can be produced by two F1 parents.

Use arrows to show how the genetic material moves from the F1 to the F2 generation.



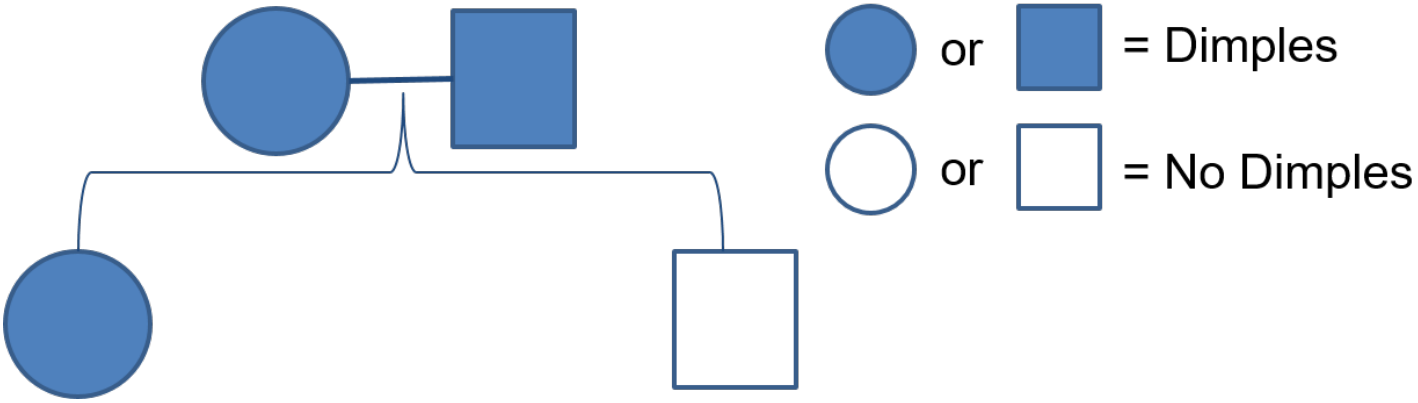
4. F1, Heterozygous individuals have ___1_____ copy(ies) of the mutant gene. So they have only about 50% of the enzyme activity. These hybrids do not show the Tay-Sachs symptoms. The Tay-Sachs trait is considered …
    a. Recessive
    b. Dominant
    c. Co-dominant
    d. Incomplete Dominant

5. Why did you make the choice above? Just one copy of the functional Tay-Sachs allele provides enough enzyme for the person to be healthy and not show Tay-Sachs symptoms. It takes two copies of the non-functional allele for the person to show symptoms therefore the Tay-Sachs trait is considered recessive.

Dimples

Another human trait is dimples on the cheek. In one family both parents have dimples but they have one girl with dimples and one boy without dimples.

6. Create a pedigree to represent this trait.



7. Based on the pedigree is dimples dominant or recessive? Why?

Dimples is dominant. Since both parents have dimples but they have a son who does not, No Dimples must be the recessive trait. It must require two copies of a certain allele, not to have dimples.

8. Based on this pedigree, what is the chance that they will have another child with dimples?

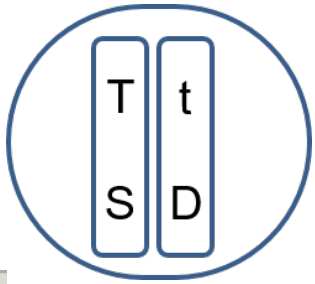
Both parents must be carriers (Dd). Dd X Dx = 75% chance of having a child with dimples.

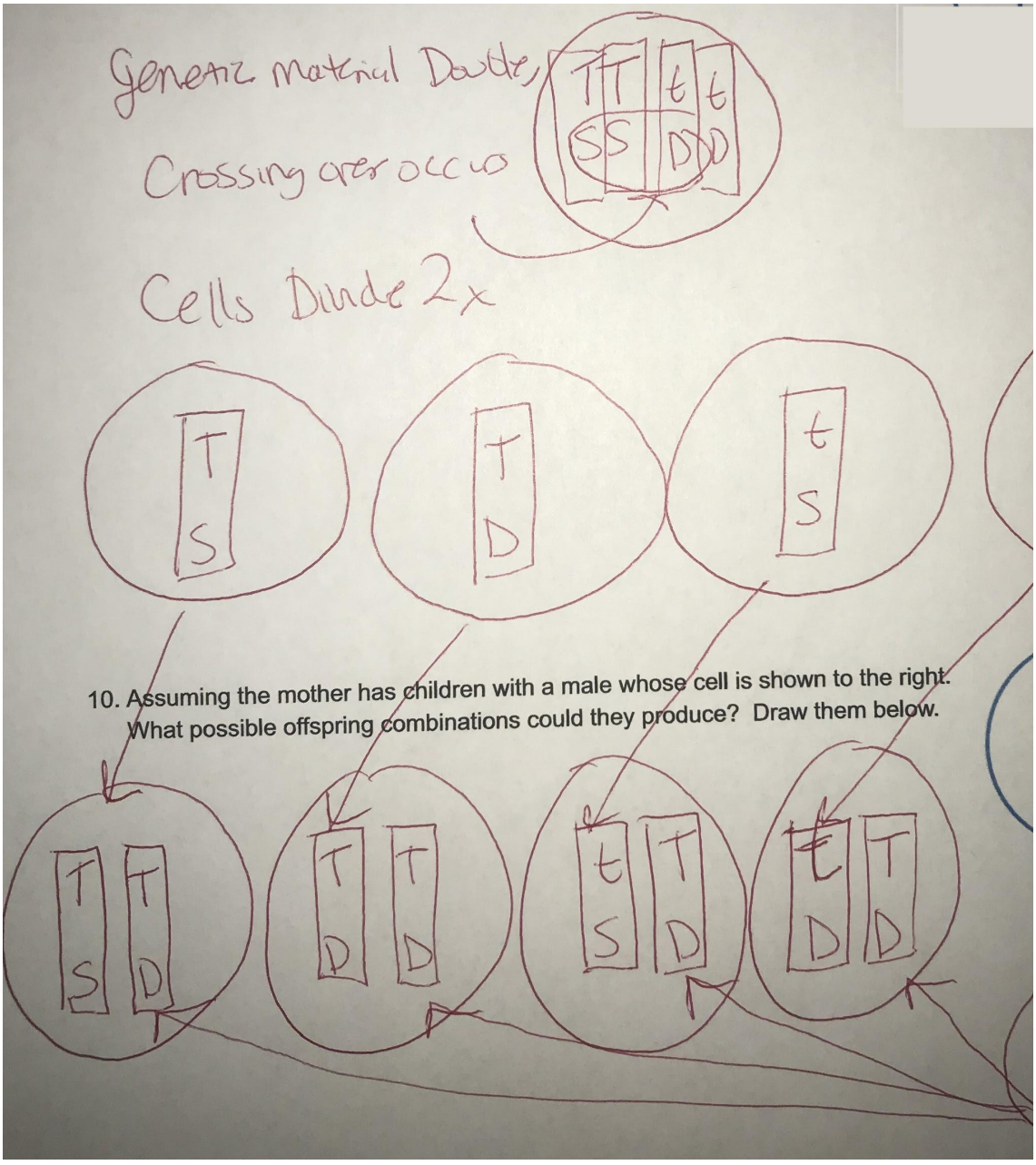
Hearing Loss

The Tay-Sachs gene is located on Chromosome 15. Another gene on chromosome 15 is STRC. The STRC gene helps code for a protein called stereocilin that helps hair cells in the ear convert sound into electrical impulses in the brain. A non-functional version of the STRC gene alters the structure of the hair cells and preventing them from reacting normally to sound waves.

T = Functional Tay-Sachs Allele **t=** Non-Functional Tay-Sachs Allele
S = Functional STRC allele **D=** Non-Functional STRC Allele

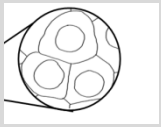
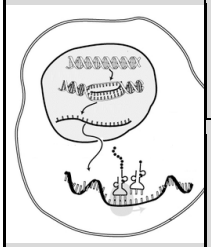
9. The diagram to the right shows a cell of a mother, only including her chromosome 15. This mother is heterozygous for both traits. Create a model, including crossing over that shows the different eggs she can produce.





Continue with the remaining questions for your Genetics Unit Review

11. Researchers are looking for a STRC gene mutations in individuals with hearing loss. They compare the STRC gene in patients who can and cannot hear. The DNA is pictured below.

Organisms		**Patient who can hear**					**Patient who has hearing loss**				
Cells How do cells influence the observable trait? 		Is the stereocilin protein functional? Yes or No					Is the stereocilin protein functional? Yes or No				
	**Amino Acids**	**Tyr**	Pro	Asp	Ser	Gly	Tyr	Asp	Ser	Gly	
	RNA	UAU	CCC	GAC	UCU	GGU	UAU	GAC	UCU	GGU	
Molecules What is happening on the molecular level?	**STRC Gene DNA**	ATA	GGG	CTG	AGA	CCA	ATA	CTG	AGA	CCA	

Second letter

First letter	U	C	A	G	Third letter
U	UUU, UUC Phe UUA, UUG Leu	UCU, UCC, UCA, UCG Ser	UAU, UAC Tyr **UAA Stop** **UAG Stop**	UGU, UGC Cys **UGA Stop** UGG Trp	U C A G
C	CUU, CUC, CUA, CUG Leu	CCU, CCC, CCA, CCG Pro	CAU, CAC His CAA, CAG Gln	CGU, CGC, CGA, CGG Arg	U C A G
A	AUU, AUC, AUA Ile AUG Met	ACU, ACC, ACA, ACG Thr	AAU, AAC Asn AAA, AAG Lys	AGU, AGC Ser AGA, AGG Arg	U C A G
G	GUU, GUC, GUA, GUG Val	GCU, GCC, GCA, GCG Ala	GAU, GAC Asp GAA, GAG Glu	GGU, GGC, GGA, GGG Gly	U C A G

12. Does the shape of the protein change due to the mutation in DNA? Why or why not?
Yes the shape of the protein changes. There was a deletion of some of the DNA and therefore one of the amino acids (Pro) was not produced. The number and order of amino acids determines the shape of the protein. The different shape of the protein makes the stereocilin protein non-functional.

S = Functional STRC allele **D=** Non-Functional STRC Allele

13. Patients with just one copy of the non-functional STRC allele show partial hearing loss. Patients with two copies show complete hearing loss. If a parent with normal hearing has children with a parent who has partial hearing loss, what is the chance that they will have a child with normal hearing?

SS X SD = 50% SS and 50% SD

Therefore they have a 50% chance of having a child with normal hearing (SS)

Cystic Fibrosis

Cystic fibrosis (CF) causes the body to produce thick, sticky mucus that clogs the lungs, leads to infection, and blocks the pancreas, which stops digestive enzymes from reaching the intestine where they are required in order to digest food. These children struggle to maintain their body weight.

14. In 1990 Scientists were curious about how second-hand smoke at home affected children with Cystic Fibrosis. They had access to students at a Cystic Fibrosis summer camp that both were and were not exposed to secondhand smoke. Design a fair investigation that answers this question.

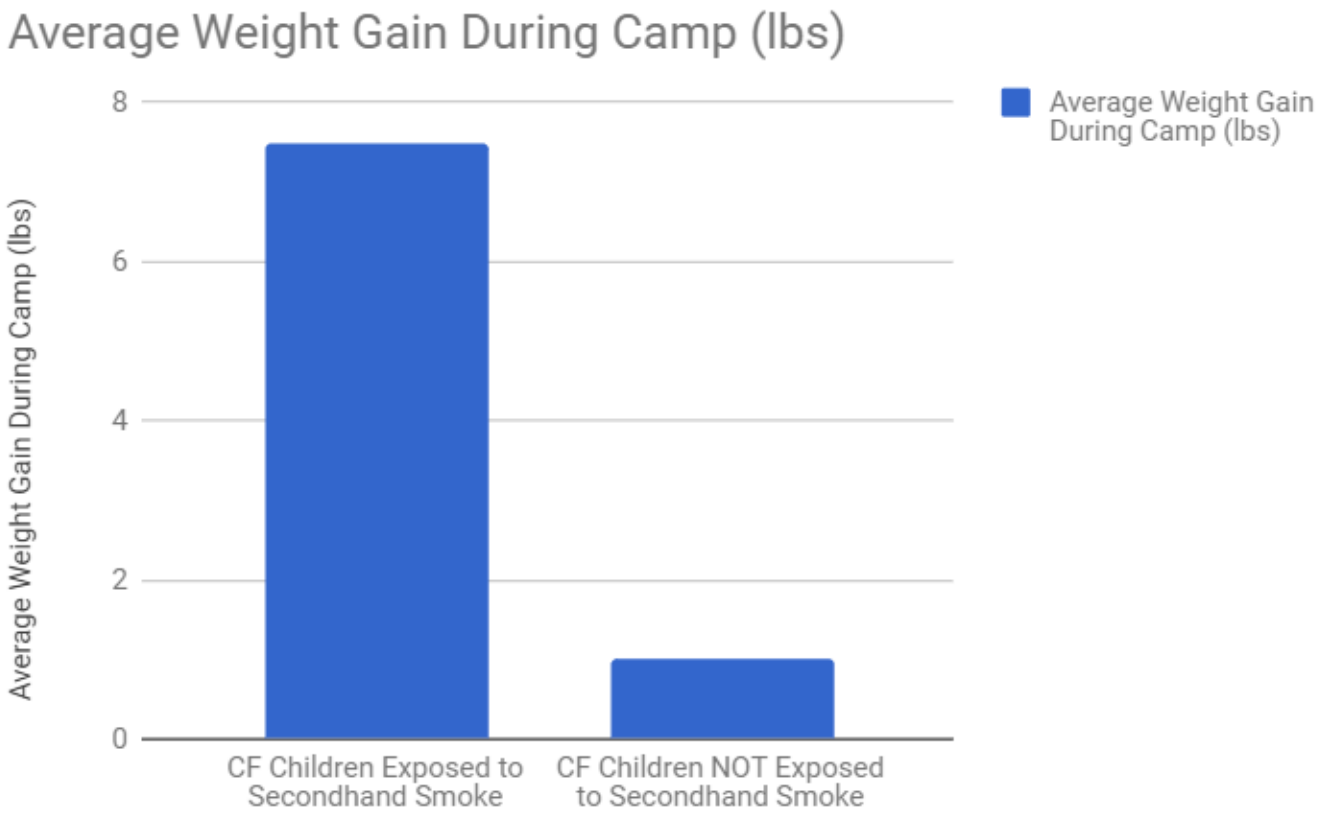
A fair investigation will measure both Cystic Fibrosis campers from second-hand smoke homes and Cystic Fibrosis campers not from second-hand smoke homes in the same way. It will measure how they entered camp and how they left camp.

For example, the actual study in 1990 measured the weight of students as they arrived at camp and at the end of the two weeks of camp. They did this in the same way for campers from second-hand smoke homes and campers not from second-hand smoke homes. Correct answers might also including examining the breathing rate or heart rate of these campers.

15. How might Cystic Fibrosis affect a person's ability to maintain homeostasis while exposed to secondhand smoke?

As we learned in first semester, Homeostasis is the body's ability to function and stay alive by maintaining a narrow range of internal conditions including nutrient and oxygen levels. The mucus caused by Cystic Fibrosis makes it harder for a person to taken in oxygen or nutrients. Secondhand smoke, may make it even harder for people to breathe in the presence of the mucus.

A negative feedback mechanism helps the body stay within this narrow limits by reversing the direction of change. For example a person with Cystic Fibrosis who is struggling to maintain the proper nutrients or oxygen levels might eat more, or breathe more often.



16. Children stayed at the camp for two weeks, and the scientists gathered the data above. Interpret the graph. What does it tell us about the effect of Secondhand Smoke on Cystic Fibrosis children?

CF children who lived in homes where they were exposed to secondhand smoke gained more weight while at camp and away from the secondhand smoke than those CF children not exposed to secondhand smoke at home.

This tells us that secondhand smoke at home may be limiting the ability for CF children to gain weight at home.

17. Is the weight of these children determined by genetics, the environment, or both? How do you know?

The weight of these children is determined by both genetics and the environment. The presence of two CF alleles and the cystic fibrosis trait makes it hard for all CF children to maintain their body weight. However, CF children in an environment with secondhand smoke have an even harder time maintaining their body weight and gain more weight when moved to an environment (like the camp) where they are not exposed to secondhand smoke.