Unit 4: Appendix D

Challenging the Immune System: diagnosis and understanding the disease

Case 3: Jackie and Daryl- a young couple worries about their baby's future

Part 1: Hearing the news

Jackie and Daryl were thrilled to be new parents. They could hardly believe she was finally here and were already enjoying every second of their time with their new daughter. Because she was born in Washington State, newborn Mikaela was taken away from excited parents for a routine newborn screening procedure only after being alive for two days. A small amount of blood was extracted from her heel and the nurse sent her blood to the lab for newborn screening tests. Mikaela did not cry and returned to the arms of her mother. Jackie was desperately in need of some sleep, so the new mother and her precious daughter cuddled while getting some much needed rest.

Over the next week at home, Jackie and Daryl were getting accustomed to their new way of life with baby Mikaela. Alternations of sleeping and eating was the routine. It was all becoming fairly normal, until they got a call from Dr. Hudson, their obstetrician. Dr. Hudson had been Jackie's doctor for many years and was there to deliver little Mikaela. Unfortunately, she got the startling results from the newborn screening and wanted to talk to the couple herself. She told them that the preliminary testing of Mikaela's blood sample gave a postivie result for the sickle cell gene. Jackie didn't know what to say, but knew that her grandfather had sickle cell disease. She was worried sick and wanted to hear more. She made an appoitnment for the family to meet with a specialist so they could hear more about the disease and figure out what to do for their daughter.

Questions: *answer the following questions in groups. Use provided resources or finish research for homework after reading the rest of this case study.*

1. What are routine screenings for newborns? Are they mandatory? What do these tests look for?

2. What do you know about sickle cell disease? Where have you heard about it? Do you know anyone who has sickle cell disease?

3. Homework/In class internet research:
    a. What are the symptoms of sickle cell disease?
    b. What is the inheritance pattern of sickle cell disease?
    c. What is the cause of sickle cell disease?
    d. What are the treatment options?

Part 2: Learning more

In their meetinng with a genetic counselor, Jackie and Daryl were able to hear more about the disease and to get information about what to do for their daughter. The counselor explained that they were high risk because they were African Americans and had family members with sickle cell disease. In the United States, sickle cell disease is most prevalent among African Americans; about one in 12 African Americans and about one in 100 Hispanic Americans carry the sickle cell trait.

The counselor explained that both Jackie and Daryl are carriers of the disease, even though they do not show symptoms. There was a one in four chance that their child would have sickle cell disease, since they are both carriers.

The good news is that early treatment for Mikaela can prevent some of the complications that may come with sickle cell anemia. Young children with the disease are especially susceptible to certain dangerous bacterial infections, such as pneumonia and meningitis. However, because they know the results from the screening, they are able to start treatment before any signs have appeared.

Studies have shown that treatment with penicillin, beginning by 2 months of age and continuing to about 5 years, dramatically reduces the risk of these infections and the deaths that result from them. Newborn screening alerts the physician to begin antibiotic treatment before infections occur.

Currently the only cure for sickle cell disease is bone marrow transplantation, but this is not something Jackie and Daryl need to be thinking about at this point. You will have to

see how Mikaela responds to preliminary treatment options before thinking ahead to the future.

Questions: Answer the following questions in groups. Jot down notes during your discussion and do research when needed.

1. What is an genetic counselor? What do they look for and how do they help families?

2. What is sickle cell disease? What is anemia?

3. What is the problem with having sickled cells? What are the risks associated with having cells in the blood that are “sickled”?

4. What other questions should the family ask the doctor or genetic counselor? Make a list of some important questions you would ask if you were this family.