

Diagnosis of a Congenital Disorder

by

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Part I – “We are expecting!”

Excited and overwhelmed, John and Shirley Reid have just found out that they are expecting a child once again. After Shirley’s previous two miscarriages, it seemed that the chances of this occurring were very slim. Due to her history, Shirley wanted to make sure she took all the prenatal precautions necessary to have a normal and healthy baby: prenatal vitamins, a healthy diet plan, and prenatal testing (not before the 9th week of pregnancy). Dr. Sheppard, their OB/GYN, had advised the Reids that although there could be some complications and harmful risks involved (fetal only) based on their history of miscarriages, it was in their best interest to have prenatal testing. John completely agreed with Dr. Sheppard, but Shirley was hesitant. She feared the risks involved in the procedures, and could not bear the thought of losing another baby prematurely. Shirley also had doubts because her mother, a devout Catholic, had instilled in her very strong feelings against prenatal procedures— that by having the procedures done they would be interfering in “God’s Plan.”



Questions

1. What are the essentials for prenatal care?
 - a. What types of prenatal vitamins are consumed? Why?
 - b. What composes a healthy prenatal diet? Why?
2. Taking into account her history of miscarriages and the risks involved in these procedures, should Shirley decide to have prenatal testing?

Part II – “Something is wrong with our baby”

Shirley decides to have an ultrasound in the 16th week (2nd trimester), which isn't known to have any negative side effects on a fetus' health. After the procedure, Dr. Sheppard informs the Reids that the ultrasound revealed some type of vascular malformation in the caudal half (lower trunk/extremity) of the fetus. Abandoning her religious views on invasive prenatal testing, Shirley decides to have amniocentesis, a test that could reveal more information about the malformation. After waiting for several weeks to have their questions answered, the Reids are informed that the fetus is female but the results are inconclusive as to the type of vascular disorder. This means that the Reids won't know what type of vascular malformation their child has until after the birth. Dr. Sheppard explains to them what options or alternatives they have, including terminating the pregnancy or waiting to see how severe their child's vascular disorder is and whether it can be corrected with surgery. John and Shirley are determined to have the baby but have serious doubts as to whether they will be able to handle what's to come.

Questions

1. Describe the procedure of ultrasound and its benefits. When is it used? What are the disadvantages?
2. Describe the procedure of amniocentesis and its benefits. When is it used? What are the disadvantages? Why is amniocentesis a better diagnostic option than ultrasound?
3. In your opinion, what are the circumstances that would lead parent(s) to abort their unborn fetus?
4. Discuss the reasons a person might have against prenatal testing or abortion.

Part III – “Your daughter has Sirenomelia”

The Reids decide to continue with the pregnancy, giving their little girl the name Emily, and hope for the best. During Emily’s birth, Dr. Sheppard immediately notices that the baby’s lower extremities are fused together, a condition known as Sirenomelia, a very rare and possibly fatal congenital defect. Children with Sirenomelia are born without external genitalia, abnormal kidneys, deficient vasculature supplying the abdomino-pelvic area, and other compromised internal organs, including the heart and lungs. Emily is born with just a portion of a kidney, prompting immediate surgery to prolong her life beyond the prognosis of a few days without the surgery. The Reids are determined to do everything to help their little girl survive and ease her pain and the challenges she faces.

Emily beats the odds and lives to celebrate her ninth birthday. Soon after Emily’s birthday, Dr. Sheppard approaches the Reids with the news that corrective lower extremity surgery is finally possible for Emily. Because Emily’s health has improved over the last few years, she is stable enough to undergo the complicated and risky surgical procedure of separating her lower extremities. The doctor warns Emily’s parents however that because Emily’s surgery will take the better part of a day, lethal complications could arise during any part of the procedure. Further complicating the situation, Dr. Sheppard add that if Emily does not have the surgery within the next few years she could develop further complications with her kidneys as she begins to grow and develop into pre-adolescence.

After considering the risks involved, John and Shirley decide that corrective lower extremity surgery is their daughter’s best chance to live a normal life. Shirley sits down with Emily a few days later to discuss the procedure and their decision. Much to Shirley’s surprise, Emily tells her that she does not want the surgery, explaining that she is happy with the way she looks. Emily tells her mother that she understands that all people are different in their own way and that she just happens to have been born “more different” than most people.

Questions

1. What are congenital disorders? What are some of the suspected causes of these disorders?
2. What are the life threatening consequences of Sirenomelia?
3. Do you agree with performing this corrective surgery knowing the risks involved? Why or why not?
4. Who has the right to decide when a child (under 18 years) should have possibly lethal corrective surgery?
5. Of the characters presented, who do you agree with most? Why?

Epilogue (optional)

Emily's parents honored her request to abstain from the corrective lower extremity surgery. Emily Reid died at the age of 10 years and 2 months due to renal and cardiac complications. Chronologically, she lived beyond the life expectancy of a young girl suffering from Sirenomelia, but she lived her life the way that she chose to live it, leaving her body looking the same way it did the day she was born.

This case study was inspired by the life of Shiloh Pepin.



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