A Sickeningly Sweet Baby Boy: A Case Study on Autosomal Recessive Inheritance

*by*

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Part I—Failure to Thrive

Emma and Jacob Miller were so excited at the birth of their baby Matthew.

“Jacob, he’s just so perfect! Just one problem though, it looks like he has your hairline!” Emma teased her husband who, though only 32, was balding.

“Emma, I spent all that time painting the baby’s room and I just hope that he’s not color blind like your father or he won’t be able to see it!” Jacob responded.

Both the pregnancy and delivery had been uneventful. But in the back of their minds, they really were worried

because their first child, Samuel, died at the age of nine days.

By the fifth day after birth, Matthew began to have trouble nursing and by the seventh day he had completely stopped

feeding. Emma and Jacob were frantic because it seemed to them that Matthew might also die.

“What is going on with our family? Another sick baby?” Jacob thought to himself.

Emma and Jacob rushed him to the emergency room. Although Mathew’s limbs were rigid and he had had a seizure,

the examination showed no infection and his x-rays were normal. The doctor also did routine lab tests on his blood

and urine.

“Doctor, do you think that this funny smell in Matthew’s diapers has anything to do with his problem?” Emma asked. “I brought one along so that you could smell it too.”

*Questions*

1. What additional information would you want to know to understand Emma and Jacob’s panic?
2. What is meant by “failure to thrive”?
3. What are some reasons why newborns fail to thrive?
4. What do you think the smell is?

Part II—Pedigree Analysis

Matthew’s urine did have a sweet, maple syrup smell and lab results revealed elevated levels of the branched chain amino acids (bcaa)—valine, isoleucine, and leucine.

Skin biopsies from the baby and his parents were taken and cultured. The ability of the cultured skin fibroblasts to metabolize bcaa was determined. While his parents’ enzyme activity levels were nearly normal, Matthew’s was less than 2% of normal.

“Given the medical information and the smell of the urine, Matthew has Maple Syrup Urine Disease (msud),” reported Dr. Morton of the Clinic for Special Children. “He will not be able to breast feed or drink regular formula. What is really important is that Matthew eats a low protein diet. Tis diet must continue for the rest of his life or else the amino acids will accumulate in the body creating a situation that leads to brain swelling, neurological damage, and death. In spite of dietary intervention, the disease may cause several complications, the most notable being mental retardation. You need to know that dietary intervention does not cure the disease.”

Emma and Jacob were Mennonites and their family history revealed that Emma’s mother had two sisters who died in their first year of life; no one knew why. Jacob’s father had a sister who died at seven months of age from unknown causes. Could the gene for msud run in both of their families?

msud is due to a recessive gene. For an individual to be affected, he or she would need to inherit a defective nonworking copy from each parent. The individual would then be described as being homozygous recessive.



*Questions*

Pedigree charts are useful tools used by genetic counselors to look for the incidence of disease within multiple generation families. Each generation is shown on a separate row.

1. Label the pedigree chart below to explain the relationships and the disease incidence within this family. Be sure to include Emma, Jacob, Samuel, Matthew, Emma’s father, Emma’s mother, Emma’s aunts, Jacob’s mother, Jacob’s father, and Jacob’s aunt.
2. Indicate on your pedigree chart the individuals who are carriers by shading half of each circle or square.
3. Define the terms genotype, phenotype, homozygous and heterozygous.
4. How could their son have inherited msud even though neither parent suffers with it?
5. What is the probability that they would have another affected child? A carrier?
6. Could Emma and Jacob have children who do not have msud (i.e. phenotypically normal)? Explain. What is the probability?
7. If msud were a dominant disorder, what would be the probability that Matthew would inherit the disease?

*BONUS Question:* Why were Emma’s and Jacobs’s enzyme levels nearly normal?

