

Explaining the Human Metabolism

EQ: What is baby Matthew's diagnosis and how can it account for his symptoms?

Metabolism

The chemical processes that occur within a living organism in order to maintain life.

What do we know about the metabolism so far?

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Case Study: Baby Matthew and his 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.”

What do we think metabolism might have to do with Baby Matthew's illness?

Let's explore the metabolism

Inborn Errors of the Metabolism

Knowing that there is one gene (segment of DNA) that codes for each enzyme of the metabolism....

Pair-Share: If an enzyme's gene has a mutation that changes its structure, **what will happen to the ability of that individual organism to carry out that enzyme's reaction?**

Pair-Share: What do you think are some possible consequences of not being able to carry out a particular reaction?

Genetic Disorders of the Metabolism--GoFormative

Your teacher will assign you one metabolic genetic disease from [this chart](#) (list form [here](#))

Assignment:

With a partner, research the genetic disorder to find out the following, then complete the GoFormative assignment question number for your assigned metabolic disease (assignment name: [Genetic Diseases of the Metabolism](#)).

1. The name of the enzyme associated with this disease
2. The substrate(s) for that enzyme
3. The product(s) of that enzyme
4. The symptoms of the disorder
5. Explanation of either:
 - a. how a build up of the substrate causes the symptoms or
 - b. how a lack of the product causes the symptoms

Discussion

Each pair of students **forms a discussion group of 4**

What did you find out about the root causes of the symptoms?

- **That the patient cannot make the product?**
 - What are the consequences of not being able to make this product?

and/or

- **That the substrate builds up to toxic levels in the body?**
 - What are the consequences of this toxicity?

Let's review baby Matthew's history and symptoms

- Baby Matthew is the second child born in the family, his sibling died soon after birth
- Both the pregnancy and delivery were uneventful.
- By the fifth day after birth, Matthew began to have trouble nursing and by the seventh day he had completely stopped feeding.
- Mathew's limbs were rigid
- He had had a seizure
- the examination showed no infection and his x-rays were normal.
- Funny smell in Matthew's diapers

GoFormative Question 16

Does baby Matthew have your assigned disease?

Write a C-E-R paragraph (Claims-Evidence-Reasoning) that uses the information you have collected and heard in your group and class discussion to answer the question.

You may use the sentence frames that are provided, or write your own paragraph.

Which Groups think their assigned disease is a candidate for Matthew's diagnosis?

Each group who has a candidate disease identified:

- Share your Claim, Evidence, and Reasoning with the class

Class discussion:

- Argue from evidence to come to consensus on which disease best fits Matthew's observed symptoms

Hope for Baby Matthew!

With a diagnosis in hand, Matthew has treatment options and will not suffer the same fate as his sibling.

Pair-share: What are your ideas on treatment options for Matthew?

How is this skill relevant to your life?
Healthcare professionals?

Understanding symptoms and their root causes - why is this important?

What do we do when multiple diseases have similar symptoms?

What are the implications for your own health advocacy?

How do health professionals practice this skill?