

Sick Kids with an Unusual Organic Aciduria

Case Study Problem No. 5 - Written by Harold B. White

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Consider your group to be an expert consulting team that takes pride in diagnosing metabolic diseases based on the abnormal organic acids excreted in a patient's urine. Over the years, for good reason, doctors from far and wide have come to rely on your team to solve their difficult cases. Typically they want to know what enzyme is defective and what therapies might ameliorate or eliminate the pathological consequence of various inborn errors of metabolism. Your team sees each new case as an intellectual challenge, but the greatest satisfaction comes from being able to save lives or improve the quality of life often through simple changes in a patient's diet.

How would you go about relating the excretion of a particular organic molecule or molecules in the urine to a specific defect in intermediary metabolism?

What are some examples of inborn errors of metabolism (genetic diseases) that cause the excretion of abnormal metabolites?