Case Study #4: The Consequences of Shared Love

Case Background
A man and three women enter the Ogden, Utah emergency department where you work as a physician with a seizing infant. A quick glance at the patient reveals slight microcephaly and widely spaced eyes. As nurses administer benzodiazepines to stop the seizures, you recognize the facial features as characteristic of Down syndrome. When you ask the parents if their child was diagnosed with any sort of developmental disorder when in infant care, they tell you that their child was a home birth.

Clinical Findings
You order routine blood tests; to your surprise, the infant has blood lactate levels of 3 mM, 25 g/dL Hb, and 3,000 white blood cells/mL (normally 0.5-1 mM, 15-20 g/dL, and 4,500-10,000, respectively). You also order the lab to perform a chromosomal karyotype to confirm your observations pertaining to the infant’s appearance.

Analysis
1. Given the blood analysis, what is a possible cause of the symptoms?

2. The lab calls back and tells you the trisomy 21 test was negative. Could there be any connection between the facial dysmorphology, the microcephaly, and a metabolic disorder?

3. You obtain a urine sample to conduct further tests; results from the urinalysis indicate high fumaric acid. Where is your patient’s enzyme deficiency?

4. Where is the patient getting oxaloacetate if they are not completing the Krebs cycle? How do you think this would impact the lactate levels?

5. How would you propose to treat the patient?

References