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"Methylcoenzyme A Mutase Deficiency (MMCOA) Combined With Homocystinuria"

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Abstract:

Background: The combined MMCOA deficiency and Hemocystinuria results from defect in cb1c & cb1d & cb1F genome. Until now only 100 cases of this disease has been reported. Most of them with cb1c deficiency. Due to this enzymatic defect, mild to moderate retention of Hemocystin (MMA) and their precursors in the body fluids (blood and urine) ensues. Although, acidemia, Hypoglycemia & megaloblastic anemia are common but hyperamonemia and hyperglycemia were not reported in these patients. Our case is a 3 years old girl with known MMCOA deficiency and Hemocystinuria who needed G.A for partial resection of the mandible because of osteomilitis. Materials and Methods: In this report we explain the preoperative preparation of the patient, induction and maintenance of Anesthesia, and discuss the biochemical, lab results during & after surgery. Results & Conclusion: Due to our result, the urine and blood hemocystin & MMCOA were not increased. This case is interesting because no theoretical or practical report on the G.A of these patients exists.

Keywords:

Hemocystinuria ، MMCOA ، G

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