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A Rare Case of Methionine Adenosyltransferase Deficiency in an 8-Month-Old Infant Presenting with Jaundice and Bleeding Episodes

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Abstract

Methionine adenosyltransferase (MAT) deficiency is a rare inherited disorder of methionine metabolism that can lead to a wide range of symptoms, including jaundice, bleeding episodes, and neurological impairment. We report the case of an 8-month-old female infant, who presented with yellowish discoloration of skin, distended abdomen, and bleeding episodes. On further evaluation, she was found to have high levels of methionine in her blood. A diagnosis of MAT deficiency was made, and was started on appropriate treatment with a methionine-restricted diet and supplements.

Index terms—

A Rare Case of Methionine Adenosyltransferase Deficiency in an 8-Month-Old Infant Presenting with Jaundice and Bleeding Episodes By Mohammad Yunus Choudhary Abstract-Methionine adenosyltransferase (MAT) deficiency is a rare inherited disorder of methionine metabolism that can lead to a wide range of symptoms, including jaundice, bleeding episodes, and neurological impairment. We report the case of an 8-month-old female infant, who presented with yellowish discoloration of skin, distended abdomen, and bleeding episodes. On further evaluation, she was found to have high levels of methionine in her blood. A diagnosis of MAT deficiency was made, and was started on appropriate treatment with a methionine-restricted diet and supplements. Introduction Methionine adenosyltransferase (MAT) deficiency is a rare autosomal recessive disorder caused by mutations in the MAT1A gene that encodes the alpha subunit of the enzyme MAT. MAT is responsible for the synthesis of S-adenosylmethionine (SAMe), a critical methyl donor involved in many essential biochemical pathways. MAT deficiency leads to an accumulation of methionine and its toxic metabolites in the body, which can cause a wide range of symptoms.

1 GJMR-F Classification

2 II.

3 Case Presentation

A 8-month-old female infant, presented with yellowish discoloration of skin and a distended abdomen. She had no history of hematemesis, rashes, blood in stools, melena, or discolored urine. She had also experienced episodes of unconsciousness and bleeding episodes in the past. On examination, she was alert and conscious with a distended abdomen. Her respiratory and cardiovascular systems were normal, and her central nervous system was intact.

Further evaluation revealed high levels of methionine in her blood, leading to a diagnosis of MAT deficiency. She was started on a methionine-restricted diet and supplements to reduce the levels of methionine in her body. Her symptoms gradually improved, and she was discharged from the hospital with regular follow-up visits.

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40 **4 Discussion**

41 MAT deficiency is a rare disorder with a wide range of clinical manifestations. Jaundice, bleeding episodes, and
42 neurological impairment are the most common symptoms. The diagnosis is usually made by measuring the levels
43 of methionine and SAME in the blood, and genetic testing can confirm the diagnosis.

44 Treatment for MAT deficiency involves a methionine-restricted diet and supplements to improve the levels of
45 SAME in the body. Early diagnosis and treatment can prevent or reduce the severity of symptoms and improve
46 the quality of life of affected individuals.

47 **5 IV.**

48 **6 Conclusion**

49 MAT deficiency is a rare inherited disorder of methionine metabolism that can lead to a wide range of symptoms,
50 including jaundice, bleeding episodes, and neurological impairment. Early diagnosis and treatment are essential
51 to prevent or reduce the severity of symptoms and improve the quality of life of affected individuals. This case
52 highlights the importance of considering rare metabolic disorders in the differential diagnosis of infants presenting
53 with jaundice and bleeding episodes.