VOLUME - 10, ISSUE - 01, JANUARY - 2021 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Paediatrics

A RARE CASE REPORT OF SCOT DEFICIENCY

Dr Ami Dabhi*	3rd Year Resident, Department Of Paediatrics, AMCMET Medical College, Maninagar, Ahmedabad. *Corresponding Author
Dr Hinali Chauhan	3rd Year Resident, Department Of Paediatrics, AMCMET Medical College, Maninagar, Ahmedabad.
Dr Priyal Halani	1st Year Resident, Department Of Paediatrics, AMCMET Medical College, Maninagar, Ahmedabad.
Dr Sagarika Upadhyay	1st Year Resident, Department Of Paediatrics, AMCMET Medical College, Maninagar, Ahmedabad.

ABSTRACT SCOT deficiency is a metabolic disorder caused by reduced or absent level of enzyme succinyl co-A 3: oxoacid co-A transferase; required in ketone body utilisation. Disorder is characterized by severe, potentially fatal intermittent episodes of ketoacidosis. It is inherited as autosomal recessive pattern. It occurs due to mutation of OXCT1 gene. Acute episodes of ketoacidosis are triggered by infection or prolonged fasting associated with hypoglycaemia & ketonemia. Treatment includes rehydration & correction of acidosis.

KEYWORDS : Ketoacidosis, Hypoglycaemia

INTRODUCTION

Few cases of SCOT deficiency have been reported. It is one of the ketone body utilisation disorders inherited as an autosomal recessive pattern. SCOT participates in the conversion of ketone bodies(acetoacetate & 3- hydroxybutyrate) generated in liver mitochondria into acetoacetyl co-A in the non-hepatic tissues. A deficiency of this enzyme results in the accumulation of ketone-bodies, ketoacidosis, increased utilization of glucose, andhypoglycaemia. A common clinical presentation is an acute episode of severe ketoacidosis in a child who had been growing and developing normally. The acute episode is often precipitated by a catabolic state triggered by an infection or prolonged fasting. Without treatment, the ketoacidotic episode can result in death. A chronic subclinical ketosis may persist between the attacks.

Development is usually normal, although severe & recurrent episodes of ketoacidosis & hypoglycaemia can predispose patients to neurocognitive impairment. Laboratory findings during acute episode are non specific and include metabolic acidosis and ketonuria with high levels of ketone bodies. Diagnosis can be established by demonstrating reduced or absent enzyme activity. Treatment of acute episode consists of rehydration with solutions containing dextrose, correction of acidosis, and the provision of a diet adequate in calories. Long term treatment should include high carbohydrate diet and avoidance of prolonged fasting and administration of dextrose before anticipated or during established catabolic states.

CASE PROFILE

A 4.5 year old Hindu male child, born of non-consanguineous marriage, coming from lower socioeconomic class, presented to paediatric emergency ward with complaint of sudden loss of consciousness with altered breathing pattern. On eliciting detailed history, we came to know that patient was suffering from high grade fever with intractable vomiting since last 2 days. Patient had complaint of reduced oral intake due to vomiting. He had past history of similar episodes thrice & was admitted in critical care unit for the same. On examination, temperature was raised (101.2), tachycardia was there with good volume pulse, RBS was 40 on admission, acidotic breathing was present with respiratory rate 52/min. On auscultation, no murmur or foreign sounds were present. On ABGA, pH was 6.8, bicarbonate value was 3.5, Pco2 was 13, and SO2 was 96.5. Hence, patient had severe metabolic acidosis.

After 1st bicarbonate correction patient's ABGA values were same with no clinical improvement. Repeat bicarbonate correction was given. Patient improved after 3 times bicarbonate correction. Meanwhile, due to high index of suspicion, urine GCMS study was sent as per advice of paediatric neurologist. Patient had 4 episodes of hypoglycaemia & ketonemia was present. Patient improved after 3 days of stay. Urine GCMS was suggestive of ketone body utilisation disorder with reduced levels of enzyme SCOT. Hence, patient was diagnosed having SCOT deficiency.

Patient was managed by rehydration therapy, correction of acidosis & ventilator support.

Patient was discharged after 6 days of hospital stay. Nature of disease & course of illness was explained to relatives. Patient was advised about sick day management & not to fast.

CONCLUSION

A 4.5 year old male child presented with severe refractory metabolic acidosis, was investigated further & diagnosed having SCOT deficiency. The disorder is not uncommon but it remains undiagnosed in many cases as after correction of acidosis patient is discharged. Hence, each & every patient presenting with metabolic acidosis must be taken into due consideration, active effort should be done to identify cause of each acidosis episode.

REFERENCES

1. Nelson textbook of paediatrics, 21st edition, chapter 103, page 715
2. http://mirror.omim.org/SCOT deficiency
3. http://rarediseases.org/SCOT deficiency