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Hepatopulmonary syndrome in a child with transaldolase deficiency: A case report

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Background

Transaldolase deficiency is a recently described inborn error of pentose phosphate pathway (PPP). This disorder is inherited as autosomal recessive, Patients may present in the neonatal or antenatal period with hydrops fetalis, hepatosplenomegaly and hepatic dysfunction. Patients has characteristic dysmorphic features . Hepatopulmonary syndrome (HPS) is considered to be one of the cardiopulmonary complications of liver disease, and it associated with portal hypertension. The triad of HPS includes liver disease or portal hypertension, hypoxemia, and intrapulmonary shunting [(1)].

CASE REPORT

We report 9-year-old girl, from Saudi Arabia was diagnosed with transaldolase deficiency based on gene test at age of 2 years, who developed respiratory distress and hypoxia in early infancy and was later diagnosed with hepatopulmonary syndrome (HPS) after history of recurrent respiratory distress and respiratory failure that required high frequency mechanical ventilation. Cardiac assessment and echocardiogram showed ASD 11, mild tricuspid and mitral valves regurgitation. The plan was to do bubble echo which will show evidence of delayed positive bubbles, indicating intrapulmonary shunting. However, the procedure was postponded because the patient was clinically unstable.

The blood gas evaluation in room air showed a pH of 7.372, PCO2t of 23.8 mmHg, PO2t of 63.7 mmHg, cHCO3- of 16.7 mmol, and base excess ecf of −11.4 mmol. The arterial-alveolar gradient (A-a gradient) was 56.28 mmHg, and the estimated normal gradient for her age is 6 mmHg, where values greater than 15 mmHg suggest the presence of a shunt. CTA pulmonary arteries revealed dilated peripheral pulmonary arteries reaching pleural margin keeping with the diagnosis of hepatopulmonary syndrome. Macro aggregated albumin perfusion lung scanning (99mTc-MAA scan) study was revealed accumulation of the tracer in the liver, kidneys and both lungs. The activity within the lung and the abdomen, suggesting that systemic shunting accounted for 43 % (this parameter is normally less than 5 %). Patient currently on oxygen 3L/min via face mask for the hypoxia, supportive management for the chronic liver disease and anemia inform of ursodeoxycholic acid, vitamin K, ADEK and iron. She has multidisciplinary follow up including pulmonology, gastroenterology and hematology. No plan for liver transplantation as the data regarding the outcome in such cases are not available Patient currently on home oxygen 3L/min via face mask for the hypoxia, and supportive management for the chronic liver disease and anemia, plus multidisciplinary follow up care. No plan for liver transplantation as the data regarding the outcome in such cases are not available. To our knowledge this will be the second case of transaldolase deficiency and hepatopulmonary syndrome to be reported.

Discussion

Hepatopulmonary syndrome (HPS) was first described in 1884 by Fluckiger, who noted a relationship between the liver and the lung . Kennedy and Knudson described hepatopulmonary syndrome in 1997 [(2)]. Eyaid et al. (2013) reported the largest case series on Transaldolase Deficiency, consisting of 12 patients from six Saudi families [(3)]. All patients had cardiac defects, dysmorphic facial features, cutis laxa, hepatosplenomegaly, thrombocytopenia, and anemia. Jassim et al (2014) reported the first case of transaldolase with hepatopulmonary syndrome in a child who was diagnosed at birth with transaldolase deficiency who subsequently developed hepatopulmonary syndrome[(4)] . This syndrome is a rare complication of liver disease acute or chronic in children, and it includes the triad of liver disease or portal hypertension, hypoxemia, and intrapulmonary shunting. Management is usually are conservative in form of supportive management for the chronic liver disease, vitamins and oxygen .Liver transplantation is not option for this child as there is no available data regarding the outcome in patients with same problem.

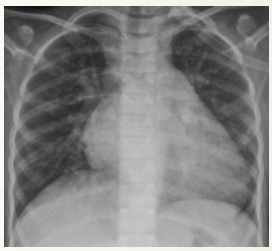

Figure 1: CXR showing bilateral infiltration , lower lobe atelectasis and cardiomegaly

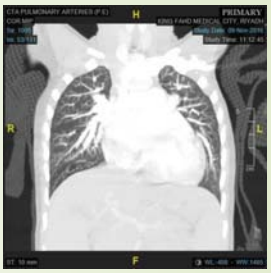

Figure 2 CTA pulmonary arteries: showing dilated peripheral pulmonary arteries reaching pleural margin

Conclusion

Hepatopulmonary syndrome should be considered as a cause of respiratory symptoms and hypoxia in patient with transaldolase deficiency, and need to rule out other possible causes. Multidisciplinary care with input from a pulmonology, gastroenterology, cardiology and hematology is crucial to the successful management of these patients, all working together with the child and the family to optimize care and quality of life. At present, there is no treatment for the condition. Some patients with transaldolase deficiency might benefit from liver transplantation, however no available data for patients with hepatopulmonary syndrome.

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