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Case Report

Paediatric autoimmune hepatitis with thalassemia trait: a case report

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ABSTRACT

Autoimmune hepatitis (AIH) is a chronic inflammatory liver condition characterized by elevated serum aminotransaminase levels, the presence of liver-associated autoantibodies, and/or hypergammaglobulinemia. Three conditions with likely autoimmune origins include AIH, autoimmune sclerosing cholangitis, and de novo AIH following liver transplantation. AIH is classified into two types based on antibody presence: Type 1 and Type 2. Systemic disorders such as hemoglobinopathies, cystic fibrosis, and histiocytic disorders have been associated with liver dysfunction. Early diagnosis requires a high level of suspicion. Paediatricians should consider AIH in patients presenting with jaundice after excluding common causes. The diagnosis is made through a combination of biochemical, immunological, and histological findings, while ruling out other liver diseases with similar serological and histological profiles. We present the case of an 8-year-old female with non-resolving jaundice, diagnosed with AIH alongside thalassemia trait.

Keywords: Autoimmune hepatitis, Thalassaemic trait, Jaundice

INTRODUCTION

Autoimmune hepatitis (AIH) is a prototypical autoimmune liver disease in both adults and children, first described in the 1950s. The prevalence of juvenile AIH is not well established, but a recent study reported an annual incidence of 0.23 per 100,000 children in a large Canadian cohor.[1] AIH is a progressive inflammatory liver disease that, if left untreated, can advance to end-stage liver disease. Its hallmark features include a female predominance, hypergammaglo-bulinemia, seropositivity for circulating autoantibodies, and the presence of interface hepatitis on histology. AIH arises in genetically predisposed individuals after an unknown trigger initiates a T-cell-mediated immune response against liver autoantigens. Potential triggers include molecular mimicry, infections, drugs, and toxins. Several HLA class II molecules, particularly DR3, DR4, and DR7 isoforms, have been linked to increased susceptibility to AIH. The clinical presentation and course of AIH are highly variable; it may be asymptomatic, mimic viral hepatitis, or manifest as chronic liver disease. Without appropriate treatment, AIH can progress to liver cirrhosis.

A key factor in diagnosing AIH is the detection of circulating autoantibodies. AIH is classified into two subtypes.[2] Based on autoantibody profiles: AIH-1, which is positive for antinuclear antibodies (ANA) and/or smooth muscle antibodies (SMA), and AIH-2, which is positive for anti-liver kidney microsome-1 (anti-LKM-1) and/or anti-liver cytosol-1 (anti-LC-1). Patients may present with nonspecific symptoms such as nausea, anorexia, malaise, pruritus, and abdominal pain. In more severe or chronic cases, symptoms may include jaundice, hepatomegaly, splenomegaly, and signs of chronic liver

disease. AIH is often associated with other autoimmune conditions, so paediatricians should be vigilant for signs of coexisting disorders such as diabetes, thyroiditis, and celiac disease.[3]

CASE REPORT

An 8-year-old female child presented with gradually progressive yellowish discolouration of body and urine since 3½ month, generalized weakness, on and off vomiting for 3-month, undocumented fever for 2 month and itching for last 10 days. The family had sought medical advice at a local hospital, and they had investigated and symptomatically treated her but no clear diagnosis had been reached; hence she had been referred to a higher centre. There was no abdominal distension or pain abdomen and no change in stool colour. It was not associated with skin rash, bruises and there was no history of joint or bone pain. There was no history of contact with patient who had jaundice, no history of blood transfusion and the patient was not on any medication. Patient had no similar complaints in past or hospitalization for the same. Regarding her neonatal history, she was delivered at full term with no neonatal intensive care admission. Her developmental history was appropriate for her age; her sibling was healthy and there were no similar conditions in family. There was history of diabetes, arthritis or chronic diarrhoea in family. There was no death or abortion in family history. Social history showed that they were living in village with low socioeconomic status and there had been no recent travel. Her vaccination was up to date. Nutrition history revealed that she was dependent on family diet.

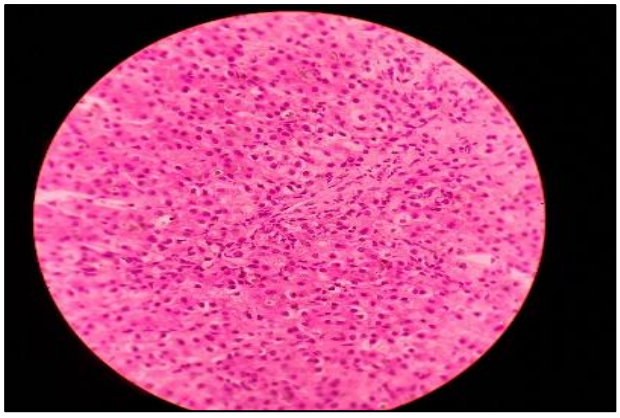

Figure 1: The image shows bile plug with feathery degeneration. There is moderate fibrosis in portal tract along with extensive infiltration by neutrophils, lymphocytes and occasional plasma cells.

On examination she was 25 kg (50th -75th percentile), height-129.5 cm (50th -75th), looked pale, icteric and well hydrated. She was afebrile with pulse rate 84/min, respiratory rate 22/min, Blood pressure was 98/64 mm hg, SpO2 was 98% on room air. Her abdomen was soft and non-tender, liver was 4 cm below right costal margin, firm in consistency with liver span of 16 cm, spleen was not palpable. CNS, chest and cardiovascular examination was unremarkable. Her slit lamp examination revealed no Kayser-Fleischer ring. The musculoskeletal and skin examination were normal.

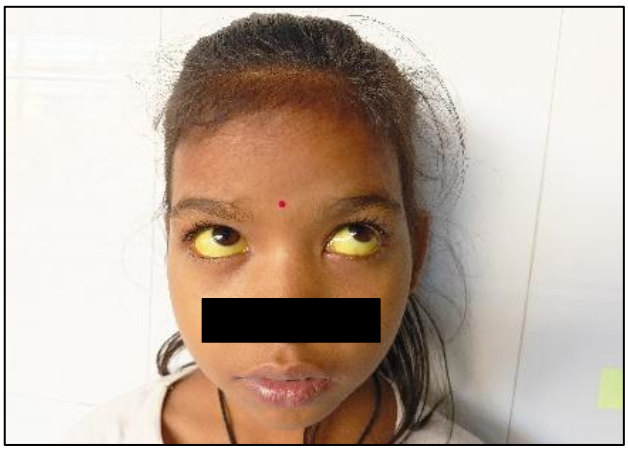

Figure 2: Image shows icterus at the time of admission.

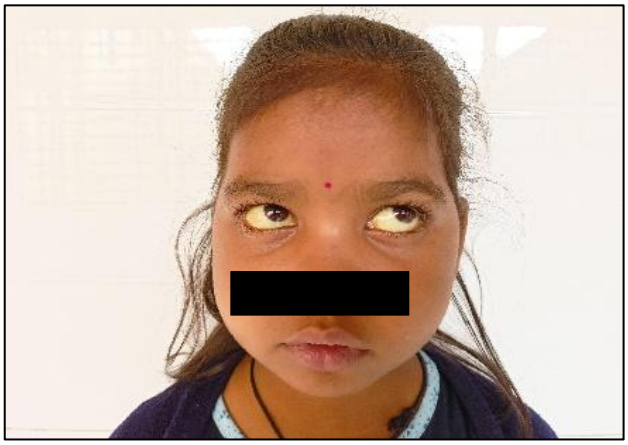

Figure 3: Image shows decreasing icterus after 6 weeks of prednisolone therapy along with Cushing facies.

Her laboratory workup was done, and her complete blood picture suggestive of anaemia with normal platelet counts. Peripheral smear showed microcytic hypochromic RBCs, Target cells, boat shaped cells with polychromatophilic cells. Hb Electrophoresis suggestive of possibility of hemoglobinopathy (Beta Thalassemia trait) and for confirmation whole exome sequencing was done. LDH was raised. DCT was positive. Her liver function was deranged [Bilirubin, SGOT, SGPT, Alkaline phosphate was elevated, with normal GGT, PT, INR and albumin]. Hepatitis serology was negative. USG whole abdomen s/o borderline hepatomegaly with normal parenchymal echotexture. Serum ceruloplasmin and 24-hour urinary copper was within normal limit. Based on the above findings, the possibility of paediatric autoimmune liver disease was considered and child was the child was evaluated for the same; her autoimmune

liver profile was positive for anti-LKM-1, anti-LC-1 and negative for ANA, anti-SMA, ANCA profile. IgG was borderline elevated. Liver biopsy showing bile plugs with feathery degeneration, moderate fibrosis in the portal tract along with extensive infiltration by neutrophils, lymphocytes and occasional plasma cells, no pseudonodule or viropathic effect identified, features suggestive of autoimmune hepatitis. MRCP done s/o hepatomegaly with oedematous GB wall, IBHR of both the lobes are normal in signal intensities. The patient was started on Prednisolone at 2 mg/kg/day. Pantoprazole, Ursodeoxycholic acid and Cholestyramine was also added. Her laboratory parameter was monitored which showed significant improvement.

DISCUSSION

Juvenile AIH can be classified into seropositive and seronegative forms. Seropositive Type 1 AIH, which accounts for 60-70% of cases, typically presents during adolescence and is more responsive to immunosuppressive therapy. In contrast, seropositive Type 2 AIH primarily affects children and young adults, often exhibiting a more severe course with frequent treatment failures. Type 2 AIH, when responsive to treatment, can also be part of the autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome. A seronegative form of AIH, responsive to steroid therapy, has been reported in paediatric studies and has been occasionally linked to the development of aplastic anaemia.[4]

We presented the case of an 8-year-old female diagnosed with autoimmune hepatitis and Beta Thalassemia trait. The diagnosis was confirmed through an autoimmune profile, liver biopsy, MRCP, and whole exome sequencing. In India, AIH is considered very rare in children, and many paediatricians may encounter it infrequently, if at all, during their practice.

In this case, we first considered common causes of jaundice, such as viral hepatitis and inherited metabolic diseases like Wilson disease. After ruling out these conditions, AIH became a consideration, and subsequent investigations revealed positivity for anti-LKM-1, anti-SMA, and elevated IgG levels, confirming Type 2 AIH. Liver biopsy confirmed the diagnosis with characteristic findings of interface hepatitis, which is typical of both AIH and autoimmune sclerosing cholangitis (ASC). However, since ASC often shows bile duct damage on MRCP, this additional factor confirmed the diagnosis of AIH in this patient, along with Beta Thalassemia trait. She was started on prednisolone at a dose of 2 mg/kg/day, and she responded well to treatment, with repeat liver function tests showing a decrease in conjugated bilirubin and improved transaminase levels.

In addition to prednisolone, Pantoprazole and ursodeoxycholic acid (UDCA) were prescribed. UDCA has choleretic and putative immunomodulatory effects, though its efficacy in this setting remains unproven. The patient remains on prednisolone, and the plan is to taper the dose over 6 to 8 weeks to a maintenance level of 5-7.5 mg/day. The ESPGHAN guidelines recommend treating paediatric AIH for at least 2 to 3 years and considering treatment withdrawal only if transaminase and IgG levels have been normal and autoantibody tests negative for at least one year.[5] Before discontinuing treatment, a repeat liver biopsy is advised, as residual inflammatory changes, even with normal blood tests, can indicate a risk of relapse.[6]

Between 9% and 55% of AIH patients may ultimately require a liver transplant.[7] The indication for transplantation is usually acute liver failure (ALF) that is unresponsive to steroid therapy. The 5-year survival rate following liver transplantation for AIH is excellent, ranging from 80% to 90%.[8] However, recurrence rates vary from 38% to 83%. Interestingly, the standard treatment for recurrent AIH post-transplant remains prednisolone, often in combination with azathioprine. In cases of treatment failure, mycophenolate mofetil (MMF) may be substituted for azathioprine in the therapeutic regimen.

There have been several reports linking Beta Thalassemia trait to autoimmune diseases such as nephritis, diabetes, arthritis, fibromyalgia, and asthma. Two possible explanations have been proposed for the increased risk of autoimmune disease in patients with Beta Thalassemia. First, the beta-globin gene locus (11p15.5) is located near several genes involved in immune regulation, including ST1MA, CD151, RRAS2, SIGIRR, LSP1, TRIM21, TOLLIP, and SLEN3. Second, alterations in hemorphin concentrations may play a role. Hemorphins, which bind to opioid receptors and exert anti-inflammatory effects, are reduced in thalassemia heterozygosity, potentially leading to a proinflammatory state and increased susceptibility to autoimmune conditions.[9]

CONCLUSION

Although AIH is rare among children, paediatricians must consider it in patients presenting with acute or chronic hepatitis, once the common causes are ruled out. AIH almost always progresses to cirrhosis, but early treatment can prolong survival and improve the quality of life.

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REFERENCES

1. Jiménez-Rivera C, Ling SC, Ahmed N, Yap J, Aglipay M, Barrowman N, et al. Incidence and Characteristics of Autoimmune Hepatitis. Pediatrics. 2015;136(5):e1237-48.

2. Kliegman R, St. Geme J. *Nelsons Textbook of Paediatrics.* Autoimmune hepatitis. 2020;2.
3. Mieli-Vergani G, Heller S, Jara P, Vergani D, Chang MH, Fujisawa T,et al. Autoimmune hepatitis. J Pediatr Gastroenterol Nutr. 2009;49(2):158-64.
4. Maggiore G, Socie G, Sciveres M, Roque-Afonso AM, Nastasio S, Johanet C et al. Seronegative autoimmune hepatitis in children: Spectrum of disorders. Dig Liver Dis. 2016;48(7):785–91.
5. Mieli-Vergani G, Vergani D, Baumann U, Czubkowski P, Debray D, Dezsofi A, et al. Diagnosis and Management of Pediatric Autoimmune Liver Disease: ESPGHAN Hepatology Committee Position Statement. J Pediatr Gastroenterol Nutr. 2018;66(2):345-60.
6. European Association for the Study of the Liver. EASL Clinical Practice Guidelines: Autoimmune hepatitis. J Hepatol. 2015;63(4):971-1004.
7. Chai PF, Lee WS, Brown RM, McPartland JL, Foster K, McKiernan PJ, et al. Childhood autoimmune liver disease: indications and outcome of liver transplantation. J Pediatr Gastroenterol Nutr. 2010 ;50(3):295-302.
8. Vergani D, Mieli-Vergani G. Autoimmunity after liver transplantation. Hepatology. 2002;36(2):271-6.
9. Altinoz MA, Gedikoglu G, Deniz G. β-Thalassemia trait association with autoimmune diseases: β-globin locus proximity to the immunity genes or role of hemorphins? Immunopharmacol Immunotoxicol. 2012;34(2):181-90.

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