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Case Report

Successful rescue of a child with Griscelli syndrome type 2 from lethal respiratory distress

Rubi Zined[1]*, Bhavna Rani[2], Aditya K. Gupta[3], Poonam Joshi[4]

[1]National Institute of Nursing Education, Post Graduation Institute of Medical Education and Research, Chandigarh, India
[2]Baba Farid University of Health Sciences, Faridkot, Punjab, India
[3]Department of Pediatrics, All India Institute of Medical Sciences, New Delhi, Delhi, India
[4]College of Nursing, All India Institute of Medical Sciences, Kalyani, West Bengal, India

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***Correspondence:**
Rubi Zined,
E-mail: Rubigurjar47@gmail.com

ABSTRACT

In this study, we report a known case of GS type 2, in a four-year-old male child. The child initially presented with fever, tachypnea, severe anemia, massive hepatosplenomegaly, wheezing on auscultation. A chest x- ray revealed bilateral perihilar infiltrates. The child was managed with high flow, heated and humidified oxygen support at 20 liters/min, inhaled corticosteroids, salbutamol nebulization and antibiotics. A respiratory viral panel PCR sent to rule out viral pathogens. There was gradual improvement, and the child was tapered off oxygen within one week. The hepatosplenomegaly gradually regressed after receiving the HLH protocol. At the time of discharge, the child was maintaining saturation on room air without any distress, had no hepatomegaly and spleen had regressed to 5 cm under right costal margin. Work up for Bone marrow transplantation (BMT), was simultaneously started. GS2 can have heterogenous clinical manifestations. Early diagnosis and rapid control of HLH by using the HLH-1994 regimen along with supportive care for the complications is the key to successful treatment.

Keywords: Griscelli syndrome, Hemophagocytic lymphohistiocytosis, Bone marrow transplantation, Packed red blood cells

INTRODUCTION

Griscelli syndrome (GS) is a rare autosomal recessive disorder initially described by Claude Griscelli in 19781. Three types of GS are identified, which are distinguished by their genetic cause and pattern of signs and symptoms GS types.[1-3] These are caused by mutations in one of three genes: RAB27A, MYO5A and MLPH. Two of these genes, RAB27A and MYO5A, are located at band 15q21. Defect of MLPH is located on band 2q37.3. RAB27A-MLPH-MYO5A form a tripartite complex facilitating intracellular melanosome transport. Deficient melanocytes of any of these genes leads to accumulation of melanosomes near the microtubule organizing center and failure to transfer to keratinocytes and pigmentary dilution of the skin and the hair (silver hair). Myosin Va is an important protein in intracellular vesicle transport. This is important for fast axonal transport in nerve cells, which may explain the neurological complications seen in GS type 12. GS type 2 is the most common type and has the most severe presentation.[3] In this type, the GTP-binding protein, which is the gene product of RAB27A appears to be involved in the control of the immune system as this gene is key effector of cytotoxic granule exocytosis, a pathway essential for immune homeostasis. Hence, most of the patients presents with episodes of Hemophagocytic lymphohistiocytosis (HLH), which is usually triggered by viral infections.[4] Although many case

reports have been published related to GS type 2, no case report has been published related to child with lethal respiratory distress in GS type 2.

CASE REPORT

A four-year-old male, known case of GS type2 presented with silver-colored hairs (Figure 1). The child, born fourth to non-consanguineous parents, is the fourth sibling. The second and third siblings died at 4 and 7 months respectively due to recurrent fever and hepatosplenomegaly. First sibling is alive and healthy. In May 2022, the child had on and off fever, diarrhea, jaundice, abdominal distension. He was initially treated at a local hospital and later referred to the tertiary care facility due to pancytopenia and hepatosplenomegaly. Suspecting thalassemia, HPLC was done, and Packed red blood cells transfusion given (HbA-96.3, HbF-2.9, HbA2-0.8).

Bone marrow examination and Ig levels were normal and the child was referred to pediatric gastroenterology for persistent hepatosplenomegaly. Tests for Hepatitis A and E viruses and immunodeficiency were conducted. Hepatitis A was confirmed on 16/6/2022 (anti HAV IgM-4.52). Primary immunodeficiency disorders (PID) workup indicated B cell deficit.

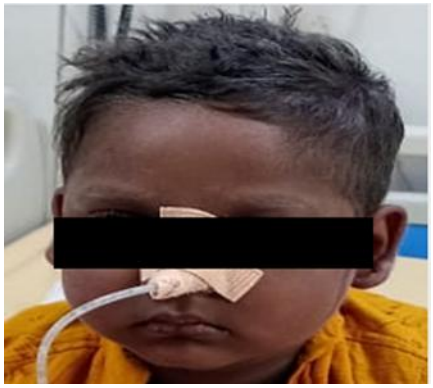

Figure 1: Showing silver colored hairs of child.

Whole Exome Sequencing (WES) identified a mutation in RARB27 gene consistent with phenotypic correlation, leading to diagnosis of GS. Child was referred to pediatric oncology and is planned for Hematopoietic stem cell transplantation. During the HSCT workup, the HLH workup came positive. The child was subsequently started on HLH 1994 regime including. Tab Dexamethasone Inj. Etoposide, Tab. septran and Fluconazole. Following this treatment, the child's fever, cytopenia and splenomegaly improved. Fifteen days after initiating the HLH 1994 protocol, the child developed respiratory distress, presented with tachypnea, body ache and high-grade fever for two days. He was admitted to the pediatric emergency department and placed on H3FNC support due to respiratory distress. Then, he was admitted into the pediatric department for further management. A system wise management of the child is presented here:

Respiratory

Upon admission, the child's respiratory rate was 70/min with wheezing on auscultation. Chest X ray revealed bilateral perihilar infiltrates, (Figure 2). And the respiratory viral panel PCR was also negative for COVID-19, influenza A and B.

Management

The child was put on high flow, heated and humidified oxygen support at 20 liters/min, inhaled corticosteroids along with salbutamol nebulization for wheezing and antibiotics for infections. Child's respiratory status improved, and he was tapered off oxygen by the end of first week. At discharge the child was maintaining saturation on room air without distress.

Infection

The child presented with fever and a respiratory focus but was non-neutropenic.

Management

As the child was presented in emergency department with tachypnea and fever. He needed H3FNC support to maintain SpO2 between 90 to 95%. His TLC was 19,400/micro liter, CRP 94.2 mg/dl, procalcitonin on second day of hospitalization was 0.79 ng/ml and serum galactomannan on the third day was 0.531ng/ml. Hence, he started on Inj. Piperacillin-tazobactam and Amikacin. Etoposide was initially withheld due to ongoing infection, but Tab Septran and Fluconazole continued. Inj. Etoposide was administered after the child became afebrile by fourth day. Despite initial improvement, fever recurred after 72 hours, with high spikes up to 1020F. Follow-up tests showed negative galactomannan (0.531ng/ml) and decreasing procalcitonin levels (0.79 to 0.33 ng/ml), and blood and urine cultures remained sterile.

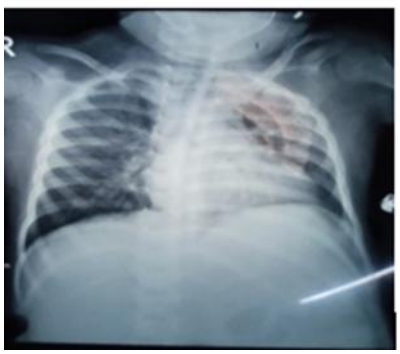

Figure 2: Bilateral perihilar infiltrates, right>left.

The patient developed loose stools, prompting an upgrade to meropenem and teicoplanin. Stool workup revealed Giardia lamblia, and treatment with nitazoxanide resolved the gastrointestinal symptoms. Possibility of other opportunistic infections was kept in view of ongoing chemotherapy and immunosuppressed status so further investigations carried out to ruled out the CMV and other opportunistic infections. Persistent fever was attributed to primary HLH, and dexamethasone dose was increased to 10 mg/m², resulting in the child becoming afebrile by the 18th day of hospital stay. At the time of discharge the child was afebrile, on oral Septran and fluconazole prophylaxis and continuing on HLH-94 protocol.

Hematological

Child had severe anemia (Hb=5.1 g/dl) at presentation.

Management

He was transfused with 20 ml/kg of packed red blood cells (PRBC) over two days, increasing hemoglobin to 8.5 g/dl. Initially, he was given stress-dose hydrocortisone for infection, later switched to dexamethasone at 5 mg/m² and etoposide as per the HLH-94 protocol. The child developed pancytopenia (Hb:7.6 g/dl, platelet count:76,000/mm³, ANC:370/mm³) due to chemotherapy, necessitating another PRBC transfusion. After pancytopenia resolved, the HLH protocol continued. Preparations for bone marrow transplantation (BMT) were initiated simultaneously.

Metabolic

Upon admission the child had persistent hypokalemia with associated metabolic alkalosis and hypertension. Trans tubular potassium gradient (TTKG) was 6.39 indicating renal potassium loss, attributed to high-dose dexamethasone with probable mineralocorticoid activity.

Management

Child was discharged with oral potassium supplementation at 4 mEq/kg/day, with plans to taper the dose during follow-up.

Other issues

Child also had transaminitis with predominant elevation of alkaline phosphatase (ALP), attributed to liver infiltration due to HLH. Abdominal ultrasound showed normal liver echotexture. Child had massive hepatosplenomegaly which gradually regressed after receiving the HLH protocol. At the time of discharge, the child had no hepatomegaly and spleen also regressed to 5 cm under right costal margin. Total duration of hospitalization was 21 days.

DISCUSSION

GS is an autosomal-recessive congenital disorder caused by mutations in RAB27A gene located at 15q21 and it is further divided into three types, based on the point of mutation.[5,6] Clinical, laboratory, and mutational characteristics of Indian GS2 patients have been reviewed in previous studies. Consistent features include fever, pallor, silvery gray hair, eyelashes and eyebrows, hepatosplenomegaly, pancytopenia, and RAB27A mutation.[6] Delayed diagnosis of GS can lead to poor prognosis. It is essential to diagnose the specific type early, as each type requires different treatments. Palliative and supportive treatments are suggested for GS1 patients, while GS3 patients require no treatment and have a good prognosis.[6,7] Due to different mutations in the RAB27. A gene, GS2 clinical symptoms can differ from case to case.[8] Children with GS2 present with partial albinism, immunodeficiency, organomegaly, pancytopenia, and lymph histiocytic infiltrates in various organs.[9] In our case the child presented with symptoms of respiratory infection was likely du e to immunosuppression from the HLH-94 protocol.[7] If the infections in these children are not identified early and managed timely, it can lead to adverse outcomes.

CONCLUSION

GS type 2 can have heterogenous clinical manifestations. Early diagnosis and rapid control of HLH using the HLH-1994 regimen along with supportive care for the complications is the key to successful treatment.

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