IOSR Journal of Nursing and Health Science (IOSR-JNHS)
e-ISSN: 2320–1959.p- ISSN: 2320–1940 Volume 11, Issue 1 Ser. V (Jan. – Feb. 2022), PP 07-09
www.iosrjournals.org

Case study on Hematopoietic stem cell transplant in Griscelli syndrome Type 2

Ms. Manju Joseph [1] Dr.Gaurva Kharya[2] Dr.Atish Bakane[3] D.Maryline Flinsi
[1](Charge nurse ,BMT and cellular Therapy Unit ,Indraprastha Apollo Hospital Delhi)
[2](Clinical lead and senior consultant Bone Marrow Transplant and cellular therapy, Indraprastha Apollo Hospital Delhi)
[3](Consultant ,Bone Marrow Transplant and Cellular therapy, Indraprastha Apollo Hospital Delhi)
[4](Principal,Apollo school of Nursing , Indraprastha Apollo Hospital Delhi)

Date of Submission: 06-02-2022 Date of Acceptance: 20-02-2022

I. Introduction

Griscelli syndrome is rare disease, with a prevalence of less than one per million. It is characterized by a silvery-grey sheen of hair and hypopigmentation of the skin, which can be associated to primary neurological impairment and immunologic impairment. As of date there are 3 known forms of GS:

Griscelli syndrome type 1 (GS1) which is characterized by severe problems with brain function (cognitive dysfunction) in addition to the distinctive skin and hair colour[1]. The affected individuals have developmental delayed, intellectual disability, seizures, and hypotonia.[2]

Griscelli syndrome type 2 (GS2) is a rare, inherited condition which affects the skin, hair, and immune system. People with GS2 have abnormally light skin and silver-colored hair.[3]

Patients with GS2 have immune system abnormalities along with hypopigmented skin and hair. The affected individuals are likely to get recurrent infections and may also develop an immune disorder called hemophagocytic lymphohistiocytosis (HLH), in which the immune system produces too many activated immune cells called T-lymphocytes and macrophages (histiocytes).

Hyperactivity of these cells can damage organs and tissues throughout the body and cause life-threatening complications if the condition is untreated[1]. Griscelli syndrome type 2 (GS2) is a rare autosomal recessive disease triggered by mutations in the RAB27A gene.[4]

The RAB27A gene provides instructions for making a protein which is involved in a process called vesicle trafficking. Vesicle trafficking moves proteins and other molecules within cells in sac-like structures called vesicles. Though RAB27A protein is found in cells and tissues throughout the body, it appears to be most important in pigment-producing cells called melanocytes and also in certain immune cells.[4]

Griscelli syndrome type 3 (GS3) is characterized by unusually light skin and hair colour. People with GS3 do not have neurological abnormalities or immune system problems[1].

II. Case Description

A 3-year-old child presented to a pediatric clinic with high spikes of fever and symptoms of URTI at 9 months of age. On evaluation he was found have hypopigmented skin and hairs. Lab workup was suggestive of HLH. Based on the clinical findings and laboratory workup, genetic evaluation for possible familial HLH was done which showed presence of RAB27A mutation suggestive of GS2. He was treated on HLH protocol 2004 and had achieved remission. The pediatrician there had suggested them to undergo bone marrow transplant as that is the only curative treatment for this fatal disease. Subsequently they approached multiple hospitals for a possible BMT as a cure for GS2 but did not get any positive response as the child did not have HLA identical family or unrelated donor and the experience of haploidentical transplant for such a rare disease is not there and the expected outcomes are poor.

At the age of 2.6 years the mother noticed that the child has regression of his mile stones and brought him to Indraprastha Apollo Hospitals for further management. After a lot of discussion about the pros and cons, he was taken up for a Haploidentical family donor stem cell transplant using his younger brother as a donor who was screened for the presence of same mutation and was found to be negative.

At presentation to IP Apollo hospital, on examination he found to have head lagging, hypotonia of all four limbs, unable to swallow, sit and walk. On further evaluation he was found to have isolated CNS HLH. He was treated with HLH 2004 protocol (CNS directed therapy) following which he achieved remission. While waiting for the transplant the patient got infected with COVID 19 and also had features of aspiration pneumonia

which was treated symptomatically. After recovery from the same, the child was admitted for the Allogenic Hematopoietic Stem Cell Transplant.

After the scheduled conditioning regimen which lasted for 7 days, he was infused with the GCSF mobilized peripheral blood stem cells collected from brother. From day +1, post BMT he started to have high spikes which was managed symptomatically considering this to be haploidentical fever. He received post-transplant cyclophosphamide for GvHD prophylaxis along with Tab.Sirolimus (Rapamycin) and Myfortic (Mycophenelate sodium) which were started on Day 5.

In view of persistence of fever with high IL6 levels, he received Inj.Tocilizumab following which his fever decreased. He had severe mucositis which manifested by red and swollen mucosa and ulcers in the mouth and loose stools. After a short period of defervescence, his fever reappeared His fever spikes continued and the blood culture showed Gram Negative Bacilli hence the antibiotics were upgraded

He was supported with prophylactic antimicrobials, granulocytes , platelets and packed red cells . Gardually he has started regaining his mile stones he has started sitting without support ,started walking and even had better swallowing and started talking few words .

He was observed for few more days in the BMT for Veno Occlusive Disease, engraftment syndrome and GVHD. A post engraftment Chimerism was sent on day +21 which showed 100% Donor DNA.

Nursing management

Highly skilled and specialized nursing care is essential for all the nurses who works in bone marrow transplant unit**.** The nurses in the Team plays an integral role during the entire transplant process, during the pre-transplant phase, nursing expertise is exemplified in the administration of chemotherapy, management of side effects and complications, teaching of transplant procedures to patient and family and supportive care which starts on the day the patient and the family decides to go for transplant on admission to the unit, and that continues throughout their journey.

Infection Prevention: Any personnel who enters the transplant unit adhered to the infection control practices. However, the nurses in the department holds the baton to ensure everyone in the department followed the practices to 100% compliance

Strict aseptic technique, personal and environmental hygiene was maintained

Regular mouth wash for oral care was given

All care bundles of central line insertion and maintenance was followed

Neutropenic food was provided to maintain adequate nutrition

Proper Isolation practices and maintaining the required positive pressure and proper maintenance of HEPA filter was done

Drugs: The patient was advised to continue immunosuppressant (Tab Sirolimus, Tab Myfortic)Antiviral Prophylaxis(Valcyclovir),Antifungal prophylaxis Voriconazole),VOD prophylaxis (Ursodeoxycholicacid) ,PCP prophylaxis (Chlotrimoxazole) ,Antibiotic (Ciprofloxacin). The patient was advised to do the Sirolimus trough levels weekly and to maintain a level of 6-12ug/L

Care of Central Line: A PICC (Peripherally inserted central Catheter) was inserted prior to the transplant. Complete aseptic technique maintained throughout the stay. The patient sent to home with the advice of weekly dressing and flushing with Hepsaline by the BMT staff.

Nutritional Support: The course of transplant is long, and the side effects like nausea and vomiting is unavoidable during this time. The food intake became lesser as the patient had mucositis. A Ryle's Tube was inserted and feeding given as per his calorie requirements. Diversional therapies like music and television helped to some extent.

Management of pain: Pain is assed every hourly and managed conservatively. A systematic escalation of analgesics done to alleviate the symptoms. Most of the time the patient might require an application of subdermal patches of fentanyl or a continuous infusion of morphine or fentanyl for the pain control.

Management of Side Effects and complication: A BMT nurse should be skillful and knowledgeable about the transplant and the disease condition in observing for possible side effect, early identification and management of any side effects and complications.

- **Nausea, vomiting, diarrhea** was managed by the use of regular anti emetics. Diversional therapy helped to a certain extend. Maintaining the intake output balance was the major issue so initiation of IV fluids and electrolytes management played the key role of management.
- **Mucositis** was managed with magic mouth wash. Magic mouth wash is a special preparation which contains Glycerine, Fluconazole,Metronidazole,and Dexamethazone which the patient needs to apply thoroughly every 2 hourly so that the symptoms are relieved

- **Bacterial, Fungal and Viral Sepsis**: Strict aseptic technique and hand hygiene practices were followed. Close monitoring of vital signs and watching for and managing the warning signs like hypotension, tachycardia, tachypnea and changes in oxygen saturation was ensured.
- **Cytokine Release syndrome** is an acute systemic inflammatory syndrome characterized by fever, raised IL6 levels and multiple organ dysfunction which is commonly associated with haploidentical transplant. He had developed CRS on Day +5 and was treated with one dose of Tozilizumab and supportive management .
- **Hepato Veno Occlusive Disorder (VOD)** or veno-occlusive disease with immunodeficiency is a potentially life threatening condition in which some of the small veins are obstructed. The patient was under continuous monitoring of weight , abdominal girth , intake and output to rule out any early signs and he never had VOD , although weight gain and fluid retention was treated with diuretics
- **Graft versus Host disease (GVHD)**. It is a life threatening syndrome which might occur as a complication of allogenic bone marrow transplant which results from a complex interaction between the immune cells present in the donor and the patient. The nursing role in management in the prevention, assessment of symptoms and managing a patient with GVHD is huge. A well experienced and expert BMT staff can easily diagnose GVHD by the keen observation of skin, mainly the palms and soles in the initial stages for skin GVHD, the color and consistency of every motion passed is assessed and measured to rule out the stage and grade of Gut GVHD, any discoloration of the sclera and skin is noted for the presence of Liver GVHD. A fine balance is supposed to be maintained between the intake, output, the nutritional needs and various other aspects.

He had a Grade 1 skin GVHD which was manifested by Erythematous rashes on his face and that was treated by the local application of Steroid**.**

Follow Up: On Day 23 the child has been discharged from the bone marrow transplant unit in a stable condition with all necessary instructions.

The patient was instructed to visit BMT unit weekly once for a central line dressing and blood sampling, thereafter for a regular check up with the transplant physician to have detailed evaluation and monitoring for any complications which might arise at this time .

Repeat Chimerism on day +45, day +60 and Day +100 showed 100% Donor DNA and the child continues to show improvements in his milestones.

The parents were advised to avoid crowded places, dusty areas and contact with people who has symptoms of infections.

Patient and Family Education: Patient and family education is a team effort and that starts on the day the patient or the family approaches the transplant physician or any of the transplant team, and it continues whole throughout the transplant journey. The transplant nurse shoulders the responsibility of teaching the patient in every transplant unit as they are available all the time with the patient.

The nurse plays a major role in enhancing the family's level of understanding about The process of bone marrow transplant and its possible side effects. The isolation practices and policies and its significance Hand hygiene and personal hygiene practices and its importance. The importance of sits bath and how it prevents blood stream infections, possible side effects and actions of each chemotherapy, immunotherapy and other supportive drugs.

Educating the family and preparing them for the discharge towards the recovery starts on the day of admission itself.

III. Discussion:

This case illustrates the accurate diagnosis, prompt treatment and effective nursing management of Griscelli syndrome Type 2 which is a rare disease

Reference

[1]. https://medlineplus.gov/genetics/condition/griscelli-syndrome/
[2]. https://www.ncbi.nlm.nih.gov
[3]. http://www.malacards.org/card/griscelli_syndrome_type_2_2.
[4]. https://medlineplus.gov/genetics/gene/rab27a/

Ms. Manju Joseph, et. al. "Case study on Hematopoietic stem cell transplant in Griscelli syndrome Type 2." *IOSR Journal of Nursing and Health Science (IOSR-JNHS)*, 11(01), 2022, pp. 07-09.