discontinued on day +90 post-transplant. This case highlights the diagnostic challenges in differentiating aGVHD in the post-HSCT setting from SJS/TEN-like presentations. It emphasises the importance of rapid intervention and the potential efficacy of JAK inhibitors in steroid-resistant cutaneous GVHD.

https://doi.org/10.1016/j.htct.2024.11.015

**Adult Hematology Abstract Categories**

**Other Diseases**
**OP 15**

## RAB27A MUTATION AND EBV INFECTION ASSOCIATED HEMOPHAGOCYTIC SYNDROME: A CASE REPORT

Ayşe Nur Akınel [1,*], Nihal Boz [2]

[1] *Gaziantep Cengiz Gökçek Gynecology And Pediatrics Hospital*
[2] *Adana City Hospital*

**Objective:** Hemophagocytic lymphohistiocytosis (HLH) is a nonmalignant immune regulation disorder within the histiocytosis group of diseases. It is a clinical condition in which there is fever, hepatosplenomegaly and cytopenia due to dysfunction of cytotoxic T-lymphocytes and natural killer (NK) cells, activation of macrophages and T-lymphocytes, excessive production of proinflammatory cytokines and hemophagocytosis. It can be primary (familial) and secondary. HLH may also be seen in the course of some immune deficiencies. We aimed to present a case of HLH that developed due to EBV infection and Griscelli syndrome with RAB27A mutation. **Case Report:** An 11-month-old male patient who presented with complaints of fever and swelling in the neck was hospitalized with respiratory distress and poor general condition. On physical examination hepatosplenomegaly, cervical lymphadenopathy and silver-gray hair color was detected. It was learned that the patient had a sibling who died at 3 months old with a similar phenotype. In his tests; wbc:10600/mm3, neu:5000/mm3, lympho:5100/mm3, hb:6.3 gr/dl, plt:29,000/mm3, ALT:167 IU/l, AST:410 IU/L, total bilirubin:2.7 mg/dl, direct bilirubin:2.6 mg/dl, sodium:126 mmol/L, albumin:2.3 g/L, LDH:765 U/L, fibrinogen:69 mg/dl, ferritin: 59334 ng/ml were detected. Hemophagocytosis was observed in the bone marrow. The patient was started on the HLH 2004 chemotherapy protocol, but died within the first 24 hours of treatment. The patient's tests at the time of admission showed EBV VCA IgM: 7.77 (positive), EBV PCR: 72,000 copies. A homozygous c.149delG (p. Arg50Lysfs*35) mutation was detected in the RAB27A gene. **Conclusion:** Griscelli syndrome is a rare autosomal recessive disease that can be accompanied by silver-gray hair, hypopigmentation, recurrent fever and infections, immune deficiency and neurological disorders. Primary HLH is common in our country due to the high prevalence of consanguineous marriages. The patients who cannot be diagnosed early and develop HLH can be fatal, so early diagnosis of these patients is of vital importance.

https://doi.org/10.1016/j.htct.2024.11.016

**OP 16**

## A CASE OF HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS IN A CHILD: A RARE AND CHALLENGING DIAGNOSIS

Metin Çil [1,*], Vusal HASANOV [2], Merve KILIÇ ÇİL [3], Özlem GÜNDEŞLİOĞLU [4], Derya ALABAZ [4]

[1] *Adana City Training and Research Hospital, Clinic of Pediatric İnfectious Diseases*
[2] *Çukurova University, Faculty of Medicine, Department of Child Health and Diseases*
[3] *Adana City Training and Research Hospital Clinic of Pediatric Hematology and Oncology*
[4] *Çukurova University, Faculty of Medicine, Department of Pediatric İnfectios Diseases*

**Case report:** Hemophagocytic lymphohistiocytosis (HLH) is an aggressive and life-threatening increased inflammatory syndrome characterized by over-activation of the immune system. Although it is more common in the first years of life, it can be seen in children and adults of all ages. HLH is divided into primary (familial) and secondary (sporadic). Secondary HLH may be associated with malignancy, rheumatologic diseases, chemotherapy or immunosuppressive therapies, but the most common cause is infections. Many viral, bacterial, fungal and protozoa infections can be triggers for both secondary and familial HLH. A 7-year-old male patient was admitted to our clinic with complaints of fever, malaise and anorexia. Blood tests revealed WBC:2260/mm$^3$, Hb:7.1 g/dL, ANS:920/mm$^3$, Platelets:41.000/mm$^3$, Fibrinogen:127 mg/dL, Triglycerides:247 mg/dL, Ferritin:687 ng/mL and splenomegaly on physical examination. Bone marrow aspiration (BMA) examination revealed hemophagocytosis. HLH was diagnosed according to HLH-2004 criteria and HLH-2004 protocol including dexamethasone and cyclosporine treatment was started. Fever, cytopenia and splenomegaly developed again during outpatient follow-up of the patient who responded adequately to the treatment. Ferritin level increased above 3000 ng/mL and another BMA was performed. This time, amastigotes were seen in addition to hemophagocytosis in the BMA evaluation. Leishmania rapid diagnostic test (RK-39) and PCR were positive. Liposomal amphotericin B treatment failed to elicit an adequate response. Meglumine Antimoniate (Glucantime) treatment was then initiated. No mutation was detected in the HLH genetic examination sent at the time of

the initial diagnosis. At the end of 4 weeks of treatment, PCR was negative and clinical improvement was seen. Visceral leishmaniasis (kala-azar) is an infectious disease caused by *Leishmania donovani* and *Leishmania infantum* and causes secondary HLH. In cases where there is no response to HLH treatment, evaluation for leishmania should be performed if it has not been done before. If diagnosed early, patients may recover with antileishmanial treatment alone. In cases of visceral leishmaniasis presenting with the hemophagocytic syndrome, if no response to amphotericin B treatment is obtained, Meglumine Antimoniate treatment should be considered for complete clinical recovery improvement.

https://doi.org/10.1016/j.htct.2024.11.017

OP 17

## CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE I: PATIENT WITH ANEMIA AND SKELETAL ANOMALY

Sevde Yazıcı Şahin [1,*], Şeyma Yılmaz [1], Haşim Atakan Erol [2], Esra Terzi Demirsoy [1], Ayfer Gedük [1], Özgür Mehtap [1], Pınar Tarkun [1], Abdullah Hacıhanifioğlu [1]

[1] *Kocaeli University*
[2] *Başakşehir Çam ve Sakura City Hospital*

**Objective:** Congenital dyserythropoietic anemias (CDAs) are inherited anemias that affect the erythroid lineage. CDAs are classified into the 3 major types (I, II, III) according to morphological, clinical and genetic features. [(1)] Before genetic diagnostic methods, bone marrow morphological abnormalities were the key diagnostic features of CDAs, such as erythroid hyperplasia with thin internuclear chromatin bridges between erythroblasts, binuclearity or multinuclearity of erythroblasts.[(2)] Next generation sequencing (NGS) has revolutionized the field of diagnosis. CDA type I (CDAI) is characterized by severe or moderate anemia and congenital anomalies such as skeletal abnormalities, chest deformity and short stature with identification of biallelic pathogenic variants in *CDAN1* or *CDIN1*. [(3)] The standard clinical management of CDA patients is measurement of hemoglobin and iron, transferrin saturation and serum ferritin concentration to monitor iron overload every three to six months. **Case report:** 28 year old female patient presented to our clinic with normocytic anemia (hb: 6,50 g/dL), splenomegaly, short stature, limb and vertebral deformities. Clinically, the patient was evaluated for anemia. All routine blood investigations were done (Table 1). Bone marrow biopsy showed hypercellularity for age with increased rate in the erythroid series with marked dysmorphism findings. (figure 1) Next-generation sequencing was performed for diagnosis in the patient with dyserythropoiesis and morphological anomaly. Homozygous variant of CDIN1 gene was detected. Detailed NGS and karyotype analysis of the patient are shown in table 2 and 3. The patient diagnosed with congenital dyserythropoietic anemia type 1 and followed up with deferasirox and erythrocyte replacement. **Conclusion:** CDAs are characterized by clinical and genetic heterogeneities. NGS based testing allows diagnosis. The increased knowledge of the genetic features and the detailed phenotyping of these patients will allow for the earliest start of the necessary treatment for the affected patients, as well as the monitoring of hemoglobin and iron levels. References:(1) Iolascon A, Andolfo I, Russo R. Congenital dyserythropoietic anemias. Blood. 2020 Sep 10;136(11):1274-1283. doi: 10.1182/blood.2019000948. PMID: 32702750. (2) Roy NBA, Babbs C. The pathogenesis, diagnosis and management of CDA type I. Br J Haematol. 2019. doi: 10.1111/bjh.15817. (3) Heimpel H, Kellermann K, Neuschwander N, Högel J, Schwarz K. The morphological diagnosis of congenital dyserythropoietic anemia: results of a quantitative analysis of peripheral blood and bone marrow cells. Haematologica. 2010;95(6):1034-1036

**Table 1 – Laboratory investigations**

| Laboratory Tests | Results | Normal Value |
|---|---|---|
| Hemoglobin | 6,50 g/dl | 12,1 - 16,6 |
| Total Leukocyte Count | 4,09 × $10^3/\mu l$ | 3,46 - 10,04 |
| Platelets | 232 × $10^3/\mu l$ | 172 - 380 |
| Mean Corpuscular Volume | 96,20 fl | 81,8 - 98 |
| Reticulocytes | 0,0747 × $10^6/\mu l$ | 0,0188 - 0,1086 × $10^6/\mu l$ |
| Total Bilirubin | 1,15 mg/dl | < 1,2 |
| LDH | 126 | 135-214 |
| İndrect Coombs | Negative | - |
| Haptoglobulin | <0,1 g/l | 0,3 - 2 |

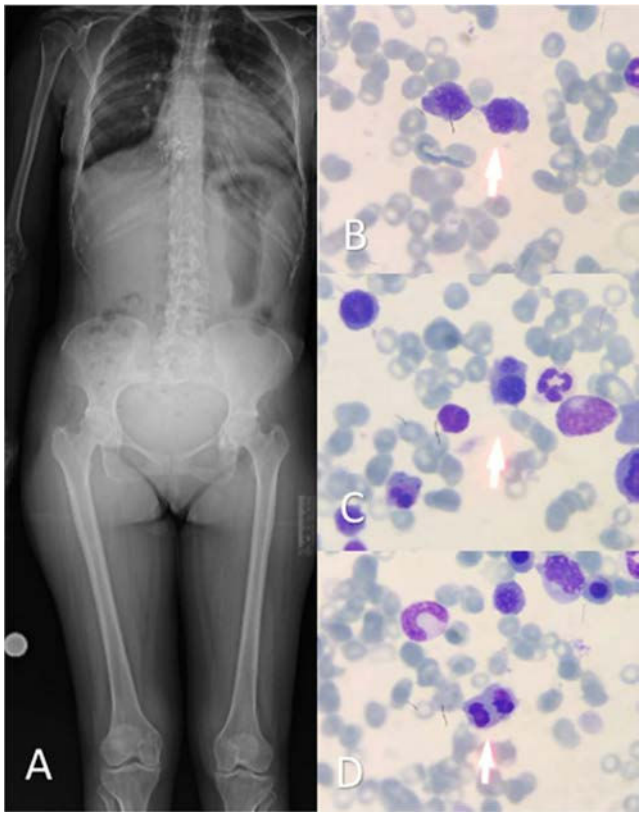


**Figure 1: A)** The patient's height was measured as 140 cm and scoliosis was detected in the skeletal survey. Bone marrow aspiration findings: **B)** Nuclear chromatin bridges between