

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.

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OP 17

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

Sevde Yazıcı Şahin^{1,*}, Şeyma Yılmaz¹,
Haşim Atakan Erol², Esra Terzi Demirsoy¹,
Ayfer Gedük¹, Özgür Mehtap¹, Pınar Tarkun¹,
Abdullah Hacıhanıoğlu¹

¹Kocaeli University

²Başakşehir 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	$4,09 \times 10^3/\mu\text{l}$	3,46 - 10,04
Platelets	$232 \times 10^3/\mu\text{l}$	172 - 380
Mean Corpuscular Volume	96,20 fl	81,8 - 98
Reticulocytes	$0,0747 \times 10^6/\mu\text{l}$	$0,0188 - 0,1086 \times 10^6/\mu\text{l}$
Total Bilirubin	1,15 mg/dl	< 1,2
LDH	126	135-214
İndirect Coombs	Negative	-
Haptoglobulin	<0,1 g/l	0,3 - 2

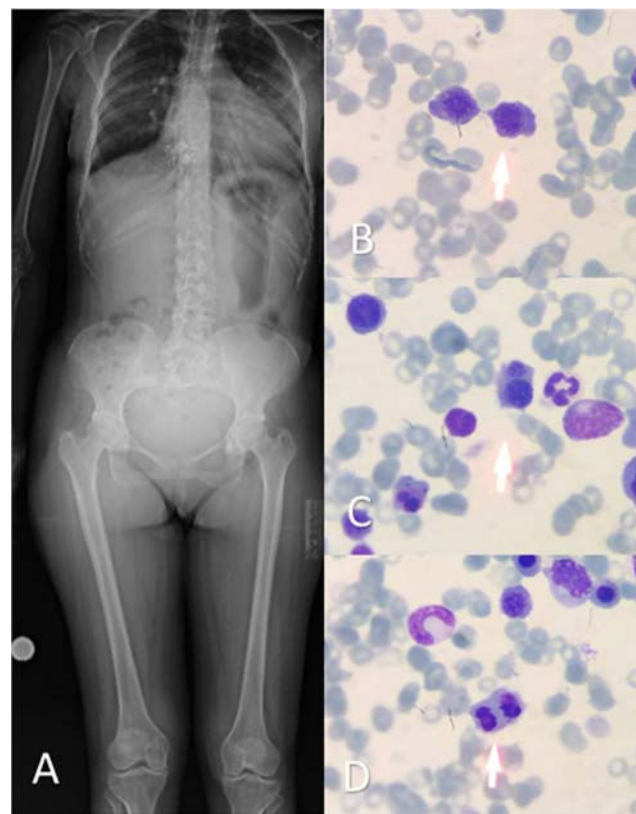


Figure 1: A) The patient's height was measured as 140 cm and scoliosis was detected in the skeletal survey. Bone marrow aspiration findings: **B)** Nuclear chromatin bridges between

erythroblasts. C) Binucleate erythroblasts D) Multinuclear erythroblasts.

Table 2 – Results of NGS analysis: Homozygous variant of CDIN1 gene was detected. (AR: Autosomal recessive, CDIN1: congenital dyserythropoietic anemia type 1, PRF1: perforin 1, TAF6: TATA-Box Binding Protein Associated Factor 6)

Gene transcript	Position	Inheritance	Genotype
CDIN1	Chromosome 15	AR	Homozygous
PRF1	Chromosome 10	AR	Heterozygotic
TAF6	Chromosome 7	AR	Heterozygotic

Table 3 – Cytogenetic analysis of the patient: With the cytogenetic analysis of the patient, tetraploid chromosome formation was detected in 17 of 18 of the metaphases as 92XXXX. (DEB: clastogenic effect of diepoxybutane, G6PD: Glucose-6-Phosphate Dehydrogenase)

Chromosomal analysis	92,XXXX	TP53 deletion	%85
Del20q12	%85	Del7q31.2	%83
Del5q31.2	%85	Tetrasomy7/tetrasomy8	%85
DEB	%0	G6PD	20,3 µg Hb (normal)
Pyruvate kinase	397,9 (normal)	Osmotic Fragility of Erythrocytes	Normal

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OP 18

DETERMINATION OF FREQUENCY AND RISK FACTORS OF SECONDARY MALIGNANCY DEVELOPMENT IN HEMATOLOGICAL MALIGNANCIES

Ennur Ramadan ^{1,*}, Güven Çetin ², Özge Pasin ³

¹ Bezmialem Foundation University, Faculty of Medicine

² Bezmialem Foundation University, Faculty of Medicine, Hematology Department

³ Bezmialem Foundation University, Faculty of Medicine, Biostatistics Department

Objective: Cancer remains a significant challenge within the healthcare system. According to the 2022 GLOBOCAN report, approximately 20 million people were diagnosed with cancer, and 10 million people passed away due to the disease. A significant improvement in the prevention, diagnosis, and treatment of cancer has resulted in a greater chance of overall survival for patients. Although survival rates for these patients have improved, they may be at risk for secondary malignancies. Secondary malignancy (SM) is defined as a tumor that differ from the primary tumor in terms of location, histopathology and genetics.

Secondary malignancies could be classified into two categories based on the time of occurrence: synchronous tumors occur within six months of an initial primary cancer, while metachronous tumors occur after six months. Despite various genetic and environmental factors being implicated, the pathogenesis remains unclear. There are no standard protocols for screening, prevention, diagnosis, or treatment. Additionally, most studies on this topic conducted on data from the SEER (Surveillance, Epidemiology, and End Results) database. Although this database has the advantage of including many patients, it is insufficient to examine potential risk factors because it doesn't include individual medical information such as personal and family medical history, or alcohol and smoking use. In this study, we aimed to reveal the incidence of secondary malignancy in hematological cancer patients, analyze the potential risk factors and determine which factors are associated with the development of secondary malignancies. **Methodology:** This retrospective study was conducted on 2,003 patients diagnosed with Hodgkin Lymphoma (HL), Non-Hodgkin Lymphoma (NHL), Chronic Lymphocytic Leukemia (CLL), Essential Thrombocytosis (ET), Chronic Myeloid Leukemia (CML), Primary Myelofibrosis (PMF), Polycythemia Vera (PV), Myelodysplastic Syndrome (MDS) and Multiple Myeloma (MM) who applied to the Hematology Clinic of Bezmialem Vakif University Hospital between February 2012 and May 2024. Patients aged above 18 years and had adequate medical records were included in the study. Patients with a prior history of cancer or an inadequate medical history were excluded. The study group consisted of patients with secondary malignancies. Control subjects were matched to the study group based on age, gender, and diagnosis. Clinical parameters compared between the study and control groups included age, gender, presence of B symptoms, stage (early or advanced), primary involvement (nodal or extranodal), extranodal involvement, relapse, hematopoietic stem cell transplantation (HSCT), treatments received (radiotherapy and chemotherapy), modifiable risk factors (diabetes, hypertension, smoking, alcohol use), and a family history of cancer. For statistical analysis, occurrences of SM by the site of diagnosis were described by counts and frequencies. Differences between groups were evaluated by the pearson chi-square or fisher exact test. Logistic regression models were used to determine predictors of occurrence of SM in patients with hematological malignancies. Survival probabilities and relapse were estimated using the Kaplan-Meier method. Cox regression analysis was performed to evaluate the factors affecting survival times and relapse. The hazard ratio (HR) with corresponding 95% confidence interval was determined based on the Cox proportional hazards model. Statistical significance level was set as 0.05 and SPSS (version 28) package program was used in calculations. All p values < 0.05 were considered statistically significant. This study was approved