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OLGU SUNUMU / CASE REPORT

Rare coexistence of chronic myeloid leukemia and albinism: a case report

Kronik myeloid lösemi ve albinizmin nadir birlikteliği: vaka sunumu

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Abstract

Albinism is a genetic disorder characterized by depigmentation. In the pediatric age group, coexistence of albinism and chronic myeloid leukemia (CML) is very rare. Understanding the pathophysiological mechanisms is crucial because splenomegaly and hematological symptoms, which may be observed in syndromic types of albinism, can mimic fatal immunodeficiency disorders and complicate the diagnostic process. 14-year-old female patient with congenital albinism presented with abdominal mass and abdominal pain. Bone marrow aspiration and biopsy were performed due to laboratory results showing leukocytosis and severe thrombocytosis. With cytogenetic and molecular testing confirming the t (9;22) translocation, the syndromic types of albinism, Chediak-Higashi and Griscelli syndromes, and essential thrombocythemia were excluded. For preventing tumor lysis syndrome and leukostasis, cytoreductive treatment and antiaggregant prophylaxis were started. Targeted imatinib was started after molecular confirmation. The patient's splenomegaly improved and deep molecular response achieved. Over a 26-month follow-up no dermatological side effects of imatinib were seen. Although the patient developed secondary amenorrhea, menstrual irregularities persisted after remission was achieved. This case highlights the diagnostic challenges of CML in patients with albinism and necessity of ruling out syndromic types of albinism. Additionally, detailed follow-up of potential side effects of treatments, especially considering the patient's albinism, is crucial for providing comprehensive care.

Keywords: Albinizm, pediatri, kronik myeloid lösemi, chediak-higashi

Öz

Albinizm melanin eksikliğiyle karakterize nadir bir genetik bozukluktur. Pediatrik yaş grubunda albinizm olgularında kronik myeloid lösemi (KML) birlikteliği son derece nadir bir durumdur. Albinizmin sendromik türlerinde görülebilecek splenomegali ve hematolojik belirtilerin, ölümcül immün yetmezlik bozukluklarını taklit edebilmesi ve tanı sürecini karmaşıklaştırması nedeniyle patofizyolojik ilişkilerin anlaşılması oldukça önemlidir. Konjenital albinizmli 14 yaşındaki kadın hasta, karın ağrısı ve karında kitle şikayetiyle başvurdu. Laboratuvar bulgularında lökositoz ve aşırı trombositoz bulguları nedeniyle kemik iliği aspirasyonu ve biyopsisi yapıldı. Sitogenetik ve moleküler testlerin t(9;22) translokasyonunu doğrulamasıyla albinizmin sendromik türleri olan Chediak-Higashi ve Griscelli sendromları ve esansiyel trombositemi dışlandı. Lökostaz ve tümör lizis sendromu risklerini hafifletmek için sitoredüktif tedavi ve antiagregan profilaksi başlandı. Moleküler doğrulama ile birlikte hedefe yönelik imatinib başlandı. Hastanın splenomegalisi geriledi, derin moleküler yanıt elde edildi. 26 aylık takip süresinde imatinibe bağlı dermatolojik etkiye rastlanmadı. Hastada sekonder amenore gelişmiş olsa da remisyon sağlandıktan sonra düzensizlikler devam etti. Bu vaka, albinizmli hastada KML'nin tanısal zorluklarına değinmekte ve sendromik albinizm türlerinin dışlanması gerekliliğini vurgulamaktadır. Ayrıca tedavi ajanlarının oluşturabileceği yan etkilerin albinizm perspektifinden detaylı şekilde takip edilmesi, hastanın kapsamlı bir tedavi sürecinden geçmesi için oldukça kritiktir.

Anahtar kelimeler: Albinism, pediatrics, chronic myeloid leukemia, chediak-higashi

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INTRODUCTION

Chronic myeloid leukemia (CML) is a clonal myeloproliferative neoplasm that occurs in hematopoietic stem cells. CML is caused by the Philadelphia chromosome, which is formed as a result of a reciprocal translocation [t(9;22) (q34;q11)] between the long arms of chromosomes 9 and 22, encoding the BCR-ABL1 oncoprotein. CML accounts for 2-3% of leukemia diagnoses in children[1,2].

CML has a clinically insidious course. Approximately 50% of patients may be asymptomatic. In asymptomatic cases, diagnosis is made during routine blood tests. In symptomatic patients, anemia and splenomegaly are frequently present in the clinical picture. The most common presenting complaints are fatigue, weight loss, night sweats, early satiety, and upper quadrant pain due to splenomegaly. Splenomegaly is one of the most characteristic findings, seen in 20-40% of patients[3].

Since tyrosine kinase inhibitors (TKIs) became widely used in the 2000s, CML has changed from a deadly disease into a chronic condition with near-normal life expectancy. In first-line treatment, in addition to the first-generation TKI imatinib, the second-generation TKIs dasatinib, nilotinib, and bosutinib, which are reported to have more potent effects, are used. For resistant cases or patients with T315I mutations, the third-generation TKI ponatinib or the STAMP inhibitor asciminib is used[4].

Albinism, on the other hand, is a heterogeneous group of diseases developing in the skin, hair, and eyes as a result of genetic defects in melanin biosynthesis. There are also forms of the disease seen with hypopigmentation only in the eye. Although most of its forms are non-syndromic, syndromic forms of albinism such as Chediak-Higashi Syndrome (CHS) and Hermansky-Pudlak Syndrome can be seen with certain systemic and hematological disorders along with pigmentation loss. CHS, originating from mutations in the *LYST* gene, presents especially with immune deficiency, an increase in bleeding tendency, and neurological findings[5,6].

Findings such as splenomegaly and leukocytosis, which can be seen in CML cases developing on the background of albinism, make differential diagnosis mandatory. The pathognomonic giant granules that can be seen in CHS patients can be confused with leukemic presentations[7]. Furthermore, the congenital lack of pigmentation masks the dermatological side effects of TKIs used for CML, making patient follow-up more challenging.

This case report presents a rare case of Philadelphia-positive CML and albinism occurring together. The aim of this study is to emphasize the importance of accurate diagnostic classification in hematological diseases accompanied by hypopigmentation and to discuss the management of this rare condition in light of current literature.

This study was written with informed consent obtained from patient's parents in accordance with the CARE Reporting Guidelines criteria[8].

CASE

A 14-year-old female patient presented to the outpatient clinic with abdominal swelling. She had previously visited an outside emergency room for abdominal pain. Referred to us with suspected acute leukemia, she also had congenital oculocutaneous albinism (OCA) and horizontal nystagmus. She reported no bleeding, fatigue, rash, or redness. Family history revealed consanguineous parents and a sibling who died of congestive heart disease. No specific genetic screening had been performed.

The patient's physical examination revealed that her general condition was stable, but hypopigmentation consistent with albinism was observed in her skin, hair, and body hair. Her eye examination showed bilateral nystagmus. An abdominal exam revealed tenderness on deep palpation and a palpable firm, mobile mass (splenomegaly) measuring about 4x5 cm that extended below the ribs to the left lower quadrant. Although her neurological examination was normal, incidental cerebellar atrophy was detected in the cranial MRI imaging report.

Peripheral blood smear (PBS), bone marrow aspiration (BMA), biopsy and molecular testing were used for diagnostic procedures. A cranial MRI was performed for neurological assessment. Because the patient had splenomegaly and leukocytosis in addition to albinism, initial evaluations focused on CHS and Griscelli syndrome due to their risk of progressing to macrophage activation syndrome. The discovery of cerebellar atrophy on the cranial MRI also made it necessary to consider a neurodegenerative syndrome.

BMA findings, which revealed approximately 3-4% myeloblasts and increased megakaryocytes, and the t (9;22) translocation detected in the analysis, excluded the syndromic evaluations and confirmed the chronic phase of CML. Although the significant thrombocytosis (>1.000.000/μL) was evaluated for essential thrombocythemia, genetic tests excluded the diagnosis.

Initially cytoreductive therapy was started for the patient, and appropriate TKI treatment was planned with addition to antiaggregant prophylaxis. During the diagnostic phase, hydroxyurea 2x500mg/day and methylprednisolone 16mg/day were started and titrated according to the white blood cell count to reduce the risk of leukostasis and tumor lysis syndrome due to marked leukocytosis and thrombocytosis. Acetylsalicylic acid 100mg was started for thrombocytosis and discontinued upon hematological recovery. Once the acute clinical risk subsided, methylprednisolone was systematically tapered (16 mg for two days, 8 mg for two days, and 4 mg for two days) and stopped. After determining t(9;22), imatinib 400mg/day was started directed to molecular target (adjusted to approximately 8.7 mg/kg/day or 300 mg/m2/day, based on the patient's weight of 46 kg and height of 138 cm). Subsequent to imatinib start hydroxyurea was gradually stopped.

The patient's complaints of splenomegaly and abdominal pain have regressed. Hematological remission was achieved with imatinib treatment. The BCR-ABL translocation analysis result of 0,008% showed that patient achieved deep molecular response (DMR) target. The clinical timeline from patient's initial application to achieving DMR response is summarized in Table 1. The patient was followed in remission for 26 months. Notably, no new skin issues related to imatinib were noted. Secondary amenorrhea developed in the patient, particularly during the treatment process. Although menstrual cycles returned with the disease going into remission, irregularities persisted.

Table 1. Diagnosis and treatment schedule

Date	Clinical	Laboratory/Imaging Findings
August 31	Referral from an external center; initial presentation.	4x5 cm palpable mass in the left quadrant (splenomegaly).
Sept 01.	BMA performed.	BMA results consistent with CML (~3-4% myeloblasts and increased megakaryocytes).
Sept 06	Cranial imaging.	Cranial MRI: Cerebellar Atrophy.
Sept 16	Initiation of cytoreductive chemotherapy.	WBC: 100.000/μL → 20.000/μL, Plt: 1.500.000/μL → 1.000.000/μL.
Sept 26	Discontinuation of methylprednisolone.	Tapered off over 10 days (16 mg → 8 mg → 4 mg).
Nov 21	Molecular verification and decision for targeted therapy (imatinib 400 mg/day; ~8.7 mg/kg/day).	Positive for t(9;22) Philadelphia chromosome.
March 16	Discontinuation of hydroxyurea.	Stabilization of blood counts.
June 10	9-month follow-up under Imatinib therapy.	BMA findings indicate complete hematologic remission.
Nov 01.	26-month evaluation of molecular response and resistance.	BCR-ABL transcript analysis: %0,008 DMR.

BMA: Bone marrow aspiration, CML: Chronic myeloid leukemia, MRI: Magnetic resonance imaging, DMR: Deep molecular response

DISCUSSION

The rare coexistence of CML and OCA have multifaceted situation for the clinical diagnosis and treatment. When hepatomegaly or splenomegaly, fever, and pancytopenia are detected in a patient with the albinism phenotype, CHS syndrome should initially be considered as a critical differential diagnosis. CHS caused by mutations in the LYST gene is an autosomal recessive disorder, characterized by giant lysosomal granules in leukocytes. 50-85% of CHS patients may be going an accelerated phase. This phase involves fever, jaundice, hepatosplenomegaly and widespread lymphohistiocytic infiltration. This situation may mimic leukemia or lymphoma, leading to a misdiagnosis[9-11].

CML diagnosis in our case was clarified with presence of Philadelphia chromosome and the detection of

BCR-ABL1 gene mutation. However, the accelerated phase of CHS can also present with cytopenia and splenomegaly, making the PBS crucial. The absence of pathognomonic granules and LYST gene mutations rules out the diagnosis of CHS. Amenorrhea observed in our case was evaluated as a potential side effect of cytoreductive/TKI agents in the endocrine system. Patient's DMR result confirmed that patient's pharmacological response was at the target level. In cases of CML and acute myeloid leukemia, 'pseudo–Chediak-Higashi' granules have been described as a morphological anomaly mimicking the giant granules observed in CHS. It is caused by the fusion of azurophilic granules, mostly in myeloblasts or myeloid precursors, and is a rare finding in hematological malignancies. Unlike true CHS granules, pseudo-CHS granules are not usually seen in lymphocytes. Making this distinction prevents the patient from being misdiagnosed with immunodeficiency[9,12,13].

TKIs used in CML treatment can have non-target effects on pigmentation pathways. In addition to inhibiting BCR-ABL1, imatinib may also inhibit the c-KIT receptor, which plays a crucial role in melanocyte proliferation and function. In the literature, vitiligo-like depigmentation associated with imatinib use has been reported. This condition may increase photosensitivity or mask potential side effects in patients who already have impaired melanin synthesis[14]. Even if excluded from our syndromic diagnosis, bleeding diathesis due to platelet granule deficiency may be observed in forms of albinism such as Hermansky-Pudlak syndrome. It should be noted that the adverse effects that second-generation TKIs such as dasatinib may have on platelet function can increase the tendency to bleed[14,15].

The lack of specific gene testing (e.g. LYST gene), that could not be done due to financial constraints, is a limitation of our study. Despite these limitations, the detection of Philadelphia chromosome and the BCR-ABL1 mutation showed a definitive diagnosis of CML, ruling out syndromic variants of albinism with absence of pathognomonic giant granules in the PBS. In addition cerebellar atrophy was evaluated as an incidental finding it did not present with typical deterioration seen in albinism variants.

In the literature the coexistence of CML and albinism in the pediatric age group was not reported. This case report highlights diagnostic difficulties regarding common features of pigmentation disorders and hematological malignancies that physicians may face. When hematological abnormalities are detected in patients with an albinism phenotype, syndromic albinism types must be ruled out. Genetic verification must be used to prevent misdiagnosis. The treatment and follow-up must be planned with ophthalmology and dermatology. Follow-up is essential for both hematological control and albinism.

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