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Case Report

When Immunodeficiency Meets Malignancy: A Case Report of Acute Lymphoblastic Leukemia in Probable Griscelli Syndrome

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Abstract:

Griscelli syndrome is a rare autosomal recessive immunodeficiency characterized by immune dysfunction and increased susceptibility to infections, with very limited reports describing its association with hematologic malignancies. We report a 14-year-old female who presented with fever, vomiting, and watery diarrhea and was found to have pancytopenia; bone marrow aspiration revealed 90% blast cells consistent with acute lymphoblastic leukemia. She was classified as intermediate risk and treated according to the Total XV protocol, achieving early minimal residual disease negativity. However, her clinical course was complicated by recurrent severe infections, cellulitis requiring skin grafting, persistent direct hyperbilirubinemia, delayed methotrexate clearance, and significant hepatotoxicity necessitating chemotherapy dose reductions and omission of consolidation cycles. These disproportionate toxicities, along with characteristic hair abnormalities identified by dermoscopy, raised suspicion of an underlying immunodeficiency, and a probable diagnosis of Griscelli syndrome was made, with whole exome sequencing planned for confirmation. This case highlights the importance of considering primary immunodeficiency disorders in pediatric leukemia patients who exhibit excessive treatment-related toxicity, as early recognition can significantly influence therapeutic decisions, reduce morbidity, and improve overall management

Keywords: griscelli syndrome; acute lymphoblastic leukemia; immunodeficiency; hepatotoxicity; case report

Introduction

To report a rare association between Suspected Griscelli syndrome Acute lymphoblastic leukemia and to highlight diagnostic and therapeutic challenges

Griscelli syndrome represent a rare autosomal recessive immunodeficiency characterize by Partial albinism, immune dysregulation & High infection risk.

It was first described in 1978 by Griscelli et al as partial albinism associated with different degrees of pancytopenia [1]. Most cases were described in the Turkish and Mediterranean populations. Major differential diagnoses of GS are Chediak-Higashi syndrome, Elejalde syndrome, Hermansky-Pudlak syndrome, and neutrophil functional abnormalities conditions such as Wisckott Aldrich, chronic childhood granulomatosis, and hyper IgA syndrome. The disease flares up due to macrophage and T lymphocytes activation. The disease is usually diagnosed between 4 months and 7 years of age [2].

Case scenario

A 14-year-old female presented with a history of persistent fever, frequent large-volume watery diarrhea, and vomiting. She initially sought care at a private clinic, where she received symptomatic treatment and antibiotics; however, complete blood count revealed pancytopenia, prompting referral to Al-Noor Hospital in Babil for further evaluation. At Al-Noor Hospital, she received two units of blood, and a peripheral blood film confirmed pancytopenia. She was subsequently referred to Babil Hospital, where bone marrow aspiration, ordered by primary pediatrician, suggested a diagnosis of acute lymphoblastic leukemia. During her course, she received an additional two units of blood and two doses of filgrastim before being referred to our center for further management. Review of systems was unremarkable across neurological, cardiovascular, respiratory, genitourinary, and musculoskeletal domains. Her past medical and surgical histories were insignificant, apart from prior blood transfusions, with no chronic illnesses or previous surgeries reported. She had no history of medication use or drug allergies and was not vaccinated. Family history was negative for similar conditions, chronic or malignant diseases, sudden death, or consanguinity; she has three healthy sisters. Psychosocially, she resides in a rural area with a low socioeconomic and educational background, and her father is a farmer. On examination, she was conscious, alert, and oriented, with notable pallor and jaundice but otherwise in stable condition; vital systems examination was unremarkable,

with no organomegaly, normal bowel sounds, and no skin lesions. Growth parameters showed a weight of 51 kg (75th percentile), height of 153 cm (10th percentile), and body surface area of 1.465 m². Initial laboratory workup on 12/11/2024 demonstrated severe anemia (Hb 5 g/dL), leukopenia (WBC 1 ×10⁹/L), and thrombocytopenia (platelets 123 ×10⁹/L), consistent with pancytopenia.

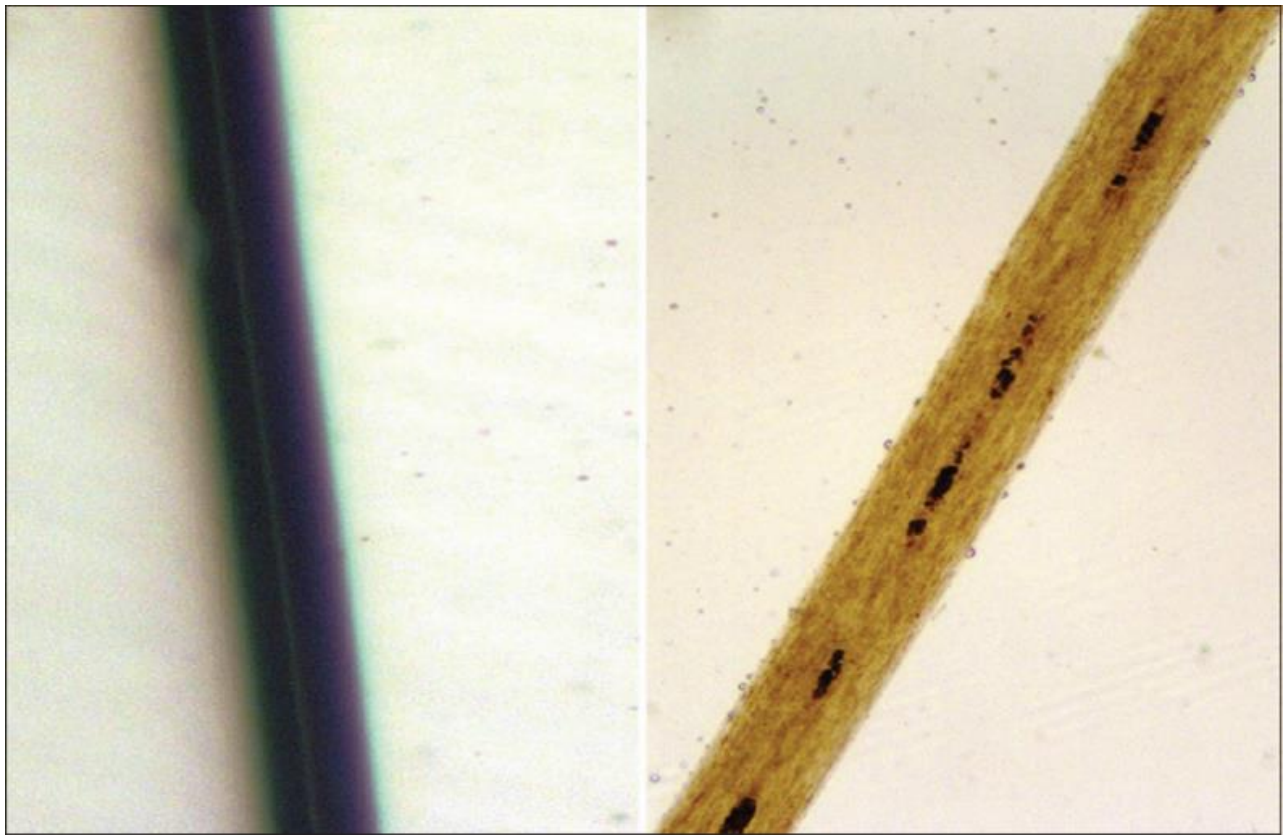

Figure 1: The figure illustrates the difference in hair shaft appearance between Griscelli syndrome and normal hair as observed under dermoscopy.

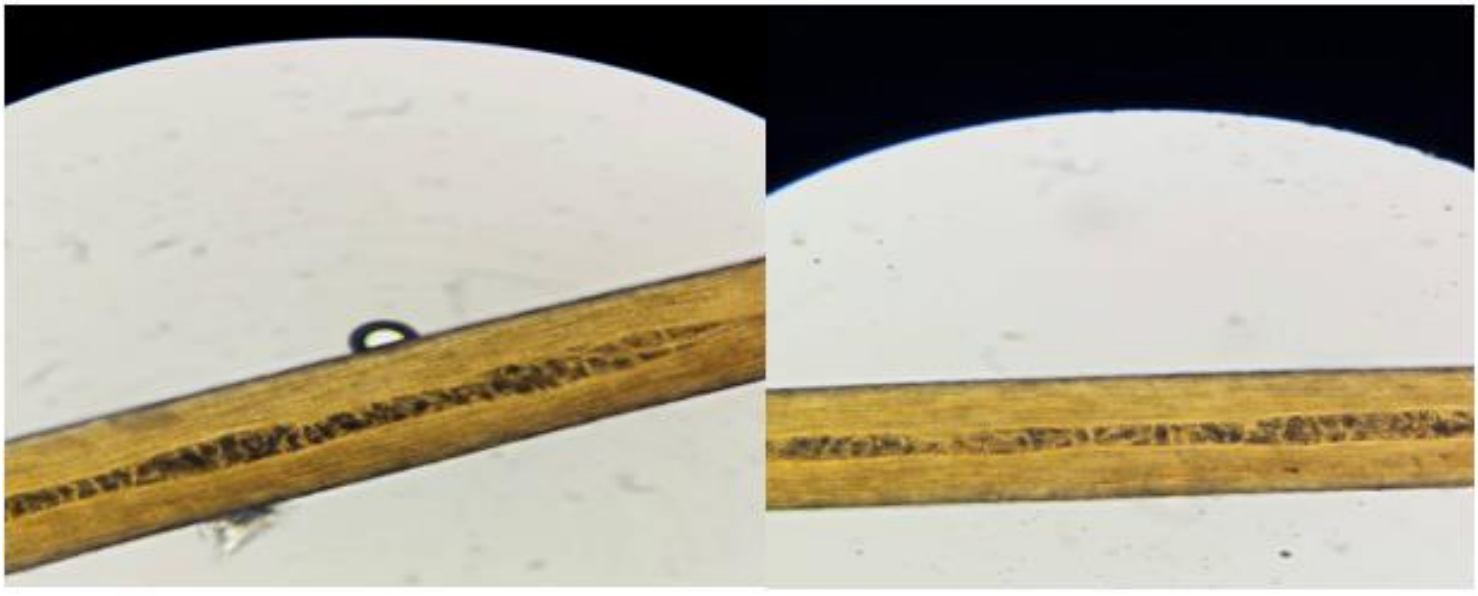

Figure 2: illustrates the dermoscopic differences in hair shaft morphology observed in our patient with Griscelli syndrome compared to normal hair.

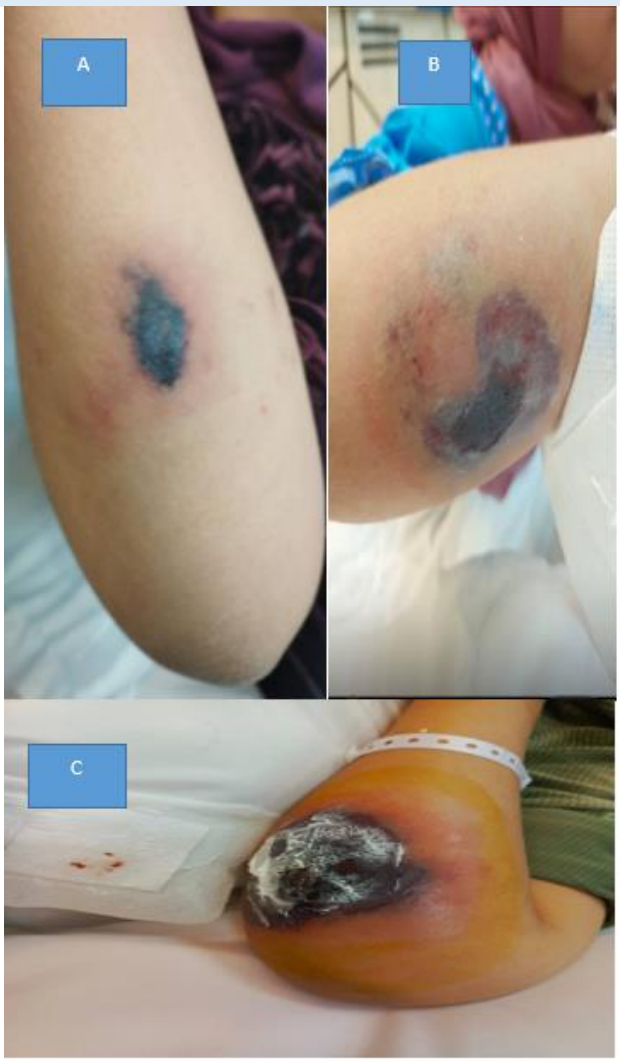

Figure 3 (A,B,C): illustrates rapidly progressive cellulitis with associated fasciitis.

Discussion:

The management of this patient highlights several important clinical considerations supported by current literature. Dose modification of chemotherapy and omission of certain intensive phases, such as consolidation, may be necessary in patients with underlying vulnerabilities, particularly when treatment-related toxicity is significant; studies have shown that individualized dose adjustment can reduce morbidity without significantly compromising outcomes in selected cases [3]. In our patient, reduction of chemotherapy intensity and early transition to maintenance were guided by hepatic toxicity and suspected immunodeficiency, aiming to balance treatment efficacy with safety. Importantly, not all observed toxicities should be attributed solely to chemotherapeutic agents, as underlying conditions such as primary immunodeficiency may mimic or exacerbate treatment-related complications [4]. Griscelli syndrome, a rare immunodeficiency disorder, can complicate the clinical picture by increasing susceptibility to infections and altering drug metabolism, thereby masking or overlapping with malignancy-related findings. This underscores the importance of a multidisciplinary approach, which has been shown to improve diagnostic accuracy and optimize therapeutic strategies in complex pediatric oncology cases [5]. Griscelli syndrome, a rare immunodeficiency disorder, can complicate the clinical picture by increasing susceptibility to infections and altering drug metabolism, thereby masking or overlapping with malignancy-related findings. This underscores the importance of a multidisciplinary approach, which has been shown to improve diagnostic accuracy and optimize therapeutic strategies in complex pediatric oncology cases [6]. Furthermore, early recognition of underlying genetic or immunologic disorders through advanced diagnostics, such as whole exome sequencing, may significantly alter management decisions and prognosis (Posey et al., 2019). In this case, clinical judgment played a critical role in modifying treatment intensity and prioritizing supportive care measures, including ursodeoxycholic acid and strict infection control, which are essential in minimizing complications. Overall, personalized treatment strategies, guided by careful clinical assessment and supported by evolving evidence, are key to improving quality of life and survival outcomes in complex cases involving malignancy with coexisting immunodeficiency [7]. Overall, this case underscores the need for individualized treatment strategies in pediatric patients with hematologic malignancy complicated by suspected immunodeficiency, where traditional high intensity chemotherapy may result in excessive toxicity; evidence suggests that reduced chemotherapy dosing and omission of further consolidation cycles, when clinically indicated, can decrease toxicity while maintaining therapeutic benefit [8]. In our patient, early transition to maintenance therapy was implemented due to significant hepatic dysfunction and treatment intolerance, highlighting that supportive care measures such as ursodeoxycholic acid and rigorous infection control are essential adjuncts that can improve tolerability and reduce complications [9].

Conclusion and Recommendation:

In this case, individualized management with reduced chemotherapy doses, omission of consolidation, and early transition to maintenance, combined with supportive care, was essential to minimize toxicity and improve tolerability. Clinical judgment remains critical, as not all adverse effects are drug-related, and underlying immunodeficiency can complicate the presentation and course of malignancy. Multidisciplinary care and early recognition of contributing conditions are vital in guiding therapy, optimizing quality of life, and potentially enhancing overall survival.

Literature search strategy and selection criteria :

A structured literature search was conducted to identify relevant studies addressing Griscelli syndrome and its association with hematologic malignancies, particularly acute lymphoblastic leukemia, as well as treatment-related toxicities in pediatric patients. Electronic databases, including PubMed, Scopus, and Google Scholar, were systematically searched using a combination of Medical Subject Headings (MeSH) and free-text terms such as “Griscelli syndrome,” “acute lymphoblastic leukemia,” “primary immunodeficiency,” “pancytopenia,” “chemotherapy toxicity,” and “hepatotoxicity,” with Boolean operators (AND, OR) applied to refine the results. The search was limited to English-language publications and primarily focused on pediatric populations, with particular emphasis on studies published within the last 10–15 years to ensure clinical relevance. Titles and abstracts were screened for relevance, followed by full-text review of potentially eligible articles, and reference lists of selected studies were manually searched to identify additional pertinent publications. Studies were included if they reported cases of Griscelli syndrome (confirmed or suspected), explored its association with hematologic malignancies or immunodeficiency, or discussed chemotherapy-related toxicities in pediatric oncology patients; eligible study designs included case reports, case series, observational studies, and review articles. Studies were excluded if they were not published in English, lacked sufficient clinical detail, focused on unrelated immunodeficiency disorders without relevant discussion, were limited to adult populations without applicability to pediatric patients, or were conference abstracts, editorials, or letters without available full-text data.

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