JHI Journal of Human Immunity

Meeting Abstracts

JHI Journal of Human Immunity

Abstracts from 5th Conference of the Arab Society for Primary Immunodeficiencies (ARAPID) in Conjunction with the 9th International Conference of the Jordanian Society of Allergy & Immunology (JSAI)

April 8–10, 2026

Amman, Jordan

All abstracts were reviewed and approved by the ARAPID scientific committee, which held full responsibility for the abstract selections

CONNECT WITH ARAPID

arapid2025 ARAPID

CONNECT WITH JHI

@jhumimmunity.org @jhumimmunity @rockefeller_university_press Rockefeller University Press jhi@rupress.org

JHI Journal of Human Immunity

ARAPID MEETING ABSTRACTS 2026

https://doi.org/10.70962/ARAPID2026abstract.1
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.1

Deficiency of Adenosine Deaminase 2 (DADA2): A Single-Center Experience with Diverse Clinical Phenotypes and Outcomes

Raed Alzyoud, Motasem Alsuweiti, Hiba Maaitah, Mohammed Noubani, Hamza Alnsour, and Sura Hnifat

Pediatric Allergy, Immunology, and Rheumatology Division at Queen Rania Children's Hospital, Amman, Jordan

Background: Deficiency of adenosine deaminase 2 (DADA2) is an autosomal recessive autoinflammatory and immunodysregulatory disorder characterized by vasculopathy, cytopenias, and variable immune dysfunction. Its expanding phenotypic spectrum continues to challenge early recognition and timely intervention.
Objective: To describe the clinical phenotypes, laboratory features, genetic variants, treatment patterns, and outcomes of patients with DADA2 at a national referral center.
Methods: We conducted a retrospective review of all genetically confirmed DADA2 cases evaluated over a 5-year period at the Pediatric Allergy, Immunology, and Rheumatology Division at Queen Rania Children's Hospital (Amman, Jordan). Demographic, clinical, laboratory, and genetic data were extracted, and therapeutic exposures and outcomes were analyzed.
Results: 10 patients (7 females, 3 males) were identified, all with homozygous pathogenic variants. The c.1471_1472dup mutation was predominant (7/10). The mean age at presentation was 63.7 ± 59.2 months, and the median diagnostic delay was 24.5 months (interquartile range 64.5). Fever was universal, and immune dysregulation was common, including vasculopathy, skin involvement, lymphoproliferation, and cytopenias. Recurrent infections occurred in only three patients. The mean ADA2 level was 1.5 mU/g protein. Two previously unreported phenotypes were observed: nonimmune hydrops fetalis and chronic pancreatitis. Treatments included corticosteroids (10/10), intravenous immunoglobulin (6/10), cyclosporine (6/10), G-CSF (7/10), mycophenolate mofetil (2/10), sirolimus (1/10), anti-TNF therapy (8/10), and hematopoietic stem cell transplantation (3/10). Overall mortality was 40%, with deaths due to sepsis (2), stroke (1), and transplant-related complications (1).
Conclusion: DADA2 demonstrates marked phenotypic heterogeneity with substantial diagnostic delays. Recognition of atypical presentations, including hydrops fetalis and chronic pancreatitis, may broaden the known disease spectrum. Early diagnosis and timely initiation of anti-TNF therapy or hematopoietic stem cell transplantation remain critical for improving outcomes.

https://doi.org/10.70962/ARAPID2026abstract.2
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.2

Inborn Errors of Immunity in Arab Countries, Access to Care: Arab Society for Primary Immunodeficiency Disorders (ARAPID) Project

Raed Alzyoud[1,2], Hind Ouair[3], and ARAPID Collaborators (ARAPID Collaborators: H. Al-Mousa[4], M. Ouederni[5], F. Ailal[3,6], N. Radwan[7], Y. Barass Ali[8], H. Shendi[9], S. Alhammadi[10], M. Alnesf[11], R. Arnaout[12], M. Ben Khaled[5], I. Benhsaien[3,6], M. Alahmad[13], and A.A. Bousfiha[3,6])

[1]ARAPID President; [2]Pediatric Allergy, Immunology Consultant, Queen Rania Children's Hospital, Amman, Jordan; [3]Laboratory of Clinical Immunology, Infection and Auto-Immunity, Faculty of Medicine and Pharmacy, Hassan II University, Casablanca, Morocco; [4]Department of Pediatrics, King Faisal Specialist Hospital & Research Center, Riyadh, Saudi Arabia; [5]Pediatric Immuno-Hematology and Hematopoietic Stem Cell Transplant Department of the National Bone Marrow Transplant Centre in Tunis, Tunisia; [6]Clinical Immunology and Infectious Pediatrics Department, Abderrahim Harouchi Hospital, Ibn Rochd University Hospital, Casablanca, Morocco; [7]Pediatric Allergy, Immunology, & Rheumatology Unit, Ain Shams University, Cairo, Egypt; [8]Pediatric Infectious Disease & Clinical Immunology, Benghazi Children's Hospital, Benghazi, Libya; [9]Sheikh Khalifa Medical City, Abu Dhabi, UAE; [10]Mohammed Bin Rashid University of Medicine and Health Sciences, Dubai, UAE; [11]Allergy and Immunology Division at Hamad Medical Corporate, Doha, Qatar; [12]Allergy and Immunology Consultant, King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia; [13]Allergy and Immunology Consultant, Al-Rashed Allergy Center, Kuwait

Introduction: Inborn errors of immunity (IEI) comprise more than 580 genetically defined disorders with heterogeneous clinical manifestations, including recurrent infections, immune dysregulation, autoinflammation, allergy, lymphoproliferation, and malignancy. Despite major diagnostic advances, timely recognition remains challenging because of limited provider awareness and overlapping or subtle presentations. In addition, access to specialized diagnostic tools and effective therapies varies globally, contributing to delayed diagnosis and suboptimal outcomes. These challenges are particularly relevant in the Arab region, where IEI prevalence is higher, partly due to high consanguinity rates.

Objectives: To evaluate access to IEI care across Arab countries using the Arab Society for Primary Immunodeficiency Disorders (ARAPID) survey, focusing on immunology workforce availability, diagnostic and genetic testing capacity, treatment options, and funding mechanisms. A structured survey was reviewed and approved by the ARAPID board members and was conducted by ARAPID country members.

Results: Eight countries participated: Kuwait, Saudi Arabia, Morocco, Tunisia, Jordan, Egypt, Libya, and the United Arab Emirates. Among the surveyed centers, 62.5% provided care for both pediatric and adult patients, while 37.5% treated pediatric patients exclusively. Basic immunologic testing was universally available, and 88% reported access to advanced cellular and functional assays. In-country genetic testing was available in 37.5% of canters, whereas 62.5% relied on overseas laboratories and 12.5% reported no access. Intravenous immunoglobulin was universally available, although only 37.5% reported a stable supply. Hematopoietic stem cell transplantation was available in 75% of countries, often requiring referral abroad, and in-country gene therapy was unavailable. Major funding gaps were identified, disproportionately affecting non-citizen patients.

Conclusions: Although the Arab region demonstrates strong foundational diagnostic capacity and broad access to core IEI therapies, critical gaps persist in adult immunology services, genetic testing, funding, and advanced curative therapies. Strengthening regional genomic services, fostering pan-Arab collaboration, and revising funding policies are essential to improving IEI care and patient outcomes.

https://doi.org/10.70962/ARAPID2026abstract.3
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.3

Clinical Phenotype and Hematopoietic Stem Cell Transplantation Outcomes in AK2-Related Reticular Dysgenesis: A Single-Center Experience

Bothainah Alaqeel[1], Faiz Aljohani[1], Nora Alrumayan[1], Ali Al-Ahmari[2], Reem Mohammed[1], Hawazen Alsaedi[2], Mouhab Ayas[2], Sultan Albuhairi[1], Sahar Elshorbagi[1], Rand Arnaout[1], Anas Alazami[3], Bander Alsaud[1], and Hamoud Al-Mousa[1]

[1]Section of Pediatric Allergy and Immunology, Department of Pediatrics, King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia; [2]Section of Stem Cell Transplantation, Department of Pediatric Hematology/Oncology, King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia; [3]Department of Translational Genomics, Genomic Medicine Center of Excellence, King Faisal Specialist Hospital & Research Center, Riyadh, Saudi Arabia

Background: Reticular dysgenesis (RD) is the most severe and rarest form of severe combined immunodeficiency (SCID), characterized by profound defects in both lymphoid and myeloid lineages and caused by biallelic pathogenic variants in AK2. Due to its rarity, data on the clinical spectrum and hematopoietic stem cell transplantation (HSCT) outcomes remain limited.

Methods: We conducted a retrospective single-center study of ten patients with genetically confirmed AK2-related RD managed at King Faisal Specialist Hospital and Research Center between 2005 and 2025. Clinical features, immunologic and hematologic findings, genetic results, transplant characteristics, immune reconstitution, and outcomes were systematically reviewed.

Results: Ten patients from eight unrelated families were included; parental consanguinity was present in eight cases. All patients presented in the neonatal period with severe bacterial infections and profound neutropenia refractory to granulocyte colony-stimulating factor, and all had bilateral sensorineural hearing loss. Nine patients shared the same homozygous missense variant (AK2 c.524G>C; p.Arg175Pro), suggesting a founder effect, while one patient carried a start-loss mutation (c.1A>G). Seven patients underwent HSCT at a median age of 4 months using matched sibling, haploidentical, or umbilical cord blood donors. Six of seven transplanted patients survived, with a post-transplant survival rate of 85.7% after a median follow-up of 9 years. All transplanted survivors achieved full engraftment of donor myeloid and lymphoid cells, with robust immune reconstitution.

Conclusion: AK2-related RD presents with a distinctive neonatal phenotype and high pre-transplant mortality. Early HSCT enables durable engraftment and favorable long-term outcomes. These findings highlight the critical importance of early diagnosis and support newborn screening for SCID in high-consanguinity populations.

https://doi.org/10.70962/ARAPID2026abstract.4
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.4

Autoimmune Hepatitis as the Initial Presentation of A20 Haploinsufficiency with Multisystem Autoimmunity and Immunodeficiency: A Case Report from Qatar

Maryam Ali Al-Nesf[1], Salma Taha[1], Yasmin El-Khatib[1], Asaad Omar[1], Yasser Medhat Kamel[2], Honar Cherif[3], and Daniella Muallem Schwartz[4]

[1]Allergy and Immunology Division, Department of Medicine, Hamad Medical Corporation, Doha, Qatar; [2]Department of Gastroenterology and Hepatology, Hamad Medical Corporation, Doha, Qatar; [3]Hematology-BMT, National Center for Cancer Care and Research, Doha, Qatar; [4]Division of Rheumatology, Departments of Medicine and Immunology, University of Pittsburgh, Pittsburgh, PA, USA

Background: Haploinsufficiency of A20 (HA20) is a rare, complex autosomal-dominant immune dysregulation disorder first described in 2016, caused by loss-of-function variants in TNFAIP3. Autoimmune hepatitis (AIH) is an uncommon manifestation and rarely the index presentation; in our case from Qatar, AIH was the first presentation.
Case Presentation: A 26-year-old male with genetically confirmed HA20 (2020), first presented at 9 years of age with AIH complicated by cirrhosis (Child–Pugh A) and portal hypertension. He subsequently developed warm autoimmune hemolytic anemia and autoimmune enteropathy with chronic refractory diarrhea despite multiple antimicrobials, immunomodulators, and prolonged systemic corticosteroids. His course was complicated by recurrent severe infections, including recurrent esophageal candidiasis and recurrent lower respiratory tract infections with bronchiectasis, leading to multiple admissions for sepsis, pneumonia, and hepatic encephalopathy. Primary immunodeficiency (PID) was suspected based on pan-hypogammaglobulinemia. Intravenous immunoglobulin (IVIG) (40 g every 3 weeks) and steroid were initiated. Recently admitted twice with sepsis, pancytopenia (lymphocyte 0.1 × 10^3/uL, platelet [PLT] 13 × 10^3/uL), severe colitis, and marked weight loss (30 kg). Given advanced liver disease, vedolizumab was started for immune-mediated colitis without adequate response; total parenteral nutrition was added. Baricitinib was introduced cautiously after completing essential vaccinations (including recombinant zoster vaccine). After one week, preliminary laboratory results (though still early) suggested improvement in hematologic parameters (lymphocyte 0.7 × 10^3/uL, PLT 28 × 10^3/uL).
Conclusion: Childhood-onset AIH can be the initial manifestation of HA20 evolving into severe multisystem autoimmunity with immunodeficiency. Early recognition of monogenic immune dysregulation is essential. Targeting interferon signaling via the JAK–STAT pathway was suggested in HA20, and baricitinib may confer greater benefit when introduced earlier.

https://doi.org/10.70962/ARAPID2026abstract.5
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.5

Congenital Neutropenia: A Retrospective Analysis of a Tunisian Cohort

Mohamed Ali Loukil, Samia Merdassi Rekaia, Aicha Amani Bettaieb, Ilhem Ben Fradj, and Monia Ouederni

Pediatric Hematology Department, Bone Marrow Transplantation Center, Tunis, Tunisia

Background: Congenital neutropenia comprises a group of rare disorders marked by persistent or intermittent neutropenia and a high risk of severe infections. This study describes the clinical, biological, cytological, genetic, and outcome characteristics of Tunisian patients.
Methods: We retrospectively reviewed patients diagnosed with congenital neutropenia before age 18, either genetically confirmed or strongly suspected based on clinical, biological, or cytological criteria, followed at the Pediatric Hematology Department, National Bone Marrow Transplant Center, Tunis, between January 2005 and December 2023.
Results: 37 patients were included. A positive family history was present in 56% of cases. Associated malformations were observed in 34% of children. Chronic digestive disorders occurred in 46%, and oral manifestations affected 81%, mainly oral aphthosis (57%). Organomegaly was noted in 38% of cases. Patients experienced a median of three hospitalizations per year for severe infections, predominantly caused by *Staphylococcus aureus*. Neutropenia was persistent in 23 patients and intermittent in 14. Monocytosis (47%), eosinophilia (30%), and other cytopenias (33%) were common. Bone marrow aspiration revealed maturation arrest in 43% of cases, especially in non-syndromic forms ($p = 0.011$). Genetic analysis was performed in 65% of patients, identifying ELANE and CN-UNC (9 cases), G6PC3 and SBDS (6 and 3 cases), rarer entities including GATA2, SHP2, GSD1b (2 cases each), and CXCR4, CXCR2, JAGN1, ADA2 (1 case each). G-CSF was administered to 62% of patients, with a median maintenance dose of 1 µg/kg/day; three patients showed G-CSF resistance. 12 malignant transformations occurred, and six patients underwent hematopoietic stem cell transplantation. Overall mortality was 19%.

Conclusion: Management of congenital neutropenia remains challenging due to heterogeneous clinical presentations and therapeutic requirements. Establishing a national registry is essential to better characterize these patients, optimize follow-up, and improve therapeutic strategies.

https://doi.org/10.70962/ARAPID2026abstract.6
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.6

Acute Graft-versus-Host Disease Following Allogeneic Hematopoietic Stem Cell Transplantation for Inborn Errors of Immunity: A Single-Center Tunisian Cohort

Samia Rekaia, Ilhem Ben Fradj, Tayssir El Mallakh, Aicha Bettaieb, Ghofrane Bedoui, Monia Ben Khaled, and Monia Ouederni

Pediatric Hematopoietic Department, National Bone Marrow Transplantation Center, Tunis, Tunisia

Background: Acute graft-versus-host disease (aGVHD) remains a major cause of morbidity and mortality after allogeneic hematopoietic stem cell transplantation (HSCT) for inborn errors of immunity (IEI).
Methods: We retrospectively analyzed all pediatric patients who underwent allogeneic HSCT for IEI at the Pediatric Hematology Department, Bone Marrow Transplantation Center, Tunis, between January 2018 and March 2023. GVHD prophylaxis consisted of cyclosporine with short-course methotrexate for HLA-matched transplants, and post-transplant cyclophosphamide with mycophenolate mofetil and cyclosporine for haploidentical transplants. aGVHD was graded according to modified Glucksberg criteria.
Results: 37 children (median age 12 months) were included: 23 with combined immunodeficiencies, 7 with congenital phagocyte defects, 4 with immune dysregulation, and 2 with osteopetrosis. 10 patients had active infection at transplantation. Donors were haploidentical in 20 and genoidentical in 17; 26 cases involved sex mismatch. Conditioning was myeloablative in 35 patients and reduced-intensity in 2. Serotherapy with anti-lymphocyte globulin was given in 12 cases. Median $CD34^+$ cell dose was 6.1×10^6/kg. Four patients had primary graft failure. Among evaluable patients, grade II–IV aGVHD occurred in 12 (36.6%), including 2 grade III–IV cases. Incidence was 37.5% in haploidentical and 35.2% in genoidentical transplants. 10 patients responded to corticosteroids; one required ruxolitinib. No clinical or transplant-related factors were significantly associated with aGVHD. Overall survival was 50% in patients with grade II–IV aGVHD versus 100% in those without ($p = 0.4$).
Conclusion: aGVHD remains a frequent complication after HSCT for IEI, affecting one-third of patients, with substantial impact on survival. Early recognition and prompt management are essential, and new strategies to prevent and treat severe aGVHD are needed in this high-risk population.7.

https://doi.org/10.70962/ARAPID2026abstract.7
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.7

Outcomes of Allogeneic Hematopoietic Stem Cell Transplantation in Leukocyte Adhesion Deficiency Types I and III: Expanded Single-Center Experience

Bashayr Alanazi and Reem Mohammed

King Faisal Specialist Hospital & Research Center, Riyadh, Saudi Arabia

Background: Leukocyte adhesion deficiency (LAD) is a rare inborn error of immunity characterized by defective leukocyte trafficking leading to severe bacterial infections and early mortality without hematopoietic stem cell transplantation (HSCT). In 2011, our center reported outcomes of 11 patients with LAD type I. We now present an expanded cohort including LAD-I and -III, with extended follow-up and contemporary transplant approaches.
Methods: We performed a retrospective analysis of patients with genetically or immunophenotypically confirmed LAD who underwent allogeneic HSCT at a tertiary immunodeficiency center between 2010 and 2026. Data collected included LAD subtype, donor source, conditioning regimen, graft characteristics, graft versus host disease (GVHD) incidence, survival, and immune reconstitution including CD18 expression and lineage-specific chimerism.
Results: 13 patients underwent HSCT (10 LAD-I, 3 LAD-III). Median age at transplant was 6 months (interquartile range 4–14). Donors included matched sibling (n = 4), other family donors (n = 5), unrelated donors (n = 2), and cord blood (n = 2). All patients received busulfan

(Bu)-based conditioning (Bu/cyclophosphamide ± antithymocyte globulin [ATG] or Bu/fludarabine ± ATG). With a median follow-up of 10.2 years among survivors, overall survival was 84.6% (11/13). Survival was 80% in LAD-I and 100% in LAD-III. Two deaths occurred in LAD-I, including one graft failure requiring second HSCT and one late infectious mortality. Acute GVHD occurred in 15% and was limited; no severe chronic GVHD was observed. Durable immune correction was achieved in the majority of survivors. Among evaluable LAD-I patients, post-HSCT neutrophil CD18 expression was high (median 96.5%), though two long-term survivors demonstrated stable mixed chimerism with partial CD18 correction (41–49%) and remained clinically well.

Conclusions: In this expanded single-center experience, allogeneic HSCT provides durable survival and sustained immune correction in LAD types I and III. Compared to our earlier report, outcomes remain favorable with low rates of severe GVHD and acceptable graft failure risk. Mixed chimerism with partial CD18 expression may be sufficient for long-term clinical stability, underscoring the importance of functional immune reconstitution rather than full donor chimerism alone.

https://doi.org/10.70962/ARAPID2026abstract.8
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.8

Hematopoietic Stem Cell Transplantation in 47 Patients with DOCK8 Deficiency: A Single-Center Experience

Bashayr Alanazi and Hamoud Almousa

King Faisal Specialist Hospital & Research Center, Riyadh, Saudi Arabia

Background and Aims: DOCK8 deficiency is a combined immunodeficiency characterized by profound atopy, chronic viral skin disease, recurrent bacterial infections, and malignancy risk. Allogeneic hematopoietic stem cell transplantation (HSCT) is the only curative therapy. We aimed to evaluate transplant outcomes, graft characteristics, immune reconstitution, and survival in a large genetically confirmed cohort.

Methods: We conducted a retrospective analysis of 47 genetically confirmed DOCK8 deficiency patients who underwent HSCT between 2012 and 2025. Data collected included donor type, conditioning regimen, graft versus host disease (GVHD) prophylaxis, survival, lineage-specific short tandem repeat (STR) chimerism, viral reactivation, IgE reduction, and time to intravenous immunoglobulin (IVIG) discontinuation.

Results: Median age at HSCT was 6 years (interquartile range [IQR] 3–10). Donors were matched sibling donor (MSD)/family matched donor (FMD) (n = 37), matched unrelated donor (MUD) (n = 7), haploidentical (n = 2), and cord blood (n = 1). Conditioning was predominantly busulfan-based regimen 66% combined with fludarabine (Bu/Flu) and 21% with cyclophosphamide (Bu/Cy). Antithymocyte globulin (ATG) was used in 55%. GVHD occurred in 12/47 patients (25.5%) and was more frequent with Bu/Cy than Bu/Flu. Overall survival was 91.5% (43/47). Among 43 patients with available STR data, 41 achieved 100% donor lymphoid and myeloid chimerism. IgE levels decreased by a median of 98%. CMV detection decreased from 54% pre-HSCT to 6% post-HSCT, and EBV from 37.5% to 2%. Median time to IVIG discontinuation was 7.7 months (IQR 4.6–11.4).

Conclusions: HSCT achieved consistent full donor engraftment and excellent survival in DOCK8 deficiency. Significant reduction in infectious and atopic manifestations supports HSCT as definitive therapy with durable immune correction.

https://doi.org/10.70962/ARAPID2026abstract.9
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.9

Demographic Influences on Quality of Life and Patient Satisfaction in Primary Immunodeficiency: Predominant Emotional and Social Associations

Mariam Al-Mansour[1], Asmaa Ali[2], and Mona Al-Ahmad[2,3]

[1]Faculty of Medicine, Kuwait University, Kuwait City, Kuwait; [2]Department of Allergy, Al-Rashed Allergy Center, Ministry of Health, Kuwait; [3]Department of Microbiology, College of Medicine, Kuwait University, Kuwait City, Kuwait

Background and Objectives: Primary immunodeficiency (PID) is a chronic condition that can substantially affect patients' health-related quality of life (HRQoL) and their experience with healthcare services. While HRQoL and patient satisfaction have been previously

studied in PID, limited data exist on how demographic factors interact with specific HRQoL domains to influence patient satisfaction. This study aimed to evaluate HRQoL and patient satisfaction in adults with PID, focusing on demographic influences and the relationship between HRQoL and satisfaction domains.

Methods: This cross-sectional study included adult patients diagnosed with PID. HRQoL was assessed using the Medical Outcomes Study 36-Item Short Form Health Survey (RAND-36), and patient satisfaction was evaluated using the Patient Satisfaction Questionnaire Short Form (PSQ-18). Descriptive statistics summarized participant characteristics and outcome measures, while correlation analyses and general linear models examined associations between age, sex, HRQoL domains, and patient satisfaction domains.

Results: A total of 34 adult patients were included, with a male-to-female ratio of 2.1:1, and a mean age of 34.9 ± 13.6 years. Participants reported moderate to high HRQoL across most domains, with preserved physical and social functioning. Energy/fatigue was the most impaired domain, indicating a persistent disease burden, while emotional well-being and bodily pain showed intermediate scores. Overall patient satisfaction was high across most PSQ-18 domains, with comparatively lower satisfaction related to accessibility and convenience of care. Increasing age was associated with poorer social functioning and general health. Female patients reported a greater emotional burden, higher satisfaction with physicians' interpersonal manners, and lower satisfaction with financial aspects of care. Significant associations between patient satisfaction and HRQoL were primarily observed in emotional well-being and social functioning, whereas physical HRQoL domains showed no significant correlations.

Conclusion: Adults with PID generally report good physical and social functioning and high satisfaction with healthcare services. However, fatigue, emotional well-being, and social functioning remain key areas of impairment, particularly among female and older patients. The strong association between patient satisfaction and emotional and social HRQoL domains underscores the importance of integrating psychosocial support into routine PID management to enhance patient-centered outcomes.

https://doi.org/10.70962/ARAPID2026abstract.10
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.10

Autosomal Dominant CARD11-Associated Immunodeficiency Presenting with Refractory Atopic Dermatitis and Chronic Mucocutaneous Fungal Infection in an Adult

Latifah Kutbi[1] and Shuayb Elkhalifa[2,3]

[1]King Abdulaziz Hospital, Jeddah, Kingdom of Saudi Arabia; [2]Cleveland Clinic Abu Dhabi, Abu Dhabi, United Arab Emirates; [3]University of Manchester, Manchester, UK

Introduction: CARD11 encodes a scaffold protein essential for antigen receptor signaling in B and T lymphocytes through activation of the nuclear factor kappa B pathway. Pathogenic variants disrupt lymphocyte activation and immune regulation, resulting in a spectrum of immune dysregulation characterized by atopic dermatitis, recurrent infections, and variable immunodeficiency. Recognition of CARD11-associated disease in adults with refractory eczema and chronic fungal infection remains limited.

Case Presentation: A 35-year-old male with persistent atopic dermatitis, bronchial asthma, and recurrent mucocutaneous fungal infections presented with chronic erythematous, pruritic eczema affecting the dorsum and soles of the feet for five years, with recent worsening causing fissuring, bleeding, and impaired quality of life. Family history was significant for eczema and fungal infections affecting his father, brother, and grandfather. Patch testing was negative. Genetic testing identified a heterozygous CARD11 frameshift variant (c.2891delG, p.Ser964fs*101), classified as likely pathogenic and consistent with autosomal dominant CARD11-associated immunodeficiency. Topical corticosteroids and tacrolimus provided limited benefit. Dupilumab was initiated, resulting in marked clinical improvement. Posaconazole was discontinued due to intolerance.

Discussion: This case demonstrates adult presentation of CARD11-associated immunodeficiency with prominent atopic dermatitis and chronic fungal susceptibility. The strong family history supports autosomal dominant inheritance. Targeted biological therapy with dupilumab provided effective control of severe eczema despite underlying immune dysregulation.

Conclusion: CARD11-associated immunodeficiency should be considered in patients with refractory atopic dermatitis, fungal infections, and relevant family history. Early genetic diagnosis enables appropriate management and targeted therapy, improving clinical outcomes and guiding long-term follow-up.

https://doi.org/10.70962/ARAPID2026abstract.11
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.11

Epstein-Barr Virus–Driven Lymphoproliferation Due to Interleukin-2–Inducible T Cell Kinase Deficiency: A Case Report

Ghofrane Bedoui[1], Nour Nebli[1], Samia Rekaya[1], Ilhem Ben Fraj[1], Aicha Ben Taieb[1], Meriem Tira[1], Manel Gouja[1], Ameni Merdassi[1], Hajer Ben Belgacem[1], Nejla Mekki[2], Imen Ben Mustapha[2], Monia Ben Khaled[1], and Monia Ouederni[1]

[1]Department of Pediatric Immunology and Hematology, National Bone Marrow Transplant Center, Faculty of Medicine, University of Tunis El Manar, Tunis, Tunisia; [2]Laboratory of Immunology, Institute of Pasteur of Tunis, Tunis, Tunisia

Introduction: Interleukin-2–inducible T cell kinase (ITK) deficiency is a rare autosomal recessive combined immunodeficiency that predisposes patients to Epstein-Barr virus (EBV) infections and severe lymphoproliferation. Early diagnosis is crucial to guide management and prevent serious complications.
Case Presentation: We report the case of an 11-year-old boy, born to a consanguineous marriage, with a history of autoimmune hemolytic anemia and a sister who died at the age of four from lymphoma, hospitalized for prolonged fever, weight loss, and productive cough. On clinical examination, the patient had hepatomegaly without peripheral lymphadenopathy or splenomegaly and presented with respiratory distress. Initial laboratory tests showed microcytic hypochromic anemia associated with lymphopenia. Bone marrow examination revealed no abnormalities. Blood cultures and microbiological tests for *Mycobacterium tuberculosis*, aspergillosis, and HIV were negative. EBV serology was discordant, with positive anti-viral capsid antigen (VCA) IgG and negative anti–Epstein-Barr Nuclear Antigen (EBNA) IgG, while blood EBV PCR was initially negative. Immunological workup revealed an expansion of double-negative T cells. Thoracic imaging showed bronchiectasis and a right basal consolidation, and abdominal ultrasound demonstrated homogeneous hepatomegaly with intra-abdominal lymphadenopathy. Despite empirical treatment for community-acquired pneumonia, the patient's condition rapidly deteriorated, with worsening respiratory failure. Liver biopsy with EBV PCR confirmed high viral load and histopathological features consistent with lymphoma. Genetic analysis identified a pathogenic mutation in the ITK gene, confirming EBV-driven lymphoma secondary to ITK immunodeficiency. Unfortunately, the patient died before targeted therapy could be initiated.
Conclusion: This case highlights the development of EBV-associated lymphoma in patients carrying ITK gene mutations. Early genetic diagnosis is critical to guide therapeutic management, including consideration for hematopoietic stem cell transplantation, to reduce mortality in this rare but potentially fatal condition.

https://doi.org/10.70962/ARAPID2026abstract.12
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.12

Late Hemophagocytic Lymphohistiocytosis Relapse in Chediak–Higashi Syndrome Following Hematopoietic Stem Cell Transplantation: Case Report

Mona Jefri and Rand Arnaout

Adult Allergy/Immunology, Department of Medicine, King Faisal Specialist Hospital and Research Centre, Riyadh, Saudi Arabia

Background: Chediak–Higashi syndrome (CHS) is a rare autosomal recessive disorder caused by biallelic LYST variants, characterized by partial albinism, recurrent infections, bleeding, and progressive neurologic decline. Majority of classic cases develop hemophagocytic lymphohistiocytosis (HLH), the main cause of early mortality. Hematopoietic stem cell transplantation (HSCT) is the only curative therapy for hematologic and immune complications, but long-term data on graft stability and late relapse are limited.
Case: We report a 29-year-old female with CHS who experienced HLH relapse and worsening neurological symptoms nearly 18 years after haploidentical HSCT, with a history of mixed donor chimerism. Lab work showed neutropenia and hyperferritinemia, and bone marrow biopsy confirmed hemophagocytic activity. Treatment with systemic steroids and intrathecal hydrocortisone/cytarabine led to clinical improvement, with a second HSCT planned. Published series indicate favorable engraftment and survival outcomes in CHS, with no higher risk of graft failure than other genetic HLH forms. Mixed chimerism can sustain remission, suggesting full donor chimerism is not always required. However, long-term graft durability and late relapse are poorly documented.
Conclusion: HLH relapse can occur decades after HSCT in CHS, underscoring the need for lifelong monitoring of chimerism and immune function. Extended follow-up studies are necessary to understand late outcomes and guide management.

https://doi.org/10.70962/ARAPID2026abstract.13
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.13

Development and Validation of B Cell and Tfh Deep Immunophenotyping for Evaluation of Inborn Errors of Immunity

Y. El Bsat[1], R. Mackeh[1], B.A. Cheaib[2], M.Y. Karim[2,3], and B. Lo[1,4]

[1]Research Branch, Sidra Medicine, Doha, Qatar; [2]Division of Hematopathology, Sidra Medicine, Doha, Qatar; [3]College of Medicine, Qatar University, Doha, Qatar; [4]College of Health and Life Sciences, Hamad Bin Khalifa University, Doha, Qatar

Background: B cell targeted therapies (BCTT) are used as effective treatment options to eliminate auto-reactive B cells in autoimmune diseases. Adverse effects of BCTT can include hypogammaglobulinemia (HG) with concomitant infection risk. A subset of children developing persistent HG (pHG) post-BCTT may have an undiagnosed inborn error of immunity (IEI) that can potentially be unveiled through deep immunophenotyping of peripheral blood mononuclear cells (PBMCs) from patients. Immune cell subsets that are altered/impaired, but not directly secondary to BCTT, may indicate underlying IEIs.
Methods: Patients receiving BCTT at Sidra Medicine were recruited pre-/post-BCTT treatment. The 24-color deep immunophenotyping panel was designed and optimized on the Cytek Aurora flow cytometer. The panel is now being implemented on patient samples along with age-matched healthy donors for phenotype characterization. The gating strategy was refined for effective data analysis using FlowJo Software. Patients developing pHG will be selected for IEI investigation through whole genome sequencing and a primary immunodeficiency (PID) gene panel.
Results: The high-dimensional 24-color panel was successfully optimized on healthy donor samples, including antibody titration. Samples analyzed simultaneously on two flow cytometers in Sidra's Research and Pathology departments (College of American Pathologists [CAP] accredited) were relatively comparable, demonstrating reproducibility and cross-instrument consistency. From 74 BCTT-treated study patients, 8 (10.8%) developed pHG. In the deep immunophenotyping conducted on a lupus patient with pHG, the B cell subsets displayed a significant reduction in the plasmablast population, which are not directly targeted by the anti-CD20 BCTT treatment. This may suggest an underlying immunodeficiency that requires further investigation through functional and genetic studies for confirmation.
Conclusion: Successful troubleshooting and cross-platform validation of the 24-color panel sets the stage for its implementation in our patient cohorts, where it may uncover immunophenotypic alterations indicative of IEIs. Further examples for the utilization of this extended panel on the suspected IEI cases are still ongoing and will be presented in the upcoming stages.

https://doi.org/10.70962/ARAPID2026abstract.14
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.14

Beyond Infection: Could Infantile Colitis Signal a Rare Immunodeficiency?

Ghofrane Bedoui[1], Ghada Ben Hlima[1], Aicha Ben Taieb[1], Manel Gouja[1], Samia Rekaya[1], Ilhem Ben Fraj[1], Ameni Merdassi[1], Hajer Ben Belgacem[1], Meriem Tira[2], Nejla Mekki[2], Imen Ben Mustapha[2], F. Mellouli[1], Monia Ben Khaled[1], and Monia Ouederni[1]

[1]Department of Pediatric Immunology and Hematology, National Bone Marrow Transplant Center, Tunis, Tunisia; [2]Immunology Laboratory, Pasteur Institute of Tunis, Faculty of Medicine, University of Tunis El Manar, Tunis, Tunisia

Introduction: Infantile colitis represents a diagnostic challenge due to its heterogeneous etiologies, including infectious, inflammatory, and immunological causes. While most cases are benign, some may reveal rare primary immunodeficiencies requiring thorough investigation and specialized management.
Case Presentation: We report a 17-month-old infant, born to second-degree consanguineous parents, with severe asymmetric intrauterine growth restriction. At 4 months, he was admitted for febrile abdominal distension, diarrhea, and vomiting. Surgical exploration revealed peritonitis secondary to a punctiform gastric perforation, with intraoperative cultures growing two non-groupable streptococcal species. At 8 months, the child was readmitted for chronic diarrhea, associated with grade III anitis, failure to thrive, and moderate malnutrition. Stool culture yielded extended-spectrum beta-lactamase–producing *Klebsiella pneumoniae*. Gastrointestinal endoscopy appeared macroscopically normal, but histopathology showed inflammatory and dystrophic changes of the duodenal and intestinal mucosa without identifiable pathogens, suggesting a noninfectious, likely immunodeficiency-related etiology. Despite transient

improvement with targeted antibiotics, intravenous immunoglobulins, and enteral nutrition, relapse occurred six months later. The patient also exhibited facial dysmorphism, congenital cytomegalovirus fetopathy, recurrent severe infections (multidrug-resistant urinary tract infection, nosocomial sepsis with pulmonary abscess), persistent neutropenia, and microcytic hypochromic anemia. Immunological workup revealed combined immunodeficiency, and genetic analysis confirmed a mutation in the HELLS gene, establishing immunodeficiency, centromeric instability, and facial dysmorphism (ICF) syndrome type 4. The child currently receives regular intravenous immunoglobulin replacement and antimicrobial prophylaxis and is a candidate for allogeneic hematopoietic stem cell transplantation.
Conclusion: Chronic infantile colitis associated with severe recurrent infections and dysmorphic features should prompt evaluation for rare primary immunodeficiencies such as ICF4, as early diagnosis is crucial for management and prognosis.

https://doi.org/10.70962/ARAPID2026abstract.15
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.15

Immunodeficiency, Centromeric Instability, and Facial Dysmorphism (ICF) Syndrome: A Rare Cause of Severe Early-Onset Infections

Ghofrane Bedoui[1], Ghada Ben Hmida[1], Samia Rekaya[1], Ilhem Ben Fraj[1], Aicha Ben Taieb[1], Meriem Tira[1], Manel Gouja[1], Ameni Merdassi[1], Hajer Ben Belgacem[1], Nejla Mekki[2], Imen Ben Mustapha[2], Monia Ben Khaled[1], and Monia Ouederni[1]

[1]Department of Pediatric Immunology and Hematology, National Bone Marrow Transplant Center, Faculty of Medicine, University Tunis El Manar, Tunis, Tunisia; [2]Immunology, Pasteur Institute of Tunis, Tunis, Tunisia

Introduction: Immunodeficiency, centromeric instability, and facial dysmorphism (ICF) syndrome is a rare autosomal recessive primary immunodeficiency caused by mutations in the DNMT3B, ZBTB24, CDCA7, or HELLS genes. It is characterized by a predominant humoral immune deficiency, centromeric instability involving chromosomes 1, 9, and 16, and variable facial dysmorphism. Patients typically present with severe and recurrent infections beginning in early childhood, which may be life-threatening.
Results: We report the case of a 5-year-old girl born to first-degree consanguineous parents, hospitalized since infancy for recurrent severe infections. She developed bilateral bullous pneumonia due to *Escherichia coli*, *Pneumocystis jirovecii* pneumonia, disseminated Bacillus Calmette-Guerin (BCG) infection with pulmonary and bone involvement, candidemia caused by *Candida tropicalis*, and cytomegalovirus infection complicated by macrophage activation syndrome and hepatic involvement. Her clinical course was marked by psychomotor delay, persistent axial hypotonia, and failure to thrive. No obvious facial dysmorphism was noted. Hematological evaluation revealed chronic neutropenia and normocytic normochromic aregenerative anemia without evidence of bone marrow failure. Immunological assessment showed hypogammaglobulinemia, decreased natural killer cells, a preserved proliferative response to phytohemagglutinin, reduced response to anti-CD3 stimulation, and absent tuberculin reactivity. Genetic analysis confirmed ICF type 1 syndrome with a homozygous DNMT3B mutation. The patient is receiving regular intravenous immunoglobulin replacement and antimicrobial prophylaxis and is a candidate for allogeneic hematopoietic stem cell transplantation.
Conclusion: ICF syndrome should be suspected in infants with severe early-onset infections associated with hypogammaglobulinemia and unexplained cytopenias, even in the absence of evident facial dysmorphism. Early diagnosis is crucial to optimize supportive management and evaluate eligibility for curative hematopoietic stem cell transplantation.

https://doi.org/10.70962/ARAPID2026abstract.16
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.16

High-Grade B Cell Lymphoma in Wiskott–Aldrich Syndrome: A Diagnostic and Therapeutic Challenge

Ghofrane Bedoui[1], Mehdi Bouaziz[1], Ilhem Ben Fraj[1], Aicha Ben Taieb[1], Samia Rekaya[1], Manel Gouja[1], Meriem Tira[1], Ameni Merdassi[1], Hajer Ben Belgacem[1], Nejla Mekki[2], Imen Ben Mustapha[2], F. Mellouli[1], Monia Ben Khaled[1], and Monia Ouederni[1]

[1]Department of Pediatric Immunology and Hematology, National Bone Marrow Transplant Center, Faculty of Medicine, University Tunis El Manar, Tunis, Tunisia; [2]Immunology, Pasteur Institute of Tunis, Tunis, Tunisia

Introduction: Wiskott–Aldrich syndrome (WAS) is an X-linked primary immunodeficiency characterized by microthrombocytopenia, eczema, recurrent infections, and immune dysregulation. It confers an increased risk of hematologic malignancies, particularly aggressive B cell lymphomas, whose classification may be challenging in the setting of immunodeficiency.

Case Presentation: We report the case of a 13-year-old boy with genetically confirmed WAS, receiving regular intravenous immunoglobulin replacement along with anti-infective prophylaxis. His family history was notable for a mother treated for colon cancer with surgery and chemotherapy, currently on azathioprine for chronic inflammatory colitis. He was admitted for abdominal pain with constipation, without cessation of stool or gas. Physical examination revealed a palpable mass in the left flank and iliac fossa. Imaging revealed a 6-cm hypermetabolic left abdominopelvic mass with peritoneal infiltration and multi-organ involvement (renal, thymic, nodal, and osseous). Histopathology showed diffuse proliferation of medium to large atypical B cells. Immunohistochemistry demonstrated CD20 and CD10 positivity, heterogeneous BCL2 expression, and a Ki-67 proliferation index of approximately 10%. These findings did not allow definitive differentiation between Burkitt lymphoma and high-grade diffuse large B cell lymphoma (DLBCL). Fluorescence in situ hybridization (FISH) analysis showed no MYC rearrangement, making classical Burkitt lymphoma unlikely. Haploidentical hematopoietic stem cell transplantation (HSCT) was planned after the treatment of lymphoma.
Conclusion: This case illustrates the aggressive oncologic evolution of Wiskott–Aldrich syndrome and highlights the diagnostic complexity between Burkitt lymphoma and DLBCL in immunodeficiency-associated lymphomas. Cytogenetic analysis is essential for accurate classification and therapeutic decision-making. Early curative hematopoietic stem cell transplantation even from an alternative donor remains crucial in WAS to prevent life-threatening complications, including malignancy.

https://doi.org/10.70962/ARAPID2026abstract.17
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.17

Lipopolysaccharide-Responsive Beige-Like Anchor Protein Deficiency in Five Pediatric Patients

M. Bouaziz, S. Rekaia, E. Ben Fraj, G. Bedoui, A. Ben Taieb, M. Ben Khaled, and M. Ouederni

National Bone Marrow Transplant Center, Tunis, Tunisia

Background: Lipopolysaccharide-responsive beige-like anchor protein (LRBA) deficiency is a rare autosomal recessive primary immunodeficiency characterized by immune dysregulation and clinical heterogeneity. Diagnostic delay is frequent and contributes to significant morbidity. We report five pediatric cases managed at the Pediatric Hematology Department of the National Bone Marrow Transplantation Center in Tunis.
Methods: Clinical, immunological, and genetic data of five pediatric patients with LRBA deficiency were retrospectively reviewed.
Results: Five patients (three females, two males), all but one from consanguineous families, were included. Mean age at symptom onset was 90 months, with a mean diagnostic delay of 38 months (mean age at diagnosis, 128 months). Evans syndrome was present in all patients, being the inaugural manifestation in three. Autoimmune enteropathy with chronic diarrhea occurred in two patients and autoimmune hepatitis with primary biliary cholangitis in one. Recurrent infections were observed in two patients; no lymphoproliferation was noted. Immunological work-up showed hypogammaglobulinemia in three patients; lymphocyte subsets were normal in all. Molecular analysis identified homozygous class 4 or 5 LRBA variants in all cases. Three patients received immunoglobulin replacement therapy. All were treated with systemic corticosteroids (mean 45 months) and mycophenolate mofetil (mean 41 months); azathioprine and sirolimus were used in one patient each. One patient underwent hematopoietic stem cell transplantation. Sustained remission of Evans syndrome was not achieved in any patient. Two patients died from severe hemorrhagic complications, including one post-transplantation.
Conclusion: This series underscores the clinical heterogeneity of LRBA deficiency and the challenges of managing severe autoimmune manifestations. Care remains difficult in settings without access to targeted therapies.

https://doi.org/10.70962/ARAPID2026abstract.18
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.18

Phosphoglucomutase 3 Deficiency: Report of Three Pediatric Cases

S. Miled, S. Rekaia, E. Ben Fraj, A. Merdassi, G. Bedoui, A. Ben Taieb, M. Ben Khaled, and M. Ouederni

Pediatric Department of the National Bone Marrow Transplantation Center, Tunis, Tunisia

Background: Phosphoglucomutase 3 (PGM3) deficiency is a rare congenital disorder of glycosylation, characterized by combined immunodeficiency, severe atopy, and variable neurodevelopmental impairment.
Objective: To describe the clinical, immunological, and genetic features of three pediatric patients with PGM3 deficiency.

Methods: We retrospectively analyzed three pediatric patients with genetically confirmed PGM3 deficiency followed at the Pediatric Department of the National Bone Marrow Transplantation Center in Tunis.
Results: The cohort included two girls and one boy, with a median age at diagnosis of 15 months (range, 9–34 months). All were born to consanguineous parents. Recurrent infections began in early infancy, predominantly lower respiratory tract infections and skin infections secondary to severe eczema. Viral infections were frequent but non–life-threatening; no invasive bacterial sepsis was documented. All patients exhibited severe atopic dermatitis, persistent hypereosinophilia, and elevated serum IgE in two cases. Immunological evaluation revealed reduced total T cell counts (CD3+) in all patients, marked lymphopenia in one child, decreased CD4+ T cells, and preserved CD8+ T cells; B cell counts were normal. One patient had partially impaired functional antibody responses, without severe hypogammaglobulinemia. Genetic analysis identified a homozygous p.Glu340del variant in all patients, suggesting a founder effect. Management included antibiotic prophylaxis, topical corticosteroids, and immunoglobulin replacement in one patient. No patient received hematopoietic stem cell transplantation. Clinical follow-up was notable for persistent severe eczema with recurrent superinfections.
Conclusion: This study underscores the key features of PGM3 deficiency, including severe eczema and frequent infections. The recurrent p.Glu340del variant indicates a potential founder effect in this population. Management remains challenging, highlighting the need for improved strategies, including targeted therapies for eczema and consideration of allogeneic hematopoietic stem cell transplantation.

https://doi.org/10.70962/ARAPID2026abstract.19
J. Hum. Immun. (2026) 2 (ARAPID2026): eARAPID2026abstract.19

Malignancy in Patients with Inborn Errors of Immunity as a Presentation Beyond the Infectious Phenotype

Raed Alzyoud, Motasem Alsuweiti, Hiba Maaitah, Mohammed Noubani, Hamza Alnsour, and Sura Hnifat

Pediatric Allergy, Immunology, and Rheumatology Division at Queen Rania Children's Hospital, Amman, Jordan

Introduction: Advances in genetic sequencing have increased recognition of inborn errors of immunity (IEI), more than a quarter of which present with noninfectious features, including malignancy. Malignancy represents the initial presentation in 0.8-0.9% of cases in the U.S. and European IEI registries, most commonly in ataxia-telangiectasia, activated PI3K-delta syndrome, and DOCK8 deficiency. In addition, the incidence of cancer is 1.8-1.9 times higher in IEI patients at ages 40-50 years. The most common malignancies presenting or complicating IEI are of the lymphoreticular system: lymphomas, leukemias, malignant histiocytosis, and thymus tumors.
Methods: We retrospectively described IEI cases in which the diagnosis of IEI was established after malignancy diagnosis in the Pediatric Immunology Division IEI registry at Queen Rania Children's Hospital in Jordan over a 10-year period (2015-2025).
Results: Six patients (three males, three females) presented with malignancy at a median age (interquartile range) of 33 (35.5) months and were diagnosed with IEI at 113 (71.8) months, with a median diagnostic delay of 65 (78.5) months. These cases accounted for 1.86% of the 321 IEI patients diagnosed during the study period. Two patients had homozygous pathogenic DCLRE1C mutations and presented with lymphoma, including EBV-driven large B cell lymphoma. Other malignancies included hepatoblastoma in ataxia-telangiectasia, gallbladder adenocarcinoma in ARHGEF1 deficiency, Rosai–Dorfman disease in SLC29A3 deficiency, and Hodgkin lymphoma in RASGRP1 deficiency. IEI diagnosis was prompted by family history of DCLRE1C at the time of presentation, autoimmune cytopenias, EBV-driven lymphoproliferation, encephalopathy with elevated alpha-fetoprotein, while recurrent infections with bronchiectasis were reported in two patients (33%). Two patients (33%) died: the ataxia-telangiectasia and the H syndrome cases.
Conclusion: We have reported a higher rate of malignancy in IEI in Jordan than in the U.S. and European registries, with a diversity of malignancy types and IEI categories. Malignancy in the pediatric age group needs to be systematically evaluated for red flags of IEI, particularly noninfectious manifestations.