replacement a few years before his diagnosis of lymphoma in favor of daily amoxicillin. After the lymphoma diagnosis, he was referred to Allergy/Immunology to attempt to better characterize his immune deficiency. Genetic testing revealed a pathogenic variant in PIK3CD gene (c.3061 G>A in exon 24), associated with PIK3CD-related disorder. He initiated treatment as part of a clinical trial that included a 14-day course of parcalsib and R-CHOP. After 1 cycle, CT imaging revealed disease progression. The merits of autologous versus allogeneic transplant were discussed. There was concern that allogeneic transplant would be too high risk in the setting of refractory disease. Ultimately, in light of underlying PIK3CD-related disorder, which is associated with high risk of lymphoma recurrence, patient underwent allogeneic transplant. He is doing well post-transplant; his lymphoma is in remission.

Discussion: Genetic testing can offer greater insights and treatment direction is patients with immune deficiency disorders. Our patient underwent allogeneic, rather than autologous, transplant. This decision was driven by his genetic diagnosis and will likely lead to better outcomes.

M159
OMENN SYNDROME ASSOCIATED WITH DNA LIGASE 1
D. Matrana1, E. Smith1, M. Marble1, L. Tran1, H. Meddaugh2, A. Heifner1, A. McKernan1, L. Wall1, I. New Orleans, LA; 2. Lafayette, LA; 3. Albuquerque, NM; 4. League City, TX; 5. Houston, TX

Introduction: Pathogenic variants in LIG1, which encodes for DNA ligase 1, have recently been reported in only two cases of SCID. We present a patient with Omenn Syndrome (atypical SCID) and two heterozygous variants of uncertain significance in LIG1.

Case Description: A 28 WGA male developed a rash at approximately 1 month of age. He developed severe exfoliative dermatitis, erythrodema, hair loss, and lymphadenopathy. Due to his impaired skin integrity, he developed significant fluid volume loss, hyperkalemia, and acute renal failure. He had exceedingly low B cells, NK cells, and naïve T cells. The total CD3 count was normal, with predominance mature (CD45RO) markers. There was no evidence of maternal engraftment. He developed profound cystic encephalomalacia and succumbed to his numerous complications at 5 months of age. WES revealed two variants of uncertain significance in LIG1 which, to our knowledge, have not been previously published: deletion c.1088–2_1094delAGTCCGCA in exon 13, a canonical splice site variant; and a missense mutation c.2312 G>A in exon 24 for which in silico predictors suggest possible deleterious effect. Neither variant is present in large population cohorts.

Discussion: To our knowledge, this is the first documented case of Omenn Syndrome associated with LIG1. Based on the profound immunologic defects in this patient, recent limited reports of SCID caused by LIG1, and the fact that these specific variants are not found in the general population, we suspect that these are pathogenic variants.

M160
ATYPICAL PRESENTATION OF GRANULOMATOUS LYMPHOCYTIC INTERSTITIAL LUNG DISEASE IN A PEDIATRIC PATIENT
J. Jung1, M. Shapero1, T. Coyle1, P. Desai2, B. Aygun2, M. Santiago2, G. Ostovar1, A. Williamson1, A. Jongco1, I. Great Neck, NY; 2. New Hyde Park, NY

Introduction: Granulomatous Lymphocytic Interstitial Lung Disease (GLILD) is a dreaded complication of Common Variable Immunodeficiency (CVID). Few pediatric cases are reported. We describe a 14-year-old female diagnosed with GLILD without a previous diagnosis of CVID.

Case Description: A 14-year-old healthy female presented with cough, dyspnea, and chest pain, without fever. Her pulmonary function test demonstrated mild lower airway obstruction without reversibility but was otherwise unremarkable. CT chest with contrast showed pulmonary nodules with bilateral axillary and subpectoral lymphadenopathy. Lymph node biopsy showed non-necrotizing granulomas but was not concerning for malignancy. Laboratory evaluation demonstrated CD8 T (254 cells/µL) and CD19 B cell (102 cells/µL) lymphopenia and panhypogammaglobulinemia (IgG 336 mg/dL, IgA 33 mg/dL, IgM 30 mg/dL). Titers were protective to measles and tetanus, equivocal to rubella, HiB and pneumococcus, and negative to mumps, diphtheria, and varicella. Infection evaluation was unrevealing, including lung biopsy cultures and PCR. She had mildly elevated double negative T (DNT) cells (2.2%), B220+ DNTs (0.9%), IL-10 (8.0 pg/mL), IL-18 (944 pg/mL), soluble Fas Ligand (293 pg/mL), but normal apoptosis assay, making Autoimmune Lymphoproliferative Syndrome (ALPS) unlikely. Targeted immunodeficiency panel revealed heterozygous Variants of Unknown Significance in CSF2RB (c.2091G>A (p.Met697Ile)) and NBAS (c.1871A>G (p.Asp624Gly)), but was negative for pathogenic variants associated with dysgammaglobulinemia or ALPS. Lung biopsy revealed lymphoid infiltrates associated with non-necrotizing granulomas, chronic bronchiolitis, hyperinflammation, and zones of interstitial fibrosis with architectural remodeling, consistent with GLILD.

Discussion: Pediatric GLILD remains poorly understood. More research is needed to elucidate the history, optimal diagnosis, and management of pediatric GLILD.

M161
CAPILLARY LEAK SYNDROME IN AN EXTREME PREMATURE INFANT: NEW INSIGHTS TO PHYSIOPATHOLOGY AND TREATMENT
M. Lozano Chinga*, M. Reyes, B. Harrison, Z. Ballas, Iowa City, IA

Introduction: Systemic capillary leak syndrome (SCLS), also known as Clarkson’s disease, is characterized by episodes of increased vascular permeability followed by a phase in which fluid shifts back to the intravascular space. Most cases have been described in adults with monoclonal gammopathy. Capillary leak occurs frequently in premature infants; this condition seems to be distinct from Clarkson’s disease. We describe an extreme premature who developed SCLS and was successfully treated with anti-TNF, aminophylline, IVIG and steroids.

Case Description: A male infant born at 23 weeks gestational age developed necrotizing enterocolitis requiring bowel resection on DOL 28. After surgery, he developed severe hypotension requiring aggressive fluid resuscitation, with subsequent anasarca. Patient continued to have hypoalbuminemia and be intravascularly depleted on echocardiographic assessments despite various treatments including pressure support, plasma expanders, aminophylline and multiple doses of IVIG. Cytokine panel revealed elevated SIL-2R (S-CD25), IL-10 and IL-6. On DOL 34 a 3-day course of anti-TNF and 5-day course of high-dose methylprednisolone was started. Aminophylline target level between 15-20µg/mL was well tolerated. Urine output improved and patient tolerated self-diuresis. There was no evidence of pulmonary edema. He completed a steroid wean over 21 days with resolution of anasarca and improved cardiovascular indices.

Discussion: SCLS is a life-threatening condition. We have previously reported successful management of Clarkson’s disease with theophylline, steroids, IVIG and anti-TNF-a. This therapeutic combination is effective in neonatal capillary leak syndrome as well.
Contrarily to the classical SCLS, our patient did not develop pulmonary edema suggesting a different physiopathology in premature newborns.

**M162**

**HEMORRHAGIC BULLOUS HENOCH-SCHÖNLEIN PURPURA - AN ATYPICAL PRESENTATION OF THE MOST COMMON CHILDHOOD VASCUITIS**


**Introduction:** Henoch-Schönlein Purpura (HSP) is the most common form of systemic vasculitis in children. This immune mediated Immunoglobulin A (IgA) vasculitis can have a diverse presentation, thus thorough workup should be considered when evaluating a pediatric patient with progressive rash and arthralgias.

**Case Description:** A 4-year-old boy with a history of eczema and parvovirus B19 presented in July with a ten-day sequela of ascending palpable purpura, hemorrhagic bullous lesions, joint pain, fever, emesis, and peripheral edema. Initial symptoms were limited to an unsteady gait and single erythematous bug-bite appearing lesion. Subsequent lesions coalesced into ascending purpuric and hemorrhagic lesions from feet to ears, sparing the torso and oral mucosa. He was admitted to inpatient due to inability to bear weight. He denied abdominal pain, diarrhea, or difficulty urinating. Abnormal laboratory workup included positive rhino/enterovirus, elevated acute phase reactants, and elevated IgA. High dose steroids and Ketorolac provided immediate symptomatic improvement. He ambulated, with resolution of peripheral edema and no new lesions. At outpatient follow up, patient had resumed baseline activity with normal lab workup and much improved purpuric lesions.

**Discussion:** HSP may present as a tetrad with palpable purpura, abdominal pain, arthralgias, and renal involvement. Though this patient did not experience abdominal pain or renal manifestations, his overall clinical presentation, elevated acute phase reactants and IgA, and immediate response to steroids confirmed suspicion of the diagnosis. It is important to consider HSP in a pediatric patient presenting with an ascending rash and arthralgias to provide the most appropriate treatment.

---

**M163**

**ASYMPTOMATIC T LYMPHOPENIA IN A YOUNG ADULT WITH PTEN GENE MUTATION**


**Introduction:** PTEN Hamartoma Tumor Syndrome (PHTS) is an autosomal dominant syndrome caused by loss-of-function mutation in the phosphatase and tensin homolog (PTEN) gene. It is characterized by hamartomas, macrocephaly, malignant tumors, and developmental delay. Variable presence of immunodeficiency is an increasingly recognized feature of PTEN mutation.

**Case Description:** A 19-year-old female with PHTS and a history of follicular thyroid carcinoma was referred to Immunology clinic for persistent lymphopenia. She denied recurrent infections. Her labs revealed absolute lymphocyte counts ranging from 0.9 to 1.4 x 10^9 over the last 6 years. Subsequent immune workup revealed normal immunoglobulins, low CD T cells of 437 cells/mL, and low CD4 of 245 cells/mL, with normal numbers of NK cells and B cells. Lymphocyte stimulation to mitogens was normal, and lymphocyte stimulation to antigens was normal to Candida but low to tetanus. Tetanus antibody was protective. On retesting 4 months later, T cell counts were stably low. Patient is being monitored clinically and with periodic lymphocyte subsets.

**Discussion:** Immune dysregulation in PHTS has been reported in several case series. PTEN mutation has been listed on the IUIS classification of Inborn Errors of Immunity/Primary Immunodeficiencies since 2017, though categorized as a primarily antibody defect. In addition to hypogammaglobulinemia, it is now known that lymphopenia and inverted CD4/CD8 ratios may occur. Clinical presentations of these lymphopenic patients range from infection-free to having opportunistic infections including Pneumocystis. Patients with PTEN mutation should be monitored for immune defects, infections, and autoimmunity, with an allergist/immunologist involved in their long-term care.

---

**M164**

**NOVEL ERBIN VARIANT AND ASSOCIATED SEVERE ECZEMA IN A 3-MONTH-OLD**


**Introduction:** ERBIN deficiency has recently been reported as a cause of autosomal dominant hyper-IgE syndrome. Loss-of-function mutations in ERBIN result in dysregulation of TGF-β signaling, which leads to increased Tregs, IL-4Ra expression, and IgE. This case discusses an infant who presented with severe eczema and failure to thrive and was found to have an ERBIN mutation.

**Case Description:** Our patient is a 3-month-old male who was admitted for severe eczema since he was 2 weeks old despite emollients, hydrocortisone, and ketoconazole. Allergy & immunology were consulted for evaluation of immune deficiencies, such as hypertrophic cardiomyopathy and arterial dilatations, with periodic lymphocyte subsets. T cell counts were stably low. Patient is being monitored clinically and with periodic lymphocyte subsets.

**Discussion:** This case illustrates a novel variant in the ERBIN gene that leads to a similar immunophenotype and clinical phenotype to...