Contrarily to the classical SCLS, our patient did not develop pulmonary edema suggesting a different physiopathology in premature newborns.

**BPD with stable aeration. Stable bilateral lung expansion without pleural effusion or pneumothorax. Body wall edema.**

**M162**

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

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**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 10e9 over the last 6 years. Subsequent immune workup revealed normal immunoglobulins, low CD3 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 and failure to thrive. He had progressively worsening eczema since he was 2 weeks old despite emollients, hydrocortisone, and ketoconazole. Allergy & Immunology were consulted for evaluation of immune deficiencies and possible food allergies. His work up showed elevated CD4/CD8 ratio (3.9), mature RA/RO ratio (2.5), elevated IgE (983 IU/mL), and elevated eosinophil count (770/μL). Genetic testing revealed an ERBIN c.1879G>A(p.Val627Ile) heterozygous variant of unknown significance. Additional immunophenotyping was performed on a research basis to characterize the ERBIN mutation, which showed increased CD45RO+ Tregs (12.2%) compared to a healthy adult control (4.15%). Dupilumab was discussed as potential treatment for his eczema; however, it was deferred due to improvement with emollient, wet wraps, and topical steroids. Because cardiac complications, such as hypertrophic cardiomyopathy and arterial dilation, have been reported in ERBIN deficiency, he had a cardiac evaluation with a normal echocardiogram.

**Discussion:** This case illustrates a novel variant in the ERBIN gene that leads to a similar immunophenotype and clinical phenotype to...
ERBIN deficiency that has been reported in the literature. Through genetic testing, we were able to identify additional potential clinical manifestations and therapeutic targets.

### M165

**RECURRENT NOCARDIOSIS IN A PATIENT WITH HYPOGAMMAGLOBULINEMIA SECONDARY TO MEMBRANOUS NEPHROPATHY**

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**Introduction:** Nephrotic syndrome is a known cause of secondary hypogammaglobulinemia. The risk of infections occurs when serum IgG levels fall below 600 mg/dL. Patients present with recurrent sinopulmonary disease, typically with encapsulated organisms. I present an interesting case of recurrent nocardiosis in a patient with hypogammaglobulinemia from asymptomatic membranous glomerulopathy (MGN).

**Case Description:** A 53 year-old male with history of pulmonary membranous glomerulopathy (MGN). A 53 year-old male with history of pulmonary membranous glomerulopathy (MGN). A 53 year-old male with history of pulmonary membranous glomerulopathy (MGN). A 53 year-old male with history of pulmonary membranous glomerulopathy (MGN).

Though the patient had normal renal function and no symptoms of hypoalbuminemia, he underwent workup for renal loss of protein. Workup revealed marked proteinuria with protein/creatinine ratio of 5128 mg/g. Kidney biopsy revealed stage 2 MGN, and direct immunofluorescence was positive for anti PLA2R antibodies, consistent with primary MGN. He will soon start treatment with IVIG.

**Discussion:** Renal disease can present with isolated IgG deficiency. The underlying mechanism is theorized to be due to renal loss of the smaller IgG molecules, coupled with accelerated IgG catabolism in the kidneys. As IgG has a longer half-life than IgA and IgM, this results in an IgG deficiency. Typically, patients have abnormal renal function and clinical signs of hypoalbuminemia. Our case is unique in that our patient had no clinical manifestations of MGN, and his atypical infection prompted the workup and diagnosis of his immunodeficiency. The recurrent nocardiosis was most likely a red herring, prompting investigation.

### M166

**MORE THAN MEETS THE IGA**

D. Rosenberg*, Madison, WI

**Introduction:** A 34 year old woman was referred for an undetectable IgA level incidentally found on routine celiac disease screening.

**Case Description:** Our patient reported a history of infections including otitis media, sinusitis and pneumonia. She also reported an episode of pseudomonal infection in childhood requiring intravenous antibiotics and long-standing mild hypocalcemia. IgG and IgM levels were normal, and post-vaccine pneumococcal tiers showed appropriate response. Flow cytometry demonstrated normal T, B, and NK cell counts. A CT chest was ordered that demonstrated a right-sided aortic arch and aberrant left subclavian artery. On further evaluation, she recalled being diagnosed with mild cleft palate in childhood. Given recurrent infections, mild hypocalcemia, and history of cleft palate, we suspected a diagnosis of DiGeorge Syndrome. Microarray testing revealed a 2.55 Mb deletion of 22q11.21, consistent with this diagnosis. Otolaryngology was consulted for recurrent left otitis externa and management of cleft palate and sleep apnea. Cardiology and Endocrinology were consulted for management of cardiac anomalies and hypocalcemia, respectively. We deferred starting antibiotic prophylaxis or immune globulin replacement given reassuring labs and suspicion that otitis externa was secondary to anatomical causes.

**Discussion:** 22q11 deletion syndrome has a wide variety of immune phenotypes ranging from near-normal function to severe combined immune deficiency. In this patient, a phenotype consistent with IgA deficiency combined with anatomical abnormalities raised concern for DiGeorge syndrome, which was successfully diagnosed via microarray testing. Given the wide range of mild immune phenotypes, 22q11 deletion syndrome may elude diagnosis until adulthood, and should be carefully considered by immunology providers.

### M167

**A CASE OF EPISODIC ANGIOEDEMA AND PERIPHERAL EOSINOPHILIA (EAE) TREATED WITH ANTI-EOSINOPHIL THERAPY**

E. Kudlaty1*, L. Maurer1, F. Kuang2, 1. Chicago, IL; 2. Bethesda, MD

**Introduction:** EAE, or Gleich Syndrome, is a rare condition characterized by cyclical episodes of angioedema and weight gain associated with hypereosinophilia. This is a case treated with targeted anti-eosinophil therapy.

**Case Description:** A 34-year-old male presents with four-year history of cyclical painful swelling and firmness of his extremities, cycling every 3-4 weeks. Initial workup demonstrated hypereosinophilia, peaking at 13,434/mL. Remainder of the workup, including bone marrow biopsy, was unrevealing except IgM elevation to 647 mg/dL. His symptoms responded to intermittent prednisone during flares, but did not respond to tacrolimus or hydroxyurea. Further evaluation revealed a clonal T cell receptor gamma gene rearrangement and an atypical T-cell population (CD3negCD4pos, 2% of total T cells) on flow cytometry. A diagnosis of EAE was made. Cyclosporine (up to 125 mg twice daily) was started to target the T cell population with mild improvement in symptoms. Anti-eosinophil therapy (mopolizumab (anti-IL-5) 300 mg) was added for two doses with an improvement in blood eosinophilia (nadir 534/mL), however without further clinical benefit and therefore discontinued.

**Discussion:** EAE is a rare hypereosinophilic syndrome with symptoms and eosinophilia thought to be driven by the IL-5 producing CD3negCD4pos T cells. Emerging data suggested a role for targeting the T cell population with mild improvement in symptoms. Anti-eosinophil therapy (mopolizumab (anti-IL-5) 300 mg) was added for two doses with an improvement in blood eosinophilia (nadir 534/mL), however without further clinical benefit and therefore discontinued.

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