MICROSCOPIC POLYANGIITIS WITH SEVERE MULTI-ORGAN INVOLVEMENT IN A 12-YEAR-OLD ASIAN FEMALE WITH TYPE 1 DISTAL CHROMOSOME 22Q11.21 DELETION SYNDROME

Sharanya Joginpalli[1], Andrea A Ramirez[2], Alvar Orjuela[3], John Hicks[4]

[1] Baylor College of Medicine, Department of Pediatrics, Rheumatology

[2] Baylor College of Medicine, Texas Children's Hospital, Pediatrics, Pediatric Rheumatology

[3] Baylor College of Medicine, Texas Children's Hospital, Pediatrics, Pediatric Nephrology

[4] Baylor College of Medicine, Texas Children's Hospital, Pathology, Pathology

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Background: ANCA associated vasculitis (AAV) primarily affects blood vessels of the lungs and kidneys as well as other organ systems. The MPA subtype is a pauci-immune necrotizing small vessel vasculitis. The etiology of AAV is unknown and thought to be multifactorial. The incidence of pediatric MPA is rare with mean age of onset 9-12 years with female predominance.

Materials/Methods: A 12-year-old Asian female with history of short stature and developmental delay presented with a 3-month history of intermittent abdominal pain, anorexia, 11-pound weight loss, hematochezia and polyarthralgia. Initial diagnostic studies revealed elevated ESR of 116 mm/hr, CRP of 9.3 mg/L, hemoglobin 11.2 mg/dl, normal iron studies. Gastroenterology (GI) evaluation raised concern for inflammatory bowel disease (IBD) but stool calprotectin was negative and endoscopy did not reveal features of IBD. She presented to the ED with hematochezia and hemoglobin 6.7 mg/dl. GI team was consulted, EGD and colonoscopy revealed no bleeding source.

Results: Rheumatology evaluation revealed diffuse peripheral arthritis and generalized erythematous purpuric ulcerative lesions. Studies revealed positive antinuclear antibody titer of 1:320 with negative autoantibodies, normal complements, positive pANCA titer 1:160 and positive myeloperoxidase antibody 57 (positive > 1.0 unit). Urinalysis revealed microhematuria and proteinuria. Skin and renal tissue biopsies showed fibrinoid necrosis without granulomas and pauci-immune glomerulonephritis, cellular crescents, and sclerotic glomeruli respectively. Chest CT obtained for unexpected hemoglobin drop showed pulmonary vasculitis with possible hemorrhage. A final diagnosis of microscopic polyangiitis (MPA) was made. Due to her multiorgan involvement, she received aggressive treatment with corticosteroids, Rituximab and Cyclophosphamide (CYC). She was discharged on an oral steroid taper and monthly CYC. History of short stature, resolved heart murmur, developmental delay and subtle dysmorphic features prompted Genetics evaluation. A chromosomal microarray revealed a Type 1 distal chromosome 22q11.21 deletion syndrome distinct from DiGeorge syndrome. This was not thought to be contributory to AAV diagnosis.

Conclusions: This case highlights the serious organ involvement occurring in MPA which guides therapeutic intensity. Acute onset anemia in AAV may reveal life-threatening lung bleeding without clear clinical symptoms. Without appropriate therapy, patients may develop significant end-organ damage and die.

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