GAUCHER DISEASE TYPE 3 AND IGA VASCULITIS: A CASE REPORT

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INTRODUCTION:

Gaucher Disease (GD) is a lysosomal disorder caused by biallelic pathogenic variants in the GBA1 gene and can course with immune dysregulation. Three phenotypes have been described, and GD type 3 (GD3) is associated with chronic visceral and neurological damage. IgA vasculitis (IgAV) is a kind of immune dysregulation characterized as a non thrombocytopenic vasculitis associated with purpura, arthralgia and IgA1 immune deposition at vessel walls. Our objective is to relate the case of a 4.3-year-old male patient diagnosed with GD3 who had a benign course of IgA vasculitis.

CASE REPORT:

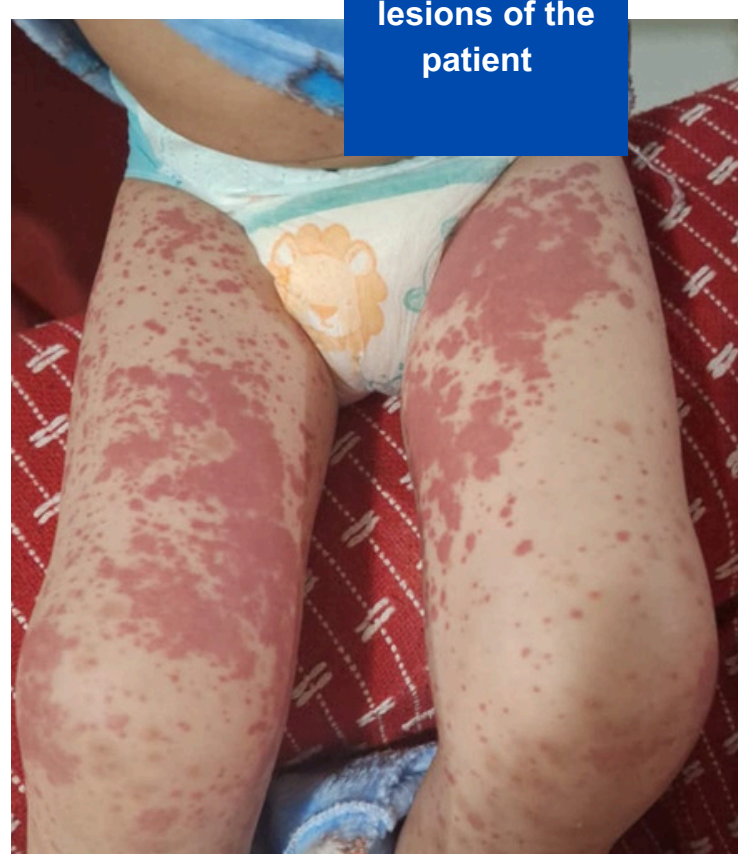
The patient was born to a non-consanguineous couple and diagnosed with GD3 at 1.8 years old due to hepatosplenomegaly, neurological impairment and strabismus. He is homozygous for the pathogenic variant c.1448T>C; p.Leu483Pro (L444P), with a a GCase activity at 1.3 nmol/h/mg protein in leukocytes (Reference value, i.e. RV, 10-45) at the time of diagnosis. He receives biweekly imiglucerase infusions of 60 IU/kg and remains stable. At the age of 4.3 years old, the patient seeked medical assistance after a week-long history of dry cough, bilateral joint pain in the knees and diffuse purpuric skin lesions in abdomen and limbs (see figures 1.A and 1.B). Blood tests at admission showed an increase in Immunoglobulin A (IgA) and C- reactive protein - 503 mg/dL (RV 21-291) and 27.1 mg/L (RV<5), respectively. There was no thrombocytopenia (platelet count was 293,000/µL, with RV>120,000/µL) nor renal function alteration (no hematuria nor proteinuria). The clinical picture along with increased plasma IgA was highly suggestive of IgA vasculitis (IgAV) diagnosis. The patient received symptomatic care, and evolved to complete resolution of symptoms.

DISCUSSION:

This case is illustrative of the benign course of IgAV in a GD3 patient, showing that even a GD3 patient who had the disease controlled by ERT could develop transient immune related alterations.

CONCLUSIONS:

This is the first report of these conditions overlap in literature to our knowledge. GD3 may be associated with the arthralgia and purpuric lesions.



Figures 1.A and 1.B: Purpuric lesions of the patient