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Gaucher disease and Lupus: A rare association?

Gaucher y lupus: ¿una rara asociación?

Dear Editor,

Gaucher disease (GD), is an autosomal recessive lysosomal storage disease that is due to mutations in the glucocerebrosidase (GC) gene, with a prevalence of 1/57,000 to 1/75,000 births worldwide[1] and significantly more common among the Ashkenazi Jewish heritage.[2] GD is categorized into three clinical types[4] and the clinical manifestations result from the accumulation of the lipid-laden macrophages in the spleen, liver, bone, bone marrow,[3] leading to impairment of central nervous system in the most severe cases.[4]

Although several reports are available related to the risk of GD patients developing other diseases like Parkinson's disease[5] and increased rates of malignancies, particularly hematologic,[6] systemic lupus erythematous (SLE) has not been described in association with GD.

We report a case of a 32-year-old Caucasian woman diagnosed with GD type 1 at 17 years-old. She had a grandmother with GD, an uncle with SLE and a cousin with rheumatoid arthritis. There is no known Jewish heritage in her family. She was medicated with velaglucerase.

With 30 years old, the patient developed malar-rash and chest eczema, associated with sun exposure. One year after she noticed worsening asthenia, anorexia, nausea, hair loss, myalgias, bilateral gonalgia, oral ulcers, Raynaud syndrome and arterial hypertension (TA 140/90 mmHg). The patient was referred to the nephrology unit with peripheral edema and the laboratory investigation showed parameters of ferropenic anemia (without signs of hemolysis), leukopenia, thrombocytopenia, elevated seric creatinine, hypoalbuminemia, active urinary sediment and nephrotic-range proteinuria observed in the 24 h urine sample (Table 1). The immune assays revealed positive antinuclear antibodies (ANA) and anti-Sjögren's-syndrome-related antigen A, elevated immunoglobulin (Ig) G (20.7 g/L) and IgA (4.64 g/L), circulating immunocomplexes (>100 μg Eq/mL) and low serum complement (C3 0.16 g/L, C4 0.021 g/L, C1q 0.208 g/L). The complementary immunologic and serologic study was negative. Renal and abdominal ultrasounds showed normal sized kidneys, increased cortical echogenicity with maintained differentiation and mild/moderate homogeneous hepatosplenomegaly. Echocardiogram revealed thickened pericardium and nuclear magnetic resonance of the inferior members showed bone alterations and moderate intra-articular left knee and mild right knee effusion.

Kidney biopsy established the diagnosis of class IV-G lupus nephritis (LN) and the treatment according to KDIGO (Kidney Disease: Improving Global Outcomes) guideline[7] was started. Hydroxychloroquine was redrawn due to gastric intolerance. She was discharged 1 month after admission with serum creatinine 1.5 mg/dL, proteinuria 4500 mg observed in the 24 h

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Table 1 – Laboratory parameters.

	Admission	Discharge	6 months	12 months
Hb (g/dL)	9.1	8.9	13.6	13.2
Leukcocytes ($\times 10^9$/L)	2900	7400	5900	4500
Platelets ($\times 10^9$/L)	87,000	168,000	237,000	367,000
Creatinine (mg/dL)	1.38 → 2.1	1.5	0.73	0.7
Albumin (g/dL)	2.1	3.1	4.1	4.2
Urinary sediment – erythrocyts (μL^{-1})	225 (15% dysmorphic)	–	8 (8% dysmorphic)	10 (4% dysmorphic)
Urinary proteins (mg/24 h)	7600	4500	341	283
ANA	Positive	–	Positive	Positive
Anti-SSA (UA/mL)	786	–	Positive	Positive
Anti-dsDNA	Negative	–	182	83
C3 (g/L)	0.16	0.42	0.78	0.90
C4 (g/L)	0.021	0.052	0.085	0.074

ANA, antinuclear antibodies; anti-SSA, anti-Sjögren's-syndrome-related antigen A; anti-dsDNA, anti-double stranded DNA; C3, complement 3; C4, complement 4.

urine sample (mg/24 h), complement levels arose and albuminemia was at the normal range (Table 1). Six months after discharge the patient is clinically stable, with normal renal function, proteinuria of 341 mg/24 h and an amelioration of complement. Anti-double stranded DNA was positive for the first time 6 months after discharge (182 U/mL) and decreased to 83 U/mL in the following 6 months (Table 1).

We describe this case of a patient with GD and SLE simultaneously with the purpose of highlighting a possible immunologic proximity between these two diseases. Although the presence of renal pathology in GD is rather rare, it consists of varying degrees of proteinuria with or without renal insufficiency and it has been described in some patients associated with the accumulation of GC in form of Gaucher bodies in glomerular, mesangial, endothelial and interstitial cells of the kidney.[8] It has been suggested that progressive accumulation of GC may trigger macrophage activation leading to chronic stimulation of the immune system.[9] Increasing clinical evidence suggests that the pathophysiology of classic GD is more complex and involves system-wide dysfunction of cell types other than macrophages.[10] Therefore, the immunological disturbances that occur in GD may function as a trigger to the development of SLE, bringing up the already existing doubt that the defects in lipid metabolism could contribute to the development of autoimmunity. In this particular case, clinical and laboratory results could led to a diagnosis of SLE: presence of 4 or more criteria, at least one clinical and one laboratorial Systemic Lupus International Collaborating Clinic (SLICC) criteria. However, and besides the clinical manifestations of malar rash associated with sun exposure, other like arthralgia, pancytopenia and hepatosplenomegaly were easily confounded with GD manifestations and the SLE diagnosis was not achieved until the renal manifestations occurred. The kidney biopsy revealing LN with positive ANA was preponderant to the diagnosis of SLE, according to SLICC criteria. Determining the class of LN was also important to guide the treatment by the histologic subtype, as the clinical presentation may not accurately reflect the severity of the histologic findings, and there was a positive clinical and laboratorial response.

However, the relationship between GD and SLE is not yet established. The involvement of immune cells has been implicated, but the underlying molecular defect is poorly understood. Further studies are necessary to highlight the possible immunologic proximity between these two rare conditions.

Declaration

Informed consent to publish individual data was obtained from the patient.

Conflict of interest

The authors declare no conflict of interest.

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Severe Parkinsonism with respiratory failure in peritoneal dialysis patient☆

Parkinsonismo severo con insuficiencia respiratoria en paciente de diálisis peritoneal

Dear Editor,

Patients with chronic kidney disease (CKD) are at an increased risk of drug-induced neurotoxicity.[1] Parkinsonism appears to be more common in patients with CKD, with a higher annual incidence rate in uraemic patients compared with non-uraemic patients.[2]

The drugs that most frequently cause Parkinsonism are calcium antagonists, orthopramides/benzamides and antipsychotics/neuroleptics.[3] Compared with Parkinson's disease, patients with drug-induced Parkinsonism are predominantly female and elderly.[3] Sulpiride is a drug used to treat dizziness, which often induces Parkinsonism.[4]

We report the case of a 52-year-old woman with systemic lupus erythematosus (SLE) and associated antiphospholipid syndrome, steroid diabetes and hypertension. The patient had CKD secondary to lupus nephropathy and began continuous ambulatory peritoneal dialysis (CAPD) in 2014, with a regimen of 4 daytime exchanges of 2000 ml: 2 Physioneal 40® 1.36% and 2 Physioneal 40® 2.27% (Baxter), with a dry night.

She went to her clinic due to a 72-h catarrhal disease and began treatment with levofloxacin 500 mg, one tablet per day. After the first dose she presented with dizziness syndrome for which she was given an intramuscular ampoule of sulpiride 100 mg, followed by 50 mg/8 h orally.

At 36 h she experienced respiratory distress with a baseline oxygen saturation of 87%. She was given oxygen therapy, intravenous (IV) corticosteroids and inhalers, without clinical improvement. During the transfer to the hospital, she presented with generalised dystonic movements. At the A&E she had serious difficulty breathing. The laboratory tests revealed mild hypocalcaemia (total corrected calcium of 8.2 mg/dl and arterial ionised calcium of 3.6 mg/dl), and she was given an IV calcium gluconate ampoule for possible tetany. No significant alterations were observed in the chest X-ray or brain CT scan.

Due to the persistence of respiratory insufficiency and generalised dystonic movements, consultation with the intensive care unit (ICU) was requested. Upon arrival, the patient presented with generalised dystonia with predominantly cervicofacial involvement and language impairment, in addition to severe ventilatory impairment.

Five (5) mg of IV biperiden were administered, which resolved the acute dystonic symptoms and improved pulmonary ventilation. Given the risk of recurrence of the symptoms and the lack of information on the elimination of sulpiride by peritoneal dialysis, it was decided to admit the patient to the ICU and to perform a haemodialysis session using a temporal–femoral catheter, with a 4008® dialysis monitor (Fresenius Medical Care), Evodial 1.6® dialyser (Gambro), for 3 h, with a blood flow of 300 ml/min and a bath flow of 500 ml/min, without ultrafiltration.

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