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CASE REPORT

Cardiac involvement in antiphospholipid syndrome associated with Sneddon syndrome: A challenging diagnosis☆

Ana Faustino[a,*], Luís Paiva[a], Ana Morgadinho[b], Emília Trigo[c], Ana Botelho[a], Marco Costa[a], António Leitão-Marques[a]

[a] *Serviço de Cardiologia, Centro Hospitalar e Universitário de Coimbra, Hospital Geral, Coimbra, Portugal*
[b] *Serviço de Neurologia, Centro Hospitalar e Universitário de Coimbra, Hospital Geral, Coimbra, Portugal*
[c] *Serviço de Medicina Interna, Centro Hospitalar e Universitário de Coimbra, Hospital Geral, Coimbra, Portugal*

KEYWORDS
Stroke;
Myocardial infarction;
Sneddon syndrome;
Antiphospholipid syndrome;
Systemic lupus erythematosus;
Livedo reticularis

Abstract Sneddon syndrome is a rare clinical entity characterized by the association of ischemic cerebrovascular disease and livedo reticularis. The authors report a case of stroke and myocardial infarction in a 39-year-old man with Sneddon syndrome and antiphospholipid syndrome who subsequently met some criteria for systemic lupus erythematosus, highlighting the complexity of cardiovascular involvement in systemic diseases.

PALAVRAS-CHAVE
Acidente vascular cerebral;
Enfarte agudo do miocárdio;

Síndroma antifosfolipídica associado a síndroma de Sneddon com envolvimento cardíaco: um diagnóstico desafiante

Resumo A síndroma de Sneddon é uma entidade rara caracterizada pela associação de doença cerebrovascular isquémica e *livedo reticularis*. Os autores apresentam um caso de acidente vascular cerebral isquémico e enfarte do miocárdio em doente com síndroma de Sneddon e

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* Corresponding author.
E-mail address: anacatarina.faustino@gmail.com (A. Faustino).

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Síndroma de Sneddon;
Síndroma antifosfolipídica;
Lúpus eritematoso sistémico;
Livedo reticularis

síndroma antifosfolipídica, que posteriormente reuniu alguns critérios de lúpus eritematoso sistémico, salientando a complexidade do envolvimento cardiovascular nas doenças sistémicas.

List of abbreviations

ANA	antinuclear antibodies
APA	antiphospholipid antibodies
APS	antiphospholipid syndrome
CT	computed tomography
ICD	implantable cardioverter-defibrillator
INR	international normalized ratio
LR	livedo reticularis
LV	left ventricular
LVEF	left ventricular ejection fraction
MI	myocardial infarction
RMCA	right middle cerebral artery
MRI	magnetic resonance imaging
MV	mitral valve
NYHA	New York Heart Association
RCA	right coronary artery
SLE	systemic lupus erythematosus
SS	Sneddon syndrome
TEE	transesophageal echocardiography
TTE	transthoracic echocardiography

Introduction

Sneddon syndrome (SS) is a rare clinical entity, with an estimated annual incidence of 4/1 000 000, that mainly affects women aged between 20 and 42 years, although cases have occasionally been reported in childhood and after the age of 65.[1] First described by Sneddon in 1965, the syndrome is characterized by the association of ischemic cerebrovascular disease and livedo reticularis (LR), caused by progressive occlusion of small- and medium-caliber arteries.[1–3] Organs and systems affected include the cardiovascular system, leading to hypertension, myocardial ischemia and valve damage, most frequently mitral but also aortic, with or without regurgitation.[1,2] There may also be involvement of other systems, particularly ocular (50–70%), gastrointestinal and renal (50–70%), as well as venous occlusions. No specific marker has been found for SS and its etiology is unclear,[1–8] but it has been suggested that its manifestations result from reduced blood flow due to increased viscosity, thrombotic and embolic events, arterial wall calcification, vasculitis and intimal hyperplasia, resulting in proliferation, recanalization and thrombosis of small- and medium-caliber arteries.[6]

Although SS was not initially diagnosed in the context of systemic diseases, the presence of antiphospholipid antibodies (APA) has been described in SS patients, with a highly variable prevalence in different series (0–85%). Criteria for systemic lupus erythematosus (SLE) and coagulation disorders including protein S deficiency and factor V Leiden have also been reported.[1–5] Three variants of the syndrome are now recognized: an idiopathic form, which is not accompanied by APA or SLE; a primary antiphospholipid syndrome (APS)-related form; and an SLE-associated form, with or without the presence of APA.[4]

A diagnosis of APS is based on the presence of at least one clinical criterion (documented recurrent arterial and/or venous thromboembolism or recurrent abortions) and one laboratory criterion (lupus anticoagulant or phospholipid-dependent antibodies, anticardiolipin and anti-β_2-glycoprotein-I, on at least two occasions at least 12 weeks apart).[9,10] Among its cardiovascular manifestations are thrombotic coronary disease, intracardiac thrombi and valve abnormalities (thickening, attached thrombotic masses, and non-infective vegetations). It is more common in women.

SLE is a systemic autoimmune disease, also more common in women, with a prevalence of 40/1 000 000.[11] Diagnosis is based on the presence of at least four of the 11 American College of Rheumatology criteria, recently updated to 17 criteria including biopsy-proven lupus nephritis together with antinuclear antibodies (ANA) or anti-double-stranded DNA antibodies.[11,12] It is associated with APS in 10–30% of cases, which raises the risk of acute coronary events 10-fold.[9] Ischemic coronary disease is one possible cardiac manifestation of SLE, caused by accelerated atherosclerosis, although it may also result from thrombosis in the context of APS due to embolization of non-infective vegetations or, less often, coronary arteritis. Other cardiac manifestations of SLE include pericarditis, pericardial effusion, myocarditis and Libman-Sacks endocarditis.[13]

Case report

A 39-year-old Caucasian man with a history of smoking (24 pack/years) and hypercholesterolemia and a first-degree relative (mother) with Sjögren syndrome, and no history of corticosteroid therapy, anabolic steroid use or drug abuse, was admitted to the neurology department on January 21, 2010 for ischemic stroke with left hemiparesis. Physical examination revealed extensive livedo reticularis (LR), predominantly on the trunk and upper limbs. No fever or