Neurofibromatosis type I and antiphospholipid syndrome

Neurofibromatosis tipo I y síndrome antifosfolípidos

To the Editor:

Neurofibromatosis type I (NF-1), an autosomal dominant disorder, can be considered as a pseudovasculitis.¹ Vascular abnormalities, mostly aneurysms or stenosis of the aortic, renal, and mesenteric circulation, are well-recognized in NF-1 patients². The relationship of NF-1 with the so-called antiphospholipid syndrome (APS)³ is enigmatic. We herein report an interesting case of NF-1 followed by typical features of APS.

The patient, a 38-year-old woman, had biopsy-proven NF-1 at the age of 16 years. In 2000, at the age of 28 years, she presented preeclampsia with subsequent fetal death. Six months after the obstetric event, an ischaemic cerebrovascular accident was diagnosed, resulting in mild hemiparesis and disartry. Antiphospholipid antibodies (APA) were not tested at that time. In January 2010 the patient had an acute myocardial infarction requiring angioplasty. IgG anticardiolipin (aCL) antibodies were present in significant levels (44 GPL). No lupus anticoagulant (LA) was found. Warfarin was therefore started. In May 2010, the patient, although anticoagulated (international normalized ratio 2.5) was re-admitted to our hospital due to a new coronary obstruction. A coronary by-pass surgery was recently performed. The IgG aCL test remained positive (37 GPL). A diagnosis of APS was therefore supported by obstetric morbidity, recurrent arterial thrombosis and laboratory criteria⁴.

The prevalence of APA in patients with NF-1 is unknown. Only one report on the association of NF-1 with APS has been described to date. The case, published in 1989, concerned a young woman with NF-1 and a history of recurrent fetal loss; the patient developed a cerebrovascular accident and was found to be positive for LA and aCL antibodies⁵.

If intrinsic vascular abnormalities proper of NF-1 predispose to APA and APS, it is plausible. Alternatively, NF-1 and APS might have coexisted by chance in these cases. Our patient with APS

developed progressive coronary disease despite of anticoagulation, which brings attention to a persistent endothelial damage perhaps linked to NF-1. The relationship of NF-1 with APS should be clarified in larger casuistics.

*Caroline Z. Xavier de Freitas¹, MD.,
Melissa Cláudia Bisi¹, MD.,
Leonardo Sinnott Silva², MD.,
Luiz Carlos Bodanese², MsC, PhD.,
Henrique Luiz Staub¹, MsC, PhD.*

¹Rheumatology Department

²Cardiology Department

Faculty of Medicine, São Lucas Hospital, Pontifical Catholic University of Rio Grande do Sul (PUCRS), Porto Alegre, Brazil

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Corresponding author:

Henrique Luiz Staub. Rheumatology Department, São Lucas Hospital, Pontifical Catholic University of Rio Grande do Sul (PUCRS).
Av. Ipiranga 6690/220. Porto Alegre, Brazil.
Telephone: (55) (51) 3320 5057 CEP: 90610-000
E-mail: reumatopucrs@gmail.com