SOCIEDADE BRASILEIRA DE REUMATOLOGIA

REVISTA BRASILEIRA DE REUMATOLOGIA

www.reumatologia.com.br

Original article

Familial autoimmunity and polyautoimmunity in 60 Brazilian Midwest patients with systemic sclerosis

Alex Magno Coelho Horimoto[a,b,c,*], *Aida Freitas do Carmo Silveira*[c], *Izaias Pereira da Costa*[c,d,e,f]

[a] *Universidade Federal de Mato Grosso do Sul (UFMS), Campo Grande, MS, Brazil*
[b] *Service of Rheumatology, Hospital Regional de Mato Grosso do Sul, Campo Grande, MS, Brazil*
[c] *Program of Medical Residence in Rheumatology, Universidade Federal de Mato Grosso do Sul (UFMS), Campo Grande, MS, Brazil*
[d] *Faculdade de Medicina, Universidade de São Paulo (USP), São Paulo, SP, Brazil*
[e] *Department of Internal Medicine, Faculdade de Medicina, Universidade Federal de Mato Grosso do Sul (UFMS), Campo Grande, MS, Brazil*
[f] *Service of Rheumatology, Hospital Universitário, Hospital Maria Aparecida Pedrossian, Universidade Federal de Mato Grosso do Sul (UFMS), Campo Grande, MS, Brazil*

ARTICLE INFO

Article history:
Received 26 December 2014
Accepted 1 September 2015
Available online 8 February 2016

Keywords:
Autoantibodies
Systemic sclerosis
Autoimmune disease
Polyautoimmunity
Familial autoimmunity

ABSTRACT

Introduction: Systemic sclerosis (SSc) is a connective tissue disease of unknown etiology, characterized by a triad of vascular injury, autoimmunity and tissue fibrosis. It is known that a positive family history is the greatest risk factor already identified for the development of SSc in a given individual. Preliminary observation of a high prevalence of polyautoimmunity and of familial autoimmunity in SSc patients support the idea that different autoimmune phenotypes may share common susceptibility variants.
Objectives: To describe the frequency of familial autoimmunity and polyautoimmunity in 60 SSc patients in the Midwest region of Brazil, as well as to report the main autoimmune diseases observed in this association of comorbidities.
Methods: A cross-sectional study with recruitment of 60 consecutive patients selected at the Rheumatology Department, University Hospital, Medicine School, Federal University of Mato Grosso do Sul (FMUFMS), as well as interviews of their relatives during the period from February 2013 to March 2014.
Results: A frequency of 43.3% of polyautoimmunity and of 51.7% of familial autoimmunity in SSc patients was found. Patients with the presence of polyautoimmunity and familial autoimmunity presented primarily the diffuse form of SSc, but this indicator did not reach statistical significance. The autoimmune diseases most frequently observed in polyautoimmunity patients were: Hashimoto's thyroiditis (53.8%), Sjögren's syndrome (38.5%), and inflammatory myopathy (11.5%). The main autoimmune diseases observed in SSc patients' relatives were: Hashimoto's thyroiditis (32.3%), rheumatoid arthritis (22.6%), and SLE (22.6%). The presence of more than one autoimmune disease in SSc patients did not correlate with disease severity or activity.

* *Corresponding author.*
E-mail: clinicaactivite@gmail.com (A.M.C. Horimoto).
http://dx.doi.org/10.1016/j.rbre.2016.01.003

Conclusions: From the high prevalence of coexisting autoimmune diseases found in SSc patients, we stress the importance of the concept of shared autoimmunity, in order to promote a continued vigilance and promptly diagnose other possible autoimmune disease in patients, or in their kin.

Autoimunidade familial e poliautoimunidade em 60 pacientes portadores de esclerose sistêmica da região Centro-Oeste do Brasil

Palavras-chave:
Autoanticorpos
Esclerose sistêmica
Doença autoimune
Poliautoimunidade
Autoimunidade familiar

RESUMO

Introdução: A esclerose sistêmica (ES) é uma enfermidade do tecido conjuntivo de etiologia desconhecida, caracterizada pela tríade de injúria vascular, autoimunidade e fibrose tecidual. Sabe-se que uma história familiar positiva representa o maior fator de risco já identificado para o desenvolvimento da ES em um determinado indivíduo. Observação prévia de alta prevalência de poliautoimunidade e de autoimunidade familiar em pacientes com ES, reforça a ideia de que fenótipos autoimunes distintos podem dividir variantes comuns de susceptibilidade.

Objetivos: Descrever a frequência de autoimunidade familiar e de poliautoimunidade em 60 pacientes com ES da região Centro Oeste do Brasil, bem como relatar as principais doenças autoimunes observadas nesta associação de comorbidades.

Métodos: Realizou-se um estudo transversal com recrutamento de 60 pacientes consecutivos, selecionados no Serviço de Reumatologia do Hospital Universitário da Faculdade de Medicina da Universidade Federal de Mato Grosso do Sul (FMUFMS), bem como entrevista de seus parentes, durante o período de fevereiro de 2013 a março de 2014.

Resultados: Foi encontrada uma frequência de 43,3% de poliautoimunidade e de 51,7% de autoimunidade familiar nos pacientes com ES. Os pacientes com presença de poliautoimunidade e de autoimunidade familiar eram principalmente da forma difusa de ES, porém este índice não atingiu significância estatística. As doenças autoimunes mais comumente observadas nos pacientes com poliautoimunidade foram: tireoidite de Hashimoto (53,8%), síndrome de Sjögren (38,5%) e miopatia inflamatória (11,5%). As principais doenças autoimunes observadas nos parentes dos pacientes com ES foram: tireoidite de Hashimoto (32,3%), artrite reumatóide (22,6%) e LES (22,6%). A presença de mais de uma enfermidade autoimune em pacientes com ES não se correlacionou com maior gravidade ou atividade da doença.

Conclusões: A partir da alta prevalência encontrada de doenças autoimunes coexistentes em pacientes com ES, salientamos a importância do conceito de autoimunidade compartilhada, de forma a promover uma vigilância constante e diagnosticar prontamente uma possível outra doença autoimune nos pacientes ou em seus familiares.

Introduction

Systemic sclerosis (SSc) is a disease of the connective tissue with an autoimmune character, and with extreme heterogeneity in its clinical presentation, with involvement of multiple systems and following a varied and unpredictable course.[1] Its etiology remains unknown, and a multifactorial cause is suggested; possibly SSc is triggered by environmental factors in a genetically predisposed individual.[2]

The hallmark of SSc is the occurrence of microvasculopathy, fibroblast activation and an excessive production of collagen.[3] This is a unique condition, because it displays features of three distinct pathophysiological processes, the so-called triad of vascular injury, autoimmunity (cellular and humoral), and tissue fibrosis, leading to cutaneous involvement, besides affecting multiple internal organs, for instance, lungs, heart and gastrointestinal tract, as well as musculoskeletal manifestations.[3,4]

The genetic component of autoimmune diseases is represented by the increased risk of developing SSc in twin brothers of affected individuals.[3,4] Basically, SSc is not a genetic disorder, but there is consensus that, actually, the disease has a genetic component based on reports of monozygotic twins with a propensity to SSc.[5]

In 1953, Rees and Bennett[6] described the first case of localized scleroderma in a father and his daughter. Later, several reports of familial scleroderma in different populations and family relationships were published.[2,5,7] Moreover, an association between HLA and SSc has been described.[5,8,9]

A positive family history is the greatest risk factor ever identified for the development of SSc in a given individual.[8,10]