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Case report

# Coexisting systemic lupus erythematosus and sickle cell disease: Case report and literature review

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ABSTRACT

*Objective:* To report a case of coexisting systemic lupus erythematosus (SLE) and sickle cell disease (SCD) with a review of the literature on the topic.
*Methodology:* Case report and literature review of the association between SLE and SCD through scientific articles in health sciences databases, such as LILACS, MEDLINE/Pubmed and Scielo, until May 2012. Descriptors used: 1. Sickle cell anemia; 2. Sickle cell disease; 3. Systemic lupus erythematosus; 4. Hemoglobinopathies.
*Results:* The authors describe an association between SLE and SS hemoglobinopathy in an eight-year-old female patient presentingarticular, hematologic and neuropsychiatric manifestations during clinical evolution. Forty-five cases of association between SLE and SCD are described in literature, mostly adults (62.2%), women (78%) and with the SS phenotype in 78% of the cases, and diverse clinical manifestations. Compared with our patient, articular, hematologic and neuropsychiatric manifestations were present in 76%, 36% and 27% of the cases, respectively.
*Conclusion:* SLE and SCD are chronic diseases that have several clinical and laboratory findings in common, meaning difficult diagnosis and difficulty in finding the correct treatment. Although the association between these diseases is not common, it is described in literature, so it is imperative that physicians who treat such diseases be alert to this possibility.


## Coexistência de lúpus eritematoso sistêmico e doença falciforme: relato de caso e revisão da literatura

*Palavras-chave:*
Anemia falciforme
Doença falciforme

RESUMO

*Objetivo:* relatar um caso de coexistência de lúpus eritematoso sistêmico (LES) e doença falciforme (DF) com revisão da literatura sobre o tema.
*Metodologia:* relato de caso e pesquisa da associação entre LES e DF na literatura, através de artigos científicos nas bases de dados de ciências da saúde, como LILACS, MEDLINE/Pubmed

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Lúpus eritematoso sistêmico
Hemoglobinopatias

e Scielo, até maio de 2012. Descritores utilizados: 1. anemia falciforme; 2. doença falciforme; 3. lúpus eritematoso sistêmico; 4. hemoglobinopatias.

*Resultados:* os autores descrevem a associação de LES e hemoglobinopatia SS em paciente do sexo feminino, oito anos, apresentando manifestações articulares, hematológicas e neuropsiquiátricas durante a sua evolução clínica. Na literatura são descritos 45 casos de associação entre LES e DF, sendo a maioria em mulheres (78%) adultas (62,2%), apresentando fenótipo SS em 78% dos casos e com manifestações clínicas variadas. Comparando com a nossa paciente, manifestações articulares, hematológicas e neuropsiquiátricas, estiveram presentes em 76%, 36% e 27% dos casos, respectivamente.

*Conclusões:* LES e DF são doenças crônicas que apresentam diversos achados clínicos e laboratoriais em comum, implicando em dificuldades diagnósticas e na correta condução terapêutica dessas doenças. A associação entre essas enfermidades não é comum, mas está descrita na literatura, por isso é importante que médicos que cuidam dessas enfermidades estejam atentos para tal possibilidade.



## Introduction

Systemic lupus erythematosus (SLE) is a multisystem autoimmune disease with an incidence of 1.9 to 5.6 per 100,000 inhabitants,[1] while sickle cell disease (SCD) is one of the most common hereditary diseases, affecting mainly black individuals.[2] SCD is characterized by a mutation in the hemoglobin beta chain with formation of abnormal hemoglobin (HbS), responsible for microcirculation obstruction, ischemia, tissue necrosis and systemic organ dysfunction.[2]

The coexistence of SCD and SLE is rarely described in literature, even in predominantly black populations, in which the prevalence of both conditions is higher.[3] Wilson et al. were the first to report this association.[4] Perhaps the defective activation of the alternative complement pathway in sickle cell patients and the increased risk of infections caused by encapsulated bacteria predispose this group to develop an autoimmune disease.[5]

The clinical features of SCD and SLE may show similar manifestations, such as the presence of fever, anemia, articular, renal, neurological and cardiopulmonary involvements and, consequently, diagnostic difficulties. Sickle cell patients showing atypical symptoms or refractory response to conventional treatment should be investigated for the possibility of coexistence of diseases.[6]

In view of the uncommon occurrence of this association, the authors describe an early childhood case and review the previously reported cases until May 2012.

## Methodology

Case report and literature review of the association between SLE and SCD through scientific articles in health sciences databases, such as LILACS, MEDLINE/Pubmed and Scielo until May 2012. Descriptors used: 1. Sickle cell anemia; 2. Sickle cell disease; 3. Systemic lupus erythematosus; 4. Hemoglobinopathies.

## Case report

Female patient diagnosed with SS hemoglobinopathy since birth, showing recurring, mild painful vaso-occlusive crises responsive to traditional hydration and analgesia. She has never received blood transfusions. At the age of nine, pain symptoms intensified, mainly characterized by repeated crises of acute and asymmetric polyarthritis of the knees, wrists, elbows and ankles, initially attributed to SCD. The clinical picture evolved with the development of photosensitivity, asthenia and intermittent fever, with an episode of generalized tonic-clonic seizure associated with transient left hemiparesis. Laboratory tests: hemoglobin, 7.9 g/dL; hematocrit, 25%; WBC, 12.000/$mm^3$; platelet count, 374.000 $mm^3$ (160-400.000); hemoglobin electrophoresis HbS 98.7% and $HbA_2$ 1.26%; positive antinuclear antibody (ANA) 1:320 (homogeneous pattern); positive anti-dsDNA antibody; erythrocyte sedimentation rate, 68 mm (<20); C-reactive protein, 71 units (< 6); rheumatoid factor, anti-Sm, anti-SSA, anti-SSB, were all negative as were viral serology. Serum $C_3$, $C_4$ and $C_{H50}$ levels were normal as were renal and liverfunctions. Cranial magnetic resonance imaging revealed an area of hypoperfusion in the right temporal lobe. The study of the cerebrospinal fluid was normal. The patient had a sibling with sickle cell anemia who died at the age of two from acute myocardial infarction, and has two maternal aunts diagnosed with cutaneous lupus. She was diagnosed with SLE according to American College of Rheumatology (ACR) criteria,[7] and the possibility of involvement of the central nervous system (CNS) by both diseases (SLE and SCD) was discussed. The patient received prednisone 2 mg/kg/day, hydroxychloroquine 5 mg/kg/day and blood transfusions in order to reduce HbS < 30%. Due to the possibility that the neurological manifestation was secondary to SCD we chose not to associate another immunosuppressive agent. There was significant reduction in joint pain, no progression of the neurological condition and improvement of hematological indices (HbS 60%; Hb 8.5 g/dL). Patient remains asymptomatic, taking hydroxychloroquine and prednisone 5 mg/day.