



Descriptive Study of Autoimmune and Systemic Diseases in Adults Patients at Niannankoro Fomba Regional Hospital in Segou From 2019 - 2023

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ABSTRACT

Introduction: Connective tissue diseases encompass a group of systemic autoimmune pathologies such as rheumatoid arthritis, systemic lupus erythematosus (SLE), inflammatory myopathies, systemic scleroderma, Gougerot-Sjögren syndrome and mixed connectivites. This study focused on six autoimmune and systemic diseases (AISDs): rheumatoid arthritis, SLE, Gougerot-Sjögren syndrome, anti-synthetase syndrome, SHARP syndrome and scleroderma, excluding organ-specific autoimmune diseases. These conditions are clinically heterogeneous, with varied manifestations depending on the patient.

Material and methods: A descriptive cross-sectional study was conducted from January 2019 to December 2023 in the internal medicine department of the Nianankoro Fomba regional hospital in Segou, Mali. All consenting inpatients and outpatients meeting diagnostic criteria for connectivitis (ACR, Kasukawa for SHARP) were included. Clinical data (age, sex, origin, socioeconomic level, diagnostic delay, joint and organ signs, comorbidities) and paraclinical data (autoantibodies, biological and imaging tests) were collected from medical records. Analysis was performed with Epi Info 7.

Results: Among 1,920 patients seen during the study period, 28 were diagnosed with MAIS, representing a hospital prevalence of 0.15%. The mean age was 47.54 ± 18.73 years (extremes: 18-87 years), with a majority of women (64.29%, sex ratio = 0.56). The mean time to consultation was 50.52 ± 33.84 months. Articular manifestations were the most frequent (96%), followed by mucocutaneous involvement (42.86%).

Discussion: MAIS remains rare and poorly recognized in sub-Saharan Africa, due to reliance on traditional practitioners and misdiagnosis. Our results confirm a low prevalence, comparable to other African countries. Rheumatoid arthritis is the most common, affecting mostly middle-aged women.

Conclusion: MAIS represent a growing public health challenge in Black Africa. Early detection and prompt management are crucial to improve patient prognosis.

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Introduction

Systemic diseases (also known as collagenoses or connectivites) are defined as a group of diseases associated with immunological and inflammatory damage to connective tissue and collagen. They are characterized by the involvement of several organs or tissues.

The term connectivitis has become established in common usage, and is usually used to describe rheumatoid arthritis, systemic lupus erythematosus (SLE), primary inflammatory myopathies (dermatomyositis, polymyositis and inclusion myositis), systemic scleroderma, primary Gougerot-Sjögren's syndrome and mixed connectivites [1]. Systemic autoimmune diseases (SADs) are increasingly described in Africa, and their prevalence is rising sharply [2-5].

The main reasons for this are a certain improvement in diagnostic tools and better knowledge of these conditions, despite a still insufficient number of specialists [6-9]. The clinical presentation of MAIS remains polymorphous, and the initial mode of revelation and reasons for admission to hospital services are diverse and varied [4,5,7,10-13].

Nonetheless, cutaneous and mucosal manifestations are among the most frequent of these conditions, and often lead to the hypothesis being put forward, thereby shortening diagnosis times. Autoimmune diseases, of which rheumatoid arthritis and autoimmune thyroiditis are the most common, affect around 5% of the population.

Autoimmune diseases may be associated in the same patient, or a combination of two or more connectivities may be observed, known as mixed connectivities or SHARP syndrome. Sjögren's syndrome (35.5%), rheumatoid arthritis (7.5%), antiphospholipid syndrome (7.5%), systemic scleroderma (5.9%), Biermer's disease (4.9%), Hashimoto's thyroiditis (3.8%) and dermatomyositis (0.5%) were associated with SLE in 52.2% of cases. Significant associations were observed. SLE associated with other IADs was associated with the presence of central neurological involvement ($p = 0.02$), thrombosis ($p = 0.02$), pulmonary hypertension ($p = 0.05$) and malar rash ($p = 0.03$). Significant associations were observed between the presence of MAI and the occurrence of central neurological impairment ($p = 0.02$), thrombosis ($p = 0.02$), pulmonary hypertension ($p = 0.05$) and malar rash ($p = 0.03$). The presence of scleroderma was significantly associated with the presence of leukopenia ($p = 0.04$), pulmonary involvement ($p = 0.009$) and corticosteroid prescription ($p = 0.03$). SAPL was associated with the occurrence of thrombosis ($p < 0.001$), renal impairment ($p = 0.04$) and lymphopenia ($p = 0.05$) [14].

An autoimmune disease can be defined as the activation of the patient's immune system against its own antigens. Activated self-reactive T and B lymphocytes will lead to the destruction of the individual's own constituents (Figure 1) via several

mechanisms, either directly by cytotoxic T lymphocytes or by the deposition of antibodies activating the complement system, or indirectly via the activation of other cells involved in the inflammatory response, as in the renal involvement of lupus.

If the autoantigens against which the immune reaction is directed are expressed in a single organ, only one organ will be affected (e.g. thyroiditis, diabetes). If these autoantigens are expressed systemically, the autoimmune disease will be systemic. Indeed, despite the ubiquitous expression of self-antigens, clinical involvement is sometimes relatively localized to certain organs in certain pathologies (e.g. ribonucleoprotein antigens SSA and SSB in Sjögren's syndrome, or tRNA synthetases in polymyositis). Problems of antigen accessibility and/or migration of immune system cells, depending on the different adhesion molecules expressed on their surface, probably explain these preferential clinical attacks [15-19].

Similarly, the expression of autoantigens may vary according to the development of the individual, thus explaining the variability of these autoimmune disorders. For example, anti-SSA antibodies bind to the cardiac conduction system in the fetus, leading to complete atrioventricular block, whereas these antibodies are not toxic to the adult heart [17,19]. Conversely, anti-desmoglein antibodies cause pemphigus in adults but not in newborns, as one of the two types of desmoglein present in newborn skin is not recognized by these antibodies [18,19].

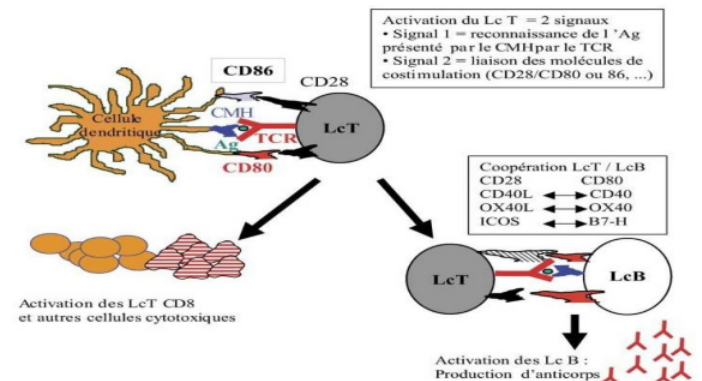


Figure 1: Reminder of the immune response (B. Bonnotte. Rev Med Int 2004; 25: 648-658).

Our study involved six autoimmune and systemic diseases (rheumatoid arthritis, SLE, Gougerot Sjögren syndrome, anti-synthetase syndrome, SHARP syndrome, scleroderma).

Patients and Methods

Methodology

Type and Setting of Study

This was a descriptive cross-sectional study carried out in the Internal Medicine Department of the Nianakoro Fomba Regional Hospital in Ségou (HNF/S), Mali.

Study Period

The study was conducted over a 04-year period (from January 2019 to December 2023).

Inclusion Criteria

All outpatients and inpatients with MAIS were included.

Non-Inclusion Criteria

All patients whose consent was not obtained or those discharged against medical advice were excluded from the study.

Diagnostic Criteria

The diagnostic criteria were those of the ACR [20]; for SHARP syndrome, the Kasukawa criteria were used.

Data Collection

Data collection was carried out on the collated consultation records of patients followed for autoimmune and systemic diseases; the following variables were collected.

Clinical Variables

Age, gender, place of residence, socioeconomic level, diagnostic delay, joint pain, swelling, joint deformities, muscle pain, lesions (skin, scalp, hands), extra-articular involvement, associated pathologies.

Paraclinical Variables

We asked for anti-NA antibodies (anti-Scl70, anti-Jo1, anti-SSA, SSB, anti-Sm, anti-U1-RNP) and anti-nuclear antibodies, anti-native DNA antibodies in all patients with suspected connective tissue diseases, anti-citrullinated cyclic peptide autoantibodies (anti-CCP antibodies) and X-rays of hands and wrists in cases with suspected rheumatoid arthritis. Other tests, depending on the type of MAIS and associated complications, included: blood count, sedimentation rate (ESR), c-reactive protein (CRP), 24-hour proteinuria, muscle enzymes (CPK), creatinineemia, blood protein electrophoresis;

Imaging

Doppler echocardiography, thoracic CT scan.

Data Entry and Analysis

Data entry was carried out using Word, and data processing was carried out using Epi info 7.

Results

During our study, we consulted 1920 patients, 28 of whom had MAIS, representing a hospital prevalence of 0.15%.

Table 1: Patient age distribution.

Age des patients (années)
26, 42, 45, 71, 80, 65, 40, 59, 66, 60, 41, 40, 66, 25, 40, 68, 87, 27, 29, 57, 36, 59, 66, 37, 25, 32, 18, 24

Modal ages were 40 and 66, mean age 47.54 ± 18.73, median age 41.5 with extremes 18-87, age range (Max-Min) = 69.

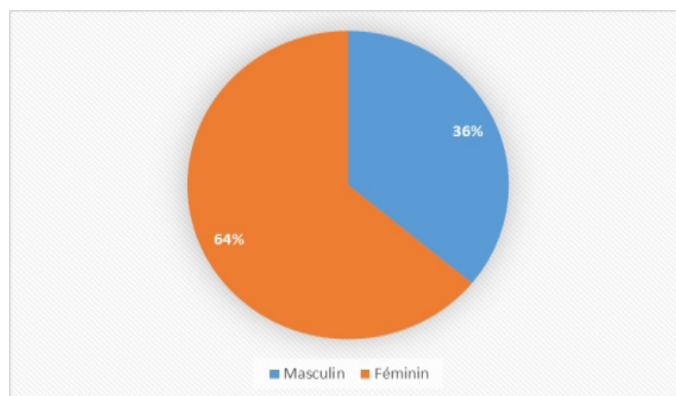


Figure 2: Distribution of patients by gender.

Females accounted for the majority of the 18 cases (64%), with a sex ratio of 0.56. The mean duration of disease progression before consultation was 50.52 months ± 33.84 months.

Reasons for consultation: polyarthralgia in 27 cases (96%), polyarthrititis in 14 cases (50%), skin rashes in 12 cases (42.86%), joint deformities in 9 cases (32.14%), muscle pain in 15 cases (53.57%), cough in 8 cases (28.57%), and altered general condition (AEG) in 12 cases (42.86%).

Immunological tests were prescribed in all patients and were positive in 27 (96%), the negative case was seronegative rheumatoid arthritis based on the presence of right carpalitis and inflammatory polyarthralgias apart from other plausible causes.

The diagnoses of MAIS retained were Systemic Lupus Erythematosus (SLE) in 8 cases (28.57%), Scleroderma in 2 cases (7.14%), Anti-Synthetase Syndrome in 1 case (3.57%), SHARP Syndrome in 1 case (3.57%), Gougerot Sjögren Syndrome in 1 case (3.57%), and Rheumatoid Arthritis in 15 cases (53.57%).

Table 2: Distribution of patients according to MAIS.

The main BUTs	Workforce N (%)
The LES	8 (28,57%)
Scleroderma	2 (7,14%)
Anti-synthetase syndrome	1 (3,57%)
SHARP syndrome	1 (3,57%)
Gougerot Sjögren's syndrome	1 (3,57%)
Rheumatoid arthritis (RA)	15 (53,57%)

The most common MAIS was RA in 15 cases (53.57%), followed by SLE in 8 cases (28.57%).

These pathologies were complicated by anemia in 11 cases (39.29%), acute renal failure in 4 cases (14.28%), inflammatory bronchopathy with bronchiectasis and nephrotic syndrome (NS) in 3 cases (10.71%); all cases of NS were complications of SLE.

Medical history of hypertension and diabetes were associated in 7 cases (25%) and 2 cases (7.14%) respectively.



Figure 3: Erythematous lupus lesions with discoid lupus lesions on the thorax and malar erythema on black skin (LES).



Figure 4: a. (Scleroderma patient); b. (PR patient, more severe bilateral carpalitis on the right).

Discussion

MAIS are rare, and their prevalence is underestimated due to under-diagnosis caused by patients being misdirected to non-specialized centers, and the use of traditional therapists. According to most of the patients interviewed, these illnesses are due to bad spells, hence the fairly long diagnosis time.

In our study we found a hospital prevalence of 0.15% for 28 cases diagnosed in 4 cases. Mijiyawa et al. (Togo) and Ouédraogo et al. (Burkina Faso) found 55 cases in 16 years and 42 cases in 6 years [6,7] in Dermatology and Rheumatology departments respectively.

Hospital prevalence remains low in most black African studies: Benin 1.3%, Togo 0.2% [4,6], which corroborates our low prevalence.

In our study, joint manifestations were the most common, with 96% of cases, followed by mucocutaneous manifestations with 42.86% of cases. This was also observed in the Burkina study, with 88% of cases [7]. Rheumatoid arthritis is usually the most common autoimmune and systemic disease, as observed by Diatou G. Dia1 et al. in Saint-Louis, Senegal [21]. The mean age was 47.54 ± 18.73 years with extremes of 18-87 years, about the same as that of Diatou G. Dia et al. with 40.12 years with extremes ranging from 7 to 69 years [21]. In most studies, more women than men were affected. The late consultation delay of $50.52 \text{ months} \pm 33.84 \text{ months}$ is also a feature of African series [21-23]. According to a systematic review with meta-analysis, the prevalence of associated GSS varies between 5 and 22% [24]; in our study, we found 3.57% or 1 case.

Conclusion

Autoimmune and systemic diseases are increasingly being studied in sub-Saharan Africa and represent a challenge for practitioners.

However, the prevalence of these MAIS remains underestimated, given the myth surrounding these diseases, and the long delay in diagnosis due to the upstream intervention of traditherapists before specialized medical consultation.

In addition, collaboration between different departments is necessary to shorten diagnosis and management times, and thus reduce the risk of patient attrition.

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