

## Adult-onset still's disease: A case report and diagnostic insights in Rwanda

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### ABSTRACT

**INTRODUCTION:** Adult-Onset Still's Disease (AOSD) is a rare, multi-systemic autoinflammatory disorder with a global incidence of 0.62 cases per 100,000 people annually. While it exhibits a bimodal age distribution affecting individuals between 15-25 years and 36-46 years, specific prevalence data in Rwanda remains limited. The disease's complex clinical presentation and unclear etiology often result in delayed diagnosis. This case study analysed and documented the clinical presentation, therapeutic management, and the diagnostic challenges of Adult-Onset Still's Disease.

**CASE PRESENTATION:** A 27-year-old female presented with a two-week history of escalating fever, multiple joint pains, and swelling. The patient met multiple Yamaguchi criteria, leading to an AOSD diagnosis. Treatment with high-dose Prednisolone and Methotrexate, supplemented with Folic acid and Omeprazole, resulted in clinical improvement. Diagnosis was challenging due to the need for extensive exclusion of infectious, malignant, and autoimmune conditions, highlighting the complexity of identifying rare rheumatological diseases in resource-limited settings.

**CONCLUSION:** This case study highlights the importance of recognizing AOSD's clinical patterns for timely diagnosis and appropriate therapeutic intervention. The successful treatment outcome demonstrates the effectiveness of standard therapeutic protocols in managing AOSD, even in resource-limited settings.

**Keywords:** Adult-Onset Still's Disease, Yamaguchi Criteria, Autoinflammatory Disorder, Clinical Presentation, Insight

### INTRODUCTION

Adult-Onset Still's Disease (AOSD) represents a rare but significant systemic inflammatory disorder of unknown etiology, characterized by a triad of persistent high fever, arthritis, and an

evanescent salmon-colored rash [1,2]. Still's disease was named after the description, in 1897, of 22 children affected by systemic onset juvenile idiopathic arthritis (sJIA), by George Still [2]. It had long been accepted that these forms of arthritis are only diagnosed in children, but almost 75

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years later, Eric Bywaters described the first 14 cases of Still's disease syndrome in female patients between 17 and 35 years with the same symptoms [3]. His report indicated that Still's disease is not an age-related version of polyarthritis. In two recent studies, the prevalence was reported to be 3.9 per 100,000 individuals in Japan and 6.77 per 100,000 individuals in Turkey [4].

Adult-onset Still's disease represents an uncommon inflammatory condition affecting approximately 0.16-0.4 individuals per 100,000 in the adult population, characterized by a distinctive dual-peak age pattern occurring during young adulthood (15-25 years) and middle age (36-45 years), with current theories suggesting its etiology stems from underlying immune regulatory abnormalities [5]. Comprehensive statistical information regarding the prevalence and distribution patterns of adult-onset Still's disease remains notably insufficient in current medical literature [3,4].

AOSD usually presents with high spiking fever, polyarthritis, evanescent salmon-pink rash, sore throat, and lymphadenopathy [3,6,7]. Polyarthritis determines joint pain, the most common symptom of AOSD, and mainly involves wrists, knees, and ankles [3,6]. Fever occurs in 60–100% of the patients, and it typically spikes once or twice daily, with the highest temperatures (>39 °C) occurring in the evening [6]. AOSD is characterized by fever, arthralgia, rash, leukocytosis, sore throat, lymphadenopathy and/or splenomegaly, liver dysfunction, and the absence of rheumatoid factor and antinuclear antibodies [3,7]. Blood analysis typically reveals generalized inflammatory markers characteristic of the condition, including elevated acute phase reactants such as C-reactive protein and sedimentation rate, alongside increased white blood cell counts with neutrophil predominance [6]. As in many other inflammatory diseases, in AOSD patients, it is possible to observe anemia, thrombocytosis, and elevated ferritin levels; however, although a 5-fold increase in the ferritin level is suggestive of AOSD, similar elevated levels can also be found in the context of infections, neoplastic conditions, and storage diseases [6].

The pathogenesis of AOSD remains poorly understood, though factors like genetic predisposition and triggers such as infections are thought to contribute. This represents the first documented case of Adult-Onset Still's Disease in

Rwanda, as no prior reports exist in the literature. In the current case report, we examined a 27-year-old female who was diagnosed with AOSD.

This case report focuses on the diagnostic process guided by widely recognized criteria and the subsequent management of the disease using a combination of conventional and disease-modifying antirheumatic drugs.

## CASE PRESENTATION

The patient, a 27-year-old female university student, presented with a two-week history of escalating fever, multiple joint pains, and swelling. Her medical history was notable for recurrent symptoms similar to those of AOSD, spanning over three years, with minimal relief provided by standard non-steroid anti-inflammatory drugs (NSAIDs) and corticosteroid treatments. She had a known congenital immune suppressive condition from her mother, who has the same disease, which was well-managed and controlled with medications, exhibiting an undetectable viral load and a CD4 count of 1130 cells/ $\mu$ L. She denies any autoimmune disease in her family. Other vital signs, the blood pressure is 97.62 mmHg, pulse rate of 108 beats/minute, respiratory rate of 22cy/min, oxygen saturation of 97% on room air, and temperature of 39.5°C.

Upon examination, the patient was acutely ill, displaying symptoms typical of AOSD, including high, recurrent fevers exceeding 39°C, a transient maculopapular rash primarily on her trunk and limbs, and severe polyarthritis. Laboratory tests revealed elevated inflammatory markers (CRP:191.2mg/dl, ESR: 117mm/h), hyperferritinemia of 469ng/ml, and leukocytosis (WBC: 18x10<sup>9</sup>), with anemia (HB: 8.64gr/dl) also present. A chest X-ray identified focal lung fibrosis. Other workup that included blood cultures, antinuclear antibody (ANA), Anti-cyclic citrullinated peptide (Anti CCP), rheumatoid factor (RF) was negative, and tumor markers tested (Alpha fetoprotein, CA125, CEA, CA19.9) were all in normal range.

The patient's clinical presentation was noted to satisfy multiple Yamaguchi criteria, and she was diagnosed with AOSD. The treatment initiated included high-dose Prednisolone (1mg/kg daily) and Methotrexate 15mg weekly, supplemented with Folic acid 5mg weekly and Omeprazole 20mg

twice daily, aimed at managing inflammation and preventing gastrointestinal side effects. This regimen led to significant improvements in her symptoms, demonstrating the efficacy of integrating disease-modifying agents in the management of AOSD.

The diagnosis of AOSD in this case presented significant challenges at CHUK, as it required extensive exclusion of other conditions with similar clinical presentations, including infectious diseases, malignancies, and other rheumatological disorders. The constellation of high fever, arthritis, rash, and elevated inflammatory markers initially raised suspicion for various differential diagnoses such as sepsis, lymphoma, systemic lupus erythematosus, and rheumatoid arthritis, necessitating a comprehensive laboratory workup including blood cultures, autoantibody panels, and tumor markers to rule out these conditions. The absence of specific diagnostic tests for AOSD, combined with its reliance on clinical criteria and exclusion of other diseases, made the diagnostic process particularly complex. Furthermore, the intermittent nature of symptoms and the requirement for prolonged observation to establish the characteristic fever pattern complicated the diagnostic process, highlighting the need for increased clinical awareness and improved access to advanced diagnostic tools for rare rheumatological conditions in resource-limited settings like Rwanda.

This case study was conducted in accordance with the ethical principles of the Declaration of Helsinki and received approval from the Institutional Review Board of the Centre Hospitalier Universitaire de Kigali (CHUK). Informed consent for participation and for publication of this case report was obtained from the patient. The patient was informed about the purpose of the study, the potential publication of their anonymized medical information, and their right to withdraw consent at any time without affecting their medical care. All patient data were anonymized to protect confidentiality in compliance with institutional and international research ethics guidelines.

This study was conducted in accordance with the ethical standards of our institutional review board and with the 1964 Helsinki Declaration and its later amendments.

## DISCUSSION

AOSD presents a complex diagnostic and therapeutic challenge due to its multifaceted and often elusive symptomatology. While not fully understood, the pathophysiology of AOSD is believed to involve a confluence of genetic predisposition and environmental triggers that activate inflammatory pathways. This autoinflammatory disorder is characterized by high spiking fevers, arthritis, and a distinctive salmon-pink rash, among other systemic symptoms. Recent research suggests a role for innate immune system dysregulation in AOSD, highlighting the involvement of pro-inflammatory cytokines such as IL-1, IL-6, and tumor necrotizing factor alpha (TNF- $\alpha$ ), which are now targets for therapeutic intervention [8]. The diagnostic process for AOSD is complicated by its nonspecific symptoms, which mimic those of other inflammatory and infectious diseases. This case employed the Yamaguchi and Fautrel criteria, which are pivotal in distinguishing AOSD from other conditions [7]. Along with ferritin and glycosylated ferritin values, the Yamaguchi criteria [7] require the presence of five features, with at least two being major diagnostic criteria. The four major Yamaguchi criteria are elevated fever lasting at least one week, arthralgias or arthritis, nonpruritic macular or maculopapular skin rash that is salmon-colored during febrile episodes, and leukocytosis with at least 80 percent granulocytes [6]. The minor Yamaguchi criteria include sore throat, lymphadenopathy, hepatomegaly or splenomegaly, abnormal liver function studies, and negative tests for antinuclear antibody (ANA) and rheumatoid factor (RF) [6]. The patient met these criteria, with the notable absence of infections and malignancies, which are key exclusion factors in the Yamaguchi criteria.

Diagnostically, AOSD is a challenge due to the lack of specific biomarkers and the necessity to exclude a wide array of mimicking conditions like infections, malignancies, and other rheumatologic diseases. The Yamaguchi criteria, despite their high specificity, lack sensitivity and require the absence of serological markers, which complicates the diagnostic process [9]. Our case employed both Yamaguchi and Fautrel's criteria, emphasizing the importance of a thorough diagnostic evaluation that includes advanced imaging and comprehensive laboratory tests to rule out other

potential causes of the symptoms. A chest X-ray identified focal lung fibrosis. A finding consistent with the pulmonary involvement occasionally seen in AOSD [6].

The management of AOSD in this patient was guided by the severity of her presentation, focusing on controlling inflammation and preventing long-term organ damage. The initial treatment regimen included high-dose corticosteroids and Methotrexate, reflecting the standard approach of using NSAIDs and glucocorticoids as first-line treatments, followed by Disease-modifying antirheumatic drugs (DMARDs) for more severe or refractory cases [4]. This strategy aligns with current recommendations that advocate for personalized treatment plans based on disease severity, patient comorbidities, and response to initial therapy.

It is also crucial to address the presence of atypical manifestations such as lung fibrosis, an uncommon feature in AOSD that may suggest an overlap with other systemic diseases or a severe disease phenotype. Lung involvement in AOSD, typically presenting as pleuritis or pleural effusion, rarely includes fibrosis, and its presence necessitates a careful evaluation to exclude other etiologies such as interstitial lung disease associated with connective tissue diseases [1].

The long-term management of AOSD requires not only controlling acute flares but also monitoring for potential complications, including macrophage activation syndrome and long-term organ damage. The evolving landscape of biologic therapies offers new hope for patients with refractory disease, with agents targeting specific cytokines showing promise in controlling disease activity and improving quality of life.

## CONCLUSION

This case report highlights the intricate interplay between the clinical presentation and the underlying pathophysiological mechanisms in AOSD, underscoring the need for a comprehensive and flexible approach to diagnosis and management. As we continue to unravel the complexities of AOSD, ongoing research and clinical trials will be pivotal in refining our understanding and treatment of this challenging disease. This case report contributes to the broader discourse

on AOSD, advocating for an integrated approach that leverages both clinical insights and emerging scientific evidence to optimize patient outcomes.

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