

# Sarcoidosis

Sarcoidosis is a multisystem inflammatory disease characterized by the formation of non-caseating granulomas, which are clusters of immune cells that form in response to chronic inflammation. Although the precise cause of sarcoidosis remains unclear, it is believed to arise from an interplay between genetic susceptibility and environmental exposures. Sarcoidosis can affect virtually any organ system, but it most commonly involves the lungs, lymph nodes, skin, eyes, liver, and less frequently, the heart, nervous system, and kidneys. The disease typically manifests in individuals between the ages of 20 and 40, with a higher prevalence in black populations and individuals of Northern European descent.

## Pathophysiology

The underlying mechanism of sarcoidosis involves the accumulation of T lymphocytes, macrophages, and other immune cells in affected tissues, leading to granuloma formation. Granulomas are structured aggregates of immune cells that attempt to contain foreign antigens or pathogens, but in sarcoidosis, they form in the absence of any clear infectious cause. These granulomas can cause tissue damage and dysfunction, which varies depending on the organs affected. The pathogenesis likely involves an exaggerated immune response triggered by environmental factors such as infections, dust, or chemicals in genetically predisposed individuals.

## Clinical Manifestations

Sarcoidosis is known for its heterogeneity in clinical presentation, which depends on the organs involved. The most common manifestations include:

### ➤ ***Pulmonary Involvement***

The lungs are the most frequently affected organ, with granulomas typically forming in the lungs' interstitial tissue. This leads to symptoms such as chronic cough, shortness of breath, wheezing, and chest pain. Pulmonary sarcoidosis can result in restrictive lung disease, pulmonary fibrosis, and, in severe cases, respiratory failure.

### ➤ ***Lymphatic Involvement***

Lymphadenopathy, particularly bilateral hilar lymphadenopathy, is a common finding in sarcoidosis. Enlargement of the lymph nodes, especially in the chest, can be detected on imaging studies, such as chest X-rays or computed tomography (CT) scans.

### ➤ ***Skin Involvement***

➤ Skin involvement occurs in approximately 25% of sarcoidosis patients, with lesions typically manifesting as erythematous, round to oval, firm, and tender nodules or plaques.

The most common locations for skin lesions are the face, arms, and shins. These lesions can vary in color from reddish-brown to purple and may persist for months or longer.

Cutaneous manifestations of sarcoidosis may include erythema nodosum, lupus pernio (a chronic, indurated skin lesion), and plaques.

➤ ***Ocular Involvement***

Ocular involvement is common and can lead to uveitis, which may cause eye pain, redness, and vision disturbances. In severe cases, it can lead to complications such as cataracts or glaucoma. Regular ophthalmological monitoring is crucial in sarcoidosis patients to prevent vision loss.

➤ ***Cardiac Involvement***

Although rare, sarcoidosis can affect the heart, leading to arrhythmias, heart failure, and even sudden cardiac death. Granulomas can infiltrate the myocardium, affecting conduction pathways and leading to ventricular arrhythmias. Cardiac sarcoidosis is a serious complication that requires early diagnosis and management.

➤ ***Neurological Involvement***

Neurological sarcoidosis can manifest as headaches, seizures, cranial nerve palsies, or meningitis. The central nervous system (CNS) involvement is uncommon but can be severe, necessitating careful monitoring.

➤ ***Renal and Endocrine Involvement***

Although less common, sarcoidosis can affect the kidneys and endocrine systems. Renal involvement typically manifests as hypercalcemia, which can lead to kidney stones, nephrocalcinosis, or renal dysfunction. Sarcoidosis may also affect the pituitary gland, causing endocrine imbalances such as diabetes insipidus.

## **Diagnosis**

The diagnosis of sarcoidosis is primarily clinical, supported by histological evidence of non-caseating granulomas obtained from biopsy of affected tissues. Common sites for biopsy include skin lesions, lymph nodes, or lung tissue. A chest X-ray or CT scan is often used to assess pulmonary involvement, particularly to detect bilateral hilar lymphadenopathy or interstitial changes indicative of sarcoidosis. Blood tests may show elevated levels of serum angiotensin-converting enzyme (ACE), although this is not specific to sarcoidosis. A thorough evaluation, including a detailed history, physical examination, and imaging, is essential to confirm the diagnosis.

## **Treatment**

The management of sarcoidosis is tailored to the severity and extent of organ involvement, as well as the patient's clinical condition. For patients with mild symptoms or limited organ involvement, observation may be sufficient. However, more extensive or symptomatic cases often require pharmacological intervention.

➤ **Corticosteroids**

The mainstay of treatment for sarcoidosis remains systemic corticosteroids, such as prednisone, which are used to reduce inflammation and granuloma formation. For patients with mild or moderate disease, corticosteroids are typically used for a short duration, with gradual tapering to minimize side effects.

➤ **Immunosuppressive Agents**

For patients with chronic or refractory sarcoidosis, immunosuppressive drugs such as methotrexate, azathioprine, or leflunomide may be used as corticosteroid-sparing agents. These drugs help control inflammation and prevent disease progression, particularly in cases with significant pulmonary, skin, or ocular involvement.

➤ **Biologic Agents**

Recent advances in treatment have led to the use of biologic agents in refractory cases. Tumor necrosis factor-alpha (TNF- $\alpha$ ) inhibitors, such as infliximab or adalimumab, have shown efficacy in treating sarcoidosis that is resistant to conventional therapies, particularly in pulmonary and cardiac involvement.

➤ **Topical and Intralesional Therapies for Skin Involvement**

For patients with limited and non-disfiguring skin lesions, topical corticosteroids or intralesional steroid injections can be effective in reducing inflammation and improving the appearance of lesions. These therapies are usually well tolerated and can offer symptomatic relief.

➤ **Surgical Intervention**

Surgical removal of skin lesions may be considered in select cases, particularly when they are causing significant cosmetic disfigurement or discomfort. However, these lesions may recur after excision, and surgery is generally not curative for more widespread disease.

➤ **Management of Specific Organ Involvement**

- **Cardiac Involvement:** Patients with cardiac sarcoidosis may require antiarrhythmic drugs, such as beta-blockers, or implantation of a pacemaker or defibrillator if arrhythmias are present. In severe cases, heart transplantation may be considered.
- **Ocular Involvement:** Ophthalmologic treatment with corticosteroids, both topical and systemic, is crucial to manage uveitis and prevent permanent vision loss.
- **Pulmonary Involvement:** In severe cases, patients may require oxygen therapy or even lung transplantation if pulmonary function is significantly impaired.

## Conclusion

Sarcoidosis is a complex multisystem disease that requires a comprehensive approach to diagnosis and treatment. Although the exact cause remains unknown, advances in understanding its pathophysiology have led to improved therapeutic options. Early diagnosis and appropriate management are essential for preventing long-term complications and improving patient outcomes. Multidisciplinary care involving pulmonologists, dermatologists, cardiologists, and

ophthalmologists is often necessary to ensure optimal management, particularly in patients with extensive organ involvement.

## References

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