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EC PULMONOLOGY AND RESPIRATORY MEDICINE

Editorial

The Diagnosis of Sarcoidosis

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Received: September 26, 2018; **Published:** October 05, 2018

Diagnosis of sarcoidosis is never secure despite the established criteria of identification [1]. If the clinical presentation is characteristic for the disease as in certain specific syndromes such as Löfgren or Heerfordt syndrome, diagnosis can be made on clinical grounds without tissue biopsy confirmation [2]. Otherwise histologic confirmation of granulomatous inflammation is required in at least two organs to confirm the diagnosis because sarcoidosis is a multisystemic disease. As the presence of granulomatous inflammation is associated with many other diseases a thorough differential diagnosis is required the for final diagnosis of sarcoidosis. Sarcoidosis is a challenge for the pulmonary clinician because neither the symptoms nor the laboratory findings are specific enough to warrant the diagnosis. Identification requires pathologic tissue examination from involved organs although the granulomas alone are not sufficient for diagnosis that the presence of a consistent clinical syndrome and a characteristic evolutionary outcome are necessary eventually. Isolated extrapulmonary sarcoidosis occurs in approximately 2% of the patients. Definitive diagnosis may not be feasible in some patients in whom sarcoidosis presents with an isolated organ involvement that requires several years of follow up to ensure the final diagnosis of sarcoidosis while another organ involvement may develop as short as six months that facilitates the diagnosis [3,4].

Confidence level for sarcoidosis diagnosis has evolved from a definite diagnosis to a highly probable diagnosis since unconditional precision in diagnosis may not be feasible [5]. Identification of sarcoidosis cannot be done on the basis of clinical and radiologic profile of the patient because similar manifestations occur in many other diseases such as tuberculosis, fungal infection, hypersensitivity pneumonitis and lymphoma. Even the presence of granulomatous inflammation in a single organ with associated clinical manifestations is not adequate for final diagnosis since many other granulomatous diseases may share similar clinical features. A compatible clinical and radiologic presentation, pathologic evidence of non-caseating granulomas and the exclusion of other disease that emerge with identical clinical or radiologic findings are required for final diagnosis.

Sarcoidosis is a complex disease that requires a diagnosis of exclusion. Because the presentation, clinical, laboratory and the radiologic manifestations are versatile and variable, every patient should be evaluated on a case to case basis using a different diagnostic approach. Half of the patients are diagnosed within three months of initial presentation while in approximately 10% of the cases the definitive diagnosis is delayed up to two years [6]. The confidence level in a diagnosis of sarcoidosis has been revised from definite to highly probable diagnosis of sarcoidosis since absolute diagnostic certainty may not be feasible in many patients [5]. Consequently, the confidence level of diagnostic accuracy has been designated as highly probable, probable, possible, or unlikely [7] even though this approach is still equivocal. Highly probable confidence level reveals a consistent clinical profile, multisystemic involvement with a positive pathology. Probable confidence level includes patients with a consistent clinical presentation with a supportive pathology from presenting and asymptomatic organ. In possible confidence level of diagnosis there is a suggestive clinical profile with an unobtainable supportive pathology. Unlikely confidence level reveals a suggestive clinical presentation with an incompatible or an absent pathology [3,7].

The diagnostic pathway for sarcoidosis is complex. The patients may be asymptomatic, may present with an isolated single organ involvement, or with an extrapulmonary organ disease may dominate the clinical picture. The clinical, laboratory and the radiologic manifestations are not specific enough to warrant a definitive diagnosis of sarcoidosis. The diagnosis is based on a compatible clinical

presentation with consistent radiologic findings, presence of non-caseating granulomas in tissue biopsy samples and the exclusion of other granulomatous diseases in the differential diagnosis. Sarcoidosis constitutes a diagnostic challenge for the pulmonary clinician thereby the identification of sarcoidosis may be delayed. Furthermore genetic variations associated with sarcoidosis may pose another difficult aspect of the disease because they promote atypical clinical manifestations. The best diagnostic pathway is the evaluation of patients on a case to case basis because no single clinical presentation, radiologic finding or an algorithm is adequately specific enough for the diagnosis of the individual sarcoidosis patient [8].

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Volume 7 Issue 11 November 2018