

Neurofibromatosis Type I With Pulmonary Involvement



Neurofibromatosis tipo I con afectación pulmonar

Dear Editor,

A 69-year-old woman with nonobstructive coronary artery disease and neurofibromatosis type I (NF1) visited a cardiologist, who noted worsening of the patient's functional class. She complained of dyspnea during physical stress, which had started years previously, and mild productive cough. She denied fever, night sweats, chills and any other symptom. She also denied a past history of pulmonary disease. She had a 20-year history of smoking 1 pack of cigarettes per week (3 pack years). A first-degree relative had also been diagnosed with NF1. Physical examination revealed multiple and widespread brownish dermatologic lesions compatible with "café au lait" spots, and multiple skin nodules compatible with neurofibromas (Fig. 1D). Chest CT showed centrilobular emphysema associated with bullae, predominating in the upper lobes of both lungs. Scattered pulmonary cysts of various sizes were also present (Fig. 1A–C). These findings are compatible with pulmonary involvement in NF1.

NF1, also known as von Recklinghausen disease, is an autosomal-dominant disorder of neuroectodermal and mesenchymal origin. It can occur as inherited or sporadic disease, the latter corresponding to 30%–50% of cases. A mutation in the NF1 gene on

chromosome 17q11.2 has been recognized as a disease promoter, and the spectrum of clinical phenotypes is wide because of the gene's complexity, size and diversity of exons. The disease is characterized by multiple tumors of ectodermal and mesodermal tissues, and its hallmarks are multiple neurofibromas, "café-au-lait" spots and pigmented hamartomas in the iris (Lisch nodules). Neurofibromas are benign nerve-sheath tumors of the peripheral nervous system. Plexiform neurofibromas occur along spinal roots and small branches and large trunks of nerves, and can undergo malignant progression, the major cause of cancer-related death in affected patients.^{1–3}

Some authors have emphasized that interstitial disease associated with NF1 is a definable clinical entity, although the true prevalence and characteristics of this association remain unknown.¹ In a literature review, Zamora et al.¹ analyzed 61 cases of interstitial lung disease associated with NF1, and the main pulmonary abnormalities on HRCT were bullae (50%), reticular abnormalities (50%), ground-glass opacities (37%), cysts (25%) and emphysema (25%). Other thoracic abnormalities described in patients with NF1 include chest-wall cutaneous and subcutaneous neurofibromas, plexiform neurofibromas, meningoceles, kyphoscoliosis, ribbon deformity of the ribs due to bone dysplasia or erosion from adjacent neurofibromas, other thoracic tumors, posterior vertebral scalloping and enlarged neural foramina.^{1–5} In conclusion, patients with NF1 may present pulmonary involvement characterized by dyspnea and tomographic evidence of interstitial lung disease, such as emphysema, bullae, cysts and reticular abnormalities.

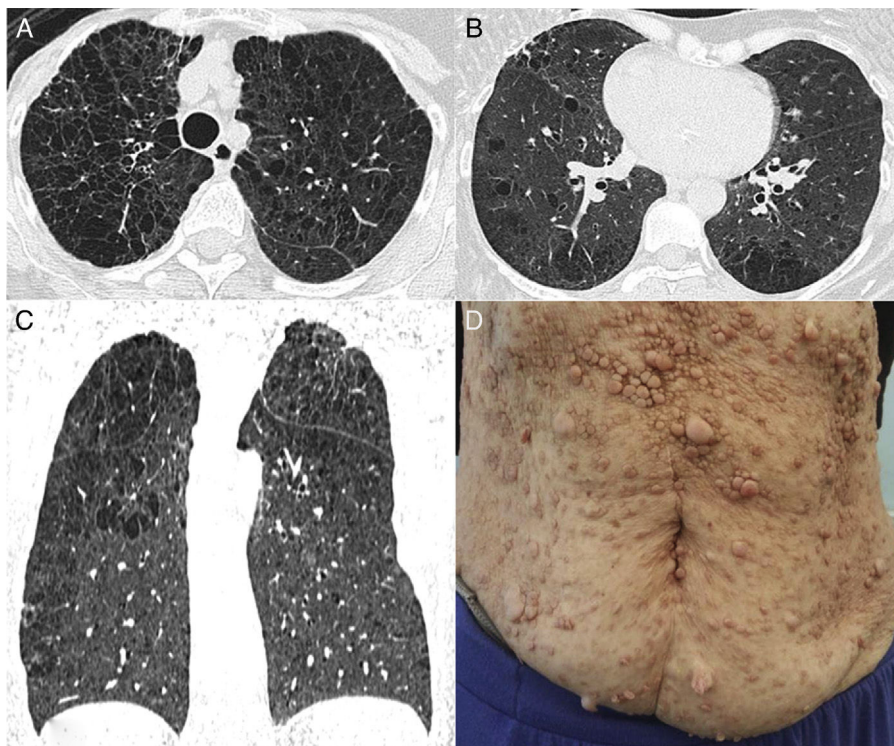


Fig. 1. Axial chest CT images obtained with lung window settings at the levels of the upper lobes (A) and lower pulmonary veins (B), and coronal reconstruction (C) showing emphysema with upper lobe predominance and multiple scattered pulmonary cysts, predominantly in the right lung. Note also in (D) multiple cutaneous neurofibromas in the anterior thoracic wall.

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Reliability of the Minimum Basic Data Set as an Epidemiological Tool in Tuberculosis[☆]



Fiabilidad del conjunto mínimo básico de datos como herramienta epidemiológica de la enfermedad tuberculosa

To the Editor,

Tuberculosis is currently the leading cause of death due to infectious disease in adults worldwide.¹ Although its incidence is decreasing in our setting, it remains a significant health problem and disease surveillance is still necessary.² However, there is no single source of information that definitively records all cases of tuberculosis, since significant underreporting has been detected in the Spanish Notifiable Diseases reporting system.³

One of the objectives of the Minimum Basic Data Set (MBDS), a data system used to collect information on hospital morbidity, is to facilitate the conduct of research studies.⁴ Its validity depends on the availability of a full clinical report and correctly recorded variables, and, as demonstrated by studies performed in Spain, the reliability of the data contained in the registry is not guaranteed.⁵ However, it is easy to access, so several authors have studied its usefulness in epidemiological studies in diseases that are managed in hospitals, included tuberculosis disease.^{6–9}

The aim of this study was to determine the reliability and usefulness of the MBDS for conducting studies in tuberculosis. To this end, we performed a retrospective study between 1994 and 2013, consulting the MBDS of the Soria Healthcare Complex that encompasses the entire province of Soria. The search was based on International Classification of Diseases (ICD-9-CM) diagnostic codes between 010.00 and 018.96 for active tuberculosis in any anatomical site, both for the primary and the secondary diagnosis. Mycobacterial records from the Microbiology Laboratory of the Soria Healthcare Complex and cases of tuberculosis notified to the Spanish Notifiable Diseases system were also consulted. The clinical records of all patients who were listed in the 3 registries were then reviewed, using the criteria described by the National Epidemiological Surveillance Network (RENAVE) for cases with clinical and/or laboratory diagnosis of tuberculosis.¹⁰

A total of 336 patients were recorded in the MBDS with a diagnosis of tuberculosis disease during the study period. After review of the clinical records, 69 patients (20.5%) listed in the MBDS were found not to have tuberculosis. In more than half of these patients (59.4%), the disease had been suspected at the time of discharge,

but the diagnosis had been subsequently ruled out on follow-up. In the remaining cases, clinical records revealed that 15.9% of patients were receiving tuberculosis prophylaxis and that 17.3% had a previous history of tuberculosis, situations that were not included in the diagnostic codes selected for the study. Another 7.2% were coding errors not associated with tuberculosis. Eight patients died in the emergency department, for whom multiple possibilities were coded, and in 5 cases, no clinical records were found, so the diagnosis could not be confirmed.

In all, 347 patients with true tuberculosis disease according to the RENAVE criteria and who figured in any of the 3 sources (MBDS, mycobacterial records of the Microbiology Laboratory, and the Notifiable Diseases system) were detected. A comparison of the registries showed that 93 of the patients with tuberculosis disease did not appear in the MBDS, accounting for 26.8% of all patients with tuberculosis detected in the province using these 3 records. On the other hand, 41 patients (11.8%) appeared exclusively in the MBDS, and did not figure in the other registries.

The individual registry that showed the greatest sensitivity, contributing most patients (78.6%), was the Microbiology Laboratory mycobacterial records. This was followed by the Notifiable Diseases system, with 77.5% of patients: only patients for whom the year of diagnosis there were obligation to notify the disease were considered. Finally, the MBDS contributed 73.1%.

The use of the MBDS in the study of tuberculosis theoretically provides access to interesting epidemiological data, but data need to be reliable if conclusions are to be drawn and trends are to be forecast. Since this registry is based on the discharge report, cases that are diagnosed *post hoc* do not appear, and, in contrast, cases that are later ruled out are included. Furthermore, errors in data entry also detract from the reliability of the registry. Results obtained in this study show that only 254 patients of the 336 included in the MBDS registry could be considered as tuberculosis cases, according to RENAVE criteria, so the positive predictive value of this registry was 75.5%.

According to these results, the epidemiological data associated with tuberculosis disease obtained from the MBDS alone are highly questionable, and the clinical records of each patient must be reviewed to confirm the diagnosis. The combined use of several registries, comparing and completing the cases provided by each one, gives a clearer picture of the real data.^{9,11} Indeed, 11.8% of the patients detected only appeared in the MBDS, since for these patients neither microbiological confirmation was available nor had they been notified.

A simpler approach would be to use only 2 data sources: the MBDS combined with the microbiological records. This approach, after the review of the clinical records, would have detected 98.5% of the patients included in this study, considerably simplifying

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