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CASE REPORT

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Spontaneous Pneumothorax in a Patient with Fabry Disease

Neumotórax espontáneo en paciente con enfermedad de Fabry

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RESUMEN

La enfermedad de Fabry pertenece al grupo de enfermedades lisosomales, poco frecuentes y de origen genético. Se comporta como una enfermedad crónica, multisistémica y progresiva, que deteriora la calidad de vida y disminuye la supervivencia del paciente. La afectación pulmonar en esta enfermedad es incierta y no existen reportes en la bibliografía sobre su incidencia. Presentamos el caso de un paciente masculino con diagnóstico de enfermedad de Fabry y compromiso pulmonar.

Palabras claves: Neumotórax espontáneo; Enfermedad de Fabry; Enfermedad lisosomal

ABSTRACT

Fabry disease belongs to the group of lysosomal diseases, which are rare and of genetic origin. It behaves like a chronic, multisystemic, progressive disease that deteriorates the quality of life and decreases patient's survival.2
Lung involvement in this disease is uncertain, and there are no reports in the literature related to its incidence. We present the case of a male patient with a diagnosis of Fabry disease and lung involvement.

Key word: *Spontaneous pneumothorax*; Fabry disease; Lysosomal disease

CASE REPORT

Medical history

22-year-old male patient, from the city of Formosa, diagnosed with Fabry disease since 2018, and currently undergoing enzyme replacement therapy. The patient denies a history of smoking or use of illicit substances. No other pathological history reported.

Reason for consultation

The patient reports having started with dyspnea and chest pain of 8/10 intensity 6 days prior to admission. He claims it was a stabbing type of pain that radiated throughout the left hemithorax and shoulder. He also stated that the pain intensity increased when lying on the affected side. And he denies any other associated respiratory symptoms. He decided to consult his primary care physician, who requested a chest X-ray.

As a previous history, the patient reports having had a pain of similar characteristics the previous year, which resolved spontaneously.

Physical examination

The patient is eupneic, with a SpO_2 of 98% on room air. There is a marked decrease in the vesi-

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cular murmur in the left hemithorax, without any added sounds. Lower excursion of the homolateral lung base.

Laboratory tests: complete blood count, liver function test, and renal function test results are normal.

Chest CAT: left pneumothorax predominantly affecting the anterior and basal regions.

No consolidative opacities or ground-glass opacities are recognized in assessable sections of the lung parenchyma.

Supplementary testing

Evolution

Given the absence of any other triggering cause, the condition is assumed to be a spontaneous pneumothorax, and the decision is made, together with the thoracic surgery team, to place a pleural drainage tube.

During hospitalization, the patient showed good clinical and radiological evolution; therefore, he was discharged.

DISCUSSION

Fabry disease is a genetic disorder with X-linked inheritance, caused by a deficiency in the activity of the lysosomal enzyme alpha-galactosidase A.[1-3]

In most cases, it is due to deficiencies in lysosomal hydrolytic enzymes, although it can also be due to alterations in lysosomal membrane proteins and those associated with the synthesis of lysosomal proteins. It is characterized by the deposition of globotriaosylceramide (Gl3 or Gb3) in different organs and tissues, with frequent involvement of the skin, the digestive system, the cornea, and more severely, in the nervous system, heart, and kidneys.[2]

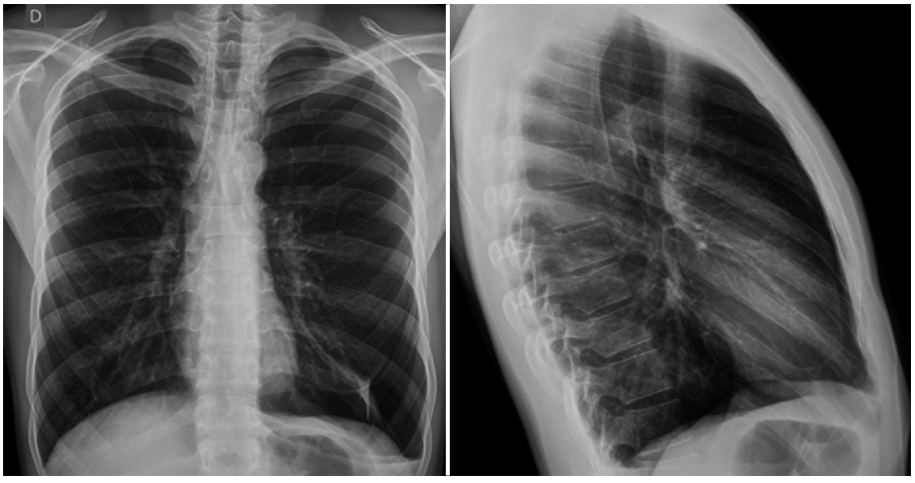

Figure 1. Chest X-ray: a grade II pneumothorax is observed in the left hemithorax.

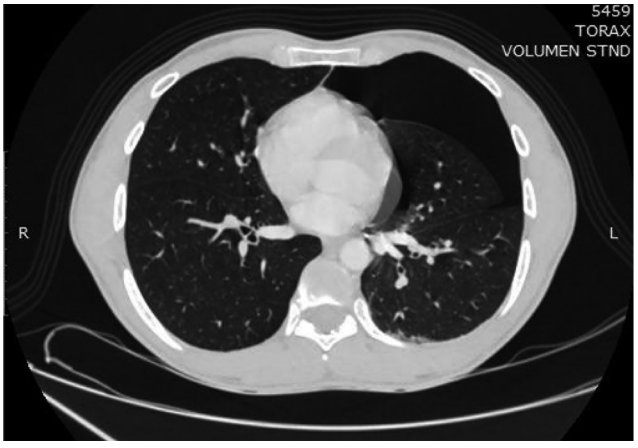

Figure 2. Chest CT: grade II pneumothorax in the left hemithorax.

Incidence: Fabry disease has an approximate incidence of 1 in every 117,000 live births and 1 in every 40,000 men, and is one of the most common lysosomal storage disorders. Recently, it has been discovered that its prevalence is higher in patients undergoing hemodialysis, young patients with cerebrovascular accident (CVA) or left ventricular hypertrophy (LVH) of unknown origin, compared to the general population.[2]

The diagnosis of this disease is made by demonstrating a decreased enzyme function, after which a confirmatory genetic study is conducted.[3]

Lung involvement in Fabry disease, and its severity, remain a controversial topic, as there are very few publications on the subject.

In a systematic review by Svensson et al, the main findings include: intermittent chest tightness, pneumothorax, hemoptysis, recurrent pulmonary infections, pulmonary thromboembolism, and pulmonary infarction.[4] In a case series published by Brown, 3 patients with spontaneous pneumothorax in Fabry disease were described: two were associated with bullous changes in chest radiography and one was associated with pulmonary emphysema in the autopsy.[5]

In the case of our patient, where no probable causes of pneumothorax were identified and it was assumed to be spontaneous, it is still possible to consider a potential association with the underlying disease. Due to the limited number of reports on the pulmonary and pleural presentation of this disease, it was decided to present this case.

Conflict of interest
The authors have no conflict of interest to declare.

REFERENCES

1. AADELFA (Asociación Argentina de estudio de enfermedad de Fabry y otras enfermedades lisosomales). Evaluation of patients with Fabry disease in Argentina. Medicina (B Aires). 2010;70:37-43.
2. Neumann P, Antongiovanni N, Fainboim A, et al. Guía para el diagnóstico, seguimiento y tratamiento de la Enfermedad de Fabry. Consenso de médicos de AADELFA Y GADYTEF. Medicina Buenos Aires. 2013;73:482-94.
3. Gonzalez-Rodriguez J, Benavidez-Villalobos J, Arroyo Villalta A Enfermedad de Fabry en el adulto: revisión bibliográfica. RHCS 2020;6:75-84. https://doi.org/10.56239/rhcs.2020.62.430
4. Svensson CK, Feldt-Rasmussen U, Backer V. Fabry disease respiratory symptoms, and airway limitation - a systematic review. Eur Clin Respir J. 2015;2. https://doi.org/10.3402/ecrj.v2.26721
5. Brown LK, Miller A, Bhuptani A, et al. Pulmonary involvement in Fabry disease. Am J Respir Crit Care Med. 1997;155:1004-10 https://doi.org/10.1164/ajrccm.155.3.9116979