

## BRONCHIECTASIS AND MEDIASTINAL NEUROFIBROMA IN A SAUDI FEMALE WITH EHLERS-DANLOS SYNDROME

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The Ehlers-Danlos syndrome (EDS) is a heterogeneous group of inherited disorders characterized by a variety of connective tissue abnormalities, occurring in at least 10 types, I to X, based on clinical, genetic and biochemical evidence, and varying in severity from mild to lethal. We present here a case of Type II EDS and discuss the clinical features and radiological abnormalities associated with the condition. To the author's knowledge, this is the first case of an association between EDS and bronchiectasis, as well as a solitary mediastinal neurofibroma.

### Case Report

A 17-year-old Saudi female presented with an eight-month history of shortness of breath which occurred after meals, and was associated with an occasional dry cough. There was no chest pain, wheezing or hemoptysis. The patient also complained of a gradual decrease in visual acuity and a generalized laxity of her joints. As a child, she had had delayed milestones, and brownish spots had occurred over her body with persistent scarring after minor traumas. She had two younger brothers and a grandmother with similar complaints.

On examination, the patient looked older than her chronological age, with hanging cheeks and freckles over her face, upper limbs and trunk but none on the lower limbs. The skin was lax and there were a few café au lait macules measuring 1.5-2 cm in diameter, ichthyosis on both legs, atrophic scars on the skin of the knees, but no superficial fibroma. Moderate hypermobility was noted in the joints of the upper, and to a lesser degree, lower limbs. Examination of the eyes revealed keratoglobus, iris nodules, and peripheral corneal thinning, as well as a mild reduction of visual acuity.

CBC, ESR, and coagulation profile were normal. Electrolytes, liver, and kidney function were within normal range. Bone profile and serum copper and ceruloplasmin were also normal. Pulmonary function tests (Table 1) showed a mixed pattern, with predominantly restrictive

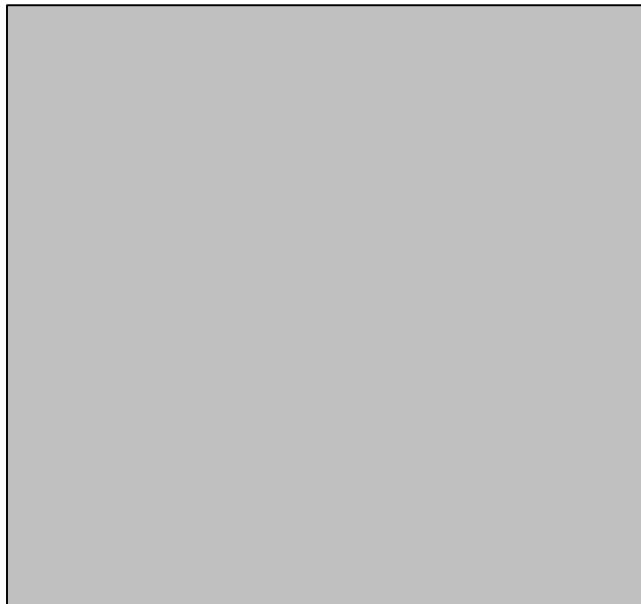


FIGURE 1. The chest radiograph of the patient showing right diaphragmatic hernia.

defect and mild airway trapping with normal diffusing capacity. Arterial blood gases were normal, and chest radiograph (Figure 1) showed evidence of right diaphragmatic hernia. CT scan of the chest (Figure 2) showed: 1) right diaphragmatic hernia, with part of the colon herniating into the chest; 2) focal bronchiectatic changes associated with mild cicatrization in the right middle lobe, lingula and anteromedial segment of the lower lobe; and 3) a left posterior mediastinal well-defined homogeneous soft tissue mass resting on the dorsal vertebrae, with widening of the underlying neural foramen, indicating a neurofibroma (Figure 3). CT scan of the brain and abdomen were normal.

### Discussion

EDS has now been established as a group of at least 10 different types of inherited connective tissue disorders.<sup>1</sup> Because the natural history, complications and mode of inheritance differ from one type to the other, reference to the syndrome is usually made by emphasis on the

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particular type.<sup>2</sup> In the case presented, type II is the most appropriate



FIGURE 2. CT scan of the chest showing the diaphragmatic hernia and the bronchiectatic changes in the right middle lobe and left lower lobe.

TABLE 1. Pulmonary function tests of patient.

Parameters	Measured	% of predicted
FVC	2.16 L	67
FEV <sub>1</sub>	2.04 L	67
Ratio		94%
TLC	3.55 L	75
RV	1.5 L	120
TLC – He	3.01 L	71
DLCO	18.8*	67
MEF <sub>50</sub>	3.15 L/S	65
FEF <sub>25-75</sub>	2.82 L/S	67
PEFR	4.66 L/S	73

S=second; \*mL/min/mmHg.

type that fits the clinical and radiological features described. In the medical literature, it is referred to as “mitis” because of the similarity to EDS type I but with less severe features. The patient presented here had only moderate joint hypermobility and limited bruising of the skin, which are typical features of the type II syndrome.<sup>3</sup>

Ayres et al.<sup>4</sup> studied the abnormalities of the lungs and thoracic cage in EDS in 20 patients with different types of the syndrome. Hemoptysis was seen in five patients and was attributed to respiratory tract infection. There was one patient with an apical bulla but none had bronchiectasis. In the patient presented, bronchiectasis was seen on a high-resolution CT scan as bilateral focal disease that was associated with cicatrization. The patient’s main complaint was shortness of breath, which could have been due to the large diaphragmatic hernia, as she had no other significant symptoms of bronchiectasis.<sup>5</sup> It is possible that bronchiectasis in the patient was not related to EDS and was due to recurrent infections. However, the clinical history did not support this possibility. It is more likely that bronchiectasis had occurred as a result of the connective tissue abnormality, with defects in the synthesis and processing of type I and III collagen, the major

proteins of the skin, ligaments, tendons, blood vessels, viscera, and the airways.<sup>6</sup> It is possible that the bronchial dilatation in EDS has a similar mechanism to the aneurysmal dilatation of



FIGURE 3. A mediastinal window cut showing the neurofibroma on the left side.

the arteries. Pulmonary function tests showing mixed obstructive and restrictive pattern are more likely due to the combined effects of both bronchiectasis and the large diaphragmatic hernia.<sup>7</sup>

No association has ever been reported between EDS and the presence of a single neurofibroma. However, one case report in the literature described a patient with EDS and von Recklinghausen neurofibromatosis.<sup>8</sup> The authors suggested that these two syndromes were generally linked through a defect on chromosome 17. In the case presented here, there was no evidence of generalized neurofibromatosis, but the finding of a single neurofibroma may strengthen this view.

Because of the tendency of these patients to have complications, e.g., poor wound healing, recurrent dislocations, and in type IV arterial ruptures, it is imperative that such patients are identified and special precautions taken during surgical or medical interventions.<sup>9</sup>

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