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Extensive Respiratory Tract Calcifications in A Case of Tracheobronchopathia Osteochondroplastica Occurring in A Patient with Spondyloepiphyseal Dysplasia Congenita: A Unique Association

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Abstract

An adult female who is a wheelchair bound since early childhood was found to have hereditary multiple skeletal deformities consistent with the rare condition of spondyloepiphyseal dysplasia congenital associated with osteoporosis as well. The patient also suffered from episodic attacks of bronchospasm, stridor and cough. They sometimes were severe enough to call for admission to hospital. Interestingly, the CT images of the chest showed extensive calcifications extending from the sub glottis to trachea and bronchial tree compatible with the diagnosis of tracheobronchopathia osteochondroplastica. This case represents a unique concurrence of these two rare conditions.

Keywords: Spondyloepiphyseal dysplasia congenita • Tracheobroncopathia osteochondroplastica • Bronchial calcifications

Introduction

Tracheobronchopathia osteochondroplastica is an idiopathic non-malignant disease of large airways featured by submucosal cartilaginous to osseous nodules overlying the cartilaginous rings, which may be focal or diffuse. It is a rare benign disease, and is under-diagnosed since it is diagnosed antemortem through bronchoscopy and CT scan, the clinical presentation is non-specific. The etiology is still unknown. The manifestations of TO may be either asymptomatic or non-specific respiratory symptoms. The incidence of TO in bronchoscopy is estimated around 0.01-4.2%. Clinical presentation varies from asymptomatic to symptoms like breathlessness, recurrent chest infections, cough and hemoptysis. Due to the lack of awareness of this disease, it remains an under recognized entity.

Case Report

A 32-year-old Arab female has been wheelchair- bound since early childhood. She and another six siblings were born to consanguineous parents. Two of her siblings had expired earlier as had been affected by the same condition, and an another affected younger brother of hers has been lying in the hospital with respiratory supportive measures for years. None of the parents is been affected.

Since birth and later in child and adulthood, she manifested with significant deformities including small stature (Height: 123 cm, BMI: 25) and retarded ossification of vertebral bodies, trunk, pelvis and extremities. Figure 1, is demonstrating the multiple skeletal deformities including deformed chest (barrel chest) in (A & B), severe scoliosis of the spine with dysplastic vertebrae in (C & D), absence of the heads of femur in (E), and deformed, short tubular bones along with widening of epiphysis in (F and G) images. Other features including, short neck, myopia, high pitched voice hypertrophic ear auricles, hypotonia of the limbs and talepus varus. The secondary sexual characters and mentation were normal. Urinary glycosaminoglycan repeatedly tested negative thus excluded Morquio's disease. The diagnosis of spondyloepiphyseal dysplasia congenital (SEDC) was furnished However, over the years she began to experience frequent episodes of stridor and bronchospasm to the extent it was severe enough times to call for admissions to the intensive care unit ward. The differential white cell count did not reveal eosinophilia and serum immunoglobulin IgE remained of normal values. Nonetheless, she was maintained on regular bronchodilator and inhaled corticosteroids as variable degree of rhonchi can always be heard on auscultation of the chest. Also, she experienced recurrent attacks of significant dyspepsia for which underwent upper gastro esophageal endoscopy. It revealed the presence of barrett esophagus and the patient was prescribed with protein pump inhibitor since. Tests for helicobacter pylori infection were negative. In further skeletal examination, the calculated Z score was -3.4 in right hip and -2.7 in the left, suggestive significant osteoporosis. The score was -2.3 in the lumbar spine and the 25(OH)D was only 13 ng/ml indicating vitamin D insufficiency (normal >30 ng/ml). PTH level was within normal. However, because of worsening of the asthma she underwent chest computed tomography scan. The outcome surprisingly, revealed the presence of extensive nodular calcification extending from the subglottis to the bronchi which is consistent with significant tracheobronchopathia osteochondroplastica (Figure 2).

Discussion

Spondyloepiphyseal dysplasia congenita (SEDC) is a rare heritable bone disorder, characterized by disproportionate dwarfism with short spine with dysplastic vertebrae shortage of trunk and short neck associated with variable degrees of coxa vara. It is caused by mutations in the *COL2A1* gene. The *COL2A1* gene is essential for the normal development of bones and other tissues that form the body's supportive framework i.e., connective tissues. Mutations in the *COL2A1* gene interfere with the assembly of type II collagen molecules, which prevents bones and other connective tissues from developing properly [1,2]. Although SEDC is typically inherited in an autosomal dominant manner a few cases of autosomal recessive forms have also been reported [3].

Tracheobronchopathia osteochondroplastica (TO) on the other hand

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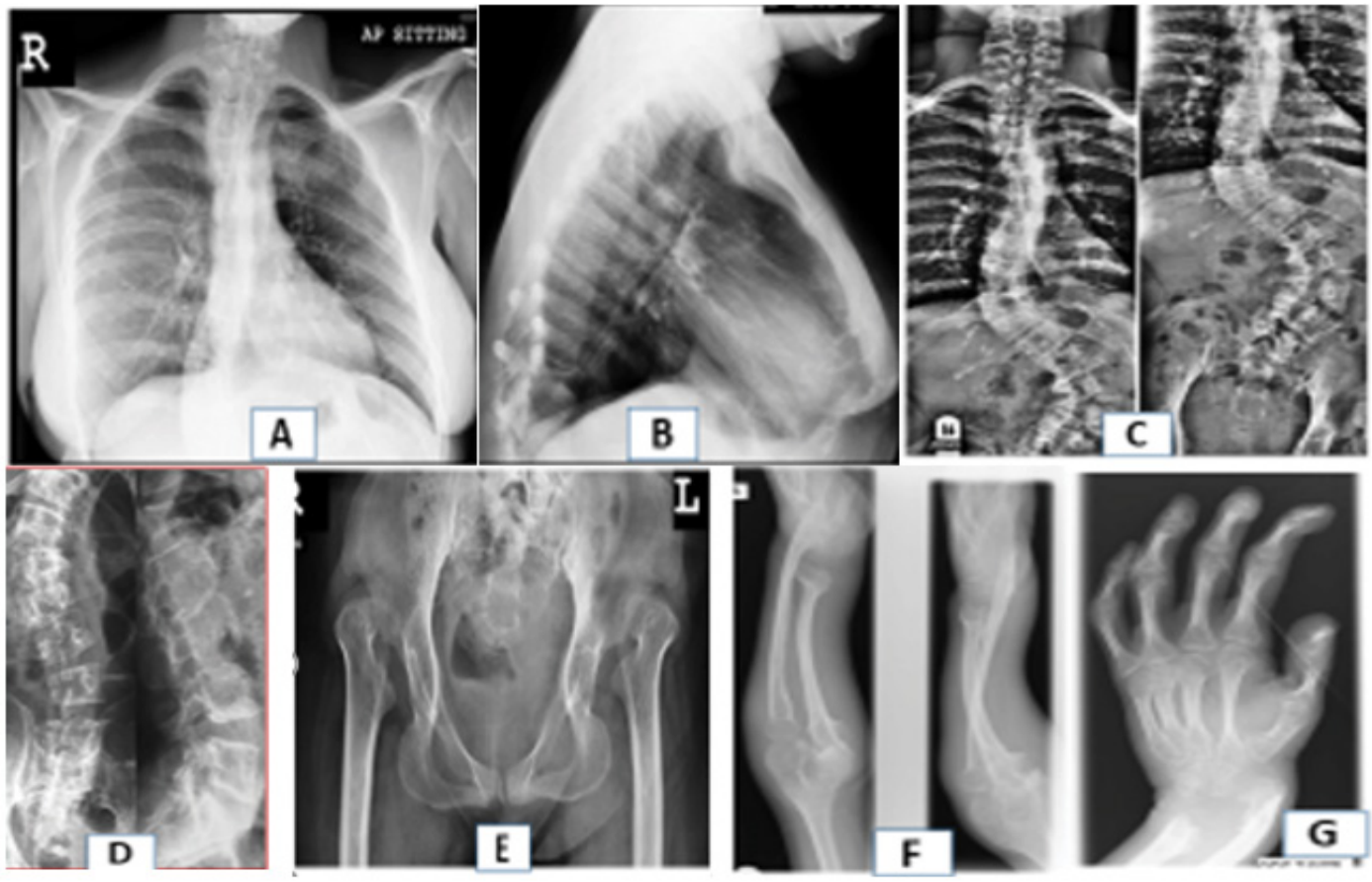

Figure 1. Kyphoscoliosis and barrel chest (A & B), severe scoliosis of the spine in (C), dysplastic vertebrae in (D), absence of the heads of femur and deformed tubular bone and enlargement of epiphysis in (F & G).

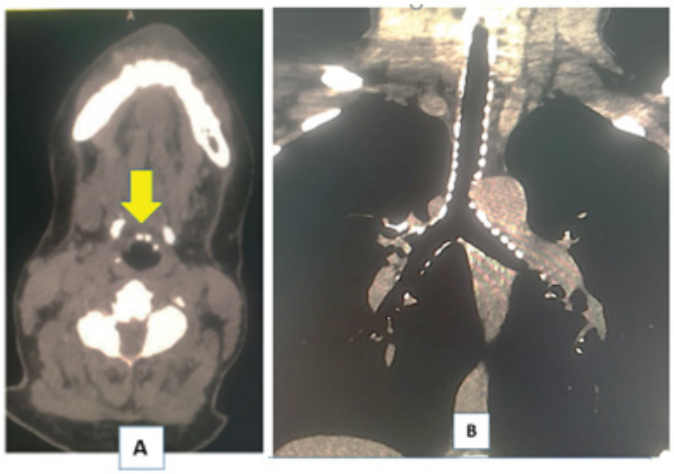

Figure 2. Computed tomography scan images showing extensive calcifications extending from the subglottis (A, arrow) to the trachea and bronchial tree (B).

is a rare benign disease, of unknown cause, characterized by numerous sessile, cartilaginous, or bony submucosal nodules distributed throughout the anterolateral walls, projecting into the laryngotracheobronchial lumen. The nodular lesions are sessile and calcified and varying from 1 to 10 mm in diameter. The diagnosis is seldom made because of the chronic and asymptomatic nature of the condition and it is diagnosed incidentally during bronchoscopy or autopsy (90% of the cases) [4]. However, symptomatic cases may have dyspnea, coughing, hemoptysis, hoarseness, and wheezing (mistaken as asthma) [4,5]. TO is not known to be associated with an any specific disease [4].

The first and perhaps the only case of a familial occurrence of TO was described in 1989 [5]. In 1997 Tajima et al. hypothesized the possible involvement of a bone morphogenetic protein (Bone Morphogenetic Protein-2, BMP-2) and of TGF-beta-1 (transforming growth factor-beta-1 in inducing the formation of submucosal osseous and cartilaginous nodules in the tracheobronchial system [6].

The patient here was a target of these two rare but significant conditions. Such co- occurrence seems rather unique and only adds further medical complications to the patient. The lack of mobility, hypovitaminosis D and the asthma or the asthma –like disease all have contributed to the development of osteoporosis in this patient. None of the parents had the SEDC while three of their children had the disease could suggest the non-autosomal dominant pattern of inheritance in this family and a recessive pattern would then be more likely.

Conclusion

As for the TO, the anatomical involvement by the calcified nodules as shown in the CT scan which is extending from the sub-glottis down to the bronchi indicates a significant respiratory disease. This is supported by the presence of continuous respiratory rhonchi also. Finally, this report may be the first to describe such concurrence of two unusually rare conditions in the English literature.

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