Congenital cystic adenomatoid malformation associated with esophageal duplication cyst

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Congenital cystic adenomatoid malformation (CCAM) of the lung is rare and is characterized by an excessive overgrowth of the terminal bronchioles.[1] Patients with CCAM may present either in the newborn period with progressive respiratory distress, or rarely in older children and adults with recurrent pulmonary infections. Intrathoracic foregut duplication cysts are also rare, and depending on their size, may be asymptomatic and discovered incidentally, or may present with a variety of symptoms, most commonly airway or esophageal obstruction.[2] The coexistence of both CCAM and esophageal duplication cyst (EDC) is extremely rare.[3,4] This is a case report of CCAM in association with an EDC in a 12-year-old child. The literature on the subject is also reviewed.

Case

A 12-year-old male child was referred to our hospital with a 6-year history of repeated attacks of chest infection. He received several courses of antibiotics and was also investigated for pulmonary tuberculosis because of persistent lung infiltrates on his chest x-ray. All investigations for tuberculosis were negative, but despite this he was treated empirically with antituberculous drugs for 6 months without improvement. At our hospital, he had a CT scan of the chest, which showed multiple cysts of variable sizes affecting the right upper lobe of the lung (Figure 1) as well as a cystic swelling in the posterior mediastinum. He underwent a diagnostic right thoracotomy. The right upper lobe was found to be firm in consistency and contained adhesions to the inner chest wall. These adhesions were released and there were multiple cysts in the right upper lobe of variable sizes (Figure 2). There was no consent for a lobectomy, so it was decided to take a large biopsy, including one of these cysts. Exploration of the posterior mediastinum revealed a cyst of about 3X2 centimeter in size overlaying the esophagus. The cyst was dissected using both sharp and blunt dissection. It was found to be adherent to the esophageal wall. The cyst was separated from the esophageal wall and excised totally. Postoperatively, the patient did well and was discharged home on the tenth postoperative day. Histopathology of the lung biopsy showed large areas of fibrosis with cystic formations of varying sizes filled with mucus and macrophages, lined by cuboidal or columnar epithelium and rarely metaplastic squamous epithelium. There was chronic inflammatory cell infiltrate in some areas with formation of lymphoid follicles, but no epithelioid or giant cell granulomas. The esophageal cyst showed a wall composed of smooth muscles with the presence of ganglion cells and lined by ciliated columnar epithelium, but no cartilage. These features are consistent with CCAM in association with an EDC. The patient was followed up in the outpatient clinic and the condition was explained to the father who agreed to a lobectomy. The patient subsequently underwent right upper lobectomy.

Discussion

Congenital cystic adenomatoid malformation is an uncommon anomaly, characterized by an exaggerated adenomatoid overgrowth of the terminal bronchioles with consequent suppression of alveolar growth and the formation of intercommunicating cysts of variable sizes.[1,3] According to Stocker et al,[5] there are 3 types of CCAM malformations: Type 1: large cysts (>2 cm in diameter and of varying sizes), Type II: cysts < 2cm in diameter, and more uniform in size, Type III: solid masses with multiple small cysts. Type II lesions are associated more frequently with other congenital malformations, while type III lesions

are known to be associated with nonimmune fetal hydrops. Our patient had a type I lesion.

Patients with CCAM commonly present early in the neonatal period with progressive respiratory distress, and may die as a result. Rarely, they may remain asymptomatic until mid- or late-childhood and then present with recurrent chest infections. The importance of this needs to be emphasized as the presentation with recurrent chest infections will ultimately lead to more fibrotic changes in the lung tissues leading to diagnostic difficulties. This was the case in our patient. He was symptomatic for 6 years prior to his presentation to our hospital, and because of diagnostic difficulties, he received several courses of antibiotics as well as an empirical antituberculous treatment for 6 months.

Duplications of the alimentary tract are rare congenital malformations. Commonly, they are classified into two types: tubular and cystic. Cystic duplications are rare and have been called enterogenous cysts, gastroenteric cysts, or alimentary duplication cysts.[6] Esophageal duplication cyst is of foregut origin, and two theories have been proposed to explain its pathogenesis. One theory suggests that it results from incomplete recanalization of the esophageal lumen between the fifth and eighth week of embryonic life,[7,8] while the other theory suggests that it is caused by an abnormal budding of the primitive foregut.[9-11] It is sometimes difficult to distinguish EDC from a mediastinal bronchogenic cyst. The epithelial lining of an EDC may be of respiratory or alimentary tract origin, or rarely, a combination of both. The close proximity of the cyst to the esophageal wall, the presence of a smooth muscle layer in the wall and the lack of cartilage help distinguish the two conditions. In our patient, the cyst was lined by ciliated columnar epithelium, and the wall contained no cartilage, but smooth muscles with the presence of ganglion cells. EDCs are usually found incidentally in asymptomatic patients, but they can present in early infancy with respiratory or esophageal obstructive symptoms if they attain a large size or if they develop complications such as infection or hemorrhage.[2] In our patient, EDC cyst was an incidental finding both radiologically and intraoperatively.

EDCs are known to be associated with other congenital anomalies such as spinal abnormalities, midgut duplications and tracheoesophageal fistula.[2,3] When foregut duplication cysts are associated with vertebral malformations, they are called neuroenteric cysts to differentiate them from simple foregut duplication cysts.[2] The coexistence of both congenital EDC and congenital bronchopulmonary malformations is rare, but has been reported.[12] From 1987 to 1992, Bogers et al treated 22 children with congenital bronchopulmonary malformations; one of them had a bronchogenic cyst combined with esophageal duplication, while another had intrapulmonary sequestration associated with esophageal duplication.[13] Kim et al reported a 13-year-old girl with an unusual, complex bronchopulmonary foregut malformation that included extralobar pulmonary sequestration and a duplication cyst of mixed bronchogenic and esophageal type.[14] DeFlice reported a case of type II CCAM

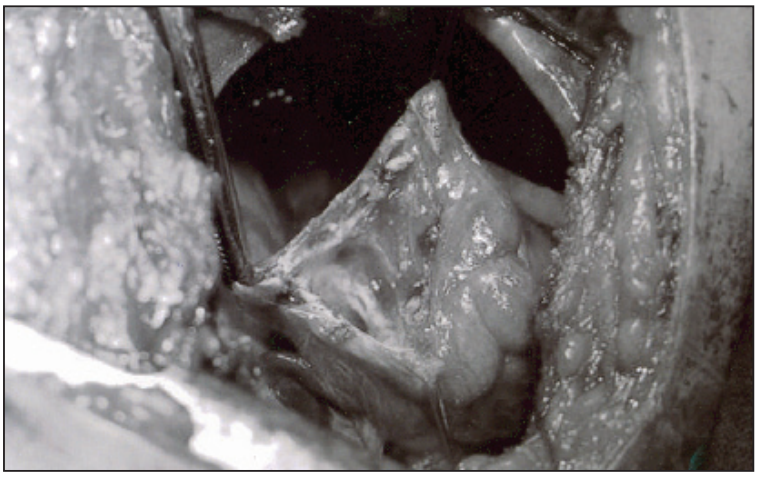

Figure 1. Intraoperative photograph showing a large, open cyst with adjacent smaller cysts.

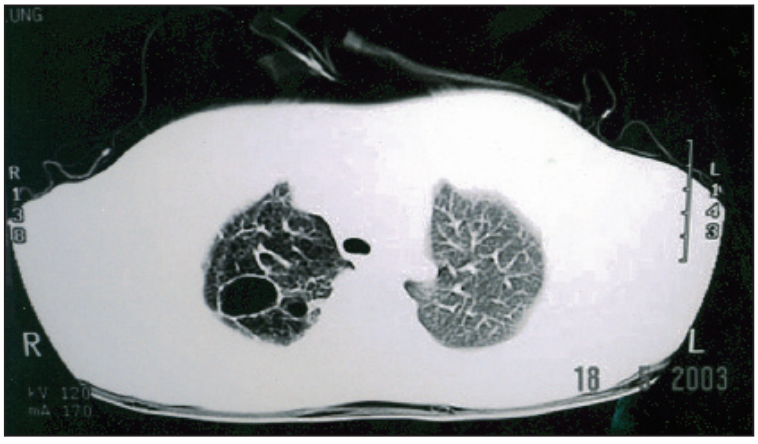

Figure 2. CT-scan of the chest showing multiple cysts of variable sizes in the right upper lobe.

of the right lower lobe associated with esophageal atresia and tracheoesophageal fistula in a full term female infant.[15] Horwitz and Lally reported a child with a bronchogenic as well as an EDC[16]. The coexistence of both CCAM and EDC on the other hand is extremely rare. In 1995, Kitano et al, in a review of the literature, found only 11 cases of various types of pulmonary cystic malformations in association with EDC, but none of them had CCAM.[12] Subsequently, Kuga et al in 2001 reported a 7-month-old girl with CCAM of the lung in association with an EDC,[4] and in 2002, Hasegawa et al reported a 6-month-old girl with CCAM and an EDC.[3] Our patient is the third of such malformations to be reported. The association of EDC or other congenital esophageal malformations such as tracheoesophageal fistula with other varieties of congenital cystic lung malformations is not a mere coincidence and like others, we think that such malformations represent a spectrum of one entity rather than separate lesions.[3,4,12] In 1968, Gerle et al proposed the term bronchopulmonary foregut malformation to represent an extralobar or an intralobar sequestration communicating with the gastrointestinal tract.[17] However, this proposal did not receive wide clinical acceptance, and in 1993, Nishijima et al proposed a new term lung-bud foregut malformation, which includes all congenital malformations derived from the foregut.[18] Such an association reported here is in support of this, and we like others feel that the malformations in our patients are the result of developmental arrest and accessory lung budding.[4]

The natural history of congenital cystic disease of the lung is not well defined, but there is a definite susceptibility to neoplastic transformation. CCAM is complicated by malignant transformation, including rhabdomyosarcoma and bronchioloalveolar carcinoma.[19,20] Bronchioloalveolar carcinoma complicating CCAM has been reported mainly in adults, but rhabdomyosarcoma has been reported in children as young as 1 year of age.[20] This calls for early diagnosis and elective excision even in asymptomatic patients. Our patient had metaplastic but no neoplastic changes.

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