Tracheobronchiomegaly*

The Mounier-Kuhn Syndrome in a Patient with the Kenny-Caffey Syndrome

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A 36-year-old woman with features of both the Mounier-Kuhn syndrome and the Kenny-Caffey syndrome is described. To our knowledge, this is the first reported case of these syndromes occurring together. Three-dimensional computed tomographic reconstruction of the upper airway revealed marked dilatation of the trachea and main-stem bronchi and several large diverticulae of the trachea.

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The Mounier-Kuhn syndrome or tracheobronchiomegaly is defined by an increase in transverse diameter of the trachea and main-stem bronchi of 3 SDs or greater. Symptoms include severe cough and chronic respiratory infections. There is increased compliance of the trachea. Diverticulae are seen from the trachea in approximately one third of patients. We report the first case (to our knowledge) of a patient with both the Mounier-Kuhn syndrome and the Kenny-Caffey syndrome.

CASE REPORT

A 36-year-old female dwarf presented to Duke University Medical Center, Durham, NC, for evaluation of a chronic cough. The patient's cough was occasionally productive and present since early childhood. Her parents and five siblings were normal in stature and without respiratory complaints.

Physical examination revealed a normally proportioned female subject 100 cm in height who weighed 16.2 kg. Vital signs were normal. Funduscopic examination and ocular pressures were normal. Hyperopia and strabismus were present. Head and neck examination revealed posterior pharyngeal lymphoid hyperplasia without lymphadenopathy. There was no secondary breast development. Expiratory wheezes and rhonchi were scattered throughout the lungs. Normal intelligence was present on informal testing.

The chest roentgenogram (Fig 1) revealed two right apical air cysts and an ill-defined opacity in the right upper lobe. Pulmonary function testing revealed a mildly reduced FEF25-75. A chest computed tomographic (CT) scan showed dilatation of the trachea, right and left main-stem bronchi with transverse diameters of 2.5 cm, 2.1 cm, and 1.6 cm, respectively. The anteroposterior diameter of the trachea was 1.9 cm. The trachea and the large cystic lesions in the right upper lobe were connected (Fig 2). Fluoroscopy revealed a 4 x 6-cm cyst in the mediastinum to the right of the midtrachea. With cough, this cystic structure moved into the neck. A ventilation study demonstrated prolonged retention of xenon 133 in the esophagus.

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FIGURE 1. Chest roentgenogram demonstrates two large cystic lesions in the right lung apex and subjacent ill-defined right upper lobe linear opacities. The trachea and main-stem bronchi are moderately enlarged. Note the small squarish, rather than rectangular, shaped thoracic vertebrae and the absence of a medullary cavity within the clavicles.

FIGURE 2. Three-dimensional (3-D) reconstructions of the trachea and attached diverticulae. The neck of the diverticula is narrowed from which the larger cysts project posteriorly. The 3-D reconstruction was prepared from serial 1.5-mm sections from the lung apex to the carina and reconstructed utilizing a reconstruction unit.
and grams esophagus mobile dwarfism, syndrome. This narrowed otherwise of bones demonstrated cysts. Routine serum electrolytes, calcium, phosphorus, magnesium, thyrocalcitonin, intact and midregion parathyroid hormone, somatomedin C level, and karyotype were normal.

Discussion

This patient has features of two unusual conditions: the Mounier-Kuhn and the Kenny-Caffey syndromes. We believe that this is the first reported case of the Mounier-Kuhn syndrome occurring in a patient with Kenny-Caffey syndrome. The Kenny-Caffey syndrome is characterized by dwarfism, normal intelligence, thickened bone cortices with small medullary cavities, ocular abnormalities, and transient hypocalcemia. This is the 22nd case reported in the English literature.

The Mounier-Kuhn syndrome or tracheobronchiomegaly is diagnosed by demonstration of an increased transverse diameter of the trachea and main-stem bronchi. Normal values for the transverse diameter of the trachea (20.2 ± 3.4 mm), right main-stem (16.0 ± 2.6 mm), and left main-stem (14.5 ± 2.8 mm) bronchi have been established by bronchography of 50 normal adult patients. Patients with the Mounier-Kuhn syndrome exceed these dimensions by 3 SDs. The cross-sectional area of the trachea correlates closely with body height. The transverse diameter of our patient's trachea and main-stem bronchi are more than 3 SDs larger than predicted for her height. Tracheobronchiomegaly has been found in 1.0 percent of 500 adult bronchographies. Chest roentgenograms may establish the diagnosis. Bronchography, chest CT, or magnetic resonance imaging may confirm the diagnosis. Fluoroscopy may demonstrate marked compliance of the trachea with narrowing on expiration and dilation on inspiration. Diverticulae of the trachea are seen in approximately one third of patients and most commonly originate from the right posterolateral wall. Pulmonary function testing may reveal increased residual volume and obstruction.

Mounier-Kuhn syndrome is hypothesized to be a congenital abnormality of the elastic and muscle fibers and enlarged cartilage usually limited to the trachea and main bronchi. The diagnosis is most often made in the fourth or fifth decade, but has been established in an 18-month-old child. It has occurred in siblings and has been reported in association with Ehlers-Danlos syndrome and cutis laxa. Severe chronic respiratory tract infections with resulting saccular bronchiectasis are common. Productive chronic cough or recurrent pneumonia often dates to childhood.

Corrective surgery has no role in the Mounier-Kuhn syndrome because of the diffuse nature of the tracheobronchial abnormality. Intubation of a patient with Mounier-Kuhn syndrome for 15 days with a high-volume-low-pressure cuff (cuff pressure of 16.9 mm Hg) has resulted in tracheal stenosis requiring surgical resection of the stenotic segment of trachea. Patients with Mounier-Kuhn syndrome who require mechanical ventilation should be intubated with an uncuffed tube followed by packing of the pharynx.

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References

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