Tracheobronchial Stenosis in Keutel Syndrome

Keutel syndrome is characterized by brachytelephalangism, abnormal cartilage calcification, peripheral pulmonary stenoses, and midfacial hypoplasia. We report the first case from East Asia in an 8-month-old boy who had the typical craniofacial appearance characterized by midfacial hypoplasia with a broad depressed nasal bridge (***Fig.* 1**). The distal phalanges of fingers were thickened. Auscultation revealed a grade 2-3/6 systolic murmur over heart, pronounced in the second and third intercostal space, and an inspiratory and expiratory stridor and wheezing over both lungs. Chest radiograph and computed tomography showed tracheobronchial cartilage calcification and tracheobronchial stenosis, confirmed on bronchoscopy. Echocardiography revealed peripheral pulmonary stenosis.

Keutel syndrome is a rare autosomal recessive disease, with 27 reported cases from 19 families in several countries; mostly from the Middle East. All of them showed tracheobronchial calcification, and five of them had stenosis of the tracheobronchial tree [1,2]. Our patient is the fifth patient with tracheobronchial stenosis, which should be emphasized as another remarkable feature in this syndrome. Meier, *et al*. [2] have reported subglottic laryngeal stenosis in two siblings, and post mortem examination in the young male revealed small cartilaginous rings with abnormal endochondral calcification causing stenosis of the trachea and the bronchi. Cartilage rings were not found in the trachea of the patient.

In recent years, the interest for endoscopic treatment modalities had increased in tracheal surgery as a possible alternative to surgical resection. Endoscopy has been suggested as the first choice for simple stenosis, and success rate of 96% has been reported. So far, this approach has rarely been used in children. Our patient accepted bronchoscopic cryotherapy and balloon dilatation four times, and the diameter of the subglottic laryngeal stenosis was expanded from 3 mm to 4.5 mm. The clinical symptoms improved after endoscopy, but he died of lung reinfection three weeks after discharge from our hospital.

FIG.1 *Midface hypoplasia is present with a depressed nasal bridge and small nose.*

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