An Atypical Case of Mounier-Kuhn Syndrome

Time to Change the Diagnostic Approach?

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Summary: **Mounier-Kuhn syndrome (MKS) is a rare disease of unknown etiology characterized by abnormal pathologic dilatation of tracheabronchial tree. The diagnosis of MKS is normally made on CT scan of chest on the basis of enlarged diameters of trachea and main stem bronchi. We are presenting histologically confirmed case of MKS, where the diameter of right main bronchus is below minimum diameter (mean + 3 SD) required for the diagnosis. We suggest that the diagnosis of MKS should not be solely based on fixed criteria such as the diameter of airways, but on the basis of the overall clinical, pathologic, and radiologic profile.**

Key Words: Mounier-Kuhn syndrome, diagnostic approach, airway diameter

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Mounier-Kuhn syndrome (MKS) is a rare disorder of unknown etiology characterized by marked dilatation of the trachea and the main bronchi. The patient usually presents with recurrent lower respiratory tract infection. The syndrome was first described by Mounier-Kuhn[1] in 1932, and till date, only a few hundred cases are reported in the medical literature.[2] Diagnosis is usually made on computed tomography (CT) scan on the basis of the diameter of the trachea and the main bronchi.

Herein, we report a case of MKS in which the diameter of the right main bronchus was <21 mm (the minimum diameter required for the diagnosis). To the best of our knowledge, this is the only case of MKS where the traditional criteria of airway diameter are not being met. We believe that the diagnosis should not be made on the basis of fixed criteria such as the diameter of airways, but on the basis of the overall clinical, pathologic, and radiologic profile.

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CASE REPORT

In August 2014, a 30-year-old man, who was a nonsmoker and a chef by occupation, presented to us with complaints of cough with an increased sputum volume and purulence and high-grade fever. He had a history of recurrent episodes of high-grade fever and cough with copious expectoration since 14 years.

The patient also had a history of recurrent episodes of childhood lower respiratory tract infection since the age of 18 months till the age of 6 years requiring multiple hospitalizations.

The patient was diagnosed with pulmonary tuberculosis on the basis of sputum microbiology 3 years ago and was treated with antitubercular treatment.

There was no history of similar illness in the family. The patient was married, and had 2 healthy children.

On general physical examination, his arm span (187 cm) was greater than his height (180 cm), and the upper body (87 cm) to lower body (93 cm) ratio was 0.94. Additional findings were a high arched palate, a positive wrist and thumb sign, and mild ptosis. There were no other significant findings on general physical examination. On respiratory system examination, the trachea was shifted to the right, and there were coarse crepitations over mammary areas on both sides on auscultation.

A chest x-ray revealed multiple ring shadows bilaterally. A pulmonary function test showed restrictive lung function, with a forced vital capacity (FVC%) of 73% and an FEV_1/FVC ratio of 80.9. The flow volume loop was normal. A CT scan of the lung demonstrated marked dilatation of the trachea (31.7 mm) and the major bronchi (the right main stem bronchus was 19.6 mm and the left main stem bronchus was 20.5 mm). There were multiple diverticulae in the trachea and the bronchi, with bilateral bronchiectatic changes predominantly in the lower lobes (Fig. 1).

On the basis of the marfanoid features, the patient was suspected to be a case of tracheobronchomegaly

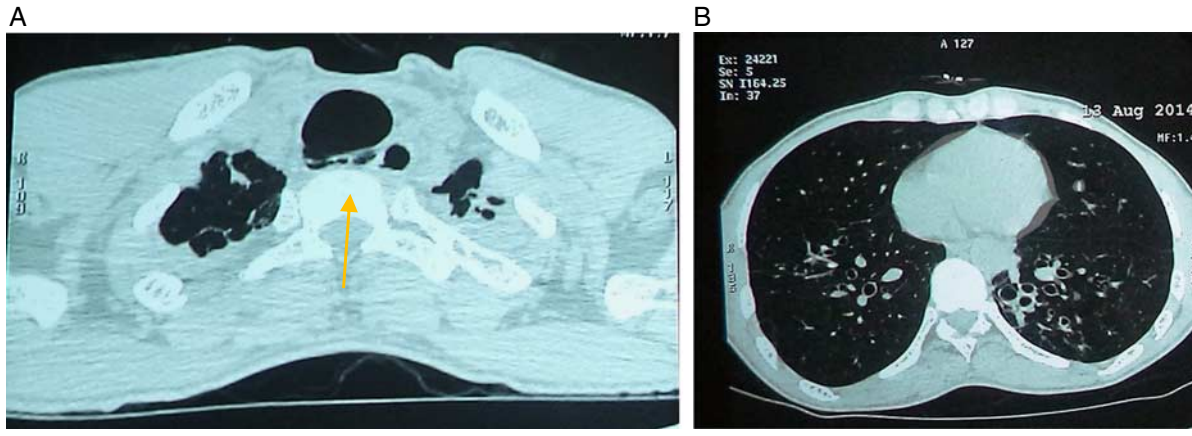

FIGURE 1. A, Marked tracheal dilatation with diverticuli (arrow). B, B/L lower lobe bronchiectasis.

secondary to Marfan syndrome and was evaluated further. Ophthalmology examination with a slit lamp showed no subluxation of the lens, and fundus examination was normal. There were no skeletal abnormalities found on detailed examination. Echocardiography was also normal.

On fiberoptic bronchoscopy examination, there were numerous outpouching in the lumen of the trachea, which extended up to the subsegmental bronchi. There was excessive collapsibility of the lumen of the trachea during expiration and coughing. A punch biopsy was taken from the anterior tracheal wall between the cartilaginous rings and examined histochemically with Verhoeff elastic stain; there was almost complete loss of elastic fibers in the tracheal wall. A culture of the BAL fluid showed pure growth of *Streptococcus pneumoniae* (Fig. 2).

On the basis of the CT scan and the bronchoscopic biopsy, the patient was diagnosed as a case of the Mounier-Kuhn syndrome, and the patient was put on antibiotics, bronchodilators, and chest physiotherapy on an outpatient basis along with flu and pneumococcal vaccination. There was rapid symptomatic improvement in the patient's clinical condition, and within 2 weeks, the patient became asymptomatic.

DISCUSSION

MKS is a rare disorder of uncertain etiology characterized by tracheobronchial dilatation secondary to atrophy of elastic and muscle fibers. This pathologic dilatation leads to ineffective cough and impairment of mucociliary clearance, leading to recurrent lower respiratory tract infection.[3]

It is more common in males and is generally diagnosed in the third and the fourth decades of life. Symptoms in MKS are nonspecific. The patient usually presents with recurrent lower respiratory tract infection, chronic cough, and

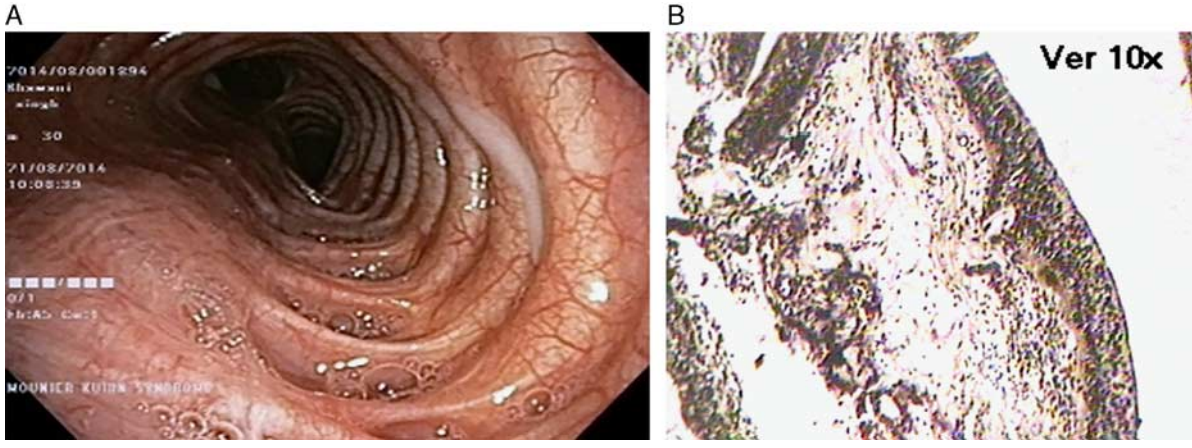

FIGURE 2. A, Bronchoscopy showing multiple diverticula. B, Elastic stain showing loss of elastic fiber.

TABLE 1. The Upper Limit of the Normal Diameter of the Trachea and the Major Bronchi

		Tracheobronchial Diameter (Mean + 3SD)					
		Trachea		RMB		LMB	
References	Method	T	S	T	S	T	S
Himalstein and Gallagher[5]	Autopsy (100 M)	28.6	24.7	25.8	17.4	22.5	16.3
Katz et al[6]	Bronchography (50M)	30.5	—	24	—	23	—
Woodiring et al[7]	Chest radiography (79 M, 121 F)	—	—	21.1 (M), 19.8 (F)	—	18.4 (M) 17.4 (F)	—
Vock et al[9]	CT scan (25 M, 25 F)	21.8 (M), 19.4 (F)	21.8 (M), 19.4 (F)	—	—	—	—
Breatnach et al[8]	Chest radiography (430 M, 378 F)	25 (M), 21 (F)	27 (M), 23 (F)	—	—	—	—

CT indicates computed tomography; F, female; LMB, left main bronchus; M, male; RMB, right main bronchus; S, sagittal; T, transverse.

occasionally with spontaneous pneumothorax, hemoptysis, finger clubbing, and pneumonia.[4] Our patient also presented with recurrent infection and chronic cough, but the onset of symptoms was quite early. In the absence of infective exacerbation, the patient may be asymptomatic.

Patients are often diagnosed on CT scan, which shows excessively large airways with the formation of diverticulae.

The diagnosis of MKS is made when the diameter of the trachea and the major bronchi exceeds the "normal" diameter. The upper limit of the normal airway diameter is calculated by studying the airway diameter of the normal healthy population with no tracheobronchial disease, and traditionally, it has been taken as mean + 3SD in most studies. Different modalities have been used in different studies for the assessment of the airway diameter. Himalstein and Gallagher[5] conducted an autopsy study; bronchography was used by Katz et al[6] for the measurement of tracheobronchial dimensions. Similarly, chest radiography[7,8] and CT scan[9] have been used to assess the airway diameter. Table 1 shows the upper limit of the normal airway diameter across different studies. There are no data about the bronchial diameter on CT scan; hence, for the diagnosis of MKS, the criteria proposed by Woodiring et al[7] on chest radiography are widely accepted as bronchography results at a greater magnification of the tracheobronchial tree as compared with chest radiographs. In the present case, all the mentioned criteria were met, except for the right main bronchus, which was 19.6 mm.

Pathologically, tracheobronchomegaly appears to arise as a congenital defect or atrophy in the connective tissues of the tracheobronchial tree. The few biopsy and necropsy studies of the tracheal wall under this condition show a thinning of the muscularis mucosae, with paucity of elastic fibers.[10] The Verhoeff elastic stain confirms the loss of elastic fibers around the respiratory tract.[3] Although there is a paucity of data regarding the sensitivity and the specificity of histopathology for the confirmation of the diagnosis of MKS, a combined approach including clinical, radiologic, and histopathologic data surely provides more specific results.

Secondary causes of tracheobronchial enlargement are connective-tissue diseases, ataxia-telangiectasia, ankylosing spondylitis, Ehlers-Danlos syndrome, Marfan syndrome, Kenny-Caffey syndrome, Brachmann-de Lange syndrome, and cutis laxa.[11,12] Hence, these conditions should be considered in the differential diagnosis. On physical examination, our patient had marfanoid features, but did not fit into the criteria of Marfan syndrome.[13]

In our case, all the traditional criteria for the diagnosis of MKS were not met (right main bronchus < 21 mm), but due to a suggestive radiographic, bronchoscopic, and histopathologic picture, there was no doubt over the diagnosis of MKS.

There is no uniformity in the literature over the diagnostic criteria of MKS, and different studies/case reports have used different criteria for the diagnosis.[5–7,9,14] This may be due to the different populations studied by them.

Using the same airway diameter cutoff across all the populations and ethnicities may not be justified. Although there is a difference in normal pulmonary function test values among various races/ethnicities, the issue of variability

of the airway diameter among them has not been investigated properly. We believe that the cutoff airway diameter for the diagnosis of MKS may be different in different ethnicities, and further studies are required to look into this.

We believe that there can be 2 distinct approaches for the diagnosis of MKS; first, on the basis of radiology alone, using a "population-specific" airway diameter cutoff. This approach does not need further invasive studies, and makes diagnosis easy in primary care settings. The drawback of this approach lies with the fact that, presently, there are hardly any data on the airway diameter in different ethnic populations. Second, on the basis of the clinical, radiologic, and pathologic picture in borderline case. This approach is difficult to follow in primary care settings, but makes diagnosis more specific.

We believe that till the time that population-specific cutoff criteria for the diagnosis of MKS are formulated, it is more prudent to use a clinical, radiologic, and pathologic approach.

In conclusion, the diagnosis of MKS should not be solely dependent on the airway diameter on radiology. Bronchoscopy and histopathologic examination should be given equal importance for the diagnosis of MKS.

REFERENCES

1. Mounier-Kuhn P. Dilatation de la trachée: Constatations radio-graphyques etbroncoschopiques [Dilatation of the trachea: radiographic and bronchoscopic findings]. *Lyon Med*. 1932;150:106–109.
2. Krustins E. Mounier-Kuhn syndrome: a systematic analysis of 128 cases published within last 25 years. *Clin Respir J*. 2014. [Epub ahead of print].
3. Celik B, Bilgin S, Yuksel C. Mounier-Kuhn syndrome: a rare cause of bronchial dilation. *Tex Heart Inst J*. 2011;38:194–196.
4. Genta PR, Costa MV, Stelmach R, et al. A 26-yr-old male with recurrent respiratory infections. *Eur Respir J*. 2003;22:564–567.
5. Himalstein MA, Gallagher JC. Tracheobronchomegaly. *Ann Otol Rhino Laryngol*. 1973;82:223–227.
6. Katz I, LeVine M, Herman P. Tracheobronchomegaly: the Mounier-Kuhn syndrome. *Am J Roentgenol Radium Ther Nucl Med*. 1962;88:1084–1094.
7. Woodiring JH, Howard RS II, Relun SR. Congenital tracheobronchomegaly (Mounier–Kuhn syndrome): a report of 10 cases and review of the literature. *J Thorac Imaging*. 1991;6:1–10.
8. Breatnach E, Abbott GC, Fraser RG. Dimensions of the normal human trachea. *AJR Am J Roentgenol*. 1984;142:903–906.
9. Vock P, Spiegel T, Fram EK, et al. CT assessment of the adult intrathoracic cross-section of the trachea. *J Comput Assist Tomogr*. 1984;8:1076–1082.
10. El-Mallah Z, Quantock OP. Tracheobronchiomegaly. *Thorax*. 1968;23:320–324.
11. Huete-Garin A, Sagel SS. Mediastinum. In: Lee JKT, Sagel SS, Stanley RJ, Heiken JP, eds. *Computed Body Tomography With MRI Correlation*, 4th ed. Philadelphia: Lippincott Williams & Wilkins; 2006: 311–410.
12. Fortuna FP, Irion K, Wink C, et al. Mounier Kuhn syndrome. *J Bras Pneumol*. 2006;32:180–183.
13. Loeys BL, Dietz HC, Braverman AC, et al. Revised Ghent criteria for the diagnosis of Marfan syndrome (MFS). *J Med Genet*. 2010;47:476–485.
14. Lee JK. *Computed Body Tomography With MRI Correlation Vol 1*, 4th ed. 2006;1:408–409.