

Imaging diagnosis

Case 440

1. Relapsing polychondritis (VEXAS syndrome) or Trachea-bronchomalacia

【Progress】

He was given steroid pulse treatment in our hospital. He got temporary relief. However, as steroid volume was getting reduced, the respiratory symptoms reoccurred. He visited the medical college and was made diagnosis of VEXAS syndrome. Lately, he was scheduled to be admitted to the medical college hospital for further investigation.

【Discussion】

Tracheabronchomalacia indicates weakness of the mural of main bronchial tract, trachea and main bronchus. Most cases with Tracheabronchomalacia occurs in infant but can occur in adult with relapsing polychondritis (1,2). The origin of relapsing polychondritis was not clarified. In these days, relapsing polychondritis is thought to belong with VEXAS syndrome (3).

The presence of VEXAS syndrome was acknowledged in the year of 2020. In the fields of hematology, rheumatology, autoimmunology, VEXAS syndrome is, still high topic. The term of VEXAS is derived from the initial of Vacuoles, E1 enzyme, X-linked chromosome, Autoinflammatory disease, Somatic. Vacuoles appear histologically in cells of bone marrow system. E1 enzyme which works to dissolve waste protein produced in cells. The deficit of E1 enzyme bring out the disability of protein induced in bone marrow system cells, inducing excess protein in cells, leading cell death (3). The abnormality of X-linked chromosome is the origin of VEXAS. Male has only one X-linked chromosome, while female has two. Then, this syndrome occurs predominantly in males, especially of 50ies and 60ies. Autoinflammation is the different from autoimmune disease that is caused by antibody production such as allergy: Allergy 1 anaphylaxis, caused by IgE. Allergy 11 caused by antibody, IgG, Allergy III caused by complex of antigen-antibody, Allergy IV caused by delayed immune response. All allergy diseases are related to lymphocytes or mast cells. Apart from these, in VEXAS syndrome, waste protein produced by cell death, remain in cells, leading cell death (3). Macrophages and neutrophils response to proteins, inducing the origin of inflammation. Autoinflammatory diseases occur by the acquired abnormality of genes and related to macrophages and neutrophils with activation of interleukin 6 (IL6). This phenomenon occurs somatically.

The symptoms of VEXAS syndrome are as follows: rheumatoid arthritis, fever, skin erythema, trachea-bronchus chondritis, medium-sized arteritis. Elevation of IL6 and CRP is acknowledged in laboratory test (3). As the results, the symptoms of VEXAS syndrome are various.

The treatments of VEXAS syndrome are reported to be effective for the relief of symptoms in use of tocilizumab, ruxolitinib or azacytidine associated with steroid and/or intravenous immunoglobulin (IVIG) (3).

The discussion below is the same as that in Case 420 about relapsing polychondritis.

Bronchial stenosis arises from various diseases: malignancy, inflammation, auto-immune diseases. As malignancy, bronchial cancer, compression from malignant swollen lymph nodes of small cell carcinoma, malignant lymphoma. As inflammation, intrabronchial tuberculosis, bronchial pneumonia, and bronchial aspergillosis are listed. Others include auto-immune diseases, trachea-bronchopathy osteo-chondro-plastics, relapsing chondritis, trachea-bronchus-malacia, and amyloidosis (4-6).

Trachea and bronchus are composed of ciliary epithelium, mucosal proximal layer, submucosal layer, chondritic layer, tunica adventitia. Chondritic layers exist in anterior and lateral side murals of trachea and bronchus, but not in posterior murals. In trachea-bronchopathy osteo-chondro-plastics, chondritic layers at anterior and lateral murals contain multiple calcified cartilages, inducing stenosis of trachea and bronchus. In relapsing chondritis, thickening of anterior and lateral murals are found in CT (7, 8). Patients with relapsing chondritis are often associated with other chondritis such as ear chondritis and/or joint arthritis. Then, illness history might be useful for evaluating diagnosis. Trachea-bronchus malacia is categorized into two types: crescent type and saber-sheath type (9-11). Crescent type of trachea-bronchus malacia occurs thin-walled membrane of posterior mural of trachea probably because of atrophy of elastic fiber, while saber-sheath type occurs by injuries of lateral or anterior cartilage. It might be difficult to differentiate relapsing chondritis from trachea-bronchus malacia because their images are mere stenosis of trachea and bronchus. Meanwhile, in amyloidosis and granulomatosis with polyangiitis, bronchial stenosis with bronchial wall thickening is reported to be found at the whole murals, irrespective of anterior, lateral and posterior (10-13).

Relapsing chondritis occurs various cartilage parts: auricular cartilage, nasal cartilage, joint cartilage and tracheal and/or bronchus cartilage. Diagnostic criteria of relapsing polychondritis are as follows: three of the five symptoms are positive of relapsing polychondritis; inflammation of bilateral auricular cartilage, inflammation of nasal cartilage, inflammation of joint cartilage, inflammation of trachea or bronchus, and inflammation of peri-eye ball (7, 8).

In our case, he suffered from joint rheumatoid arthritis, bronchial stenosis, suspicious poly relapsing chondritis. He was given pulse-steroid therapy that induced him to bronchiolitis infected by mycotic infection. He was recovering to reduce gradual decrease of steroid volume and proper antibiotics.

【Summary】

We presented a fifty-year-old male with cough and dyspnea. He suffered from rheumatoid joint arthritis. Right main bronchus stenosis was depicted on chest CT, diagnosed as probable relapsed chondritis. Later, relapsed chondritis is one of the diseases of VEXAS syndrome. It is borne in mind that when stenosis of trachea and bronchus in adults is encountered on chest CT, trachea-bronchomalacia due to relapsing chondritis is listed. Relapsing chondritis belongs to VEXAS (Vacuoles, E1 enzyme, X-linked chromosome, Autoinflammatory disease, Somatic), according to the recent report.

【References】

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