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SCIENTIFIC LETTER

Mutations in *CCNO* Result in Primary Ciliary Dyskinesia Complicated with Diffuse Bronchiolitis

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To the Editor: Reduced generation of multiple motile cilia caused by the *CCNO* gene, which plays a key role influencing the pathways controlling the ciliogenesis of multiciliated cells (MCCs), is one of the subtypes of primary ciliary dyskinesia (PCD) [1, 2]. Patients with *CCNO* mutations develop various clinical manifestations [3, 4]. Here we presented one case with combined neonatal respiratory distress and diffuse bronchiolitis.

An eight-year-old girl was admitted to our hospital for recurrent cough and wheeze after birth with intolerance to physical activity. She presented with dyspnea at birth and treated with mechanical ventilation for 10 d. Thereafter, recurrent cough with accompanying wheeze occurred three to four times annually and was diagnosed with bronchiolitis obliterans in a local hospital. Upon physical examination, she looked ill and had growth retardation. She had tachypnea with cyanosis, a tri-retraction sign, and tubbiness thorax with clubbing finger. Wheezing and rales were heard in both lungs. The HRCT showed diffuse centrilobular small nodules throughout both lungs with a tree-in-bud sign, chronic maxillary sinusitis and ethmoid sinusitis; and slightly widened lateral ventricle. Bronchoscopy revealed endobronchial inflammation with secretions, and biopsy with no ciliary structure of the bronchial mucosa tissues. The gene sequence analysis identified compound heterozygous mutation of *CCNO* with c.263_267dupAGCCC and c.258_262dupGGCCC mutation from her mother and father respectively.

This study describes the clinical features of one *CCNO*-related PCD patient, and indicates that it should be considered in term infants with neonatal respiratory distress syndrome, persistent cough, and wheeze after birth. *CCNO* variants are rare in PCD patients but cause more severe phenotypes than other genes, while heterotaxy was never seen. We overviewed the genotype and phenotype-oriented literature of *CCNO*-related PCD (43 patients from 30 families) and compared to the findings of our patient. Diffuse bronchiolitis could be the hallmark radiologic feature of *CCNO*-related PCD patients.

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Declarations

Conflict of Interest None.

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