Case Report: Filamin A deficiency lung disease recognized in an eleven-year-old child

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Abstract

The loss of function of the FLNA gene may result in impairment of the filamin A protein. Of the many clinical syndromes, this condition may produce lung disease. This usually presents itself and is diagnosed in the infant/toddler age group that may mimic broncho-pulmonary-dysplasia. It is part of the entities included in Childhood Interstitial Lung Disease (chILD) group of disorders. We are reporting on a patient that was diagnosed at eleven years of age. This case provides a unique insight into the long-term course of lung disease in this illness and broadens our understanding of the spectrum of its presentation. Although the patient had symptoms very early in life, the diagnosis may not have been entertained because of the rarity of the disorder, its atypical presentation, and discontinuous care due to parents moving to different cities for reasons of employment. Her initial presentation to our institution was for pneumonia. Due to the highly unusual chest x-ray images, asthenia, and early clubbing, an extensive work up was undertaken that included further imaging and a lung biopsy. The final diagnosis was confirmed by the detection of FLNA LOF gene mutation.

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