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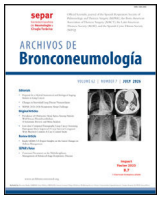

Clinical Image

Subpleural Cysts in Kabuki Syndrome

Carlos Martín-de Vicente [a,*], Martín Bahón-Fauro [b], Ruth García-Romero [c]

[a] *Pediatric Pulmonology Department, Hospital Universitario Miguel Servet, Zaragoza, Spain*
[b] *Department of Pediatrics. Hospital Universitario Miguel Servet, Zaragoza, Spain*
[c] *Pediatric Gastroenterology Department, Hospital Universitario Miguel Servet, Zaragoza, Spain*

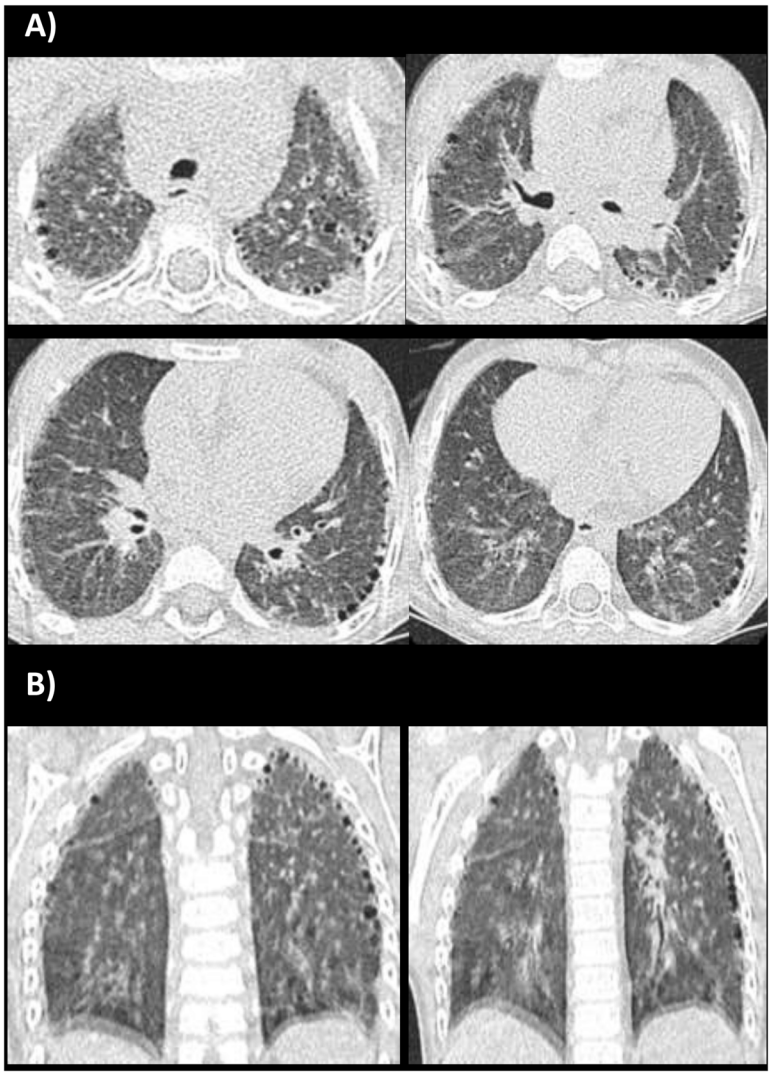

Fig. 1. Pulmonary computed tomography showing multiple subpleural cysts. (A) Axial sections; (B) coronal sections.

* Corresponding author.
E-mail address: clmartin@salud.aragon.es (C. Martín-de Vicente).

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A 3-year-old girl with Kabuki syndrome, KMT2D variant *(NM_003482.4):c.6595delT;p.(Tyr2199Ilefs*65)*, diagnosed on the basis of typical facial features and cleft palate, presented in the first weeks of life with vomiting, moderate-severe oropharyngeal dysphagia and gastro-oesophageal reflux. A high-resolution pulmonary computed tomography (CT) scan at 2 months of age, due to hypoxaemia and cough with feeding, revealed generalized interstitial lung findings suggestive of aspiration syndrome, which improved following gastrostomy and Nissen anti-reflux. Since both procedures, she has normal oxygen saturation and no chronic symptoms. A pulmonary CT scan at 3 years of age revealed multiple diffuse bilateral subpleural cysts, with some cysts of peribronchovascular distribution (Fig. 1). Subpleural cysts are typical findings in Down's syndrome [1,2], but also in other diseases as described by Singh et al., who report the only case of Kabuki syndrome to date, leading them to suggest these cysts as a result of growth abnormalities of the lung [3]. There are cases of interstitial lung disease in this syndrome, caused by variants clustered within a 40-aminoacid region of the KMT2D gene [4], a region in which our patient's variant is not found, or associated with immunodeficiency [5]. Given the patient's current favourable respiratory status, a lung biopsy was not deemed necessary; however, clinical and radiological follow-up will be carried out to monitor any progression of the pulmonary lesions.

Generative AI

No material has been partially or totally produced with the help of any artificial intelligence software or tool.

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Conflicts of interest

The authors declare not to have any conflicts of interest that may be considered to influence directly or indirectly the content of the manuscript.

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