Personalized therapy for interstitial lung diseases in caused by ABCA3 mutations

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Introduction

Interstitial lung disease (ILD) in infants and children comprises a large spectrum of rare respiratory disorders that are mostly chronic and associated with high morbidity and mortality.(1)

A four-year-old child with severe recurrent pulmonary infections from birth and a marked growth deficit was conducted to our attention. Her exome was sequenced and resulted in a double heterozygous on the ABCA3 gene compatible with a diagnosis of an autosomal type 3 recessive surfactant disease.

The aim of this work is to describe the management of therapy for a child with ABCA3 mutations focusing on the safety and effectiveness of the drugs prescribed.

GANTT

DRUGS						
Hydroxychloroquine				6mg/kg		
Omeprazole		1 mg/kg				
Prednisone		0.5 mg/kg				
Azithromycin		10mg/kg				
Methylprednisolone		10mg/kg				
Oxygen						
Fluticasone	100mcg/kg					
TIME	set-20	nov-20	giu-21	lug-21	set-21	ott-21

Start Therapy

Stop Therapy

Methodology

Data were collected from the Hospital committee for off-label use and from clinical records.

Results

Sep-2020: Oxygen therapy was started because of persistent hyposaturation in association with Fluticasone 100 mcg bid

Nov-2020: The following drugs were added: Methylprednisolone ev 10 mg/kg/d for 3 days, every month until July 2021, azithromycin os 10 mg/kg/d for 3 days/week, Prednisone os 0.5 mg/kg/d once every 48h and Omeprazole 1 mg/kg/d os.

Jun-2021: the registered saturation was normalized and the oxygen therapy was interrupted.

July-2021: Hydroxychloroquine was started (6 mg/kg/die) and Azithromycin and Methylprednisolone were discontinued.

Sep-2021: Both Prednisone and Omeprazole were discontinued.

Oct-2021: The child was clinically well, growth data improved and vital parameters were better at nocturnal oximetry. No adverse event was reported.

Conclusion

Every change in drug therapy was designed to contrast the progression of disease and to limit the use of steroids and their collateral effects.

The rationale behind the use of hydroxychloroquine was based on immune-modulating effects, as largely reported in the literature.

A strict monitoring led to a target therapy designed around the clinical need of the patient.

Bibliography

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