

**CASE REPORT**

**SAETTINI ET AL.,2021**

**NIEHUES ET AL., 2020**

PATIENT	New Patient	P1	P2	P3	P1	P2	P3
ANCESTRY	Unrelated parents	Consanguinity	Unrelated parents	Unrelated parents	Consanguinity	Consanguinity	Consanguinity
INHERITANCE	Compound heterozygous	Homozygous	Uniparental isodisomy	Compound heterozygous	Homozygous	Homozygous	Homozygous
VARIANT	c.3334C>T/large deletion	c.C868T	c.3306+1G>A	c.3218delT /del ex9-ex18	c.3353G>A	c.3353G>A	c.1289del
AGE AT EVALUATION	30 yrs	3 yrs	9 yrs	25yrs	7 mo	14.5 yrs	8 mo
INFECTIONS	Upper and lower respiratory tract infections	Upper and lower respiratory tract infections, and sepsis, gastroenteritis	Respiratory infections, diarrhea, and failure to thrive	Pneumonia, respiratory infections with recurrent sinusitis, gastrointestinal infections, Crohn disease	Laryngitis, pneumonia, bronchitis	Otitis media, pneumonia, bronchitis	Otitis media, obstructive bronchitis
HEART	Hypertrophic Cardiomyopathy (left ventricle)	Hypertrophic cardiomyopathy	Hypertrophic cardiomyopathy Persistent interatrial communication	Dysplastic, mildly apically displaced tricuspid valve, supraventricular tachycardia	Pre-excitation syndrome, Hypertrophic non-obstructive cardiomyopathy	Pre-excitation syndrome, Hypertrophic non-obstructive cardiomyopathy	Pre-excitation syndrome, Hypertrophic non-obstructive cardiomyopathy
RESPIRATORY TRACT	-	-	Bilateral bronchiectasis	-	Asthma	Asthma, bronchiectasis	Interstitial lung disease
SKELETAL MUSCLE	Deformation of the left foot (congenital convex)	-	No dysmorphic features	-	Not tested	Metabolic Myopathy	
CNS	Psychomotor developmental delay	Psychomotor developmental delay	-	-	Microcephaly Motoric developmental delay	Motoric developmental delay	
IMMUNOLOGICAL FEATURES	Agammaglobulinemia, absence of B cells. neutropenia	Intermittent or severe neutropenia, Hypogammaglobulinemia	Agammaglobulinemia, absence of B cells.	Severe hypogammaglobulinemia, absence of B cells. Severe neutropenia,	T cell lymphocytosis Hypogammaglobulinemia	T cell lymphocytosis	

Supplemental information Table 1. Summary of clinical features of FNIP1 patients