

Table 1: Variants of unknown significance (VUS) identified in Case 1 (blue) and Case 2 (green)

Case	Chr.	Gene	OMIM phenotype	Inheritance pattern	gnomAD MAF [Highest]
Case 1	1	<i>CR2</i>	Immunodeficiency, common variable	AR	0.00416% African
	1	<i>WDR26</i>	Skraban-Deardorff syndrome	AD	0.00%
	11	<i>EXT2</i>	Exostoses, Seizures, scoliosis, and macrocephaly syndrome	AD	0.00580% East Asian
	11	<i>RNASEH2C</i>	Aicardi-Goutieres syndrome	AR	0.00%
	X	<i>SEPT6</i>	X-linked Intellectual Disability	X-linked	0.00%
	12	<i>POLE</i>	FILS syndrome IMAGE-I syndrome	AR	0.00%
	16	<i>HYDIN</i>	Ciliary dyskinesia, primary, 5	AR	0.00%
	5	<i>C6</i>	C6 deficiency; Combined C6/C7 deficiency	AR	0.000899%
	5	<i>C6</i>	C6 deficiency; Combined C6/C7 deficiency	AR	0.00416%
	9	<i>DNAI1</i>	Ciliary dyskinesia, with or without situs inversus	AR	0.000895%
	9	<i>C5</i>	C5 deficiency	AR	0.0845%
Case 2	22	<i>TUBA8</i>	Cortical dysplasia, other brain malformations	AR	0.0149%
	6	<i>PSMB8</i>	Proteasome-associated autoinflammatory syndrome 1	AR	0.00%
	9	<i>DOCK8</i>	Hyper-IgE recurrent infection syndrome	AR	0.0942%