

**Suppoting Table 1.** Comparison of the clinical characteristics and pituitary function tests of our patient with other previously reported patients with *RNPC3* mutations.

Reference	Sex	<i>RNPC3</i> mutation (NM_017619.3)	Pituitary hormones							Pituitary MRI	Associated findings
			GH	PRL	TSH	ACTH	FSH	LH	ADH		
Argente <i>et al</i> 2014, Akin <i>et al</i> 2022	F	Compound heterozygous for c.1420C > A (P474T) / c.1504C > T (R502X)	↓	Low N	N	N	↑	N	NR	Hypoplasia	Mild microcephaly, §neuropathy
	F <sup>§</sup>		↓	Low N	N	N	↑	↑	NR	Hypoplasia	
	F		↓	Low N	N	N	↑	N	NR	Hypoplasia	
Gucev <i>et al</i> 2015, Akin <i>et al</i> 2022	M <sup>¶</sup>	Compound heterozygous for c.613C > T (R205X) / c.1420C > A (P474T)	↓	NA	N	N	N	N	NR	NR	Red hair, obesity, ¶myopathy
	M		↓	NA	N	N	N	N	NR	NR	
Akin <i>et al</i> 2022	F	Homozygous for c.1449A > T (L483F)	↓	↓	N	N	↑	N	NR	Hypoplasia	‡Neuropathy
	F		↓	↓	N	N	↑	N	NR	Hypoplasia	
	F		↓	↓	↑	N	↑	N	NR	Hypoplasia	
	F		↓	↓	↑	N	↑	N	NR	Hypoplasia	
	F		↓	↓	N	N	↑	N	NR	Hypoplasia	
	M		↓	↓	N	N	N	N	NR	Hypoplasia	
	M <sup>‡</sup>		↓	NA	↑	N	N	N	NR	Hypoplasia	
	M		↓	↓	N	N	N	N	NR	Hypoplasia	
Akin <i>et al</i> 2022	F	Compound heterozygous for c.1420C > A (P474T) / c.624+1G > T	↓	N	N	N	↑	↑	NR	Hypoplasia	
Akin <i>et al</i> 2022	M	Compound heterozygous for c.1421delC (P474Lfs*10) / c.1420C > A (P474T)	↓	↓	↓	N	N	N	NR	Hypoplasia	Dysmorphic face, clinodactyly, mild developmental delay, seizures, neuropathy
Verberne <i>et al</i> 2020	M	Compound heterozygous for c.259C > T (Q87X) / c.443G > C (G148A)	↓	↓	↓	N	NA	NA	NR	NA	Congenital cataract, intellectual disability
	M		↓	↓	↓	N	↓	↓	NR	NA	
	F		↓	↓	↓	N	↓	↓	NR	NA	
Yamada <i>et al</i> 2021	F	Compound heterozygous for c.261dup (L88Tfs*11) /	↓	↓	↓ (GD)	N	N	N	NR	N	Dysmorphic face, severe microcephaly, intellectual

		c.1228T > G (F410V)										disability, Graves' disease
Patient in this report	F	Homozygous for c.1328A > G; (Y443C)	↓	↓	↓	N	N	↓	N	Hypoplasia		Severe microcephaly, intellectual disability, diffuse cerebral and cerebellar atrophy, dysmorphic face, spasticity in extremities (neuropathy?)

F: female, M: male, GH: growth hormone, PRL: prolactin, TSH: thyroid stimulating hormone, ACTH: adrenocorticotrophic hormone, FSH: follicle stimulating hormone, LH: luteinizing hormone, ADH: antidiuretic hormone, GD: Graves' Disease, N: normal, NA: not assessed, NR: not reported. § Female patient having neuropathy ¶ Male patient having myopathy ‡ Male patient having neuropathy

