

	defect	number of affected children	OFC at birth	OFC after age 1 yr	short stature	mental retardation	language	psychomotor delay	hyperkinesia	seizures	facial features	other features	MRI
Mir et al. (2009)	c.1423C>T, p.R475X	8		1 pt normal, 2 pts around the fifth centile, 3 pts -2/3SD	no	moderate to severe	few words	yes		pt2: yes		pt3: mild kyphosis	diminished cerebral white matter volume with sulcal enlargement, thinning of the corpus callosum, and mildly reduced cerebellar volume ; several areas of T2 hyperintensity in the subcortical white matter
Mochida et al. (2009)	c.1423C>T, p.R475X	3	-1,3SD; -0,3SD; -2SD	-4,1SD; -3,3SD; -5,8SD		moderate to severe	few words	no	pt1: bruxism and hand-flapping movements				thin corpus callosum and reduced volume of the cerebral white matter
Abou Jamra et al. (2011)	c.1423C>T, p.R475X	6		<<3rd centile	moderate to severe (15th centile to <3rd centile)	moderate to severe	absent	severe	neonatal hypotonia, stereotypic movements, hand-flapping	pt1: yes	<i>low frontal hairline, synophrys</i>	pt4,5,6: spontaneously losing their teeth and losing weight after 20y	
Giorgio et al. (2016)	c.1423C>T, p.R475X	1		<3rd centile	no	severe	absent	severe	stereotypic movements (hand shaking, waving, and body rocking)	no	<i>brachycephaly, round face, thin and horizontal eyebrows, synophrys, deep set eyes, wide nasal bridge, thin upper lip</i>	obesity	severe corpus callosum thinning, clear reduction of the white matter with poor myelination, normal cerebellum
Mir et al. (2009)	c.2311-2314 delTGTT, p.Leu772TrpfsX7	4		-1 to -3SD		severe	absent	yes		no			
Philippe et al. (2009)	c.1708C>T, p.R570X	3	normal	-2SD; -1SD (later -3SD); -2SD	no	severe	speech delay	yes	hyperactivity		<i>hypertelorism, short neck, prominent upper central incisors, short and smooth philtrum; pt2: unilateral labial cleft</i>	truncular obesity; pt2: long and thin fingers	unusual white matter abnormalities with discordance between T2 and FLAIR sequences (normal myelination on T2 sequence, but important white matter abnormalities on FLAIR sequence)
Kakar et al. (2012)	c.1024+1G>T	4		little below 3rd centile	no	severe	few words	yes	pt2: muscular weakness	no			
Marangi et al. (2013)	c.2851-2A4 C, p.T951Y fsX17	2		10th centile; <2nd centile	no	severe	absent	yes	hypotonia after birth; pt1: tapering fingers	pt1: yes	<i>peculiar craniofacial appearance, including brachycephaly, round face, hypertelorism, straight eyebrows, synophrys, wide nasal bridge, and thin upper lip with downturned mouth</i>	pt1: clinodactily of the fifth finger	diminished cerebral white matter volume, with marked sulcal enlargement, thinning of the corpus callosum, and reduced cerebellar volume ; several areas of T2 hyperintensity were present in the subcortical white matter
Present patients	c.533T>C, p.Leu178Pro	2	microcephaly at birth (OFC	-4SD; -2,8SD	no	severe	pt1: no; pt2: few words	severe	yes	pt1: yes	<i>pt1: sloping forehead</i>	no	atrophy of the corpus callosum, pt1 white matter abnormalities

			not known)											
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Supplementary File 2. Clinical features in reported cases of TRAPPC9 defects and in the present probands