Table S1. Summary of genetic and clinical features in individuals with *SHROOM4* variations

Variation type	Genetic change	Neurocognitive/developmental phenotype	Other manifestations	Reference
SNV: missense	NM_020717.4: c.3266C>T; p.(S1089L)	four-generation pedigree with multiple affected members: severe ID, delayed or no speech, seizures, periods of depression, aggressiveness and hyperactivity, short stature	bilateral congenital hip luxation, scoliosis	Stocco dos Santos et al. [3-4], Hagens et al. [5]
	NM_020717.4: c.436C>T; p.(R146W)	dyspraxic gait, speech delay, eye pointing, peripheral vasomotor disturbances, microcephaly	kyphosis, small cold hand and feet	Lopes et al. [6]
	NM_020717.4: c.1913C>G; p.(S638C)	Stocco dos Santos syndrome		Farwell et al. [7]
	NM_020717.4: c.1201C>T; p.(H401Y)	myoclonic atonic epilepsy		Routier et al. [16]
	NM_020717.4: c.13C>A; p.(P5T), c.3236C>T; p.(E1079A), c.3581C>T; p.(S1194L), c.4288C>T; p.(R1430C), c.4303G>A;p.(V1435M), c.4331C>T; p.(P1444L)	six unrelated male individuals with idiopathic epilepsy		Bian et al. [17]
SNV: nonsense	NM_020717.4: c. 2050C>T; p.(R684*)	complete corpus callosum agenesis, Blake's pouch cyst	Turner syndrome	Heide et al. [18]
Structural: deletions	Xp11.23p11.22; 2.86 Mb	mild ID		Honda et al. [8]
	Xp11.22; 0.14 Mb and 2.6 Mb	hydrocephalus, severe growth and psychomotor retardation	Dent's disease: renal proximal tubulopathy with low-molecular-weight proteinuria, hypercalciuria, hyperaminoaciduria, hypophosphatemia and hyperuricemia	Armanet et al. [9]
	Xp11.23p.11.22; 0.7 Mb	mild ID, speech delay, microcephaly, short stature, developmental delay	short fingers with bilateral clinodactyly V, short toes, Dent's disease: proteinuria, nephrocalcinosis and	Danyel et al. [10]

			hypophosphatemia, facial dysmorphology	
Structural: duplications	Xp11.23p11.22; 4.4 - 8.9 Mb	12 affected individuals: 12/12 mild-severe ID, 12/12 language impairment, 11/12 psychomotor delay, 8/12 behavious disorder (autism, aggressiveness, hyperactivity), 7/12 sleeping disorder, 6/12 epilepsy, 5/12 hypotonia, 4/12 abnormal brain MRI	5/12 extremity abnormalities, 7/12 early onset of puberty, 1/12 shawl scrotum	Nizon et al. [11]
	Xp11.23p11.22; 0.34 - 4.6 Mb	9 affected individuals: 9/9 mild to severe ID, 8/9 speech delay, 7/9 attention deficit disorder, 7/9 motor delay, 4/9 autism	6/9 hand abnormalities (clinodactyly V and/or tapering fingers), 6/9 foot abnormalities, 4/5 early onset of menses in females, 5/9 synophrys, 5/9 constipation	Grams et al. [12]
	Xp11.23 3.1 Mb	developmental delay, stereotypic movements, poor communication, autistic features	broad thorax, wide-spaced nipples	Froyen et al. [13]
	Xp11.22; 0.26 Mb (possibly disrupting *SHROOM4* locus)	developmental delay	choanal atresia, ventricle septum defect and camptodactyly	Isrie et al. [14]
Structural: complex rearrangements	individual 1: 46,X,t(X;8)(p11.2;p22.3); individual 2: 46,X,t(X;19)(p11.2;p13.3)	two female individuals with mild to moderate ID (2/2) individual 1: seizures, aggressive behavior, delayed motor skills, normal brain MRI. individual 2: intrauterine growth delay, short stature, hypotrophic limbs	individual 1: mild hyperextensibility of fingers, mild clinodactyly V, cutaneous syndactyly II/III individual 2: foot abnormalities, both individuals: discrete facial dysmorphology individual 2: right cardiac ventricular dysplasia of unknown origin	Hagens et al. [5]
	dup(X)(p11.22p11.21); 4.6 Mb duplication and insertion	moderate developmental delay, autism spectrum disorder		Dong et al. [15]