Supplementary Table 1. Clinical details of patients with *KMT2B* mutations

Patient	Variant	Age at assessment (yr)/Sex	AAO (yr)	Duration (yr)	Phenotype	Site of on set	Latency to generalization (yr)	Current distribution of body parts affected	Severity of dystonia (motor scores)	Disability	Additional phenotype	Perinatal history	Brain MRI	Current management	Pre-genetic diagnosis
1	g.36228077A>G	12/M	10	2	Generalized dystonia	Right LL	2	Bilateral LL, UL, neck, trunk, oro- bucco- lingual dystonia, larynx and blepharospasm	BFMDRS-M 86	BFMDRS-D 19	Facial dysmorphism: thick lips, coarse facial features, chalky white dentition, thoracic kypho-scoliosis	LBW	SWI hypointensity of bilateral GP and posterior putaminal atrophy	THP 12 mg CLO 0.75 mg BAC 40 mg LD 150 mg	NBIA spectrum
2	g.36221499T>C	18/M	5	13	Generalized dystonia	Oro-mandibular	1	Oro-mandibular, lingual, jaw opening dystonia, blepharospasm, larynx, bilateral UL, LL, and trunk	BFMDRS-M 92	BFMDRS-D 26, Anarthria	Bulbous nose tip, everted ears, high arched palate, genu varum, hammer toes, drooling of saliva; varus deformity, head thrusts, impaired vertical gaze, and high-stepping gait Mild intellectual disability, psychiatric symptoms	Normal	Symmetric hypointensity of bilateral GP	THP 2 mg TET 75 mg CLO 0.5 mg LD 300 mg BOT 75 U (2 trials) QUE 100 mg VAL 300 mg ARI 2 mg	NBIA spectrum
3	g.36210725delC	43/F	3 months	43	Generalized dystonia	Neck	4	Lower face, larynx, neck, distal arm, trunk, and LL	GDS 37 UDRS 37 BFMDRS 53	BFMDRS-D 10	-	Normal	Nonspecific T2- FLAIR hyperintensities involving bilateral subcortical white matter and mild cerebellar atrophy	BAC 45 mg TET 25 mg CLO 1.5 mg	Post-encephalitis dystonia
4	g.36214765delC	16/M	11	5	Generalized dystonia	Right LL	3	Neck, lower face, jaw, trunk, bilateral UL (proximal > dista), and bilateral LL (proximal > distal)	GDS 121 UDRS 86 BFMDRS 106 Dystonic spasms present	BFMDRS 23	Microcephaly, bulbous nose tip, short stature, scoliosis, upgaze restriction and choreiform movements	Normal	Hypointensities of BL GP	THP 6 mg TET 50 mg CLO 4 mg BAC 20 mg PRA 0.750 mg Initial improvement with LD followed by worsening	DRD
5	g.36227669A>G	19/F	2 months	19	Generalized dystonia	LL	2	Oro-mandibular, larynx, lingual, neck, trunk, bilateral UL and LL	BFMDRS 87.5	BFMDRS 29 Anarthria	Delayed milestones, elongated facies, bulbous nose tip, high arched palate, left LL varus, right LL valgus, ankle contractures (Rt > Lt) Upgaze and downgaze restriction	Poor feeding in the neonatal period	Normal	TET 50 mg DIA 6 mg BAC 30 mg CLO 0.5 mg THP 12 mg	Genetically determined generalized dystonia
6	g.36223433G>A	20/M	6 months	20	Generalized dystonia	LL	6 months	Neck, trunk, UL, and LL	BFMDRS-M 61 Dystonic spasms	BFMDRS-D 21	Global developmental delay, facial dysmorphism: dolichocephaly, bulbous nose tip Psychotic symptoms	Delayed cry at birth	Normal	BAC 60 mg CLO 4 mg THP 18 mg PRA 4.5 mg QUE 25 mg BOT 200 U	Post-HIE sequalae vs. DRD
7	g.36218074T>A	13/F	4	9	Generalized dystonia	Left foot	2	Neck, trunk, bilateral arms, tongue mild, LL	GDS 54 UDRS 49 BFMDRS 56	BFMDRS 11	Microcephaly, bulbous nose tip	Normal	Normal	THP 12 mg TET 75 mg CLO 0.5 mg LD 300 mg	DRD

AAO, age at onset; MRI, magnetic resonance imaging; M, male; F, female; LL, lower limb; UL, upper limb; BFMDRS, Burke-Fahn-Marsden Dystonia Rating Scale; LBW, low birth weight; SWI, susceptibility weighted imaging; GP, globus pallidus; THP, trihexyphenidyl; CLO, clonazepam; BAC, baclofen; LD, levodopa; NBIA, neurodegeneration with brain iron accumulation; TET, tetrabenazine; BOT, botulinum toxic-A; QUE, quetiapine; VAL, valproate; ARI, aripiprazole; GDS, Global Dystonia Rating Scale; UDRS, Unified Dystonia Rating Scale; FLAIR, fluid-attenuated inversion recovery; BL, bilateral; PRA, pramipexole; DRD, dopa-responsive dystonia; Rt, right; Lt, left; DIA, diazepam; HIE, hypoxic ischemic encephalopathy.