Supplementary Table 1. Summary of the reported cases of *DHDDS* mutations

Author	Number of patients (ancestry)	Clinical phenotype	Genotype	Inheritance
Hamdan et al.[1]	5	Global developmental delay, seizures, hypotonia, short stature, tremor, ataxia, myoclonus, bradykinesia, myokymia	c.110G>A, p.(Arg37His) c.632G>A, p.(Arg211Gln)	Denovo
Kim et al.[2]	1 (Korean)	Cortical myoclonus, ataxia, intellectual disability	c.109 C>T {p. Arg37Cys}[het]	Denovo
Ware et al.[8]	1	Myoclonus, seizures, global developmental delay, visual impairment	c.632G>A p.Arg211Gln	Denovo
Wood et al.[3]	2	Global developmental delay, hypotonia, myoclonus, tremor, ataxia, orofacial dyskinesia	c.614G>A(p. Arg205Gln)	Denovo
Kim et al.[4]	1	Epilepsy, myoclonus, ataxia, tremor	c.638G>A (p. Ser213Asn) (het)	Denovo
Courage et al.[5]	3	Myoclonus (3), ataxia (3), tonic clonic seizure (2), cognitive impairment (1), bilateral deafness (1)	c.632G>A (p.Arg211Gln) c.283G>A (p.Asp95Asn) c.614G>A (p.Arg205Gln)	Denovo (1) Unknown (2)
Galosi et al.[6]	25 (22 new patients)	Global developmental delay, ataxia, myoclonus, tremor, hypotonia, joint laxity	c.632G>A, p.(Arg211Gln) c.110G>A, p.(Arg37His) c.109C>T, p.(Arg37Cys) c.698C>G, p.(Pro233Arg) c.104G>A, p.(Gly35Glu) c.638G>A, p.(Ser213Asn) c.614G>A, p.(Arg205Gln) c.124_126del, p.(Lys42del)	Denovo
Jiao et al.[7]	10	Seizures (10), myoclonus (8), tremor (6), ataxia (5), hypertonia (3), global developmental delay (10)	c.109C>T (p.R37C) c.614G>A (p.R205Q) c.632G>A (p.R211Q) c.110G>A (p.R37H) c.113G>C (p.R38P)	Denovo
Togashi et al.[9]	1 (Japanese)	Cortical myoclonic tremor, rare seizures, intension tremors, dystonia, intellectual disability	c.110G>A{p. Arg37His}	Denovo
Piccolo et al.[10]	1 (Italian)	Mild intellectual disability, impaired speech, myoclonus, chorea, stereotypies, refractory epilepsy, facial dysmorphism, skeletal anomalies	c.632G>A {p.Arg211Gln}	Denovo
Our study	1	Myoclonus, seizures, ataxia, intellectual disability, chorea	c.724 G>A (p.Glu242Lys)	Similar mutation present in father also

Parentheses at 'Clinical phenotype' and 'Inheritance' column indicate the number of patients.

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