Supplementary Table 6. Summary of all the available 33 protein sequences from the stop-gain and frameshift mutations and their involved isoforms

No.	Mutation	Phenotype	Impacted isoforms¶	Reference
1	p.R85X§	CA plus†	1, 4, 5, 6, 7, 10	Cheng et al. (2021)[8]
2	p.G185R§	CA plus†	1, 4, 5, 6, 7, 10	Izumi et al. (2013)[1]
3	p.N641Mfs*20§	Pure CA	1, 4, 5, 6, 7, 10	Our study
4	p.K1094X§	CA plus*	1, 4, 5, 6, 7, 10	Cheng et al. (2021)[8]
5	p.Q1295X§	Pure CA	1, 4, 5, 6, 7, 10	Our study
6	p.Q1498Rfs*3§	CA plus†	1, 4, 6, 7, 10	Duan et al. (2021)[7]
7	p.Q1741X§	Pure CA‡	1, 4, 7, 10	Kim et al. (2019)[5]
8	p.Q2282Sfs*3	Pure CA	1, 4, 7, 10	Yoshinaga et al. (2017)[2]
9	p.S2526Sfs*8	CA plus*	1, 4, 7, 10	Qian et al. (2022)[9]
10	p.I2995V§	Pure CA	1, 4, 7, 10, 12	Kim et al. (2019)[5]
11	p.E3053fs	Pure CA	1, 4, 7, 12	Cheng et al. (2021)[8]
12	p.R3479X§	CA plus‡	1, 4, 7	Cheng et al. (2021)[8]
13	p.R3597X	Pure CA	1, 4, 7	Izumi et al. (2013)[1]
14	p.L4224Pfs*16	Pure CA	1, 4, 7	Duan et al. (2021)[7]
15	p.D4368Tfs*2§	Pure CA	1, 4, 7	Duan et al. (2021)[7]
16	p.Y4534fs4539X	Pure CA	1, 4, 7	Izumi et al. (2013)[1]
17	p.E5112D§	Pure CA	1, 4, 7	Kim et al. (2019)[5]
18	p.E5273fs§	CA plus*	1, 4, 7	Cheng et al. (2021)[8]
19	p.H5844fs§	CA plus	1, 2, 4, 8	Cheng et al. (2021)[8]
20	p.Arg5982Ter§	CA plus	1, 2, 4, 8	Peng et al. (2018)[6]
21	p.R6156L§	Pure CAǁ	1, 2, 4, 8	Duan et al. (2021)[7]
22	p.Trp6228Ter§	CA plus	1, 2, 4, 8	Peng et al. (2018)[6]
23	p.R6380*	CA plus	1, 2, 4, 8	Kume et al. (2019)[3]
24	p.L6946fs§	CA plus‡	1, 2, 4, 8	Cheng et al. (2021)[8]
25	p.Arg7155*§	CA plus	1, 2, 4, 8	Naruse et al. (2021)[4]
26	p.Arg7190Ter	CA plus	1, 2, 4, 8	Peng et al. (2018)[6]
27	p.Q7319*§	CA plus	1, 2, 4, 8	Duan et al. (2021)[7]
28	p.R7486fs7488X§	CA plus	1, 2, 4, 8	Izumi et al. (2013)[1]
29	p.Lys7612Serfs*28§	CA plus	1, 2, 4, 8	Naruse et al. (2021)[4]
30	p.L7747Afs*28	CA plus	1, 2, 4, 8, 11	Duan et al. (2021)[7]
31	p.V7875Afs*47§	CA plus	1, 2, 3, 4, 8, 9, 11	Duan et al. (2021)[7]
32	p.T7926K§	CA plus	1, 2, 3, 4, 8, 9, 11	Duan et al. (2021)[7]
33	p.R8652*§	CA plus‡	1, 2, 3, 4, 8, 9, 11	Duan et al. (2021)[7]

The number order of these mutations are arranged according to their position from the N-terminus to the C-terminus. *the patients harbor no mutation involving in those isoforms near the C-terminus, but they exhibit ataxia plus syndromes; †the patients harbor another mutation involving in those isoforms near the C-terminus; ‡the patients harbor another splice site mutation; §heterozygous mutations or co-existing mutations on the same patient; ǁthe patients harbor a mutation involving in those isoforms near the C-terminus, but they exhibit pure cerebellar ataxia; ¶the names of nesprin-1 isoforms are based on UniProt ID: Q8NF91 (https://www.uniprot.org/, accessed on September 17, 2022). The synonyms of these isoforms: 1, Nesprin-1 Giant or Enaptin; 2, Nesprin-1 beta; 3, Nesprin-1 alpha; 4, Nesprin-1 isoform 4; 5, Nesprin-1 isoform 5; 6, Nesprin-1 isoform 6; 7, Nesprin-1 isoform 7; 8, Nesprin-1 beta 2; 9, Nesprin-1 alpha 2; 10, Drop 1; 11, Myne-1; 12, GSRP-56. Pure CA, pure cerebellar ataxia; CA plus, ataxia plus syndromes.