Sequence variant	KCNA1 Segment	Primary phenotype	Other Clinical Observations
L155P	N-terminal	EA1	
R167M; A170S; V174F; I176R; I177N	Lower S1	EA1	
F184C	Upper S1	EA1, seizures	
C185W	Upper S1	EA1, hyperthermia	short sleeper
T226A; T226M	S2	EA1	
T226K	S2	myokymia	
T226R	S2	EA1, epilepsy	sleep, apnea, sleep latency, developmental delay
R239S	S2	EA1	
A242P	S2	neuromyotonia, seizures	
P244H	S2–3 linker	myokymia	
F249C	S2–3 linker	EA1 + Hyperthermia	
F249I	S2–3 linker	EA1	
FF>F250	S2–3 linker	EA1	breathing difficulties during attacks
N255D	S3	hypomagnesemia	
N255K	S3	paroxysmal kinesigenic dyskinesia	
I262T; I262M	S3	EA1	
E283K	S3–4 linker	EA1	
V299I	S4	EA1, paradoxical myotonic congenita	
F303V; L305F; R307C	S4	EA1	
G311D; G311S; I314T	S4–5 linker	EA1	
L319R	S4–5 linker	paroxysmal kinesigenic dyskinesia, seizures	
R324T	S5	EA1, epilepsy	
E325D	S5	EA1	
L328V	S5	hypomagnesemia	
L329I	S5	EA1	
S342I	S5	EA1, seizures	
V368L	S5–S6 pore loop	Epileptic encephalopathy	severe intellectual disability
A395S	S6	EA1	
P403S	S6 proline-valine-proline motif	EA1, epilepsy	very loud breathing at night before age 2, developmental delay, moderate intellectual disability
V404I	S6 proline-valine-proline motif	EA1	mild intellectual disability
P405S	S6 proline-valine-proline motif	epileptic encephalopathy	developmental delay, macrocephaly
P405L	S6 proline-valine-proline motif	epileptic encephalopathy	pervasive developmental disorder
I407M	S6	EA1	
V408A	S6	EA1	
V408L	S6	EA1, seizures	global developmental delay
F414C	C Terminus	EA1	
F414S	C Terminus	EA1, epilepsy	
R417stop	C Terminus	EA1	

Supplementary Table 1. Genotype-phenotype correlations for human KCNA1 sequence variants.

Adapted from Paulhus, K., Ammerman, L., and Glasscock, E. (2020). Clinical Spectrum of KCNA1 Mutations: New Insights into Episodic Ataxia and Epilepsy Comorbidity. *Int J Mol Sci* 21(8): 2802.

L155P (the first variant listed) is reported for the first time in the current study.