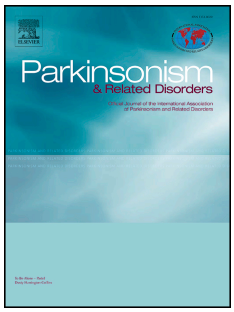
Accepted Manuscript



Phenotype variability and allelic heterogeneity in *KMT2B*-Associated disease

Toshitaka Kawarai, Ryosuke Miyamoto, Eiji Nakagawa, Reiko Koichihara, Takashi Sakamoto, Hideo Mure, Ryoma Morigaki, Hidetaka Koizumi, Ryosuke Oki, Celeste Montecchiani, Carlo Caltagirone, Antonio Orlacchio, Ayako Hattori, Hideaki Mashimo, Yuishin Izumi, Takahiro Mezaki, Satoko Kumada, Makoto Taniguchi, Fusako Yokochi, Shinji Saitoh, Satoshi Goto, Ryuji Kaji

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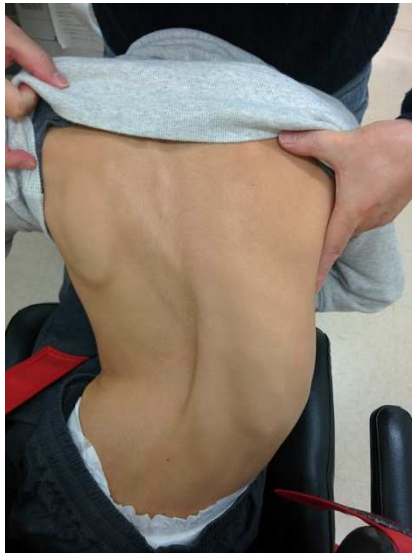
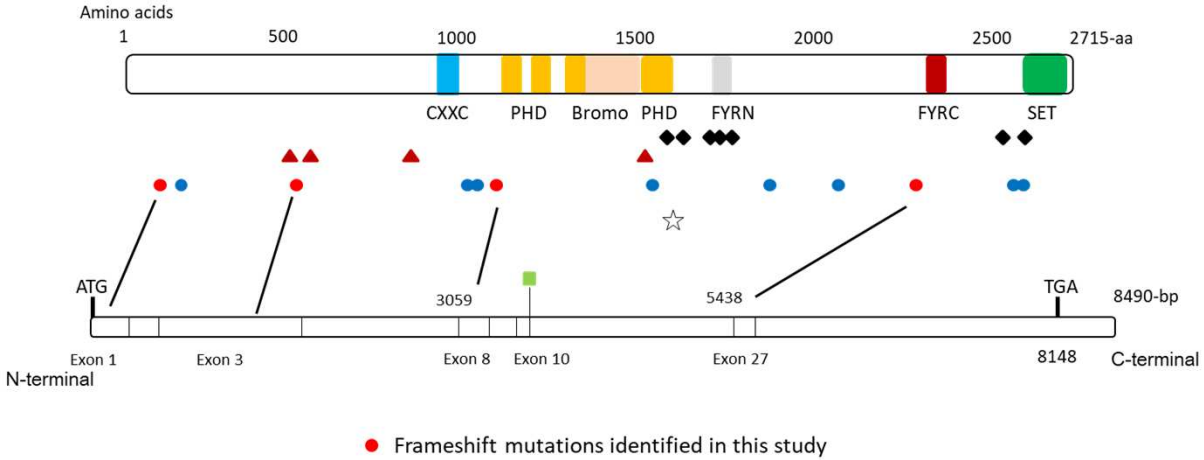
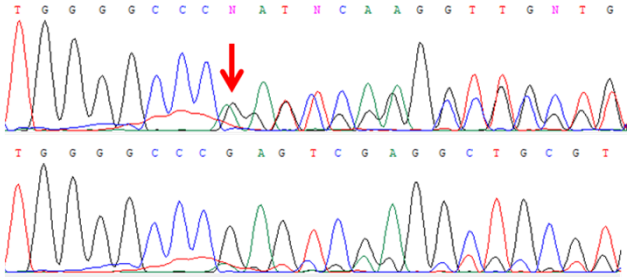


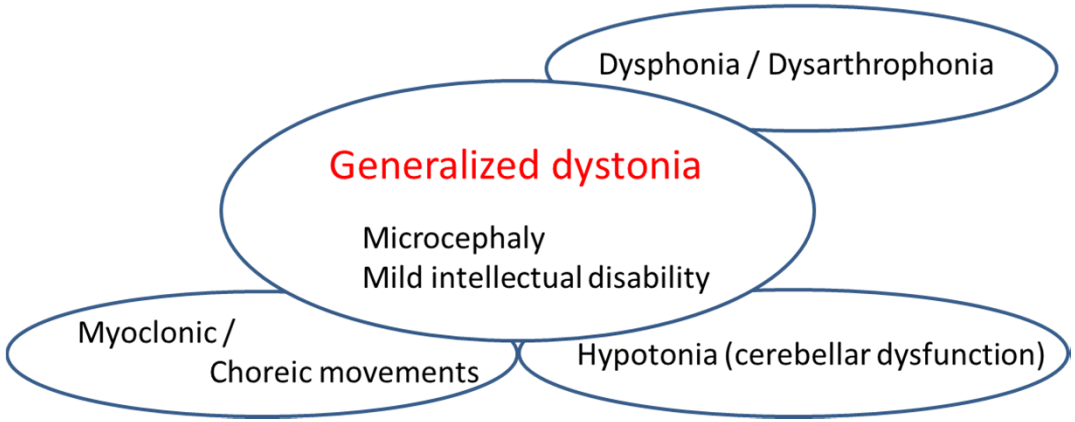


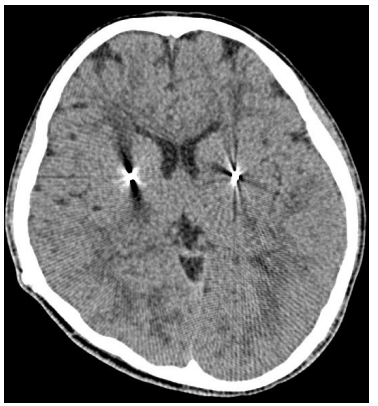
Four novel frameshift mutations in *KMT2B* identified in dystonia patients



Haploinsufficiency via Nonsense-mediated mRNA decay

Clinical features of KMT2B-associated disease





Excellent or partial response to GPi-DBS