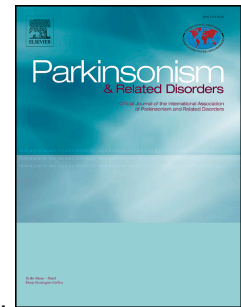


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Phenotype variability and allelic heterogeneity in *KMT2B*-Associated disease

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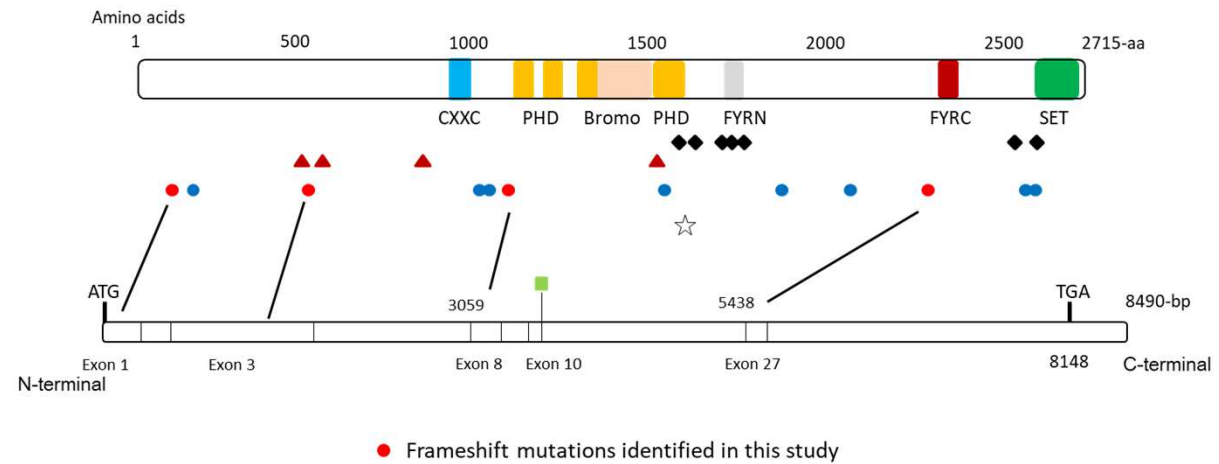
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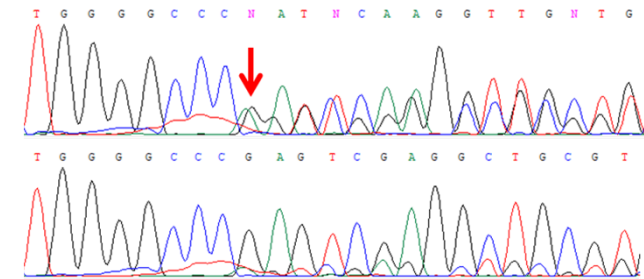
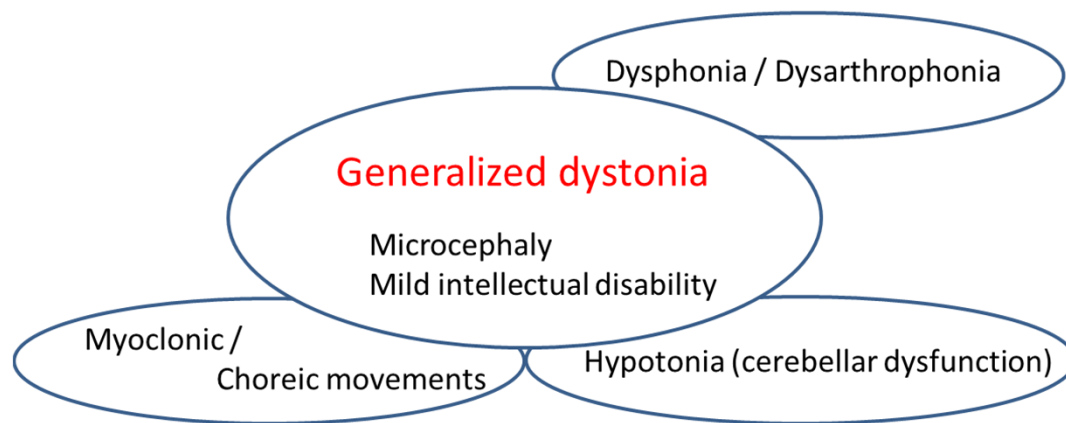
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Four novel frameshift mutations in *KMT2B* identified in dystonia patients

Clinical features of *KMT2B*-associated disease



Haploinsufficiency via Nonsense-mediated mRNA decay



Excellent or partial response to GPI-DBS

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