

Segregation and potential functional impact of a rare stop-gain PABPC4L variant in familial atypical Parkinsonism

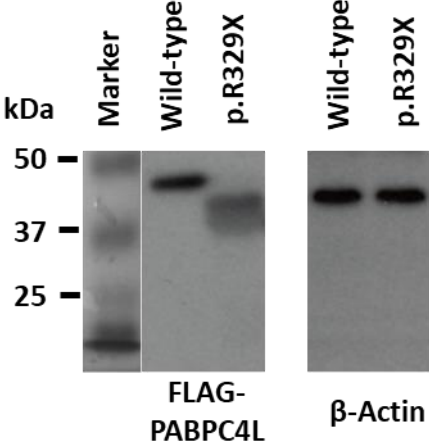
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Supplementary table: Genotyping by Sanger sequencing of the three candidate causative variants located in three different genes to validate the disease cosegregation

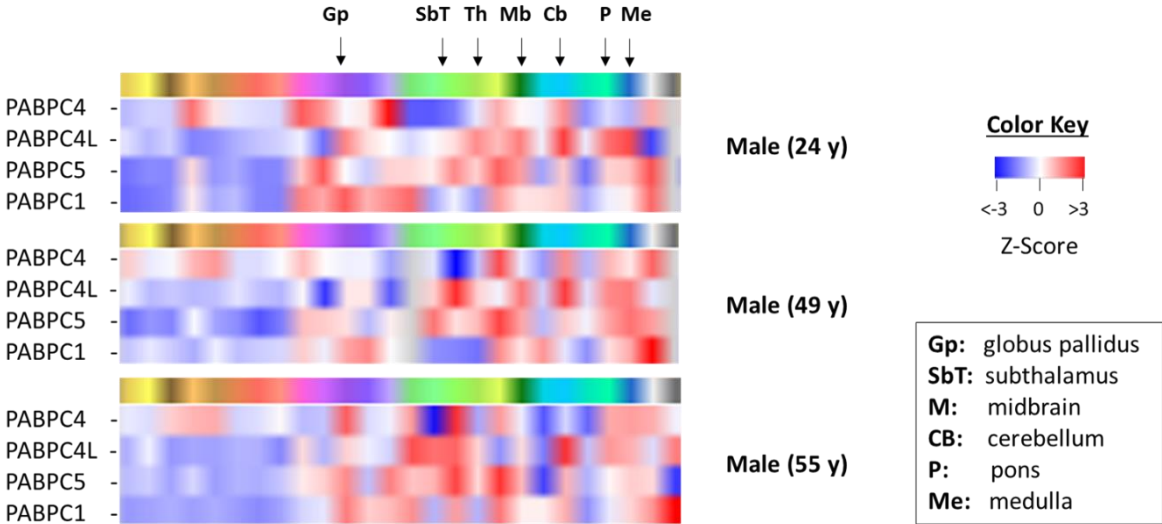
Gene		PABPC4L	ZNF292	C6orf163
Ensemble transcript ID		ENST00000421491.3	ENST00000369577.3	ENST00000388923.4
NCBI reference sequence ID		NM_001114734.2	NM_001351444	NM_001010868
UniProt ID		POCB38	O60281	Q5TEZ5
Variation (cDNA; Protein)		SNV (c.C811T:p.R271X)	SNV (c.G4918C:p.A1640P)	SNV (c.C215T:p.A72V)
GRCh37 Chrom_pos_ref_alt		4_135121364_G_A	6_87968265_G_C	6_88058596_C_T
Status	Pedigree ID	Genotyping by Sanger sequencing		
Affected	III:1*	C/T	G/C	C/T
	III:3	C/T	G/C	C/T
	IV:4	C/T	G/G	C/T
	IV:7	C/T	G/G	C/C
	IV:9	C/T	G/G	C/C
	IV:11*	C/T	G/C	C/T
Unaffected	III:2*	C/C	G/G	C/C
	III:6	C/C	G/C	C/T
	III:7	C/C	G/G	C/T
	III:8	C/C	G/G	C/C
	III:12	C/C	G/G	C/C
	III:13	C/C	G/G	C/C
	IV:1	C/C	G/G	C/C
	IV:3	C/C	G/C	C/C
	IV:5	C/C	G/C	C/C
	IV:6	C/C	G/G	C/T
	IV:8	C/C	G/G	C/C
	IV:10	C/C	G/C	C/C
	IV:12*	C/C	G/G	C/C

*Individuals for whom WGS has been performed

Supplementary figure S1: Full blots for Extended Data Figure 2c



Supplementary figure S2: Heatmap representation of transcript expression profiles of the four type I cytoplasmic PABP family members in adult human brain (image credit: Human microarray data from the Allen Brain Atlas).



Heatmaps showing normalized Z-score in "Coarse" mode representing transcript expression profiles of the type I cytoplasmic PABPs in different brain regions (multi-color ribbon). Regions with detectable PABPC4L transcript expression are highlighted. Expression data from three different donors is shown covering the age of onset of clinical symptoms in the investigated family.

Image credit: Allen Institute https://human.brain-map.org/microarray/search/show?search_type=user_selections&user_selection_mode=1.