Overview of PPMI iPSC Lines

As patient-derived cells present a valuable model for disease research, iPSCs were generated within a PPMI sub-study. PBMCs were reprogrammed into iPSCs by Cellular Dynamics International (CDI) and deposited in the PPMI Indiana University (IU) biorepository. Gene-revertant controls with mutations edited back to WT were generated by PPMI iPSC recipients and deposited back in the biorepository for community access, as per PPMI policy. Multiple clones have been generated for some patients, leading to a higher number of lines vs patients in the table in some cases. Quality control testing was performed at CDI, including karyotype analysis, identity confirmation (genotyping of 38 SNPs), pluripotency testing (gene expression of 48 mRNAs), and mycoplasma testing. Each patient who contributed PBMCs also contributed clinical, imaging, and biosample data.

All lines are available through the PPMI biospecimen request process. License fees only apply to industry users and are tiered based on company size and screening plans. Additional fibroblast-derived iPSCs from a separate study are available through PPMI as well. Visit https://www.ppmi-info.org/access-data-specimens/request-cell-lines for more information and to access the lines.

PD Status	Cohort	Subgroup	Sex	iPSC Lines		Gene-Revertant Controls	
				# Patients	# Lines	# Patients	# Lines
Not Manifesting PD	Healthy Control	N/A	Male	7	15		
			Female	7	8		
	SWEDD	N/A	Male	1	1		
			Female	0	0		
	Prodromal	RBD	Male	2	2		
			Female	0	0		
		Hyposmic	Male	3	4		
			Female	2	2		
	GBA Mutation	N370S (p.N409S)	Male	8	11		
			Female	11	16		
	LRRK2 Mutation	G2019S	Male	7	10		
			Female	11	17		
		G2019S + N2081D	Male	1	1		
			Female	1	1		
		R144G	Male	1	1		
			Female	0	0		
	GBA+LRRK2 Mutation	N370S (p.N409S) + G2019S	Male	1	1		
			Female	1	2		
		T369M (p.T408M) +	Male	1	1		
		G2019S	Female	0	0		
	SNCA Mutation	A53T (p.G209A)	Male	0	0		
			Female	2	3	1	3
Manifesting PD	Idiopathic PD	N/A	Male	36	53		
			Female	14	24		
	GBA Mutation	N370S (p.N409S)	Male	10	18		
			Female	6	10		
		T369M (p.T408M)	Male	1	1		
			Female	0	0		
		A456P (p.A495P)	Male	1	1		
			Female	0	0		
		E326K (p.E365K)	Male	0	0		
			Female	1	2		
	LRRK2 Mutation	G2019S	Male	10	15	2	2
			Female	3	7	2	2
		N2081D	Male	1	1		
			Female	0	0		
		R144G	Male	1	1		
			Female	1	2		
	SNCA Mutation	A53T (p.G209A)	Male	2	4		
			Female	1	1	1	3