NYSCF The New York Stem Cell Foundation Research Institute

BEYOND BATTEN DISEASE FOUNDATION

BBDF iPSCs

	NYSCF ID	Family Group	Relation	Sex	Age at Biopsy	Known Genotypes	Self-Reported Disease	Self-Reported Ethnicity	SNP Genotyping Ethnicity: Admixed American	African	East Asian	European	South Asian
Family Group: 882													
	051191-01-MR	882	Male Sibling	Male	25	Heterozygous for E13 c.1001G>A, p.Arg334His mutation		Hispanic, White	50%	0%	0%	50%	0%
	051198-01-MR	882	Paternal	Male	66	Heterozygous for E13 c.1001G>A, p.Arg334His mutation	hypertension, kidney disease	Hispanic	52%	0%	0%	48%	0%
	051199-01-MR	882	Paternal Uncle	Male	57		High cholesterol	Hispanic	43%	0%	0%	56%	0%
Family Group: 884													
	051205-01-MR	884	Affected Proband	Male	18	Homozygous for common (1kb) deletion	anxiety disorder, Batten disease, blindness, Epilepsy/seizures, Genetic variant - CLN3, mental depression, obsessive-compulsive disorder	White	2%	0%	1%	96%	1%
	051206-01-MR	884	Maternal	Female	48	Heterozygous for common (1kb) deletion	anxiety disorder, arthritis, Genetic variant - CLN3, mental depression, miscarriage	White	4%	0%	0%	94%	2%
Family Group: 885													
	051208-01-MR	885	Affected Proband				Batten disease, blindness, Epilepsy/seizures, genetic variant - CLN3	White	2%	0%	0%	97%	1%
Family Group: 5003													
	051104-01-MR	5003	Maternal	Female	56	Heterozygous for common (1kb) deletion		White	0%	3%	1%	93%	4%
	051105-01-MR	5003	Paternal	Male	59	Wild-Type; Carrier E13 c. 988>T, p.Val330Phe Mutation	asthma	White	3%	0%	0%	97%	0%
	051131-01-MR	5003	Affected Proband	Male	21	Compound mutation: Heterzygous for common (1kb) deletion and heterozygous for E13 c.988G>T, p.Val330Phe Mutation	Batten disease, Genetic variant - CLN3	White	0%	1%	1%	97%	2%
Family Group: 5005													
	051107-01-MR	5005	Paternal	Male	59	Heterozygous for common (1kb) deletion	asthma, blood coagulation disease	White	2%	0%	1%	96%	0%
	051106-01-MR	5005	Maternal	Female	57	Wild-Type? No known mutation		White	3%	0%	0%	96%	1%
	051132-01-MR	5005	Affected Proband	Female	22	Homozygous for common (1kb) deletion	Batten disease, Genetic variant - CLN3	White	2%	0%	0%	98%	0%
Family Group: 5009													
	051110-01-MR	5009	Maternal	Female	47	Heterozygous for common (1kb) deletion	placenta praevia	White	0%	0%	2%	98%	0%
Family Group: 5010													
	051111-01-MR	5010	Maternal	Female	48	Heterozygous for common (1kb) deletion	hypertension, polycystic ovary syndrome	White	1%	1%	1%	97%	0%
	051135-02-MR	5010	Affected Proband			Homozygous for common (1kb) deletion	attention deficit hyperactivity disorder, Autism spectrum disorder, Batten disease, Genetic variant - CLN3	White	2%	0%	1%	96%	1%
Family Group: 5014													
	051116-01-MR	5014	Maternal	Female	56	Mutation confirmation pending		White	3%	0%	5%	92%	0%
Family Group: 5020													
	051240-01-MR	5020	Maternal	Female	56	Mutation confirmation pending	bone disease, mental depression	White	0%	0%	0%	100%	0%
	051242-01-MR	5020	Affected Proband			Mutation confirmation pending	Batten disease, bone disease, Epilepsy/seizures, Genetic variant - CLN3	White	1%	0%	1%	98%	0%
Adult CLN													
	051090-01-MR	N/A	Affected Proband	Female	46		Batten disease, hypertension, neuropathy	White	1%	1%	0%	92%	6%
	051117-01-MR	N/A	Affected Proband	Male	51		Batten disease	White	2%	0%	0%	97%	1%

NOTE: Isogenic controls are in progress for the 1KB deletion from selected families and the E13 c.1001G>A, p.Arg334His mutation from family 882

* Geneotypes are confirmed by NYSCF by sequencing. All samples are genotyped with primer sets that detect the common 1kb deletion. In some samples, we additionally genotyped for status o other known mutations. ** Availability dates for gene editing are estimates based on NYSCF experince with editing other genes. However, gene editing for this locus has not been previously performed and efficiency of editing at the sites indicated is not currently known. As such, availability of gene-edited lines is subject to change and will be updated as work progresses. Culture conditions: All of our iPSC lines are derived and cultured under feeder-free conditions (see Paull D et al, Nature Methods 2015). Full culture protocol details will be provided along with the shipments. Cell lines are available upon request and subject to a fully executed MTA before shipment.