NYSCF The New York Stem Cell Foundation Research Institute



	NYSCF ID	Family Group	Relation	Sex	Age at Biopsy	Known Genotypes	Reprogramming Method	Status
Family Gro	oup: 882					·		
	051190-01-MR	882	Maternal	Female	55	Heterozygous for E13 c.1001G>A, p.Arg334His mutation	mRNA	Available
	051191-01-MR*	882	Male Sibling	Male	25	Heterozygous for E13 c.1001G>A, p.Arg334His mutation	mRNA	Available
	051192-01-MR*	882	Proband	Male	22	Homozygous for E13 c.1001G>A, p.Arg334His mutation	mRNA	Available
	051198-01-MR	882	Paternal	Male	66	Heterozygous for E13 c.1001G>A, p.Arg334His mutation	mRNA	Available
	051199-01-MR	882	Paternal Uncle	Male	57		mRNA	Available
Family Gro	oup: 884							
	051205-01-MR	884	Proband	Male	20	Homozygous for common (1kb) deletion	mRNA	Available
	051206-01-MR	884	Maternal	Female	50	Heterozygous for common (1kb) deletion	mRNA	Available
Family Gro	oup: 885							
	051208-01-MR	885	Proband	Male	24		mRNA	Available
Family Gro	oup: 5003	•						•
	051104-01-MR	5003	Maternal	Female	56	Heterozygous for common (1kb) deletion	mRNA	Available
	051105-01-MR*	5003	Paternal	Male	59	Wild-Type; Carrier E13 c. 988>T, p.Val330Phe Mutation	mRNA	Available
	051131-01-MR	5003	Proband	Male	21	Compound mutation: Heterzygous for common (1kb) deletion and heterozygous for E13 c.988G>T, p.Val330Phe Mutation	mRNA	Available
Family Gro	oup: 5005				•			•
<u> </u>	051107-01-MR	5005	Paternal	Male	59	Heterozygous for common (1kb) deletion	mRNA	Available
	051106-01-MR	5005	Maternal	Female	57	Wild-Type? No known mutation	mRNA	Available
	051132-01-MR*	5005	Proband	Female	22	Homozygous for common (1kb) deletion	mRNA	Available
Family Gro	oup: 5009				•			•
	051110-01-MR	5009	Maternal	Female	47	Heterozygous for common (1kb) deletion	mRNA	Available
Family Gro	oup: 5010	•			•	<u> </u>		•
	051111-01-MR	5010	Maternal	Female	48	Heterozygous for common (1kb) deletion	mRNA	Available
	051135-02-MR	5010	Proband	Female	13	Homozygous for common (1kb) deletion	mRNA	Available
Family Gro	oup: 5014							
	051115-01-MR	5014	Paternal	Male	61	Mutation confirmation pending	mRNA	Pending
	051116-01-MR	5014	Maternal	Female	55	Mutation confirmation pending	mRNA	Available
Family Gro	oup: 5020							
	051240-01-MR	5020	Maternal	Female	56	Mutation confirmation pending	mRNA	Available
	051242-01-MR	5020	Proband	Male	23	Mutation confirmation pending	mRNA	Available
Adult CLN								
	051090-01-MR	N/A	Proband	Female	46		mRNA	Available
	051117-01-MR	N/A	Proband	Male	50		mRNA	Available

NOTE: Isogenic controls are in progress for the lines denoted with an asterisk (*). These lines are a work in progress and estimated availability dates for the isogenic controls will be updated on this table once the edit has been confirmed and the cells are in final expansion and QC.

NOTE: Genotypes 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 of other known mutations.

NOTE: Culture conditions: 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 cells distributed. Cell lines are available upon request and subject to a fully executed MTA before shipment.