

Patient Number	AGS 1	AGS 2	AGS 3	AGS 4	AGS 5	AGS 6	AGS 7	AGS 8	AGS 9	AGS 10	AGS 11	AGS 12	AGS 13	AGS 14	AGS 15	AGS 16	AGS 17	AGS 18	AGS 19
Age of Onset	UNK	3 mo.	< 1 mo.	1 mo.	3 mo.	< 1 mo.	UNK	UNK	1 mo.	1 mo.	< 1 mo.	1 mo.	< 1 mo.	1 mo.	1 mo.	7 mo.	3 mo.	3 mo.	< 1 mo.
Sex	M	M	M	F	M	M	M	M	F	F	F	F	M	F	M	M	F	M	
Age at MRI	3 mo.	4 mo.	2 mo.	2 mo.	13 mo.	2 wks.	2 yrs.	1 mo.	5 mo.	6 mo.	1 mo.	3 mo.	1 wk.	3 mo.	4 mo.	7 mo.	2 mo.	5 mo.	4 mo.
Gene	RNase H2C	TREX1	RNaseH 2C	RNase H2B	TREX1	TREX1	RNase H2C	RNase H2C	TREX1	TREX1	TREX1	RNase H2C	TREX1	RNase H2B	SAMH D1	TREX1	TREX1	RNase H2B	SAMH D1
Outcome (Developmental Delay; G-tube dependent; ambulatory; verbal)	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	
	UNK	+	+	UNK	+	-	UNK	+	UNK	+	UNK	-	UNK	-	+	-	+	-	+
	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	
	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	

Patient Number	AGS 20	AGS 21	AGS 22	AGS 23	AXD 1	AXD 2	AXD 3	AXD 4	AXD 5	AXD 6	AXD 7	AXD 8	AXD 9	CMV 1	CMV 2	CMV 3	CMV 4
Age of Onset	<1 mo.	7 mo.	1 mo.	3 mo.	6 mo.	18 mo.	2 mo.	UNK	22 mo.	23 mo.	1 mo.	4 mo.	1 mo.	UNK	UNK	UNK	UNK
Sex	F	F	M	F	F	F	M	F	M	F	F	F	F	M	M	F	M
Age at MRI	3 mo.	9 mo.	1 mo.	4 mo.	22 mo.	23 mo.	8 mo.	3 mo.	25 mo.	27 mo.	3 mo.	4 mo.	7 mo.	3 yrs.	4 yrs.	2 yrs.	6 yrs.
Gene	SAMHD 1	RNase H2B	TREX1	TREX1	GFAP	GFAP	GFAP	GFAP	GFAP	GFAP	GFAP	GFAP	GFAP	N/A	N/A	N/A	N/A
Outcome (Developmental Delay; G-tube dependent; ambulatory; verbal)	+	+	+	+	+	+	+	+	+	+	+	+	UNK	+	+	+	+
	+	+	+	+	+	+	-	-	UNK	-	-	UNK	-	-	-	-	+
	-	-	-	-	-	-	-	-	UNK	-	-	UNK	+	+	-	-	-
	-	+	-	-	-	-	-	-	UNK	-	-	UNK	-	+	-	-	-

Patient Number	LCC 1	LCC 2	LCC 3	MLC 1	MLC 2	MLC 3	MLC 4	MLC 5	VWM 1	VWM 2	VWM 3	VWM 4	VWM 5	VWM 6	VWM 7	VWM 8	VWM 9
Age of Onset	<1 yr.	5 mo	2 mo.	3 yrs.	8 mo.	UNK	UNK	4 mo	8 yrs.	5 yrs.	3 yrs.	15 mo.	4 yrs.	2 yrs.	18 mo.	18 mo.	2½ yrs.
Sex	M	F	M	F	M	M	M	M	M	M	F	F	F	M	F	F	F
Age at MRI	8 yr.	19 mo.	2 mo.	3.5 yrs.	8 yrs.	10 mo	6 mo	5 mo.	9 yrs.	5 yrs.	2 yrs.	2 yrs.	7 yrs.	4 yrs.	3 yrs.	1½ yrs.	3½ yrs.
Gene	UNK	UNK	UNK	MLC1	MLC1	MLC1	MLC1	HEPA CAM	eIF2B5	eIF2B3	eIF2B5	eIF2B4	eIF2B5	eIF2B5	eIF2B5	eIF2B5	eIF2B5
Outcome (Developmental Delay; G-tube dependent; ambulatory; verbal)	+	+	+	UNK	UNK	UNK	+	+	+	+	+	+	+	+	UNK	UNK	UNK
	-	-	+	UNK	UNK	UNK	-	UNK	UNK	UNK	+	-	-	-	UNK	UNK	UNK
	+	-	+	UNK	UNK	UNK	+	+	-	-	-	-	-	-	UNK	UNK	UNK
	+	-	+	UNK	UNK	UNK	+	+	+	-	-	-	-	-	UNK	UNK	UNK