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Supplemental Data

Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene *SLC39A8*

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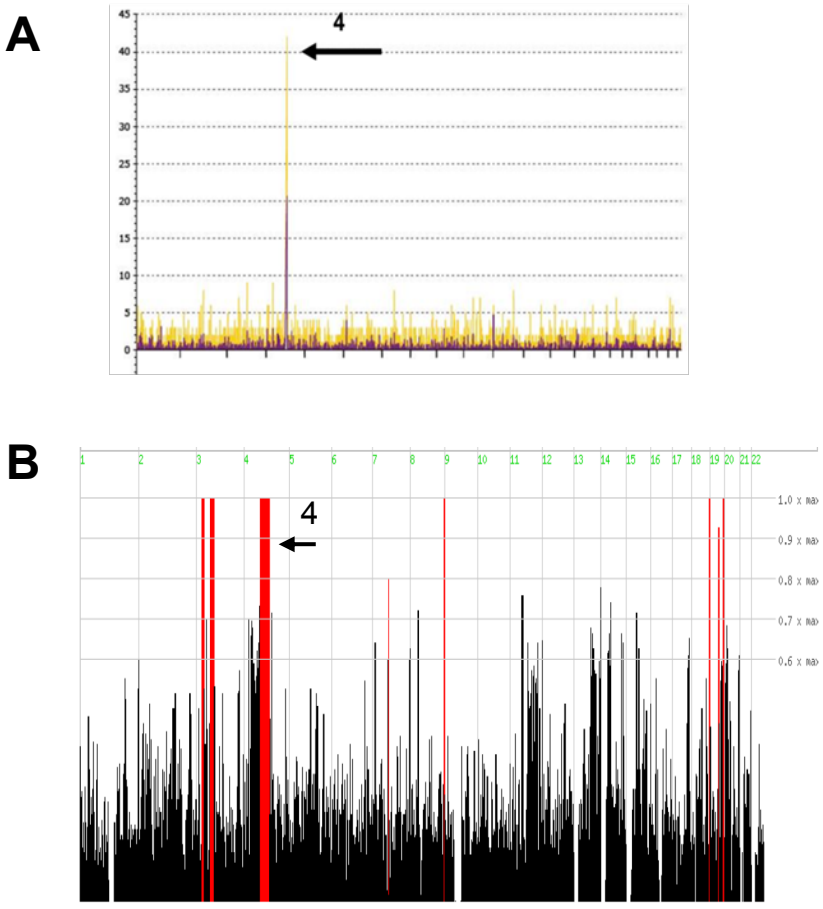

Figure S1. Genome-wide Homozygosity Mapping

(A) Identity-by-descent mapping was performed using DNA from Individuals 1, 2, and 3 (Hutterite) and GeneChip Human Mapping 10 K (Xba 2.0) and 50 K (Xba 240) arrays. We identified a single shared 11.7 Mb homozygous region at 4q22-25.

(B) Identity-by-descent mapping was performed using DNA from individuals 7 and 8 (Egyptian) and their mother using the HumanCoreExome BeadChips (Illumina) with 240 K polymorphic variants and analyzing using the web-based application HomozygosityMapper. Six candidate regions spanning a total length of 72 Mb were identified.

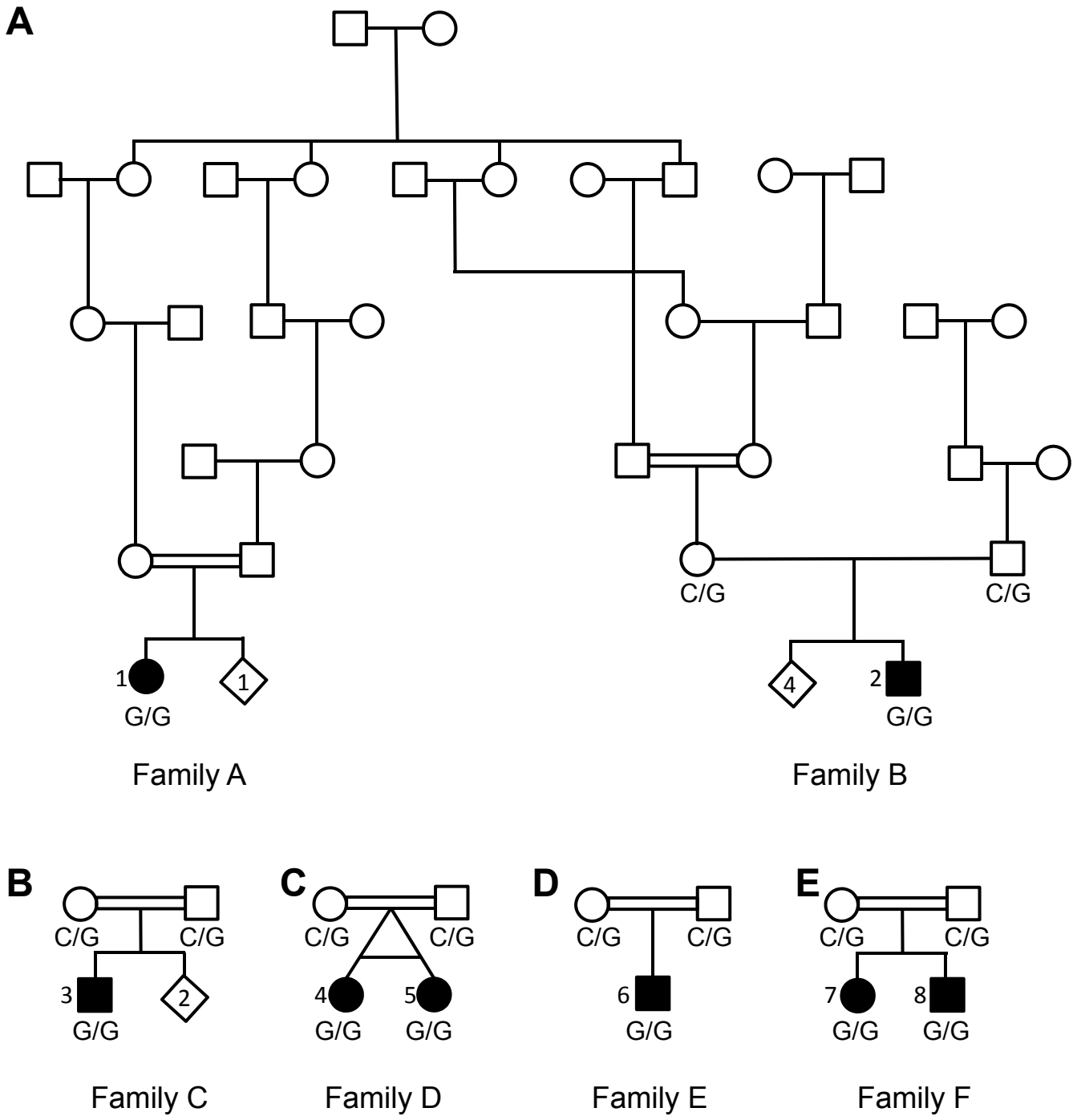

Figure S2. Sanger Segregation of the c.112G>C, p.Gly38Arg SLC39A8 Variant

(A) Pedigree and segregation indicating the relationship between Families A and B, individuals 1 and 2; (B-E) Pedigree and variant segregation of families C,D,E and F, individuals 3-8.

Table S1. Haplotype Analysis of Egyptian and Hutterite Probands Reveals Maximum 181,077bp Region

Position	dbSNP ID	Reference	Hutterite	Egyptian
Chr4:103174819	192068	G	G/G	A/A
Chr4:103180875	189215	C	C/C	T/T
Chr4:103184089	151393	A	G/G	G/G
Chr4:103189416	13114343	G	A/A	A/A
Chr4:103216782	62327949	A	G/G	G/G
Chr4:103225255	985989	A	A/A	C/C
Chr4:103225946	397692179		A/A	A/A
Chr4:103228545	1462947	G	A/A	A/A
Chr4:103265343	1462943	A	A/A	C/C
Chr4:103265708	**SLC39A8 mutation***	***C***	***G/G***	***G/G***
Chr4:103446420	4647983	TG	-	TG/TG
Chr4:103458825	230526	A	A/A	G/G
Chr4:103458877	230525	G	G/G	A/A
Chr4:103503824	4699030	G	C/C	G/G
Chr4:103506095	1598858	A	G/G	A/A
Chr4:103514445	1020760	C	G/G	C/C
Chr4:103514658	1609993	T	C/C	C/C
Chr4:103514741	4648050	T	C/C	C/C

*Region of maximum haplotype sharing extends from SNP 1462943 to SNP 4647983, a distance of 181kb. The discordant SNP 1462943 is located within the first intron of SLC39A8, demonstrating that there is no extended common haplotype across the disease gene shared by the Hutterite and Egyptian patients.