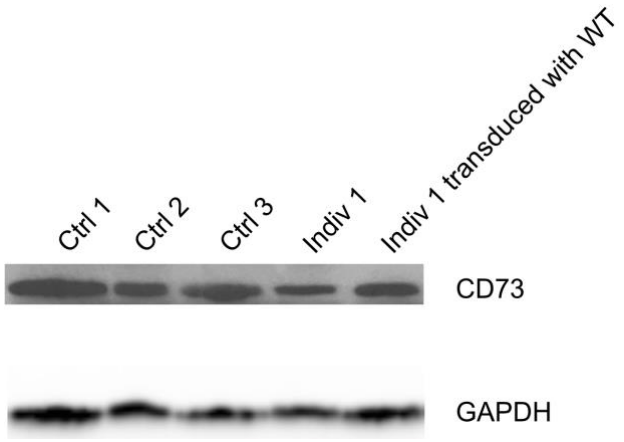
Supplementary Table 1. Bi-allelic variants in GPI biosynthesis genes in initial DOORS syndrome cohort[1].

Patient number in previous study[1]	**Gene**	**Variant 1**	**Variant 2**	**Comment**
11	*PIGM* (NM_145167.2)	c.*122A>G (3'UTR)	c.238C>G, (p.Leu80Val)	CADD score for variant 1 is 2 (variant is present in Rhesus macaque, thus might be benign), CADD score for variant 2 is 24 and affected amino acid is highly conserved
12a and 12b	*PIGB* (NM_004855.5)	c.212G>A, (p.Arg71Gln)	c.1162G>C, (p.Ala388Pro)	Pathogenic, see other publication for details [2]
13	*PIGN* (NM_176787.5)	c.1434_1434+ 1delGGinsAA	c.1434_1434+ 1delGGinsAA	Deletes splice donor, reported as pathogenic in ClinVar
21	*PIGN* (NM_176787.5)	c.548_549+ 6delAGGTTTGT	c.439A>T, (p.Lys147Ter)	Variant 1 reported as pathogenic in a publication[3] and ClinVar, second variant is loss-of-function (predicted to lead to nonsense-mediated decay)
22	*PIGN* (NM_176787.5)	c.782C>T, (p.Ser261Phe)	c.804G>C, (p.Trp268Cys)	CADD score variant 1 is 28.5, variant 2 is 33 and affected amino acids are highly conserved
23	*PIGF* (NM_002643.4)	c.515C>G, (p.Pro172Arg)	c.515C>G, (p.Pro172Arg)	This study

Supplementary figure 1. Western blot analysis of CD73 in fibroblasts.



Western blot analysis was performed for control, patient and lentivirus-rescued patient fibroblasts using a monoclonal antibody against CD73 (MyBioSource catalog # MBS668166). Decreased levels in individual one which increase upon wildtype protein expression suggests that CD73 proteins which are not GPI-anchored get degraded.

References

1. Campeau PM, Kasperaviciute D, Lu JT, et al. The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurol. 2014;13(1):44-58.
2. Murakami Y, Nguyen TTM, Baratang N, et al. Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. Am J Hum Genet. 2019;105(2):384-394.
3. Fleming L, Lemmon M, Beck N, et al. Genotype-phenotype correlation of congenital anomalies in multiple congenital anomalies hypotonia seizures syndrome (MCAHS1)/PIGN-related epilepsy. American journal of medical genetics Part A. 2016;170A(1):77-86.