

Table S3. List of nonsynonymous *DBR1* variants for which homozygous carriers have been reported in the GnomAD database, Related to Figure 1.

Position	RSID	Reference	Alternative	Protein consequence	Number of homozygotes	Allele frequency	CADD RawScore	CADD PHRED	PolyPhenCat	PolyPhenVal	SIFTcat	SIFTval
Chr3:137881084	rs36061810	C	G	p.Glu428Gln	25	0.008266	4.896726	24.9	Probably_damaging	0.968	Tolerated	0.06
Chr3:137881029	rs34889464	C	T	p.Gly446Asp	5	0.00284	1.541015	13.53	Benign	0.013	Tolerated	0.28
Chr3:137881275	rs117574050	T	C	p.Asn364Ser	17	0.002757	-0.159478	1.328	Benign	0.314	Tolerated	0.06
Chr3:137881014	rs114451412	G	A	p.Ser451Leu	3	0.001999	2.993518	22.2	Benign	0.006	Tolerated	0.07
Chr3:137890531	rs191578745	C	T	p.Arg116Gln	2	0.001144	3.323458	22.9	Benign	0.067	Tolerated	0.35
Chr3:137881320	rs146690949	T	C	p.Asp349Gly	1	0.0003935	5.589105	26.5	Possibly_damaging	0.462	Deleterious	0.02