

Additional File 1

A multi-species comparative structural bioinformatics analysis of inherited mutations in α -D-Mannosidase reveals strong genotype-phenotype correlation

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Table S1 - Non-splicing sequence mutations in the MAN2B1 gene

All mutations are those characterized in humans unless otherwise mentioned in parentheses.

No.	Mutation	Sequence change		Effect on coding sequence
		Position	Base	
1.	Missense H72L	215	CAT-CTT	His72Leu substitution
2.	H200L	599	A>T	His200Leu substitution
3.	H200N	597	C>A	His200Asn substitution
4.	R220H (cow)	662	G>A	Arg220His substitution
5.	R227W (guinea pig)	679	C>T	Arg227Trp substitution
6.	F320L (cow)	961	T>C	Phe320Leu substitution
7.	T355P	1063	ACC-CCC	Thr355Pro substitution
8.	P356R	1067	CCC-CGC	Pro356Arg substitution
9.	E402K	1204	GAG-AAG	Glu402Lys substitution
10.	S453Y	1358	A>C	Ser453Tyr substitution
11.	L518P	1553	T>C	Leu518Pro substitution
12.	W714R	2140	TGG-CGG	Trp714Arg substitution
13.	R750W	2248	CGG-TGG	Arg750Trp substitution
14.	G801D	2402	G>A	Gly801Asp substitution
15.	L809P	2426	CTG-CCG	Leu809Pro substitution
16.	R916S	2746	C>A	Arg916Ser substitution

No.	Mutation	Sequence change		Effect on coding sequence
		Position	Base	
	Nonsense			
17.	E53X	157	G>T	Termination at codon 53
18.	W77X	231	TGG-TGA	Termination at codon 77
19.	R188X	562	C>T	Termination at codon 188
20.	Y359X	1077	TAC-TAA	Termination at codon 359
21.	Y461X	1383	C>G	Termination at codon 461
22.	E563X	1687	GAG-TAG	Termination at codon 563
23.	Q639X	-	CAG-TAG	Termination at codon 639
24.	R760X	-	CGA-TGA	Termination at codon 760
	Insertions			
25.	293-294insA	Frame-shift at codon 99		Termination at codon 160
26.	322-323insA	Frame-shift at codon 108		Termination at codon 160
27.	1076–1077insA	Frame-shift at codon 359		Termination at codon 359
28.	1197-1198insA	Frame-shift at codon 399		Termination at codon 438
29.	1153–1154insCC	Frame-shift at codon 384		Termination at codon 476
	Deletions			
30.	965delAT	Frame-shift at codon 321		Termination at codon 322
31.	1815delA	Frame-shift at codon 605		Termination at codon 623
32.	1748del4 (cat)	Frame-shift at codon 583		Termination at codon 645
33.	2548delC	Frame-shift at codon 850		Termination at codon 924
34.	2660delC	Frame-shift at codon 887		Termination at codon 932