

Online Supplementary Table S1

Table S1 Pathogenic *GLA* mutations (n=46, all associated with the classic phenotype of Fabry disease) and baseline plasma α -Gal A activity levels for the 52 patients included in the 10-year agalsidase beta outcome analysis

Patient #	<i>GLA</i> Mutation	Category	Plasma α -Gal A activity (nmol/h/mL)*
1	c.1088delG	Frameshift	0.90
2	p.Asp92His	Missense	0.58
3	N/A	N/A	1.50
4	c.954_955ins5	Frameshift	0.27
5	c.893_894insG	Frameshift	0.40
6	c.975_976insG	Frameshift	1.50
7	p.Cys52Arg	Missense	1.50
8	c.1277_1278del	Frameshift	0.00
9	N/A	N/A	1.50
10	p.Pro259Leu	Missense	0.30
11	p.Trp287Gly	Missense	0.21
12	p.Asp92His	Missense	0.37
13	p.Gln327Lys	Missense	0.30
14	p.Arg112Cys	Missense	0.20
15	p.Asn272Lys	Missense	1.50
16	p.Asn272Lys	Missense	1.50
17	Large deletion	Deletion	0.40
18	c.1081_1100del	Frameshift	1.50
19	N/A	N/A	0.30
20	c.1071_1076del	Frameshift	0.60
21	c.1016_1026del	Frameshift	0.02
22	V281_T282delinsA	Unknown	0.18
23	p.Trp204*	Nonsense	0.37
24	p.Leu120Pro/p. Ala121Thr	Missense	0.00
25	p.Arg227*	Nonsense	0.14
26	p.Ser148Arg	Missense	0.40
27	p.Arg342Gln	Missense	1.50
28	p.Trp262*	Nonsense	1.50
29	c.640-2A>T	Splice site	1.50
30	Large deletion	Deletion	0.10
31	p.Arg227*	Nonsense	1.50
32	p.Arg227*	Nonsense	0.00
33	p.Gly411Asp	Missense	0.00
34	p.Arg227*	Nonsense	0.00
35	p.Gly328Glu	Missense	0.00

36	p.Ser78*	Nonsense	0.00
37	p.Asn224Asp	Missense	0.00
38	N/A	N/A	1.50
39	c.639+4A>T	Splice site	0.00
40	p.Arg342Gln	Missense	0.30
41	p.Trp226*	Nonsense	0.50
42	p.Tyr88Asp	Missense	0.53
43 ^F	p.Tyr88Asp	Missense	1.30
44	c.716_717insT	Frameshift	0.50
45	c.1175_1178delGGAA	Frameshift	0.06
46	p.Arg227*	Nonsense	0.00
47 ^F	N/A	N/A	1.10
48	c.304delT	Frameshift	0.30
49	p.Arg227*	Nonsense	0.40
50	IVS5	Splice site	0.20
51	p.Arg220*	Nonsense	0.00
52	N/A	N/A	1.50

F: heterozygous female; N/A: not available

* Normal mean, 8.1 nmol/h per mL of plasma [SD, 3.2] [n = 218]¹⁹