

Supplementary Table. Genotype, phenotype and serum Lyso-Gb3 levels of the included Fabry patients

A. Classic Males

Age at ERT start	GLA Mutation	Predicted Enzyme Protein Change	Serum Lyso-Gb3 level (ng/ml)
35	c.744_745delTA	p.F248LfsX7	92.2
40	c.744_745delTA	p.F248LfsX7	74.8
27	c.370-2A>G	Cons. Splice Site	36.8
31	c.370-2A>G	Cons. Splice Site	57.6
38	Deletion exon 2		115.0
46	c.581C>T	p.T194I	41.0
55	c.581C>T	p.T194I	46.1
39	c.581C>T	p.T194I	40.0
24	c.581C>T	p.T194I	40.4
44	c.899T>A	p.L300H	83.0
49	c.899T>A	p.L300H	40.1
14	c.125T>C	p.M42T	39.0
24	c.125T>C	p.M42T	35.3
28	c.827G>A	p.S276N	45.6
26	c.1033T>C	p.S345P	24.7
46	c.1033T>C	p.S345P	55.2
26	c.1055_1057dupCTA	p.A352_M353insT	71.7
38	c.613C>T	p.P205S	17.2

B. Classic Heterozygotes

Age at ERT start	GLA Mutation	Predicted Enzyme Protein Change	Serum Lyso-Gb3 level (ng/ml)
	c.1147_1149del	p.F383del	10.1
30	c.744_745delTA	p.F248LfsX7	5.2
	c.744_745delTA	p.F248LfsX7	8.6

	c.581C>T	p.T194I	9.7
	c.581C>T	p.T194I	10.1
22	c.581C>T	p.T194I	8.3
53	c.581C>T	p.T194I	17.8
	c.581C>T	p.T194I	6.1
52	c.581C>T	p.T194I	12.0
	c.1033T>C	p.S345P	7.8
	c.704C>A	p.Ser235Tyr	8.2
21	c.125T>C	p.M42T	8.0
17	c.125T>C	p.M42T	7.3
20	c.125T>C	p.M42T	7.5
16	c.125T>C	p.M42T	8.6
55	c.796G>T	p.D266T	8.9
28	c.640-3C>G	Cons. Splice Site	10.0
	c.154T>C	p.C52R	5.9
46	c.1033T>C	p.S345P	16.5
36	c.1235_1236delCT	p.N122IfsX8	12.6
62	c.1033T>C	p.S345P	16.1
48	c.870G>C	p.M290I	0.8
54	c.796G>T	p.D266Y	15.9
	c.1033T>C	p.S345P	7
52	c.72G>A	p.Y24X	9.3
	c.613C->T	p.W24*	13.5
43	c.365delA	p.N122IfsX8	23.1
35	c.901C>T	p.R301X	10.7

C. Later-Onset Males

Age at ERT start	GLA Mutation	Predicted Enzyme Protein Change	Serum Lyso-Gb3 level (ng/ml)
62	c.644A>G	p.N215S	5.3
	c.1196G>C	p.W399S	2.0

36	c.902G>A	p.R301Q	7.5
72	c.902G>A	p.R301Q	15

D. Later-Onset Heterozygotes

Age at ERT start	GLA Mutation	Predicted Enzyme Protein Change	Serum Lyso-Gb3 level (ng/ml)
68	c.902G>A	p.R301Q	4.9
	c.902G>A	p.R301Q	5.2
	c.337T>C	p.F113L	1.6