

**Table S1. Relationship between *GLA* gene mutations and clinical manifestations of patients of different sexes with classical and late-onset Fabry disease**

	Truncated mutation group (N=62)						Nontruncated mutation group (N=101)					
	Classical phenotype			Late-onset phenotype			Classical phenotype			Late-onset phenotype		
	All patients (N=56)	Males (n=40)	Females (n=16)	All patients (N=6)	Males (n=2)	Females (n=4)	All patients (N=80)	Males (n=60)	Females (n=20)	All patients (N=21)	Males (n=13)	Females (n=8)
<b>Acroparesthesia (limb pain)</b>	51/56 (91.07)	39/40 (97.50)	12/16 (75.00) <sup>a</sup>	0/6 <sup>d</sup>	0/2	0/4	70/80 (87.50)	58/60 (96.67)	12/20 (60.00) <sup>b</sup>	0/21 <sup>e</sup>	0/13	0/8
<b>Anhidrosis or hypohidrosis</b>	43/56 (76.79)	36/40 (90.00)	7/16 (43.75) <sup>a</sup>	0/6 <sup>d</sup>	0/2	0/4	63/80 (78.75)	52/60 (86.67)	11/20 (55.00) <sup>b</sup>	2/21 (9.52) <sup>e</sup>	2/13 (15.38)	0/8
<b>Angiokeratomas</b>	33/56 (58.93)	29/40 (72.50)	4/16 (25.00) <sup>a</sup>	0/6 <sup>d</sup>	0/2	0/4	43/80 (53.75)	38/60 (63.33)	5/20 (25.00) <sup>b</sup>	0/21 <sup>e</sup>	0/13	0/8
<b>Renal involvement</b>	40/56 (71.43)	33/40 (82.50)	7/16 (44.75) <sup>a</sup>	3/6 (50.00)	1/2 (50.00)	2/4 (50.00)	51/80 (63.75)	46/60 (76.67)	5/20 (25.00) <sup>b</sup>	12/21 (57.14)	10/13 (76.92)	2/8 (25.00)
Proteinuria	35/56 (62.50)	28/40 (70.00)	7/16 (43.75)	2/6 (33.33)	1/2 (50.00)	1/4 (25.00)	45/80 (56.25)	41/60 (68.33)	4/20 (20.00) <sup>b</sup>	12/21 (57.14)	10/13 (76.92)	2/8 (25.00)
Increased serum creatinine	24/56 (42.86)	22/40 (55.00)	2/16 (12.50) <sup>a</sup>	2/6 (33.33)	0/2	2/4 (50.00)	26/80 (32.50)	26/60 (43.44)	0/20 <sup>b</sup>	6/21 (28.57)	5/13 (38.46)	1/8 (12.50)
<b>Cardiovascular involvement</b>	40/56 (71.43)	30/40 (75.00)	10/16 (62.50)	4/6 (66.67)	1/2 (50.00)	3/4 (75.00)	60/80 (75.00)	48/60 (80.00)	12/20 (60.00)	10/21 (47.62) <sup>e</sup>	5/13 (38.46)	5/8 (62.50)
Palpitation	21/56 (37.50)	16/40 (40.00)	5/16 (31.25)	4/6 (66.67)	1/2 (50.00)	3/4 (75.00)	30/80 (37.50)	23/60 (38.33)	7/20 (35.00)	3/21 (14.29) <sup>e</sup>	1/13 (7.69)	2/8 (25.00)
Chest tightness	8/56 (14.29)	7/40 (17.50)	1/16 (6.25)	1/6 (16.67)	0/2	1/4 (25.00)	9/80 (11.25)	6/60 (10.00)	3/20 (15.00)	1/21 (4.76)	0/13	1/8 (12.50)
Chest pain	3/56 (5.36)	3/40 (7.50)	0/16	1/6 (16.67)	0/2	1/4 (25.00)	6/80 (7.50)	4/60 (6.67)	2/20 (10.00)	0/21	0/13	0/8
Hypertension	14/56 (25.00)	11/40 (27.50)	3/16 (18.75)	0/6	0/2	0/4	21/80 (26.25)	19/60 (31.67)	2/20 (10.00)	4/21 (19.05)	2/13 (15.38)	2/8 (25.00)
Left ventricular hypertrophy	24/56 (42.86)	19/40 (47.50)	5/16 (31.25)	1/6 (16.67)	0/2	1/4 (25.00)	32/80 (40.00)	28/60 (46.67)	4/20 (20.00)	6/21 (28.57)	3/13 (23.08)	3/8 (37.50)
Valve disease	12/56 (21.43)	10/40 (25.00)	2/16 (12.50)	2/6 (33.33)	1/2 (50.00)	1/4 (25.00)	19/80 (23.75)	14/60 (23.33)	5/20 (25.00)	2/21 (9.52)	1/13 (7.69)	1/8 (12.50)
Abnormal electrocardiogram	17/56 (30.36)	11/40 (27.50)	6/16 (37.50)	1/6 (16.67)	0/2	1/4 (25.00)	21/80 (26.25)	14/60 (23.33)	7/20 (35.00)	4/21 (19.05)	2/13 (15.38)	2/8 (25.00)
<b>Neuropsychiatric involvement</b>	39/56 (69.64)	30/40 (75.00)	9/16 (56.25)	2/6 (33.33)	0/2	2/4 (50.00)	57/80 (71.25)	42/60 (70.00)	13/20 (65.00)	9/21 (42.86) <sup>e</sup>	6/13 (46.15)	3/8 (37.50)
Decreased attention	22/56 (39.29)	18/40 (45.00)	4/16 (25.00)	1/6 (16.67)	0/2	1/4 (25.00)	38/80 (47.50)	30/60 (50.00)	8/20 (40.00)	7/21 (33.33)	5/13 (38.46)	2/8 (25.00)
Anxiety and depression	22/56 (39.29)	17/40 (42.50)	5/16 (31.25)	2/6 (33.33)	0/2	2/4 (50.00)	35/80 (43.75)	30/60 (50.00)	5/20 (25.00)	1/21 (4.76) <sup>e</sup>	1/13 (7.69)	0/8
Dizziness	24/56 (42.86)	17/40 (42.50)	7/16 (43.75)	1/6 (16.67)	0/2	1/4 (25.00)	34/80 (42.50)	28/60 (46.67)	6/20 (30.00)	3/21 (14.29) <sup>e</sup>	1/13 (7.69)	2/8 (25.00)
Headache	2/56 (3.57)	2/40 (5.00)	0/16	0/6	0/2	0/4	0/80	0/60	0/20	0/21	0/13	0/8
Cerebral infarction	11/56 (19.64)	8/40 (20.00)	3/16 (18.75)	0/6	0/2	0/4	11/80 (13.75)	10/60 (16.67)	1/20 (5.00)	0/21	0/13	0/8
Cerebral hemorrhage	3/56 (5.36)	2/40 (5.00)	1/16 (6.25)	0/6	0/2	0/4	4/80 (5.00)	4/60 (6.67)	0/20	0/21	0/13	0/8
<b>Digestive system involvement</b>	40/56 (71.43)	31/40 (77.50)	9/16 (56.25)	3/6 (50.00)	0/2	3/4 (75.00)	55/80 (68.75)	44/60 (73.33)	11/20 (55.00)	5/21 (23.81) <sup>e</sup>	2/13 (15.38)	3/8 (37.50)

Nausea and vomiting	19/56 (33.93)	15/40 (37.50)	4/16 (23.00)	1/6 (16.67)	0/2	1/4 (25.00)	19/80 (23.75)	18/60 (30.00)	1/20 (5.00) <sup>b</sup>	3/21 (14.29)	1/13 (7.69)	2/8 (25.00)
Stomachache	19/56 (33.93)	14/40 (35.00)	5/16 (31.25)	0/6	0/2	0/4	25/80 (31.25)	21/60 (35.00)	4/20 (20.00)	2/21 (9.52) <sup>e</sup>	1/13 (7.69)	1/8 (12.50)
Diarhea	28/56 (50.00)	24/40 (60.00)	4/16 (25.00) <sup>a</sup>	2/6 (33.33)	0/2	2/4 (50.00)	44/80 (55.00)	38/60 (63.33)	6/20 (30.00) <sup>b</sup>	2/21 (9.52) <sup>e</sup>	2/13 (15.38)	0/8
Constipation	12/56 (21.43)	8/40 (20.00)	4/16 (25.00)	1/6 (16.67)	0/2	1/4 (25.00)	14/80 (17.50)	7/60 (11.67)	7/20 (35.00)	0/21 <sup>e</sup>	0/13	0/8
<b>Eye involvement</b>	42/56 (75.00)	34/40 (85.00)	8/16 (50.00) <sup>a</sup>	4/6 (66.67)	1/2 (50.00)	3/4 (75.00)	56/80 (70.00)	46/60 (76.67)	10/20 (50.00)	8/21 (38.10) <sup>e</sup>	3/13 (23.08)	5/8 (62.50)
Vision loss	32/56 (57.14)	27/40 (67.50)	5/16 (31.25) <sup>a</sup>	4/6 (66.67)	1/2 (50.00)	3/4 (75.00)	37/80 (46.25)	31/60 (51.67)	6/20 (30.00)	6/21 (28.57)	2/13 (15.38)	4/8 (50.00)
Corneal vortex opacity	23/56 (41.07)	18/40 (45.00)	5/16 (31.25)	0/6 <sup>d</sup>	0/2	0/4	28/80 (35.00)	22/60 (36.67)	6/20 (30.00)	0/21 <sup>e</sup>	0/13	0/8
Retinal vascular tortuosity	15/56 (26.79)	12/40 (30.00)	3/16 (18.75)	0/6	0/2	0/4	14/80 (17.50)	11/60 (18.33)	3/20 (15.00)	2/21 (9.52)	1/13 (7.69)	1/8 (12.50)
Conjunctival vascular tortuosity	12/56 (21.43)	9/40 (22.50)	3/16 (18.75)	0/6	0/2	0/4	10/80 (12.50)	8/60 (13.33)	2/20 (10.00)	1/21 (4.76)	0/13	1/8 (12.50)
Posterior capsular opacification	14/56 (25.00)	10/40 (25.00)	4/16 (25.00)	0/6	0/2	0/4	9/80 (11.25)	6/60 (10.00)	3/20 (15.00)	0/21	0/13	0/8
<b>Ear involvement</b>	40/56 (71.43)	33/40 (82.50)	7/16 (43.75) <sup>a</sup>	2/6 (33.33)	1/2 (50.00)	1/4 (25.00)	45/80 (56.25)	33/60 (55.00)	12/20 (60.00)	7/21 (33.33)	4/13 (30.77)	3/8 (37.50)
Tinnitus	34/56 (60.71)	27/40 (67.50)	7/16 (43.75)	2/6 (33.33)	1/2 (50.00)	1/4 (25.00)	35/80 (43.75)	25/60 (41.67)	10/20 (50.00)	5/21 (23.81)	3/13 (23.08)	2/8 (25.00)
Deafness	4/56 (7.14)	4/40 (10.00)	0/16	0/6	0/2	0/4	6/80 (7.50)	6/60 (10.00)	0/20	1/21 (4.76)	1/13 (7.69)	0/8
Hearing loss	32/56 (57.14)	26/40 (65.00)	6/16(37.50)	2/6 (33.33)	1/2 (50.00)	1/4 (25.00)	31/80 (38.75) <sup>c</sup>	24/60 (40.00)	7/20 (35.00)	6/21 (28.57)	4/13 (30.77)	2/8 (25.00)
<b>Respiratory system involvement</b>	13/56 (23.21)	10/40 (25.00)	3/16 (18.75)	0/6	0/2	0/4	16/80 (20.00)	13/60 (21.67)	3/20 (15.00)	2/21 (9.52)	1/13 (7.69)	1/8 (12.50)
Dyspnea	7/56 (12.50)	6/40 (15.00)	1/16 (6.25)	0/6	0/2	0/4	5/80 (6.25)	3/60 (5.00)	2/20 (10.00)	0/21	0/13	0/8
Chronic bronchitis	7/56 (12.50)	4/40 (10.00)	3/16 (18.75)	0/6	0/2	0/4	9/80 (11.25)	7/60 (11.67)	2/20 (10.00)	2/21(9.52)	1/13 (7.69)	1/8 (12.50)
COPD	2/56 (3.57)	2/40 (5.00)	0/16	0/6	0/2	0/4	4/80 (5.00)	4/60 (6.67)	0/20	0/21	0/13	0/8
<b>Osteoporosis</b>	13/56 (23.21)	10/40 (25.00)	3/16 (18.75)	0/6	0/2	0/4	18/80 (22.50)	12/60 (20.00)	6/20 (30.00)	2/21 (9.52)	1/13 (7.69)	1/8 (12.50)

Note: <sup>a</sup> the difference between female and male patients in the truncated mutation classical group was statistically significant ( $P<0.05$ ); <sup>b</sup> the difference between female and male patients in the truncated mutation classical group was statistically significant ( $P<0.05$ ); <sup>c</sup> the difference between all patients in the nontruncated mutation classical group and all patients in the truncated mutation classical group was statistically significant ( $P<0.05$ ); <sup>d</sup> the difference between all patients in the truncated mutation late-onset group and all patients in the truncated mutation classical group was statistically significant ( $P<0.05$ ). <sup>e</sup> the difference between all patients in the nontruncated mutation late-onset group and all patients in the nontruncated mutation classical group was statistically significant ( $P<0.05$ ).