

Supplemental Table 3. Differences in the levels of lyso-Gb₃ and analogues in plasma and urine and Gb₃ in urine in different classes of *GLA* mutations. Lyso-Gb₃ and analogue values are expressed as median (Q1-Q3). Plasma lyso-Gb₃(+50) data could not be analyzed due to many results below the limit of detection. LOD: limit of detection. n=number of patients with available specimens/total number of patients.

| | ALL | | | | NEVER TREATED PATIENTS | | | | TREATED PATIENTS (BEFORE TREATMENT SPECIMENS) | | | |
|--------------------------------------|----------------------|----------------------|-----------|---------|------------------------|-----------------------|-----------|---------|---|--------------------|-----------|---------|
| | Missense | Nonsense | Deletions | p-value | Missense | Nonsense | Deletions | p-value | Missense | Nonsense | Deletions | p-value |
| Plasma (nM) | | | | | | | | | | | | |
| n | 47/49 | 5/6 | 1/2 | | 17/18 | 3/3 | - | | 30/31 | 2/3 | 1/2 | |
| Lyso-Gb ₃ (-28) | 0.35(0.07-0.79) | 0.13(0.11-0.62) | 1.81 | 0.25 | 0.09 (<lod -0.25) | 0.13 (0.11-0.62) | - | 0.25 | 0.58 (0.28-0.89) | 0.85 (0.07-1.63) | 1.81 | 0.28 |
| Lyso-Gb ₃ (-2) | 3.11(2.09-5.10) | 4.09(2.37-4.81) | 12.89 | 0.22 | 2.49 (1.66-2.99) | 4.09 (2.37-4.81) | - | 0.26 | 4.25 (2.57-5.56) | 5.03 (1.72-8.33) | 12.89 | 0.24 |
| Lyso-Gb ₃ | 11.19(7.19-16.00) | 10.44(7.88-18.63) | 26.73 | 0.26 | 7.68 (5.74-10.98) | 10.44 (9.40-13.18) | - | 0.26 | 12.36 (9.44-18.46) | 15.21 (6.36-24.06) | 26.73 | 0.29 |
| Lyso-Gb ₃ (+16) | 0.70(0.25-1.18) | 0.43(<lod -0.97) | 4.17 | 0.21 | 0.23 (<lod -0.42) | 0.43 (<lod -0.97) | - | 0.46 | 0.88 (0.58 -1.92) | 1.31 (<lod-2.61) | 4.17 | 0.23 |
| Lyso-Gb ₃ (+18) | 0.34(<lod-0.84) | 0.33(<lod -1.01) | 0.61 | 0.83 | <lod (<lod -0.40) | 1.01 (<lod -1.69) | - | 0.18 | 0.53 (<lod -0.86) | 0.17 (<lod -0.33) | 0.61 | 0.49 |
| Lyso-Gb ₃ (+34) | <lod (<lod -0.46) | <lod (<lod -0.66) | 1.10 | 0.19 | <lod (<lod -<lod) | <lod (<lod -0.89) | - | 0.15 | 0.36 (<lod -0.69) | 0.33 (<lod -0.66) | 1.10 | 0.26 |
| Lyso-Gb ₃ (+50) | <lod (<lod-<lod) | <lod (<lod-<lod) | <lod | - | <lod (<lod -<lod) | <lod (<lod -<lod) | - | - | <lod (<lod -<lod) | <lod (<lod -<lod) | <lod | - |
| Urine (pM/mM cre) | | | | | | | | | | | | |
| n | 28/49 | 3/6 | 1/2 | | 17/18 | 2/3 | - | | 11/31 | 1/3 | 1/2 | |
| Lyso-Gb ₃ (-28) | 11.53(5.54-24.33) | 5.99(4.37-12.54) | 30.59 | 0.29 | 7.08(3.62-14.4) | 9.27(5.99-12.54) | - | 0.84 | 20.88(10.83-25.4) | 4.37 | 30.59 | 0.14 |
| Lyso-Gb ₃ (-12) | 50.6 (42.69-113.50) | 56.39(41.30-58.78) | 281.96 | 0.23 | 44.76(35.58-81.15) | 57.59(56.39-58.78) | - | 0.42 | 110.36(45.88-133.24) | 41.3 | 281.96 | 0.09 |
| Lyso-Gb ₃ (-2) | 16.13(5.69-29.19) | 16.05(5.81-16.24) | 40.89 | 0.42 | 6.76(2.06-16.13) | 16.15(16.05-16.24) | - | 0.41 | 23.53(16.77-30.7) | 5.81 | 40.89 | 0.14 |
| Lyso-Gb ₃ | 25.83 (5.19-39.76) | 2.76(.00-14.48) | 22.45 | 0.24 | 17.16(2.72-26.56) | 8.62(2.76-14.48) | - | 0.65 | 35.89(10.8-45.81) | 0 | 22.45 | 0.23 |
| Lyso-Gb ₃ (+14) | 17.42(9.17-35.45) | 17.92(8.97-19.79) | 78.28 | 0.27 | 12.02(5.12-21.06) | 18.86(17.92-19.79) | - | 0.41 | 27.3(18.36-47.58) | 8.97 | 78.28 | 0.09 |
| Lyso-Gb ₃ (+16) | 148.52(97.44-251.77) | 123.77(63.30-189.09) | 545.93 | 0.20 | 105.46(80.95-148.52) | 156.43(123.77-189.09) | - | 0.42 | 224.04(155.17-412.28) | 63.3 | 545.93 | 0.04 |
| Lyso-Gb ₃ (+34) | 89.82(60.44-146.33) | 95.98 (51.92-114.49) | 247.47 | 0.24 | 67.9(44.04-101.32) | 105.24(95.98-114.49) | - | 0.29 | 146.33(89.82-171.23) | 51.92 | 247.47 | 0.09 |
| Lyso-Gb ₃ (+50) | 45.85(40.44-65.28) | 45.86(30.48-67.48) | 121.29 | 0.29 | 41.46(32.73-45.85) | 56.67(45.86-67.48) | - | 0.14 | 65.28(50.29-96.98) | 30.48 | 121.29 | 0.15 |
| Gb ₃ (µg/mmol creatinine) | 21.81(10.2-44.41) | 22.9(17.11-23.5) | 32.34 | 0.82 | 15.10 (10.10-49.00) | 23.20(22.9-23.5) | - | 0.42 | 31.02(20.56-43.39) | 17.11 | 32.34 | 0.66 |