

Supplementary Materials

The Expression and Secretion Profile of TRAP5 Isoforms in Gaucher Disease

Supplemental Table 1. Demographics, genotypes, and clinical characteristics of bone disease in patients with GD at the time of the study partially have been previously published [18, 21]. GD, Gaucher disease; ERT, enzyme replacement therapy; SRT, substrate reduction therapy; EM, Erlenmeyer; AVN, avascular necrosis.

| nt No. | Age yrs. | Sex M/F | ERT/SRT duration (yrs.) | Genotype | Chito nmol/hr/mL | Lyso Gb-1 ng/mL | CCL18 ng/mL | Bone Pain | Bone marrow infiltration | EM-flask deformity | Pathologic fractures/ Bone surgery | AVN |
|---------------------------------------|----------|---------|-------------------------|--------------|------------------|-----------------|-------------|-----------|--------------------------|--------------------|------------------------------------|-----|
| No bone complication. Total 11 | | | | | | | | | | | | |
| P1 | 50 | F | ERT 5-10 | N370S/N370S | 52 | 2.7 | 60.9 | No | Yes | Yes | No/No | No |
| P2 | 25 | F | ERT >10 | L444P/L444P | 273 | 15.9 | 180.4 | No | No | No | No/No | No |
| P3 | 18 | F | ERT 5-10 | N370S/F213I | 215 | 7.9 | 54.5 | Yes | Yes | Yes | No/No | No |
| P4 | 41 | F | SRT | N370S/R463C | 160 | 10.9 | 159.4 | No | Yes | Yes | No/No | Yes |
| P5 | 29 | F | SRT | N370S/N370S | 619 | 6.7 | 75 | No | No | No | No/No | No |
| P6 | 56 | F | ERT >10 | N370S/N370S | 40 | 1.1 | 41 | Yes | Yes | Yes | No/No | Yes |
| P7 | 37 | F | ERT >10 | N370S/W381X | 133 | NA | 41 | Yes | Yes | No | No/No | Yes |
| P8 | 49 | F | ERT >10 | N370S/L444P | 44 | 4 | 68 | Yes | Yes | No | No/Yes | Yes |
| P9 | 29 | F | ERT 5-10 | /L444P | 34 | 7 | 105 | No | No | No | No/No | No |
| P10 | 21 | M | SRT | N370S/L444P | 228 | 13.3 | 76 | No | Yes | No | No/No | No |
| P11 | 33 | M | ERT 1-5 | R48Q/L444P | 1022 | 11 | 1087.9 | Yes | Yes | Yes | No/No | No |
| Osteopenia (OSN). Total 14 | | | | | | | | | | | | |
| P12 | 29 | F | ERT 1-5 | N370S/N370S | 1104 | 29.6 | 170.1 | No | Yes | Yes | No/Yes | No |
| P13 | 61 | F | ERT >10 | N370S/N370S | 103 | 2.5 | 210.2 | Yes | Yes | Yes | Yes/No | Yes |
| P14 | 32 | F | ERT 5-10 | N370S/L444P | 127 | 10.7 | 63.1 | No | No | No | No/No | No |
| P15 | 52 | F | SRT | N370S/N370S | 42 | 1 | 37.3 | No | Yes | No | No/No | No |
| P16 | 40 | F | SRT | N370S/R463C | 5 | 1 | 79.1 | Yes | Yes | No | No/No | No |
| P17 | 41 | F | naive | N370S/N370S | 1194 | 49 | 407 | No | No | No | Yes/No | No |
| P18 | 51 | F | SRT 5-10 | N370S/L444P | 314 | 11.7 | 289.2 | No | No | No | No/No | No |
| P19 | 74 | F | SRT 1-5 | N370S/N370S | 283 | 1.4 | 191 | Yes | Yes | Yes | No/No | No |
| P20 | 38 | F | ERT >10 | N370S/N370S | 110 | 35 | 276 | Yes | No | No | No/No | No |
| P21 | 61 | F | 1 year | N/A | 2080 | 375 | 662 | Yes | No | No | No/No | No |
| P22 | 65 | M | SRT 5-10 | N370/N370 | 44 | 1.5 | 56.1 | Yes | Yes | No | No/No | No |
| P23 | 18 | M | naive | N370S/N370S | 164 | 5.1 | 97.5 | No | Yes | No | No/No | No |
| P24 | 37 | M | ERT 1-5 | N370S/ V394L | 493 | 12 | 103 | Yes | No | No | Yes | No |
| P25 | 20 | M | ERT >10 | L444P/P266A | 118 | 7.5 | 58.2 | No | No | No | No/No | No |
| Osteoporosis (OSR) Total 14 | | | | | | | | | | | | |
| P26 | 57 | F | SRT 5-10 | L444P/R502C | 118 | 3.4 | 95.5 | Yes | Yes | Yes | No/No | No |
| P27 | 54 | F | ERT >10 | N370S/L444P | 663 | 129.5 | 131.4 | Yes | Yes | Yes | No/No | Yes |
| P28 | 20 | F | ERT 1-5 | N370S/L444P | 665 | 36 | 145.5 | Yes | No | No | No/No | No |
| P29 | 62 | F | SRT | N370S/N370S | 7 | 1 | 61.2 | Yes | Yes | Yes | No/No | No |
| P30 | 30 | F | ERT >10 | N370S/N370S | 116 | 17.6 | 120.1 | No | Yes | No | No/No | No |
| P31 | 63 | F | naive | N370S/R496H | 38 | 2.5 | 45.7 | Yes | Yes | Yes | No/No | No |
| P32 | 44 | F | ERT/SRT >10 | L444P/R493C | 530 | 42.7 | 178.5 | Yes | Yes | No | Yes/Yes | No |

| | | | | | | | | | | | | |
|-----|----|---|----------|-------------|-------|------|-------|-----|-----|-----|---------|-----|
| P33 | 68 | F | ERT 5-10 | N370S/R463C | 63 | 2.5 | 123.8 | Yes | No | Yes | No/Yes | Yes |
| P34 | 45 | F | ERT 5-10 | N370S/N370S | 19.96 | 5.4 | 13.8 | Yes | No | Yes | No/Yes | No |
| P35 | 46 | F | SRT | N370S/N370S | 97.8 | 39 | 331 | Yes | No | Yes | No | Yes |
| P36 | 55 | M | SRT 0-1 | N370S/L444P | 376 | 15.2 | 120.2 | Yes | No | No | Yes/Yes | No |
| P37 | 36 | M | ERT >10 | N370S/Y412X | 546 | 85.6 | 562.9 | Yes | Yes | Yes | Yes/Yes | Yes |
| P38 | 39 | M | ERT 5-10 | N370S/N370S | 57 | 39.5 | 836.5 | No | No | No | No/No | No |
| P39 | 42 | M | SRT 5-10 | N370S/N370S | 254 | 35.8 | 171 | No | Yes | No | No/Yes | No |

Supplemental Table 2. The sequences of gene primers that have been used for real-time PCR.

| Gene | Primers Sequence | |
|------------|------------------|---------------------------|
| GAPDH | Forward | GAGAGGGCTGGGCTCATTTCGAC |
| | Reverse | CCATCCACAGTCTTCTGGGTGGCAG |
| CCL2/MCP-1 | Forward | GCAATCAATGCCCCAGTCA |
| | Reverse | TGCTGCTGGTGATTCTTCTATAGCT |
| ACP5 | Forward | CGTATTCTCTGACCGCTCCC |
| | Reverse | TCTTGAAGTGCAGGCGGTAG |

Supplemental Table 3. Prevalence of osteopenia and osteoporosis in female and male patients with Gaucher disease by age group. NB – normal bone mineral density, OSN – osteopenia, OSR – osteoporosis.

| | Female | | | Males | | |
|---------------------|--------|-----|-----|-------|-----|-----|
| | NB | OSN | OSR | NB | OSN | OSR |
| Patients number | 9 | 10 | 10 | 2 | 4 | 4 |
| Minimum age (Years) | 18 | 29 | 20 | 21 | 18 | 36 |
| Maximum age (Years) | 56 | 74 | 68 | 33 | 65 | 55 |
| Mean age (Years) | 37 | 48 | 49 | 27 | 35 | 43 |
| Std. Deviation | 13 | 14 | 15 | 8.5 | 22 | 8.4 |
| Std. Error of Mean | 4.3 | 4.6 | 4.8 | 6.0 | 11 | 4.2 |