

Supplemental Table 5 The creatinine-corrected concentrations of SNAG- Δ^5 -CA, SNAG- Δ^5 -CG, SNAG- Δ^5 -CT, S7B- Δ^5 -CA and S7O- Δ^5 -CA in the urine of all subjects.

Sample	Age (Years)	Sex	SNAG- Δ^5 -CA (ng/mg Cr)	SNAG- Δ^5 -CG (ng/mg Cr)	SNAG- Δ^5 -CT (ng/mg Cr)	S7B- Δ^5 -CA (ng/mg Cr)	S7O- Δ^5 -CA (ng/mg Cr)	Total (ng/mg Cr)
NPC1	0.25	Male	29900	37200	42800	44900	48400	203000
NPC2	16	Female	184	1150	66.4	695	114	2210
NPC3	13	Male	291	237	15.2	182	121	846
NPC4	12	Female	900	834	110	395	269	2510
NPC5	5	Female	719	593	89.1	554	364	2320
NPC6	22	Female	186	309	49.3	514	130	1190
NPC7	15	Male	293	194	75.4	468	119	1150
NPC8	14	Female	989	316	78.2	574	207	2160
NPC9	3	Male	21100	9390	3940	21000	9630	65100
NPC10	8	Male	0.561	0.821	0.182	1170	257	1950
NPC11	36	Male	1160	863	28.3	739	173	2960
NPC12	24	Female	543	1190	60.5	1070	174	3040
NPC13	12	Female	31600	81100	3570	52700	8870	178000
NPC14	26	Female	430	450	62.8	776	128	1850
NPC15	18	Female	613	1260	85.7	998	155	3110
NPC16	11	Female	1280	1670	120	1550	394	5010
NPC17	30	Female	N.Q.	3.46	N.Q.	748	122	875
NPC18	0.333	Male	44200	32600	22800	33200	148000	281000
NPC19	0	Male	13600	547	3960	9820	92300	120000
NPC20	11	Female	4030	5050	560	4340	680	14700
NPC21	4	Female	1820	1160	74.7	2020	796	5870
NPC22	0.67	Male	235	284	19.0	342	89.1	969

NPC23	30	Female	80.4	100	15.3	142	35.8	374
NPC24	0.6	Male	131000	132000	14900	83400	19500	381000
NPC25	6	Female	680	896	210	383	144	2310
NPC26	48	Male	655	730	52.4	394	83.3	1910
NPC27	0.5	Female	9550	4290	8380	3790	6540	32600
NPC28	9	Female	2330	1880	205	3360	627	8410
Healthy control1	0.25	Female	97.4	55.4	92.7	80.9	173	499
Healthy control2	0.33	Male	68.5	57.5	52.8	125	50.1	354
Healthy control3	0.33	Female	145	165	76.1	55.6	44.5	486
Healthy control4	0.5	Male	41.6	31.2	44.7	59.8	31.0	208
Healthy control5	0.5	Male	49.5	36.4	59.8	99.7	24.9	270
Healthy control6	0.5	Male	65.1	46.6	21.4	37.0	43.3	213
Healthy control7	1	Male	34.0	16.7	7.34	24.2	13.6	95.8
Healthy control8	1	Male	30.9	24.5	7.10	17.9	7.43	87.8
Healthy control9	1	Female	5.63	13.7	4.25	93.0	14.6	131
Healthy control10	2	Male	12.8	13.5	4.36	17.3	5.85	53.8
Healthy control11	2	Female	21.7	21.4	6.32	21.4	5.53	76.2
Healthy control12	2	Female	10.8	18.9	22.9	24.8	6.75	84.1
Healthy control13	2	Male	16.4	19.0	8.02	19.1	6.27	68.8
Healthy control14	2	Male	23.0	14.8	6.37	16.6	5.43	66.1
Healthy control15	2	Male	12.7	16.6	4.36	21.1	9.46	64.2
Healthy control16	35	Female	13.2	34.2	5.22	8.41	5.80	66.8
Healthy control17	28	Female	20.0	16.0	1.45	13.6	5.92	57.0
Healthy control18	25	Female	8.73	11.6	2.14	5.45	4.12	32.0
Healthy control19	23	Female	10.3	13.8	1.44	5.65	2.92	34.1
Healthy control20	24	Male	15.5	17.3	1.13	11.6	4.65	50.1
Healthy control21	30	Male	10.8	16.5	1.37	7.41	4.34	40.4

Healthy control22	30	Male	12.7	18.1	2.52	9.84	4.21	47.4
Healthy control23	36	Male	4.85	7.70	1.75	5.58	2.53	22.4
Healthy control24	21	Male	3.33	10.2	0.906	7.23	1.97	23.7
Healthy control25	22	Male	8.10	11.9	1.98	7.80	4.34	34.1
Healthy control26	23	Male	N.Q.	N.Q.	N.Q.	7.82	1.96	9.78
Healthy control27	44	Female	11.3	20.2	2.26	7.38	4.17	45.3
Healthy control28	29	Male	6.47	15.4	3.36	7.58	3.38	36.2
Healthy control29	47	Male	25.4	29.5	1.93	15.6	3.81	76.2
Healthy control30	31	Male	5.91	9.46	0.748	8.93	1.50	26.5
Healthy control31	36	Male	14.2	18.3	3.25	16.4	8.16	60.3
Healthy control32	36	Male	20.4	51.5	4.80	12.1	2.71	91.5
Healthy control33	34	Male	8.35	9.85	3.05	5.97	3.17	30.4
Healthy control34	22	Male	4.53	6.98	2.32	19.3	5.19	38.3
Healthy control35	23	Female	16.3	25.9	3.07	8.95	4.49	58.7
Healthy control36	21	Male	6.79	13.6	1.54	11.7	3.50	37.1
Healthy control37	21	Female	5.64	6.06	0.305	7.44	3.04	22.5
Healthy control38	26	Male	7.68	15.4	1.98	5.41	2.47	33.0
CTX01	42	Female	2.0	6.5	0.9	3.1	1.3	13.9
CTX02	56	Female	11.7	7.9	4.8	16.7	3.5	44.5
CTX03	50	Female	12.5	8.5	4.5	7.5	4.0	36.9
GAU01	18	Female	82.9	55.4	4.4	25.2	0.9	168.7
GAU02	20	Female	0.1	0.5	0.5	23.7	8.2	33.1
GAU03	15	Female	58.4	21.7	7.8	39.3	11.2	138.4
HUN01	18	Male	5.3	11.6	3.9	19.6	3.7	44.1
HUN02	8	Male	44.6	35.9	5.2	18.3	11.2	115.3
SLOS01	22	Male	67.0	85.9	33.4	328.0	77.0	591.3

N.Q., Quantitation was not carried out because it was below the lower limit of determination; Cr, creatinine; NPC, Niemann–Pick disease type C; SNAG- Δ^5 -CA, 3 β -Sulfoxy-7 β -*N*-acetylglucosaminy-5-cholenoic acid; SNAG- Δ^5 -CG, Glycine-amidated 3 β -sulfoxy-7 β -*N*-acetylglucosaminy-5-cholenoic acid; SNAG- Δ^5 -CT, Taurine-amidated 3 β -sulfoxy-7 β -*N*-acetylglucosaminy-5-cholenoic acid; S7B- Δ^5 -CA, 3 β -Sulfoxy-7 β -hydroxy-5-cholenoic acid; S7O- Δ^5 -CA, 3 β -Sulfoxy-7-oxo-5-cholenoic acid; **CTX, Cerebrotendinous Xanthomatosis; GAU, gaucher disease; HUN, hunter disease; SLOS, Smith-Lemli-Opitz syndrome.**