

Table S1 – Cohort of NP-C1 patients. Correspondence with patients in Jahnova et al [3] and full genotypes.

Patient No.	Patient No. in Jahnova et al.	Clinical Phenotype	<i>NPC1</i> cDNA sequence change	Deduced <i>NPC1</i> protein change	UC nmol/mg protein	CE nmol/mg protein
1	8. T2	Early infantile	1812dupT/3558delC	A605Cfs*1/ A1187Rfs*54	202.4±2.3	35.0±3.0
2	-	Early infantile	2746_2748 delAAT/3734_3735 delCT	N916del/ P1245Rfs*12	140.9±9.4	18.0±5.0
3	7. T2	Early infantile	3182T>C/3591+1G>A	I1061T/splice	148.3±5.4	21.2±1.2
4	11. T2	Early infantile	3557G>A/3614C>A	R1186H/T1205K	330.3±7.2	21.3±0.9
5	10. T2	Late infantile	826T>C/3557G>A	Y276H/R1186H	196.7±15.3	19.7±4.4
6	10. T3	Late infantile	826T>C/3557G>A	Y276H/R1186H	195.6±7.8	25.5±1.8
7	1. T6	Late infantile	3557G>A/3557G>A	R1186H/R1186H	212.2±33.7	18.2±5.5
8	12. T3	Late infantile	3557G>A/3557G>A	R1186H/R1186H	141.5±0.4	20.2±3.8
9	9. T3	Late infantile	3019C>G/3557G>A	P1007A/R1186H	74.7±2.3	24.0±6.3
10	17. T4	Juvenile	2861C>T/3557G>A	S954L/R1186H	47.6±10.4	14.2±0.8
11	-	Juvenile	2861C>T/3557G>A	S954L/R1186H	60.4±10.4	16.6±1.2
12	20. T4	Juvenile	2861C>T /3557G>A	S954L/R1186H	78.4±2.7	43.2±21.7
13	12. T4	Juvenile	2861C>T/3557G>A	S954L/R1186H	64.0±8.2	15.0±0.7
14	9. T4	Juvenile	1421C>T/2072C>T	P474L/P691L	43.6±8.8	16.1±1.5
15	13. T4	Juvenile	2849T>G/3019 C>G	V950G/P1007A	37.0±4.0	25.3±1.1
16	11. T4	Juvenile	1232G>C/3019C>G	R411P/P1007A	61.4±1.4	18.2±0.7
17	19. T4	Juvenile	1232G>C/2861 C>T	R411P/S954L	92.3±19.2	19.9±0.4
18	-	Juvenile	527T>G /2861C>T	L176R/S954L	66.6±4.2	19.0±1.9
19	14. T4	Juvenile	352_353delAG/3019C>G	Q119Vfs*7 /P1007A	97.2±10.2	27.9±1.7
20	-	Juvenile	808delG/2861C>T	V270Sfs*39 /S954L	55.3±12.5	13.0±0.3
21	-	Juvenile	1723delG/2861C>T	E575Rfs*17 /S954L	89.2±0.8	28.6±2.3
22	15. T4	Juvenile	1812dupT/2861C>T	A605Cfs*1 /S954L	52.6±1.6	13.4±0.8
23	3. T5	Adolescent/Adult	2712delG/2861C>T	Q905Rfs*30 /S954L	53.8±11.8	20.2±0.3
24	18. T4	Adolescent/Adult	2780C>T/2780C>T	A927V/A927V	44.1±1.9	30.7±4.1
25	5. T5	Adolescent/Adult	2780C>T/2780C>T	A927V/A927V	47.8±4.5	23.0±0.1
26	6. T5	Adolescent/Adult	2780C>T/2780C>T	A927V/A927V	61.0±10.1	18.4±3.6
Control levels (n=3)		<i>wt</i>	<i>wt</i>	<i>wt</i>	58,4 (39,6-77,6)	19,0 (10,9-25,1)

