

SUPPLEMENTAL INFORMATION:

Histone deacetylase inhibitors correct the cholesterol storage defect in most NPC1 mutant cells

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Supplemental Table S1.

p	Vorinostat treatment	Panobinostat treatment
<0.001	P166S, H215R, P532L, G535V, A605V, E612D, R615L, Y634C, G673V, L724P, Y825C, I858V, G910S, Q921P, R934Q, S940L, W942C, V950M, S954L, R958Q, D948N, P1007A, G1012D, G1034C, T1036M, R1059Q, I1061T, R1077Q, Y1088C, L1102F, W1145R, G1162A, V1165M, L1191F, T1205K, V1212L, L1213F, G1240R, L1244P	P166S, R404Q, P532L, G535V, A605V, Y634C, L648H, G673V, L724P, V780G, Q921P, I923V, R934Q, S940L, W942C, D948N, V950M, S954L, R958Q, P1007A, T1036M, R1059Q, I1061T, L1191F, T1205K, V1212L, L1213F, G1240R
0.001-0.01	C113R, D242H, S636F, L648H, V780G, G992R, 3565-66 insG, 3741-44del	V378A, E612D, R615L, S666N, S734I, G1012D, R1077Q, Y1088C, G1162A, V1165M, A1187V, L1244P
0.01-0.05	S734I, D874V, I923V, A1151T, G1195V	C113R, H215R, S636F, Y825C, I858V, D874V, G910S, G992R, L1102F, G1034C, G1195V, 3741-44del
>0.05	L80V, V378A, R404Q, H510P, R518Q, S666N, P691S, A1187V	L80V, D242H, H510P, R518Q, P691S, W1145R, A1151T, 3565-66 insG

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The effects of 10 μ M Vorinostat or 50 nM Panobinostat were tested on 60 different NPC1 mutations. DMSO was used as a solvent control. Filipin fluorescence images of the transfected cells were analyzed to obtain an LSO value as illustrated in Figure 6. Data from 15-25 fields with ~1-5 transfected cells per field were obtained and an LSO value per field was obtained. Statistical significance was measured by t-test with two-tailed distribution and two-sample equal variance (homoscedastic) type using Microsoft Excel software. The responses of are grouped according to the statistical significance of the difference between HDACi-treated and solvent treated cells.