

Supplemental online Figure Legends

Supplemental Figure 1 online : A. Bone marrow aspiration shows large foamy histiocytes with vacuolated cytoplasm (Leishman stain; x40) B. A typical storage cell with finely vacuolated cytoplasm (Leishman stain; x100).

Supplemental Figure 2 online: Exome sequencing of the trio followed by alignment of paired-end reads (gray horizontal bars) to the human genome assembly 37 (hg19) showed no reads for exon 2 and exon 3 of *NPC2* gene in the proband (red arrows). Parents had reduced coverage for both the exons (proband-upper panel, mother-middle panel, father-lower panel).

Supplemental Figure 3 online: Multiplex ligation-dependent probe amplification using the SALSA MLPA probe sets P193 identified a homozygous deletion of exon 2 and 3 in the proband (blue bars: control, green bars: test) and heterozygous carrier status for the same in both the parents (proband-upper panel, mother-middle panel, father-lower panel).