

Supplementary Figure 1. Sanger sequencing confirmed two compound heterozygous mutations

of *B3GALNT2* gene: (A) c.1421_c.1423delCTC (p.P474del) and c.979G>A (p.D327N) in patients 1;

(B) c.48dupG (p.L17fs) and c.1183G>A (p.G395R) in patient 2. All the parents were carriers.

