



Fig. S1. (a) Sanger sequencing results of the identified compound heterozygous *LAMB3* mutations in 3 families. (b) Microfluidic electrophoresis and smear analysis demonstrated the ratio of mutant (Mut)/wild-type (WT) transcript level caused by the 3 splice site mutations: 40:1 for c.373-9T>A, 1:3–1:7 for c.629-12T>A, and 3:1 for c.2137+1G>T. While only wild-type transcript was seen in the healthy control.