**Supplementary Figure 1.** Pedigrees of patients harboring novel variants and undergoing segregation study. The arrows indicate probands.

|  |  |
| --- | --- |
| Patient No.9 *SH3TC2* c.929dup, p.Ser312Valfs\*18  c.929dup/  WT  WT/  c.929dup  c.929dup/  c.929dup | Patient No.32 *PRDM12* c.570+2T>G, p.? /   c.796A>C, p.Thr266Pro  c.570+2T>G/  WT  WT/  c.796A>C  c.570+2T>G/  c.796A>C |