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

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