

Supplementary Figure S1. The age at clinical observation among different genotype groups.

(A). The types of *FBNI* mutations of all probands: inframe mutations (including missense mutations, inframe deletions/insertion, inframe intragenic deletion/duplication), PTC (including frameshift mutations, nonsense mutations, one allele deletion), and splicing mutations. Complex mutations were excluded. The age distribution was shown in violin graph. There is no age difference among groups (Kruskal-Wallis test, $P = 0.860$).

(B). The location of *FBNI* mutations of all probands: N-terminal site, C-terminal site, and middle site (Kruskal-Wallis test, $P = 0.788$).

(C). The amino acid changes of *FBNI* missense mutations: Cys-creating, Cys-eliminating, and changes involving other amino acids (Kruskal-Wallis test, $P = 0.217$).

(D). The protein domains of *FBNI* missense mutations: EGF-like domain, cb-EGF-like domain, TGF β domain, 4-Cys motif LTBP-like domain, Hybrid module (Kruskal-Wallis test, $P = 0.217$).

cb, calcium binding; EGF, epidermal growth factor; LTBP, latent transforming growth factor β binding protein; PTC, Premature termination codons; TGF β , transforming growth factor β binding protein.