



**Fig. S2.** (a) Exome sequencing identified a *PDGFRB* (c.1994T>C, p.Val665Ala) variant (red frame). (b) Sanger sequencing confirmed the *PDGFRB* variant in the proband. (c-e) Sanger sequencing showed no *PDGFRB* variants in the proband's father, mother, sister, respectively.