



Figure S1: The clinical feature and amino acid sequences of two brothers of family A and skin defect of proband of family D

The fundus photograph or anterior segmental photography of two brothers from family A (A-D) and the occipital skin defect of proband D (F). The old brother presented with visual loss and unocular blindness. The fundus images of right eye showed tessellated changes and retinal detachment occurred soon after presence (A). The anterior segmental image showed corneal leukoma of left eye (B). The fundus photograph of younger brother showed tessellated fundus appearance and macular dysplasia (C, D). The result of amino acid sequences of c.2992g>A from nine species reported in the mutation taster is shown (E). A focal occipital skin defect was observed on the occipital scalp in the proband of family D (F).