

Cerebral hypomyelination associated with biallelic variants of *FIG4*

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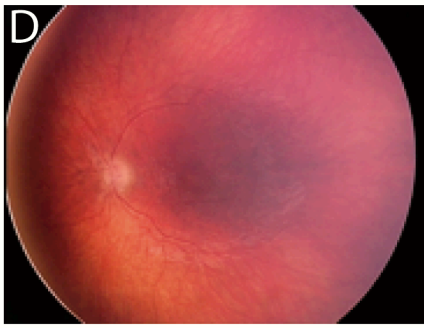
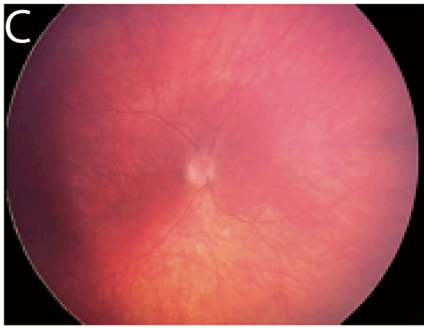
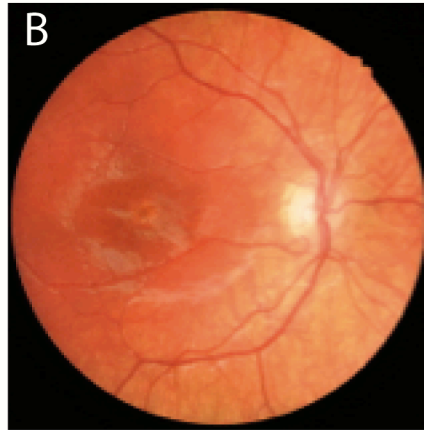
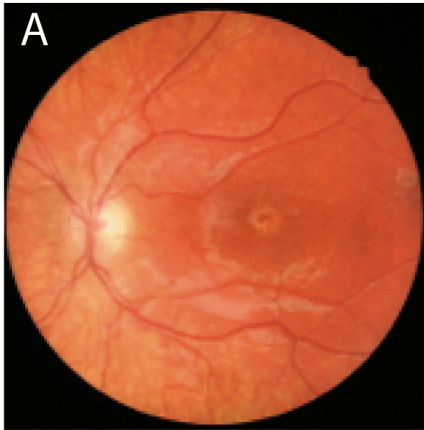
Supplemental Figure S1. Bone defects in Patient 3. X-rays show severe scoliosis and thin shafts of the tubular bones with over tubulation in Patient 3, who also exhibits thin metacarpals, coxa valga and dolicocephaly.

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Supplemental Figure S2. Ocular defects in affected individuals from Family 4.

(A, B) Fundus photography of the left and right eye, respectively, of Patient 7 at 6 years of age, showing bilateral mild Bull's eye maculopathy and temporal optic nerve pallor. (photographed 18/06/18). (C, D) Fundus photography of the right and left eyes, respectively, of Patient 6 at eight years of age, demonstrating depigmented macular change, generalized retinal atrophy, vessel attenuation and optic atrophy. (E, F) Spectral Domain (Spectralis – Heidelberg Optical) Optical Coherence Tomogram line scan of left macula, performed in lateral position under anaesthetic, of Patient 6 at 4 years, 9 months of age. Note macula outer retina / RPE pigmentary disturbance and loss of inner retinal nerve fibre layers. (G) Optical coherence tomogram of normal macula for comparison. (on following page)



OS, BAF 30° ART + OCT 20.0° (5.2 mm) ART (17) Q: 35 [HS]

