

Fig S1. Fundus photograph and OCT of SRF964. a and b: Right and left fundus pictures. A few bone-spicule and obvious salt-pepper changes were observed and retinal vessels were attenuated. c and d: Right and left eye OCT images. The IS/OS signal was disrupted.

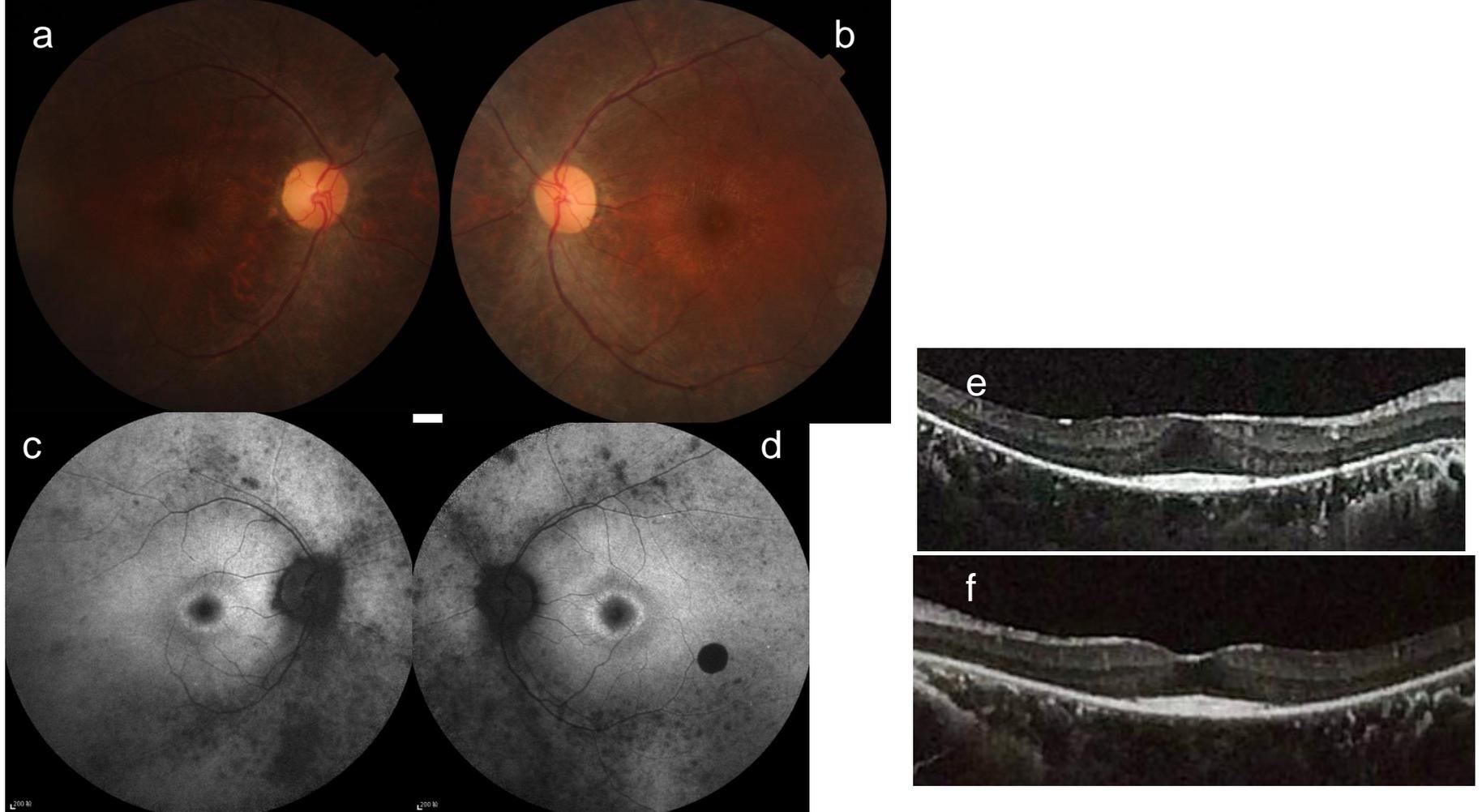


Fig S2. Fundus photograph and autofluorescence images of SRF1186. a and b: Right and left fundus pictures show Retina pigment epithelium (RPE) layer is atrophy with gray pigments in the mid-peripheral retina. c and d: Right and left eye autofluorescence images show patchy hypofluorescence changes at peripheral retina and hyperfluorescence ring around fovea. e and f: right and left OCT images showed preserved IS/OS in the central fovea and epi-retinal membrane of the right eye.

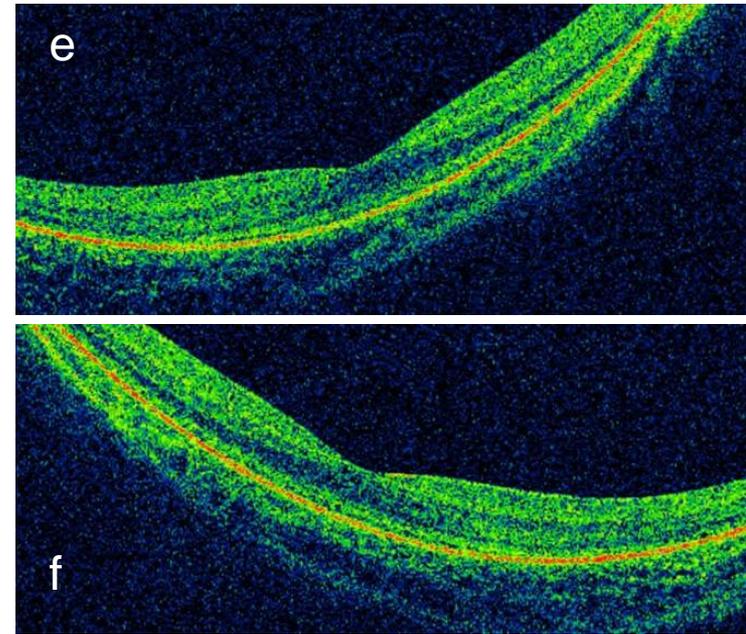
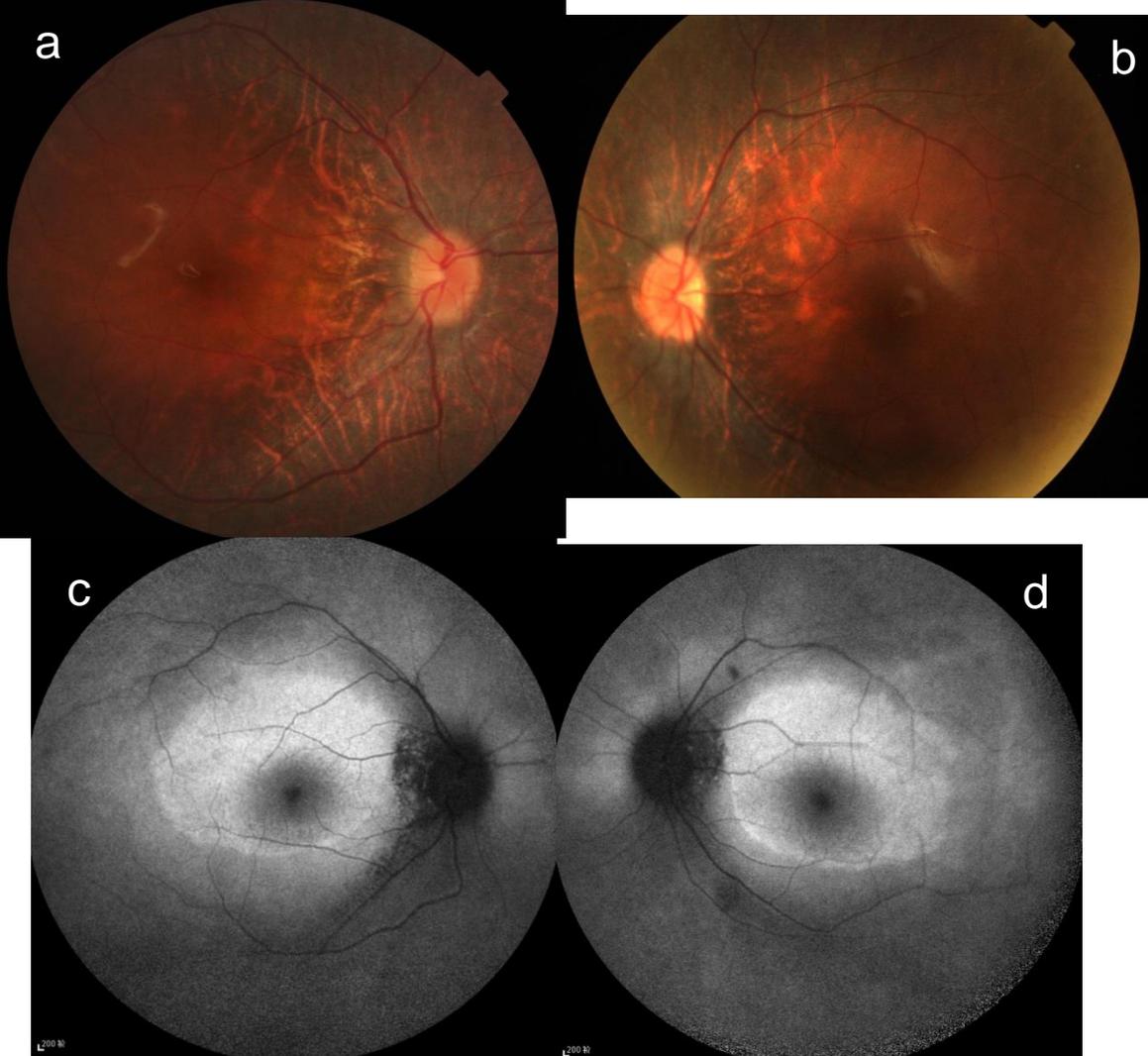


Fig S3. Fundus photograph, autofluorescence and OCT of SRF117. a and b: Right and left fundus. Retina pigment epithelium (RPE) layer is atrophy with visualization of choroid vessels. c and d: Right and left eye autofluorescence images show hyperfluorescence ring in the macular area. e and f: Right and left eye OCT image showed disruption of IS/OS and increased foveal retina thickness.



Fig S4. X-ray radiography of SRF71's hands. The patient has no cone shaped epiphysis or polydactyly, which are typical skeletal anomalies of MZSDS and JATD.