Supplementary Figure 1. Optical coherence tomography of the macular fovea in five participants with aniridia: (Grade 0) Normal characteristics of the foveal structure, including a: foveal pit (arrow); b: extrusion of plexiform layers (arrow); c: outer nuclear layer widening (arrow); d: cone outer segment lengthening (arrow). Retinal layers specified by arrows and abbreviations on left side of the image.

(Grade 1) Foveal hypoplasia marked by absent extrusion of plexiform layers (crossed b; stippled arrow), otherwise by features as in grade 0. This patient also showed elevation of retinal pigment epithelium because of drusen (black arrows).

(Grade 2) Foveal hypoplasia with all characteristics of grade 1 except presence of a foveal pit (crossed a; stippled arrow). (Grade 3) Foveal hypoplasia including all
features of grade 2 apart from lengthening of the cone outer segments (crossed d; stippled arrow). (Grade 4) Foveal hypoplasia presenting all hallmarks noticed in grade 3 other than widening of the outer nuclear layer (crossed c; stippled arrow).

ELM, external limiting membrane; EZ, ellipsoid zone; GCL, ganglion cell layer; INL, inner nuclear layer; IPL, inner plexiform layer; IZ, interdigitation zone; ONL, outer nuclear layer; OPL, outer plexiform layer; RNFL, retinal nerve fiber layer; RPE-BrM; retinal pigment epithelium-Bruch’s membrane.