**Appendix 5: Summary of fundus appearance and retinal imaging**

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| **Patient** | **Fundus** | **Fundus autofluorescence** | **OCT** |
| MEE1a | Extensive well-demarcated macular RPE atrophy with preserved foveal island; vessels mildly attenuated; periphery normal | Macula: Tiny foveal AF island with surrounding hypoAF  Periphery: Normal | Diffuse outer retinal atrophy |
| MEE2a | Foveal pigment mottling with RPE changes in inferior arcade; vessels mildly attenuated; periphery with nasal atrophy and bone spicules | Macula: Foveal hyperAF; hypoAF inferior arcade  Periphery: Nasal hypoAF | Fovea preserved with surrounding PR loss |
| MEE3 | Coarse pigment deposits in macula; vessels attenuated; periphery diffusely atrophic with bone spicules | Macula: Diffuse hypoAF  Periphery: Diffuse hypoAF | Diffuse outer retinal atrophy |
| MEE4 | Extensive well-demarcated macular RPE atrophy with preserved foveal island; vessels attenuated; periphery normal | Macula: Tiny foveal AF island with surrounding hypoAF  Periphery: Normal | Diffuse outer retinal atrophy |
| MEE5 | Extensive well-demarcated macular RPE atrophy with preserved foveal island; vessels attenuated; periphery diffusely atrophic with rare bone spicules | Macula: Foveal AF island with surrounding hypoAF;  Periphery: Diffuse hypoAF | Diffuse outer retinal atrophy with foveal island |
| MEE7 | Diffuse macular granularity; vessels attenuated; periphery atrophic with rare bone spicules | Not available | Fovea preserved with surrounding PR loss |
| MEE8 | Fovea-sparing macular atrophy; vessels attenuated; periphery atrophic with bone spicules | Macula: Parafoveal hypoAF ring  Periphery: Diffuse hypoAF | Bare preservation of foveal EZ with surrounding outer retinal loss |
| MEE9 | Macula atrophic; vessels attenuated; periphery atrophic with bone spicules | Macula: Diffuse hypoAF  Periphery: Diffuse hypoAF | Diffuse outer retinal atrophy |
| MEE10b | Macula atrophic with heavy pigmentation; vessels attenuated; periphery atrophic with bone spicules | Macula: Diffuse hypoAF Periphery: Diffuse hypoAF | Diffuse outer retinal atrophy |
| MEE11 | Well-demarcated central macular RPE atrophy; vessels attenuated; periphery atrophic with bone spicules | Macula: Prominent central hypoAF  Periphery: Diffuse hypoAF | Diffuse outer retinal atrophy |
| MEE12 | Foveal RPE atrophy; vessels attenuated; periphery atrophic with bone spicules | Macula: Foveal hyperAF  Periphery: Diffuse hypoAF | Fovea preserved with surrounding PR loss |
| MEE13 | Perifoveal RPE atrophy; vessels attenuated; inferior bone spicules | Macula: Perifoveal hypoAF, hypoAF in arcades  Periphery: Inferior hypoAF | Fovea preserved with surrounding PR loss |
| CEI23745 | Macula atrophic; vessels attenuated; rare peripheral bone spicules | Macula: Near-confluent diffuse nummular hypoAF  Periphery: Diffuse granular hypoAF | Foveal remnant with diffuse outer retinal atrophy |
| CEI26396 | Diffuse macular granularity; vessels attenuated; periphery atrophic with white depigmented spots | Macula: Parafoveal hyperAF ring  Periphery: Diffuse hypoAF | Loss of foveal EZ definition between ages 10 and 14; surrounding outer retinal atrophy |
| **Patient** | **Fundus** | **Fundus autofluorescence** | **OCT** |
| CEI26528a | Subtle parafoveal atrophy; mild attenuation of vessels; peripheral mottling with rare bone spicules | Macula: Parafoveal hyperAF ring; broad hyperAF ring in arcades  Periphery: HypoAF in far periphery | Foveal thinning with bare EZ and surrounding PR loss |
| CEI26529a | Subtle parafoveal atrophy; mild attenuation of vessels, marked depigmentation in far periphery | Macula: Para- and perifoveal hyperAF rings  Periphery: HypoAF in far periphery | ONL thinning but EZ preservation at fovea with broad ring of PR loss |
| CEI29023 | Subtle parafoveal atrophy; attenuated vessels; peripheral coarse pigment deposits and depigmented spots | Macula: Parafoveal hyperAF ring  Periphery: Diffuse hypoAF | Fovea preserved with surrounding PR loss |
| CEI24459 | Well-demarcated parafoveal RPE atrophy; vessels attenuated; periphery atrophic | Macula: Prominent parafoveal hypoAF ring with surrounding hypoAF  Periphery: Unavailable | Fovea preserved with surrounding outer retinal atrophy |
| CEI29345 | Diffuse macular atrophy extending into midperiphery with bone spicules; far periphery normal | Macula: Diffuse hypoAF  Periphery: Midperipheral hypoAF | Diffuse outer retinal atrophy |

a Siblings

b Clinical exam and imaging limited by cataract

Abbreviations: AF – autofluorescence; OCT – optical coherence tomography; EZ – ellipsoid zone; PR – photoreceptor; ONL – outer nuclear layer; RPE – retinal pigment epithelium