**Appendix 3: Summary of clinical data**

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| **Patient** | **Sex** | **Dx** | **Onset (y)** | **Initial symptom(s)** | **Age,**  **First / last exam** | **VA**  **OD first; last**  **OS first; last** | **Kinetic visual fields (V4e)c** | **Full field ERGd** |
| MEE1a | M | MD | 30 | Nyctalopia (30), ↓ vision (32) | 38 / -- | 20/100 ; --  CF ; -- | Not performed | Rods: Near-normal  Cones: Near-normal, normal timing |
| MEE2a | M | RP | 25 | ↓ dark adaptation | 37 /-- | 20/20 ; --  20/30 ; -- | Temporal scotomas | Rods: 50% normal  Cones: <15% normal |
| MEE3 | M | RP | 22 | Nyctalopia | 25 /-- | 20/100 ; --  20/500 ; -- | 20°; temporal crescents | Rods: ND  Cones: SD |
| MEE4 | F | CRD | 20 | ↓ vision | 30 /-- | 20/300 ; --  20/300 ; -- | Not performed | Rods: ~50% normal  Cones: ~50% normal |
| MEE5 | F | RP | 30 | Nyctalopia | 46 / 48 | 20/60 ; 20/60  20/20 ; 20/60 | Central & pericentral ring scotomas | Rods: ND  Cones: 20-35% normal |
| MEE7 | M | RP | 4 | Nyctalopia, ↓ peripheral vision | 10 /-- | 20/40 ; --  20/30 ; -- | 20°; temporal crescents | Rods (OD only): ND  Cones (OD): SD |
| MEE8 | M | RP | 7 | Nyctalopia | 22 /-- | 20/60 ; --  20/70 ; -- | ~25° | Rods: ND  Cones: SD |
| MEE9 | M | RP | 4 | Nyctalopia, ↓ peripheral vision | 9 / 47 | 20/400b ; LP  20/50 ; LP | 13y: V4e < 8° | Rods and cones: ND |
| MEE10 | M | RP | 5 | Nyctalopia | 13 / 56 | 20/30 ; LP  20/30 ; LP | 31y: ~18°; temporal crescents | Rods and cones: ND |
| MEE11 | F | RP | 5 | Nyctalopia, ↓ peripheral vision | 22 / 27 | 20/200 ; 20/150  20/200 ; 20/150 | ~30°; temporal crescents | Rods: ND  Cones: SD |
| MEE12 | F | RP | 4 | Nyctalopia | 13 / 15 | 20/30 ; 20/40  20/40 ; 20/40 | ≤ 20° | Not performed |
| MEE13 | F | RP | 28 | Nyctalopia | 28 / 36 | 20/20 ; 20/20  20/20 ; 20/20 | Superior constriction | Rods: ND  Cones: 25% normal |
| CEI23745 | F | RP | 27 | Nyctalopia | 30 / 45 | n/a ; 20/1000  n/a ; 20/30 | 35y: ~15°; inferior crescents | Rods: ND  Cones: SD |
| CEI26396 | F | LCA | 6 | Nyctalopia | 10 / 14 | 20/40 ; 20/50  20/50 ; 20/80 | I4e 5-10°; V4e full | Rods: ND  Cones: SD |
| CEI26528a | F | CRD | 12 | Nyctalopia | 18 / 22 | 20/25 ; 20/25  20/30 ; 20/25 | Central scotoma through I3e OU; V4e full | Rods: ND  Cones: SD |
| CEI26529a | M | CRD | 13 | ↓ vision | 15 / 19 | 20/20 ; 20/40  20/20 ; 20/30 | 16y: Central scotoma through I3e; V4e full | Rods: 50-60% normal  Cones: < 40% normal |
| CEI29023 | M | EOSRD | 10 | Nyctalopia | 13 / -- | 20/20 ; --  20/20 ; -- | I4e ~ 10°; V4e full | Rods: ND  Cones: SD |
| CEI24459 | F | RP | 15 | Not available | 20 / 27 | 20/30 ; n/a  20/30 ; n/a | Not available | Rods: ND  Cones: SD |
| CEI29345 | M | RP | 16 | Nyctalopia | 37 | n/a | Central scotomas contiguous with superior peripheral loss | Rods: ND  Cones: SD |

Results are from the first exam unless indicated. All ages are in years.

Abbreviations: Dx – diagnosis; MD – macular dystrophy; RP – retinitis pigmentosa; CRD – cone-rod dystrophy; EOSRD – early-onset severe retinal dystrophy; LCA – Leber congenital amaurosis; VA – visual acuity; OD – right eye; OS – left eye; OU – both eyes; LP – light perception; HM – hand motion; ND – nondetectable; SD – severely depressed.

a Siblings (Pairs at CEI and MEI)

b Anisometropic amblyopia diagnosed as a child.

**c** Goldmannperimetry was used at MEE; Octopus semi-automatic kinetic perimetry was used at CEI. Degrees refer to central horizontal visual field diameter. All data refers to V4e unless indicated.

d Classification of cone dysfunction based upon 30 Hz flicker amplitudes; severe depression judged as <1 µV at MEE (normal range 50-125 µV) and <5 µV at CEI (normal range 82-186 µV). Cone timing delayed unless specified otherwise.