**APPENDIX 4. The clinical features of 8 patients with unsolved cases**

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| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Patient****No.** | **Clinical Diagnosis** | **Sex** | **Age****(y)** | **Nystagmus** | **Refraction** | **BCVA** | **Fundus** | **ERG** | **Additional phenotype** |
| **RE** | **LE** |
| 43 | LCA | M | 0.4 | RBJ or vertical jerk infrequent | 4.5 | 4.5 | UCSM/UCSM | Coloboma like lesion at macula | Extinguished | Developmental delay |
| 44 | LCA | M | 2 | Wandering eye movement | -1.25 | -2.25 | UCSM/UCSM | Severe optic atrophy & Pigmentary retinopathy | Flat at photopic | Cerebellar atrophy |
| 45 | LCA | M | 2.8 | Wandering eye movement | 3.5 | 3.0 | UCSM/UCSM | Optic atrophy & Pigmentary retinopathy | Extinguished | Developmental delay, Cerebellar atrophy |
| 46 | LCA | M | 39.8 | 2Hz LBJ bilateral symmetric | -4.5 | -1.75 | 0.08/0.06 | Optic atrophy & Pigmentary retinopathy | Extinguished |  |
| 47 | LCA | M | 20.3 | 2-3Hz pendular bilateral symmetric | -7.5 | -2.75 | 0.2/0.3 |  Pigmentary retinopathy | NA |  |
| 48 | LCA | M | 1.3 | 2Hz LBJ bilateral symmetric infrequent | 3 | 3 | CSM/CSM | Pigmentation with Macula coloboma like lesion | Extinguished |  |
| 49 | LCA | F | 1 | Wandering eye movement | 6.5 | 6.5 | UCSM/UCSM | Normal | Extinguished |  |
| 50 | LCA | M | 0.5 | Wandering eye movement | 5 | 5 | UCSM/UCSM | Normal | Extinguished |  |

BCVA = best corrected visual acuity; CSM = constant, steady, and maintained fixation; F = female; IIN = idiopathic infantile nystagmus; LBJ = left beat jerk; LCA = Leber congenital amaurosis; LE = left eye; M = male; NA = not available; RBJ = right beat jerk; RE =right eye; UCSM = no constant, no steady, no maintained fixation