**Table 1 Summary of clinical features observed in affected individuals in families 1-4 with OCA**

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| --- | --- | --- | --- | --- | --- |
| Family (ID) | Nystagmus | Hair colour | Eye colour | Other ocular features | Other systemic features |
| 1 (X:1) | + | Blonde | Blue | Iris transillumination defects, depigmented fundus, foveal hypoplasia, alternating esotropia, optic disc hypoplasia | - |
| 2 (X:2) | + | NA | NA | Blunted foveal reflex, depigmented fundus.ERG limited and awaiting repeat | NA |
| 3 (X:3) | + | Blonde | Blue | Iris transillumination defects, blunted foveal reflex  | - |
| 3 (X:4) | + | Dark blonde | Blue | transillumination defects, foveal hypoplasia, strabismus  | - |
| 3 (X:5) | NA | Strawberry blonde | NA | Blunted foveal reflex | - |
| 4 (IX:9) | + | Blonde | Blue | Pale fundi, iris transillumination defects, foveal hypoplasia, myopia, strabismus.Nyctalopia, photosensitivity and peripheral VF loss with normal ERG |  |
| 4 (IX:10) | + | Pigmented | Blue | - | Mild learning difficulties |
| 4 (IX:12) | + | Light brown | NA | - | - |
| 4 (IX:14) | + | Dark brown | Blue | Pale fundi | - |
| 4 (IX:15) | + | Pigmented | Blue | - | - |
| 4 (IX:16) | + | NA | NA | NA | NA |
| 4 (IX:20) | + | Blonde | Blue | - | - |
| 4 (IX:22) | + | White/ blonde | Blue | - | - |
| 4 (X:15) | + | Brown | Brown | Myopia | Neonatal intraventricular haemorrhage |

Abbreviations: ERG, electroretinogram; NA, information not available. The (+) and (-) symbols indicate the presence of absence of a feature in an affected subject respectively