Appendix 1: Clinical features of patients

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| Patient ID | Sex/age of onset (months) | Phenotype | | Treatment | Available parental samples |
| Laterality | Sporadic(S) or Familial (Fa) |
| RB2 | F/12 | U | S | NA | Mother |
| RB5 | F | B | S | NA | Mother |
| RB6 | F | U (L) | S | NA | Mother |
| RB7 | M | B | Fa, affected brother, died | NA | Mother |
| RB8 | F | B | Fa, affected sister died | Enucleated (L) | Mother |
| RB10 | M | U (L) | S | NA | Mother |
| RB11 | M | U (R) | S | Chemotherapy | Mother |
| RB12 | F | B | S | Enucleated (L), laser (R) | Mother |
| RB13 | F | B | Fa, affected brother and sister | NA | Mother |
| Rb14 | M | U | S | NA | NA |
| RB15\* | F | B | S, affected sister (RB24) | Enucleated (L), chemotherapy | Parents |
| RB16 | M | B | S | NA | Mother |
| RB17 | M | U | Fa, affected father with U/Enucleated (R) | NA | Parents |
| Rb18 | F | B | S | NA | NA |
| RB19 | M | B | S | Enucleated (R), chemotherapy | Mother |
| RB20 | M | B | Fa, affected father with U/enucleated (R) | NA | Parents |
| RB21 | M | B | Fa, affected father with U/enucleated (NA) | NA | Father |
| RB23 | F | U | S | NA | Mother |
| RB24\* | F | U (R) | Fa, affected sister (RB15) | Laser (R) | Parents |
| RB25 | F | B | S | Enucleated (L), chemotherapy | NA |
| RB26 | F | B | S | NA | Mother |
| RB27 | M | B | S | Enucleated (L/R), chemotherapy | NA |
| RB28 | F | B | S | Enucleated (R), chemotherapy | Mother |
| RB35 | F | B | Fa, affected mother with U/enucleated (R) | Enucleated (R), laser (L) | Mother |
| RB36 | F | B | S | Enucleated (L) | Parents |
| RB38 | F | B | S | Enucleated (L), laser (R) | Parents |
| RB39 | M | B | S | Enucleated (L), laser (R) | Parents |
| RB40 | F | B | S | Enucleated (R), laser (L) | Parents |
| RB43 | M | B | S | Enucleated (R), laser (L) | Parents |
| RB44 | F | B | S | Enucleated (R), laser (L) | Mother |
| RB45 | F | B | S | Enucleated (L), laser (R) | Parents |
| RB47 | F | U | S | Enucleated (R), laser (L) | Parents |
| RB48 | F | U | S | Enucleated (R), laser (L) | NA |
| RB49 | F | B | S | Enucleated (L), laser (R) | Parents |

\*: silbling, F: female, M: male, B: bilateral, U: unilateral, Fa: family, S: sporadic, L: left eye, R: right eye, NA: not available