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| Supplementary Table 2. Clinical information of 51 new probands with pathogenic variants in ACHM-associated genes | | | | | | | |  |  |  |  |  |  |
| ID | Gene | Genotype | Initial | Gender | Age (year) at | | First | Best visual acuity | | Fundus changes | ERG responses from | | OCT |
|  |  |  | diagnosis |  | first exam | onset | symptom | right | left | right | rod | cone |  |
| 14542 | ATF6 | c.[1127G>A];[1456T>G] | CORD | F | 2.5 yrs | FBM | NYS | 0.1 | 0.1 | Normal | Mildly reduced | Extinguished | NA |
| 16187 | ATF6 | c.[1434-1G>A];[1434-1G>A] | ACHM | F | 7 yrs | EC | PV | NA | NA | PFR | Normal | Extinguished | FH |
| 8296 | GNAT2 | c.[619G>A];[124G>C] | CORD | F | 12 yrs | NA | NA | NA | NA | PFR | NA | NA | NA |
| 10668 | CNGA3 | c.62C>G(;)128C>A | CORD | M | 3 yrs | NA | NYS | NA | NA | PFR | Moderately reduced | Extinguished | NA |
| 15732 | CNGA3 | c.[62C>G];[1585G>A] | IRD | F | 3 yrs | 2 mon | NYS, PP | 0.08 | 0.08 | PFR | Severely reduced | Severely reduced | NA |
| 15890 | CNGA3 | c.[464del];[513G>T] | CORD | F | 3 yrs | 1 yrs | PP | NA | NA | Normal | Moderately reduced | Extinguished | NA |
| 15095 | CNGA3 | c.[464del];[1074G>A] | CORD | M | 1 mon | FBM | NYS, PP | NA | NA | Normal | Moderately reduced | Extinguished | NA |
| 11787 | CNGA3 | c.[512G>A];[1585G>A] | CORD | M | 2.5 yrs | FBM | NYS, PP | NA | NA |  | Severely reduced | Extinguished | NA |
| 11802 | CNGA3 | c.[513G>T];[633T>A] | ACHM | M | 7 yrs | FBM | PV, PP | 0.1 | 0.04 | Normal | Normal | Extinguished | NA |
| 14792 | CNGA3 | c.513G>T(;)1627\_1635del | IRD | M | 5 mon | FBM | NA | NA | NA | NA | Extinguished | Extinguished | NA |
| 11062 | CNGA3 | c.[542A>G];[1585G>A] | CORD | M | 4 yrs | FBM | NYS, PP | 0.03 | 0.03 | LF | Moderately reduced | Extinguished | NA |
| 18739 | CNGA3 | c.[587A>G];[872\_873del] | IRD | M | 5 yrs | 3 yrs | PV, PP | 0.07 | 0.05 | Normal | NA | NA | NA |
| 17885 | CNGA3 | c.609G>A(;)1039C>T | IRD | M | 32 yrs | EC | PV | 0.1 | 0.1 | Normal | NA | NA | EZ disruption |
| 11902 | CNGA3 | c.[661C>T];[830G>A] | CORD | M | 1 yrs | 2 mon | NYS, PP | NA | NA | PFR | Severely reduced | Severely reduced | NA |
| 5692 | CNGA3 | c.667C>T(;)1793T>G | ACHM | M | 9 mon | 6 mon | NYS | NA | NA | Normal | Normal | Severely reduced | NA |
| 16657 | CNGA3 | c.[674-2A>C];[1294G>T] | ACHM | M | 3 yrs | FBM | PP, PV | NA | NA | PFR | NA | NA | NA |
| 18874 | CNGA3 | c.[755A>C];[1306C>T] | ACHM | M | 2 yrs | FBM | NYS, PP | NA | NA | PFR | Normal | Severely reduced | NA |
| 13066 | CNGA3 | c.[829C>T];[847C>T] | CORD | F | 8 mon | FBM | NYS, PP | NA | NA | Normal | Mildly reduced | Extinguished | NA |
| 14481 | CNGA3 | c.829C>T(;)1315C>T | CORD | M | 2 yrs | 6 mon | PP | NA | NA | PFR | Severely reduced | Severely reduced | NA |
| 8405 | CNGA3 | c.[830G>A];[1642G>A] | ACHM | M | 1 yrs | 3 mon | NYS | NA | NA | TDP | Normal | Extinguished | NA |
| 11187 | CNGA3 | c.[830G>A];[1709G>A] | CORD | M | 8 mon | 3 mon | NYS, PP | NA | NA | Normal | Moderately reduced | Extinguished | NA |
| 8437 | CNGA3 | c.[833T>C];[1306C>T] | CORD | M | 1 yrs | 5 mon | NYS | NA | NA | NA | Moderately reduced | Extinguished | NA |
| 9020 | CNGA3 | c.[833T>C];[1306C>T] | CORD | F | 9 yrs | EC | PV, PP | 0.1 | 0.1 | PFR | Mildly reduced | Extinguished | NA |
| 5384 | CNGA3 | c.[952G>A]; [1706G>A] | CORD | M | 16 yrs | EC | PV, PP | 0.2 | 0.1 | Normal | Mildly reduced | Extinguished | NA |
| 16065 | CNGA3 | c.[1039C>T];[1319G>A] | IRD | M | 10 yrs | 8 yrs | PV | 0.15 | 0.2 | Normal | NA | NA | irregular EZ |
| 16653 | CNGA3 | c.[1074G>A];[1074G>A] | ACHM | F | 24 yrs | EC | NYS, PV, PP | 0.05 | 0.05 | Normal | Normal | Extinguished | NA |
| 12291 | CNGA3 | c.[1074G>A];[1982G>A] | ACHM | M | 5 yrs | 6 mon | NYS, PV | 0.1 | 0.2 | LF | Normal | Severely reduced | NA |
| 18525 | CNGA3 | c.[1074G>A];[1163G>T] | CORD | F | 4 yrs | FBM | PV | 0.1 | 0.15 | WPD | Moderately reduced | Severely reduced | FH |
| 19550 | CNGA3 | c.[1074G>A];[1627\_1635del] | CORD | M | 4 yrs | FBM | PP, PV | NA | NA | Normal | Moderately reduced | Extinguished | NA |
| 9106 | CNGA3 | c.[1116dup];[1116dup] | CORD | M | 11 mon | FBM | NYS | 0.05 | 0.05 | NA | Moderately reduced | Extinguished | NA |
| 9727 | CNGA3 | c.[1116dup];[1811del] | CORD | F | 1 yrs | 3 mon | NYS, PP | NA | NA | LF | Mildly reduced | Extinguished | NA |
| 5340 | CNGA3 | c.[1124A>G];[1585G>A] | CORD | M | 2.5 yrs | FBM | PP, NYS | NA | NA | ARA | Mildly reduced | Severely reduced | NA |
| 18659 | CNGA3 | c.1163G>A(;)1585G>A | CORD | F | 7 mon | FBM | NYS, PP | NA | NA | PFR | Moderately reduced | Extinguished | NA |
| 14308 | CNGA3 | c.1267dup(;)1306C>T | CORD | M | 1 yrs | 10 mon | PP | NA | NA | PFR | Moderately reduced | Extinguished | NA |
| 10661 | CNGA3 | c.[1306C>T];[1306C>T] | CORD | M | 4 yrs | EC | PV, PP | NA | NA | Normal | Moderately reduced | Severely reduced | NA |
| 17281 | CNGA3 | c.[1306C>T];[1687C>T] | IRD | M | 1.5 yrs | FBM | NYS, PP | NA | NA | Normal | Extinguished | Extinguished | NA |
| 19610 | CNGA3 | c.[1315C>T];[1435A>G;1436A>T] | IRD | F | 4 yrs | 1 yrs | NYS, PP | 0.05 | 0.05 | Normal | NA | NA | NA |
| 18349 | CNGA3 | c.[1569\_1577del];[1569\_1577del] | MD | M | 7 yrs | 5 mon | PV | 0.05 | 0.07 | MD, PFR | NA | NA | FH |
| 9733 | CNGA3 | c.[1585G>A];[1585G>A] | CORD | M | 29 yrs | EC | PV, PP | 0.06 | 0.06 | LF, CR, ARA | Mildly reduced | Severely reduced | NA |
| 16260 | CNGA3 | c.[1585G>A];[1585G>A] | ACHM | F | 3 yrs | FBM | NYS, PP, PV | NA | NA | ARA | NA | NA | NA |
| 6184 | CNGB3 | c.[3G>A];[680T>C] | CORD | M | 6 yrs | 6 yrs | PV | 0.3 | 0.3 | LF, PFR | Mildly reduced | Extinguished | TR |
| 10815 | CNGB3 | c.[11C>A];[11C>A] | ACHM | F | 4 yrs | FBM | NYS, PP | 0.1 | 0.08 | Normal | Normal | Extinguished | NA |
| 18060 | CNGB3 | c.[680T>C];[1155G>T] | CORD | M | 6 yrs | 1 yrs | NYS, PP | 0.2 | 0.2 | PFR | Moderately reduced | Extinguished | FH |
| 9291 | CNGB3 | c.[1063C>T];[1063C>T] | LCA | M | 1 yrs | FBM | NYS | NA | NA | NA | Extinguished | Extinguished | NA |
| 9007 | PDE6C | c.[85C>T];[85C>T] | MD | M | 25 yrs | EC | PV | 0.1 | 0.1 | MC | NA | NA | NA |
| 8430 | PDE6C | c.[421G>C];[939+2T>C] | CORD | F | 2 yrs | FBM | NYS, PV | NA | NA | Normal | Moderately reduced | Extinguished | Normal |
| 16648 | PDE6C | c.648C>G(;)1114T>C | MD | F | 32 yrs | NA | NA | NA | NA | MC | NA | NA | MA |
| 17031 | PDE6C | c.1004+1G>A(;)1771G>A | CORD | M | 2 yrs | NA | PV | 0.15 | 0.15 | Normal | Moderately reduced | Extinguished | Lost EZ |
| 19249 | PDE6C | c.[1777T>C];[2159C>T] | ACHM | M | 5 yrs | 2 yrs | NYS | 0.15 | 0.15 | Normal | NA | NA | NA |
| 15771 | PDE6C | c.[2156T>C];[2156T>C] | CORD | M | 29 | EC | NYS, PP, PV | 0.1 | 0.1 | NA | Mildly reduced | Extinguished | NA |
| 20669 | PDE6C | c.2364\_2368del(;)2377C>T | ACHM | M | 5 yrs | FBM | PP, PV | 0.2 | 0.2 | ARA | Normal | Extinguished | NA |
| ARA= Attenuated retinal arterioles; PFR= Poor foveal reflex; EC= Early childhood; FMB= First few months after birth; NYS= Nystagmus; PV= Poor vision; PP= Photophobia; | | | | | | | | | | | | | |
| PFR= Poor foveal reflex; MD= Macular Dtrophy; NA= Not available; PL=Persuing light; PM= Pigmentation in macular; PO= Pursuing object; NFR= No foveal reflex; | | | | | | | | | | | | | |
| TDP= Temporal disc pallor; TDC=PIG= Temporal disc coloboma; WPD= Waxy pale discs; RA= Retina atrophy; LF= leopard fundus; CR, crescent; | | | | | | | | | | | | | |
| FH= Foveal hypoplasia; EZ= Ellipsoid zone; TR= Thinning retina; MA= Macular atrophy. | | | | | | | | | | | | | |