Table S1. The genetic analysis of the proband after data filtering

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| --- | --- | --- | --- | --- | --- | --- | --- |
| **Gene** | **Position** | **Transcript** | **hgvs.c** | **hgvs.p** | **Genotype** | **ACMGLevel** | **ACMGEvidence** |
| YY1AP1 | chr1:155658035-155658035 | NM\_001198903.1 | c.221T>A | p.L74\* | het | Likely pathogenic | pvs1 pm2 |
| FOXRED1 | chr11:126146018-126146018 | NM\_017547.3 | c.875G>T | p.G292V | het | Likely pathogenic | pm1 pm2 pm5 pp3 |
| SLC12A3 | chr16:56904142-56904142 | NM\_000339.2 | c.736C>A | p.L246I | het | Likely pathogenic | pm1 pm2 pm5 pp3 |
| PLD3 | chr19:40872763-40872763 | NM\_012268.3 | c.186C>G | p.Y62\* | hom | pathogenic | pvs1 pm2 pm3 |
| ATR | chr3:142218530-142218530 | NM\_001184.3 | c.5319C>G | p.Y1773\* | het | Pathogenic | pvs1 pm1 pm2 pp3 |
| ADGRV1 | chr5:89954046-89954046 | NM\_032119.3 | c.4703G>A | p.S1568N | het | Likely pathogenic | pm1 pm2 ps1 |
| CFTR | chr7:117175372-117175372 | NM\_000492.3 | c.650A>G | p.E217G | het | Likely pathogenic | pm1 ps1 pp3 |
| TMEM67 | chr8:94776138-94776138 | NM\_153704.5 | c.475T>C | p.S159P | het | Likely pathogenic | pm1 pm2 ps1 |
| ASS1 | chr9:133355186-133355186 | NM\_000050.4 | c.772G>A | p.A258T | het | Likely pathogenic | pm1 pm2 pm5 pp3 |
| YY1AP1 | chr1:155658033-155658033 | NM\_001198903.1 | c.222\_223insAA | p.A75Kfs\*65 | het | Likely pathogenic | pvs1 pm2 |
| MLYCD | chr16:83932809-83932809 | NM\_012213.2 | c.60del | p.R21Gfs\*52 | het | Likely pathogenic | pvs1 pm2 |
| CYP24A1 | chr20:52779402-52779402 | NM\_000782.4 | c.845-2dup | . | het | Pathogenic | pvs1 pm2 pp3 |
| NPC1L1 | chr7:44578722-44578723 | NM\_013389.2 | c.1273\_1274del | p.L425Afs\*120 | het | Likely pathogenic | pvs1 pm2 |
| ASPH | chr8:62550924-62550924 | NM\_004318.3 | c.791-4\_791-3dup | . | het | Pathogenic | pvs1 pm3 pp3 |



Figure S1. The MRI of other family members.The T2 FLAIR image of other family members didn’t show high intensity of white and gray matter.