**Supplemental Table S9** The annotation results of *GLMN* variant identified by exome sequencing and Sanger sequencing in the individuals of the DUH family.

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **Sample ID** | **Ⅰ-2** | **Ⅱ-3** | **Ⅱ-10** | **Ⅲ-12** | **Ⅱ-11** | **Ⅲ-14** |
| **Variant** | 1:92754541-CA-C | 1:92754541-CA-C | 1:92754541-CA-C | 1:92754541-CA-C | 1:92754541-CA-C | 1:92754541-CA-C |
| **RS ID** | - | - | - | - | - | - |
| **Gene** | GLMN | GLMN | GLMN | GLMN | GLMN | GLMN |
| **Coding DNA change** | c.561delT | c.561delT | c.561delT | c.561delT | c.561delT | c.561delT |
| **Protein change** | p.F187fs | p.F187fs | p.F187fs | p.F187fs | p.F187fs | p.F187fs |
| **Zygosity** | het | het | het | het | het | het |
| **ACMG** | PVS1, PM2 | PVS1, PM2 | PVS1, PM2 | PVS1, PM2 | - | - |
| **ExonicFunc** | frameshift deletion | frameshift deletion | frameshift deletion | frameshift deletion | - | - |
| - | - |  |
| **SIFT,Polyphen2\_HVAR,Polyphen2\_HDIV,MutationTaster,CADD** | - | - | - | - | - | - |  |
|  |
| **GnomAD\_ALL\_AF** | - | - | - | - | - | - |  |
| **GnomAD\_ALL\_AN** | - | - | - | - | - | - |  |
| **GnomAD\_EAS\_AF** | - | - | - | - | - | - |  |
| **GnomAD\_EAS\_AN** | - | - | - | - | - | - |  |
| **AF in 1000G\_Chinese** | - | - | - | - | - | - |  |
| **AF in****ExAC\_EAS** | - | - | - | - | - | - |  |
| **AF in Novo-Zhonghua-WES** | - | - | - | - | - | - |  |
| **AF in Novo-Zhonghua-WGS** | - | - | - | - | - | - |  |