**Supplemental Table S5** The annotation results of *SFTA3* SNV identified by exome sequencing and the Sanger sequncing results in the affected individuals of the DUH family.

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Sample ID** | Ⅰ-2 | Ⅱ-3 | II-7 | Ⅱ-10 | II-11 | Ⅲ-6 | Ⅲ-12 |
| **Variant** | 14:36946225-AG-A | 14:36946225-AG-A | 14:36946225-AG-A | 14:36946225-AG-A | 14:36946225-AG-A | 14:36946225-AG-A | 14:36946225-AG-A |
| **RS ID** | - | - | - | - | - | - | - |
| **Gene** | SFTA3 | SFTA3 | SFTA3 | SFTA3 | SFTA3 | SFTA3 | SFTA3 |
| **Coding DNA change** | c.211delC | c.211delC | c.211delC | c.211delC | c.211delC | c.211delC | c.211delC |
| **Protein change** | p.L71fs | p.L71fs | p.L71fs | p.L71fs | p.L71fs | p.L71fs | p.L71fs |
| **Zygosity** | het | het | het | het | het | het | het |
| **ACMG** | - | - | - | - | - | - | - |
| **ExonicFunc** | Frameshift deletion | Frameshift deletion | - | Frameshiftdeletion | - | Frameshift deletion | Frameshift deletion |
| **SIFT,Polyphen2\_HVAR,Polyphen2\_HDIV,MutationTaster,CADD** | ././././11.52 | ././././11.52 | - | ././././11.52 | - | ././././11.52 | ././././11.52 |
| **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** | **.** | **.** | **-** | **.** | **-** | **.** | **.** |