

# #608161 MACULAR DYSTROPHY, VITELLIFORM, ADULT-ONSET

| OMIM Gene ID | HGNC  | UniProtAC |
|--------------|-------|-----------|
| 179605       | PRPH2 | P23942    |
| 607854       | BEST1 | O76090    |

Table 1: OMIM - UniProtAC mapping

## Legend

- N1: #input proteins associated to the significant GO term
- N2: #proteins associated to the significant GO term
- P-value: Bonferroni-corrected p-value of Fisher's exact test
- *red*: go terms not related to the input proteins
- *blue*: go terms related to the input proteins (enriched uniquely by network-based method)
- *green*: go terms ancestors of terms enriched with the standard method (enriched uniquely by network-based method)

## 1 Standard enrichment

| GO Term    | N1 | N2   | P-value    | Description                          |
|------------|----|------|------------|--------------------------------------|
| GO:0007601 | 2  | 214  | 0.00169593 | visual perception                    |
| GO:0050953 | 2  | 218  | 0.00176007 | sensory perception of light stimulus |
| GO:0007600 | 2  | 586  | 0.0127546  | sensory perception                   |
| GO:0030321 | 1  | 9    | 0.0252735  | transepithelial chloride transport   |
| GO:0050877 | 2  | 1063 | 0.0420022  | neurological system process          |
| GO:0070633 | 1  | 16   | 0.0449266  | transepithelial transport            |

Table 2: Overrepresented GO terms with the standard enrichment

## 2 Network-based enrichment

*No novel enriched terms*