|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| id | Gene | HPO | phi | gene\_sum | hpo\_sum | well\_known |
| 9 | SATB2 | HP:0000164 | 0.36282 | 506 | 74 | yes |
| 12 | DHODH | HP:0000625 | 0.352744 | 112 | 14 | yes |
| 13 | SATB2 | HP:0008872 | 0.340669 | 506 | 62 | yes |
| 15 | LRP1 | HP:0000121 | 0.33317 | 12 | 3 | yes |
| 16 | LRP1 | HP:0006530 | 0.33317 | 12 | 3 | yes |
| 17 | TMEM231 | HP:0000817 | 0.33317 | 12 | 3 | yes |
| 18 | GLB1 | HP:0008454 | 0.33317 | 12 | 3 | yes |
| 19 | IFT140 | HP:0001636 | 0.33317 | 12 | 3 | yes |
| 20 | DHODH | HP:0000202 | 0.32973 | 112 | 16 | yes |
| 21 | DHODH | HP:0000448 | 0.32973 | 112 | 16 | yes |
| 22 | SF3B4 | HP:0011800 | 0.322677 | 122 | 20 | yes |
| 23 | SS18L1 | HP:0002190 | 0.308427 | 14 | 3 | no |
| 24 | SS18L1 | HP:0002242 | 0.308427 | 14 | 3 | no |
| 25 | COG5 | HP:0100512 | 0.308427 | 14 | 3 | yes |
| 26 | COG5 | HP:0010522 | 0.308427 | 14 | 3 | yes |

Phi-coefficient – Matthew’s Correlations Co-efficient Analysis

Gene – HPO pairs with evidence of some correlation based on the phi-coefficient (phi > 0.30). Pairs with an hpo\_sum less than 3 were excluded from the list. Low frequency of use is HPO terms seems to skew the phi coefficient so they have been removed. The columns gene\_sum and hpo\_sum are the number of time that a gene or hpo occurred in MyGene2. The last column (well\_known) is if there is a seemingly known- well understood connection between the gene and HPO term or not. This is based on the the information about the gene found on <https://ghr.nlm.nih.gov/> and the HPO term descriptions.

**SS18L1 & HP:0002190**

HPO: Choroid plexus cyst : Choroid plexus cysts can be observed on prenatal ultrasound examinations and are associated with a weakly increased risk for fetal chromosome abnormalities such as trisomy 18.

Gene: The SS18L1 gene encodes a calcium-responsive transactivator (CREST) that is an essential subunit of a neuron-specific chromatin-remodeling complex (nBAF)

Unknown significance (OMIM 3 cases).

**SS18L1 & HP:0002190**

HPO: Abnormal intestine morphology

Gene: The SS18L1 gene encodes a calcium-responsive transactivator (CREST) that is an essential subunit of a neuron-specific chromatin-remodeling complex (nBAF)

Unknown significance (OMIM 3 cases).