

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).