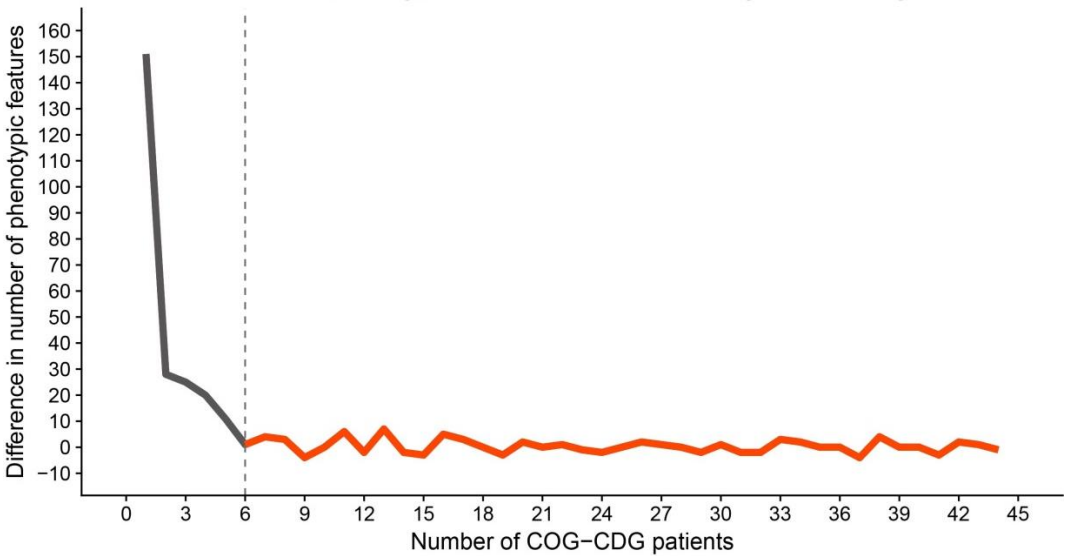
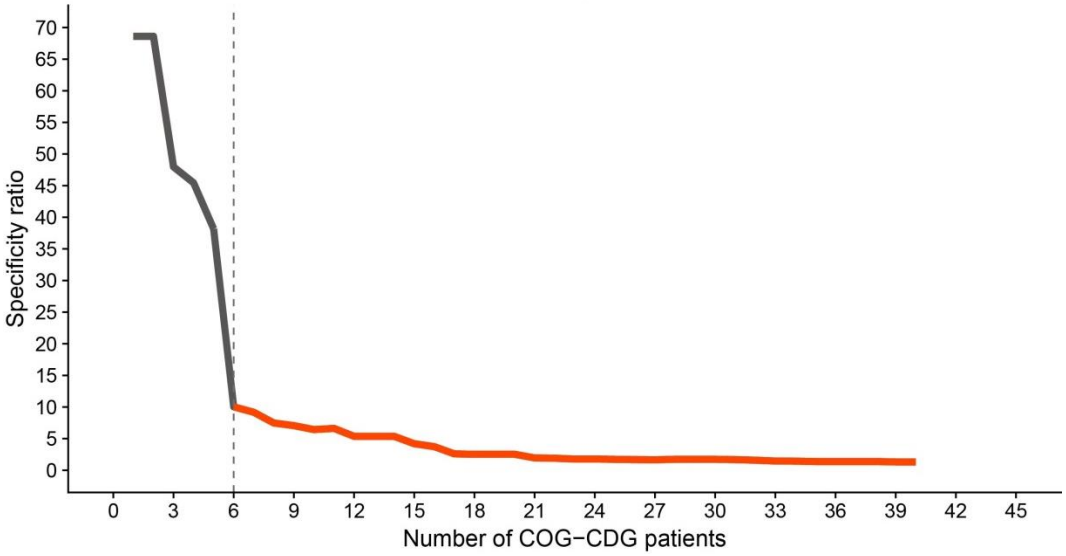
Supplementary Figure 1: Number of phenotypic features versus phenotypic frequency in COG-CDG



Patients	Features	Difference
1	253	
2	102	151
3	74	28
4	50	25
5	29	21
6	18	11
7	17	1
8	13	4
9	10	3
10	14	-4
11	14	0
12	8	6
13	10	-2
14	3	7
15	5	-2
...	...	...
Total	**666**	

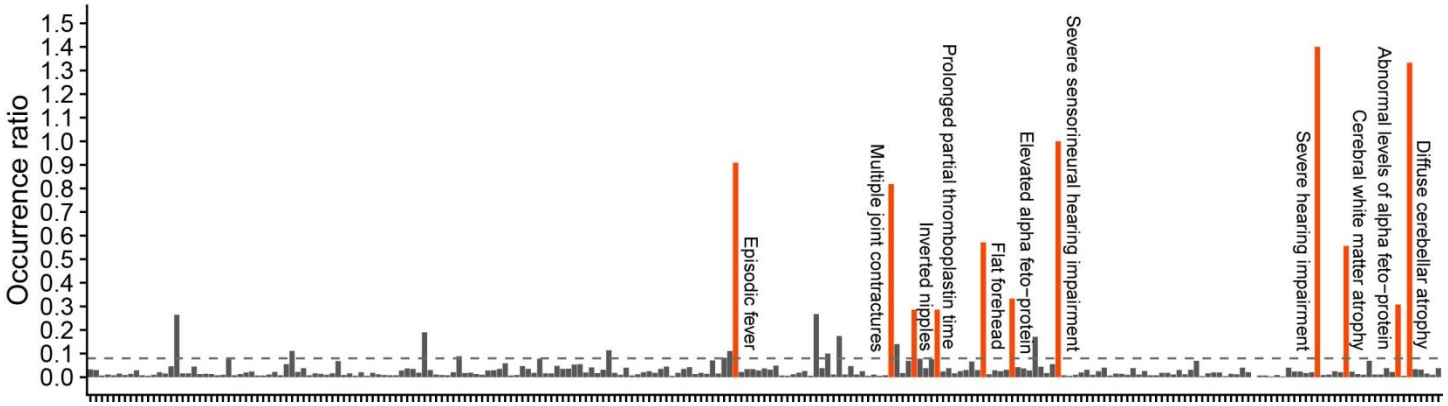
A sliding scale was noted of only one phenotypic feature that is described in all patients, towards many phenotypic features that are described in only one COG-CDG patient (table, column "features"). For features described in at least six COG-CDG patients the number of phenotypic features added to the total list per added patient frequency is very constant (Figure). This suggests that zooming in on features described in at least six COG-CDG patients provides the most relevant focus.

Supplementary Figure 2: Specificity ratio versus phenotypic frequency in COG-CDG



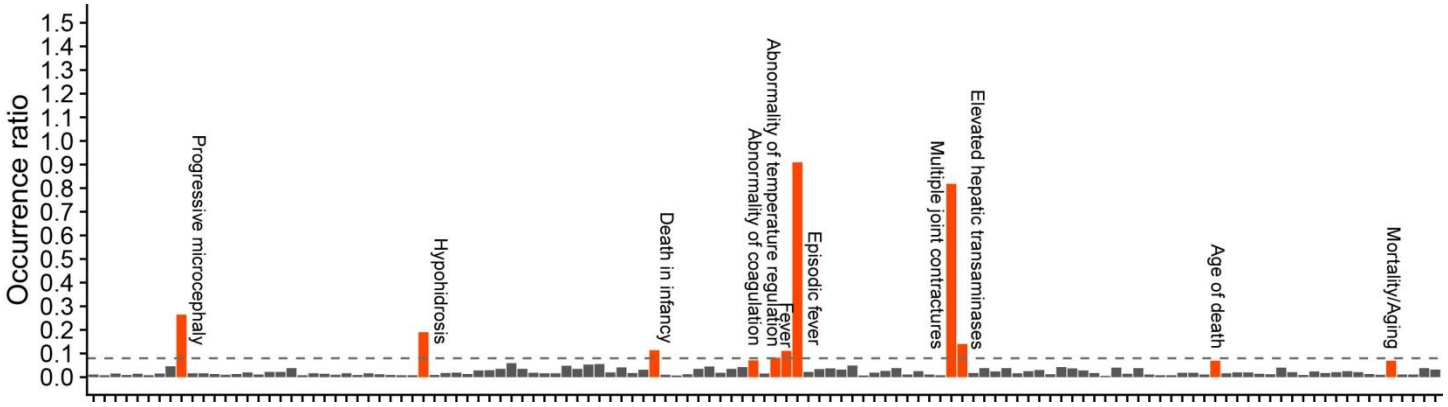
A "specificity ratio" was calculated: the ratio of the median occurrence ratio of the top 10 phenotypic features over the median occurrence ratio of all other phenotypic features. This specificity ratio in relation to the number of COG-CDG patients presenting with a phenotypic feature supported the cut-off value of at least six patients presenting with a certain phenotypic feature.

Supplementary Figure 3: Phenotypic specificity when assessing features present in at least four patients



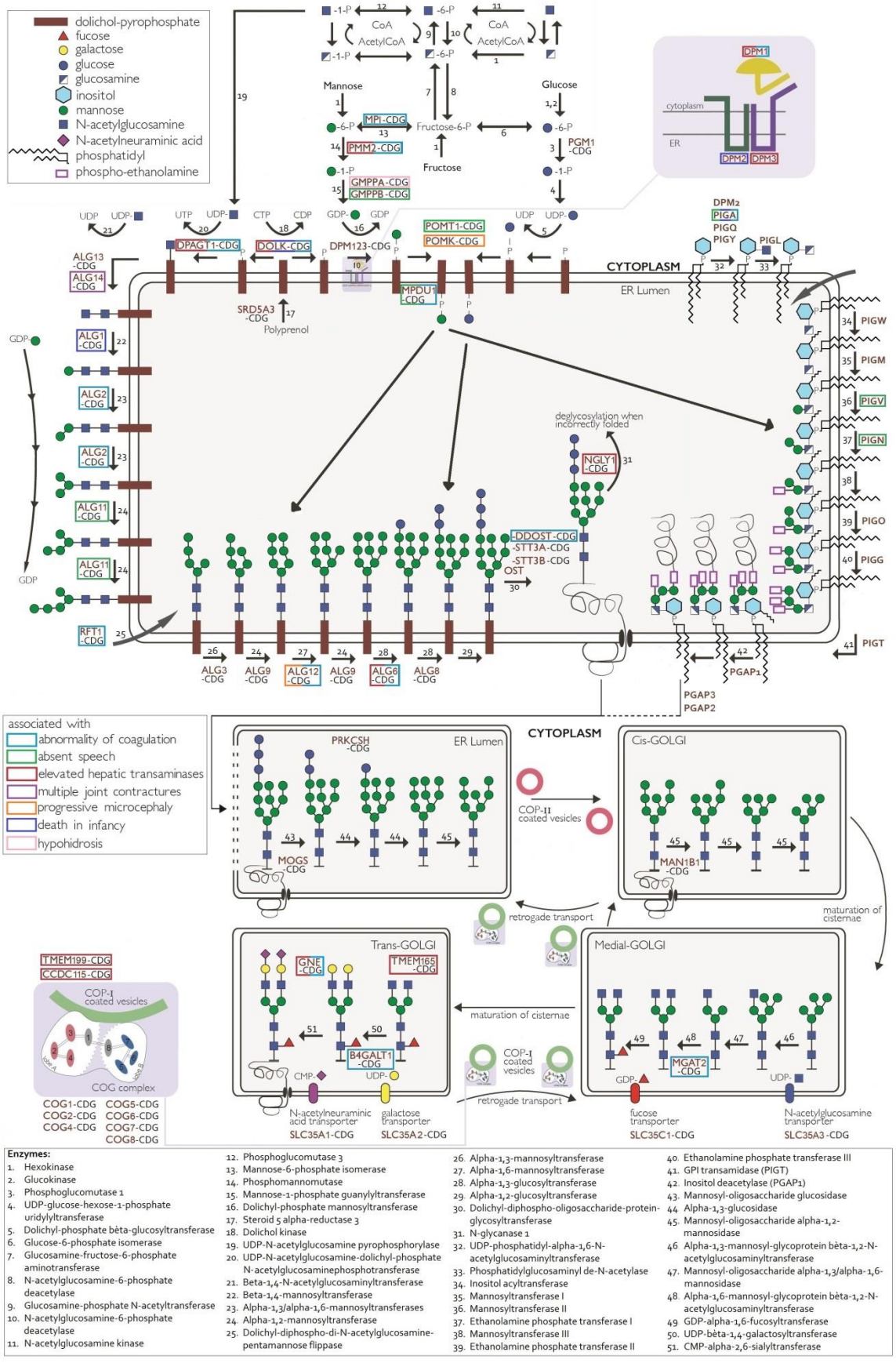
A wider zoom than the cut-off value of a phenotypic feature being present in at least six COG-CDG patients, namely being present in at least four COG-CDG patients, results in a longer list of less relevant features.

Supplementary Figure 4: Phenotypic specificity when assessing features present in at least eight patients



A narrower zoom than the cut-off value of a phenotypic feature being present in at least six COG-CDG patients, namely being present in at least eight COG-CDG patients, results in missing relevant features.

Supplementary Figure 5: Specific phenotypic features in COG-CDG associated with other CDG



Enzymes:

1. Hexokinase
2. Glucokinase
3. Phosphoglucomutase 1
4. UDP-glucose-hexose-1-phosphate uridylyltransferase
5. Dolichyl-phosphate bèta-glucosyltransferase
6. Glucose-6-phosphate isomerase
7. Glucosamine-fructose-6-phosphate aminotransferase
8. N-acetylglucosamine-6-phosphate deacetylase
9. Glucosamine-phosphate N-acetyltransferase
10. N-acetylglucosamine-6-phosphate deacetylase
11. N-acetylglucosamine kinase
12. Phosphoglucomutase 3
13. Mannose-6-phosphate isomerase
14. Phosphomannomutase
15. Mannose-1-phosphate guanylyltransferase
16. Dolichyl-phosphate mannosyltransferase
17. Steroid 5 alpha-reductase 3
18. Dolichol kinase
19. UDP-N-acetylglucosamine pyrophosphorylase
20. UDP-N-acetylglucosamine-dolichyl-phosphate N-acetylglucosaminephosphotransferase
21. Beta-1,4-N-acetylglucosaminyltransferase
22. Beta-1,4-mannosyltransferase
23. Alpha-1,3/alpha-1,6-mannosyltransferases
24. Alpha-1,2-mannosyltransferase
25. Dolichyl-diphospho-di-N-acetylglucosamine-pentamannose flippase
26. Alpha-1,3-mannosyltransferase
27. Alpha-1,6-mannosyltransferase
28. Alpha-1,3-glucosyltransferase
29. Alpha-1,2-glucosyltransferase
30. Dolichyl-diphospho-oligosaccharide-protein-glycosyltransferase
31. N-glycanase 1
32. UDP-phosphatidyl-alpha-1,6-N-acetylglucosaminyltransferase
33. Phosphatidylglucosaminyl de-N-acetylase
34. Inositol acyltransferase
35. Mannosyltransferase I
36. Mannosyltransferase II
37. Ethanolamine phosphate transferase I
38. Mannosyltransferase III
39. Ethanolamine phosphate transferase II
40. Ethanolamine phosphate transferase III
41. GPI transamidase (PIGT)
42. Inositol deacetylase (PGAP1)
43. Mannosyl-oligosaccharide glucosidase
44. Alpha-1,3-glucosidase
45. Mannosyl-oligosaccharide alpha-1,2-mannosidase
46. Alpha-1,3-mannosyl-glycoprotein bèta-1,2-N-acetylglucosaminyltransferase
47. Mannosyl-oligosaccharide alpha-1,3/alpha-1,6-mannosidase
48. Alpha-1,6-mannosyl-glycoprotein bèta-1,2-N-acetylglucosaminyltransferase
49. GDP-alpha-1,6-fucosyltransferase
50. UDP-bèta-1,4-galactosyltransferase
51. CMP-alpha-2,6-sialyltransferase

The glycosylation process. ER, endoplasmatic reticulum; COG, Conserved Oligomeric Golgi; CDG, Congenital Disorder of Glycosylation. Black text: metabolites. Red text: known CDG, caused by a deficiency of the gene encoding the required enzyme. Seven of the ten extracted highly specific phenotypic features are clearly associated with other CDG, depicted in the different colors surrounding the known CDG. For example, abnormality of coagulation and elevated hepatic transaminases are mainly seen in CDG affecting N-glycosylation (process drawn on the left of the ER), while absent speech is seen in CDG affecting O-mannosylation and glycosylphosphatidylinositol anchoring (process drawn on the right of the ER).

Supplementary Notes 1: R code to create figures

```
library('ggplot2')
library('cowplot')

setwd("~/Promotietraject/3) COG-CDG/A) Change of focus")
color<-c('TRUE'='orangered','FALSE'='gray34')

# Load files
AllRatios<-read.csv("COG_AllRatios_CSV.csv", sep=";", stringsAsFactors=FALSE)
AllRatios$HPOcode<-factor(AllRatios$HPOcode,levels=unique(AllRatios$HPOcode))
SelRatios<-read.csv("COG_SelRatios_CSV.csv", sep=";", stringsAsFactors=FALSE)
SelRatios$HPOcode<-factor(SelRatios$HPOcode,levels=unique(SelRatios$HPOcode))

# Figure 1A and 1B
p1<-ggplot()+
  geom_bar(data=AllRatios[which(AllRatios[,5]>5),],
  stat='identity',aes(x=HPOcode,y=Ratio,fill=Ratio>=0.08))+
  geom_text(data=AllRatios[which(AllRatios[,6]>=0.08 & AllRatios[,5]>5),],
  aes(x=HPOcode,y=Ratio,label=Feature),size=3,vjust=0)+
  geom_hline(yintercept=0.08,col='gray43',linetype=2)+
  scale_fill_manual(values=color)+
  scale_y_continuous(breaks=seq(0,1.5,0.1),limits=c(0,1.5))+
  labs(x='HPO code',y='Occurrence ratio', title='Phenotypic specificity in
  COG-CDG')+
  theme(axis.text.x=element_text(angle=90,hjust=1,size=7),
  legend.position='none')

p2<-ggplot()+
  geom_bar(data=SelRatios[which(SelRatios[,5]>5),],
  stat='identity',aes(x=HPOcode, y=Ratio, fill=Ratio>=0.07))+
  geom_text(data=SelRatios[which(SelRatios[,6]>=0.07 & SelRatios[,5]>5),],
  aes(x=HPOcode,y=Ratio,label=Feature),size=3,vjust=0)+
  geom_hline(yintercept=0.07,col='gray43',linetype=2)+
  scale_fill_manual(values=color)+
  scale_y_continuous(breaks=seq(0,1.5,0.1),limits=c(0,1.5))+
  labs(x='HPO code',y='Ratio COG-CDG patients over HPO associated genes',
  title='Phenotypic specificity in COG-CDG, excluding super classes of
  episodic fever')+
  theme(axis.text.x=element_text(angle=90,hjust=1,size=7),
  legend.position='none')

plot_grid(p1,p2,ncol=1,axis='rlbt',rel_heights=c(1,1))
rm(p1,p2)

# Supplementary Figure 1
Diff<-read.csv("COG_difference_CSV.csv", sep=";", stringsAsFactors=FALSE)
ggplot(Diff)+
  geom_line(aes(x=Patients,y=Difference,col=Patients<5),size=2)+
  geom_line(aes(x=Patients,y=Difference,col=Patients>=6),size=2)+
  geom_vline(xintercept=6,col='gray43',linetype=2)+
  scale_color_manual(values=color)+
  scale_y_continuous(breaks=seq(-10,160,10),limits=c(-10,160))+
  scale_x_continuous(breaks=seq(0,45,3),limits=c(0,45))+
  labs(x='Number of COG-CDG patients',y='Difference in number of phenotypic
  features',title='Number of phenotypic features versus phenotypic frequency
  in COG-CDG')+
  theme(legend.position='none')

# Supplementary Figure 2
AllRatios<-AllRatios[order(-AllRatios$Ratio),]
Spec<-setNames(data.frame(matrix(ncol=3,nrow=45)),
c('MedianTop10','MedianNoise','MedianRatio'))
for(freq in 1:40) {
  Spec[freq,1]<-median(as.numeric(AllRatios[which(AllRatios[,5]>
  (freq-1)),6][1:10]),na.rm=TRUE)
  Spec[freq,2]<-median(as.numeric(AllRatios[which(AllRatios[,5]>
  (freq-1)),6][11:length(AllRatios[which(AllRatios[,5]>
  (freq-1)),6])]),na.rm=TRUE)
  Spec[freq,3]<-(Spec[freq,1]/Spec[freq,2])
}
```

```
Spec$Patients<-as.numeric(as.character(rownames(Spec)))
ggplot(Spec)+
  geom_line(aes(x=Patients,y=MedianRatio,col=Patients<5),size=2)+
  geom_line(aes(x=Patients,y=MedianRatio,col=Patients>=6),size=2)+
  geom_vline(xintercept = 6,col='gray43',linetype=2)+
  scale_color_manual(values=color)+
  scale_x_continuous(breaks=seq(0,45,3),limits=c(0,45))+
  scale_y_continuous(breaks=seq(0,70,5),limits=c(0,70))+
  labs(x='Number of COG-CDG patients',y='Specificity ratio',
  title='Specificity ratio versus phenotypic frequency in COG-CDG')+
  theme(legend.position='none')

# Supplementary Figure 3
ggplot()+
  geom_bar(data=AllRatios[which(AllRatios[,5]>3),],
  stat='identity',aes(x=HPOcode,y=Ratio,fill=Ratio>=0.28))+
  geom_text(data=AllRatios[which(AllRatios[,6]>=0.08 & AllRatios[,5]>3),],
  aes(x=HPOcode,y=Ratio,label=Feature),size=3,vjust=0)+
  geom_hline(yintercept=0.08,col='gray43',linetype=2)+
  scale_fill_manual(values=color)+
  scale_y_continuous(breaks=seq(0,1.5,0.1),limits=c(0,1.5))+
  labs(x='HPO code',y='Occurrence ratio', title='Phenotypic specificity in
  COG-CDG')+
  theme(axis.text.x=element_text(angle=90,hjust=1,size=7),
  legend.position='none')

# Supplementary Figure 4
ggplot()+
  geom_bar(data=AllRatios[which(AllRatios[,5]>7),],
  stat='identity',aes(x=HPOcode,y=Ratio,fill=Ratio>=0.06))+
  geom_text(data=AllRatios[which(AllRatios[,6]>=0.08 & AllRatios[,5]>7),],
  aes(x=HPOcode,y=Ratio,label=Feature),size=3,vjust=0)+
  geom_hline(yintercept=0.08,col='gray43',linetype=2)+
  scale_fill_manual(values=color)+
  scale_y_continuous(breaks=seq(0,1.5,0.1),limits=c(0,1.5))+
  labs(x='HPO code',y='Occurrence ratio', title='Phenotypic specificity in
  COG-CDG')+
  theme(axis.text.x=element_text(angle=90,hjust=1,size=7),
  legend.position='none')
```

Supplementary Notes 2: Standard Operating Procedure "Determining phenotypic specificity to facilitate understanding of underlying pathophysiology in rare genetic disorders

A Standard Operation Procedure for the identification of highly specific phenotypic features in rare genetic disorders, to study shared pathophysiology.

Listing all phenotypic features

1. Perform a literature study. Identify all described cases and collect all data from case-reports and case-series. If you need additional information, do not hesitate to contact the authors.
   Example: 45 COG-CDG patients from 32 families, reported in 22 case-reports and case-series.

2. Choose a single case to start with, preferably the one described in most detail

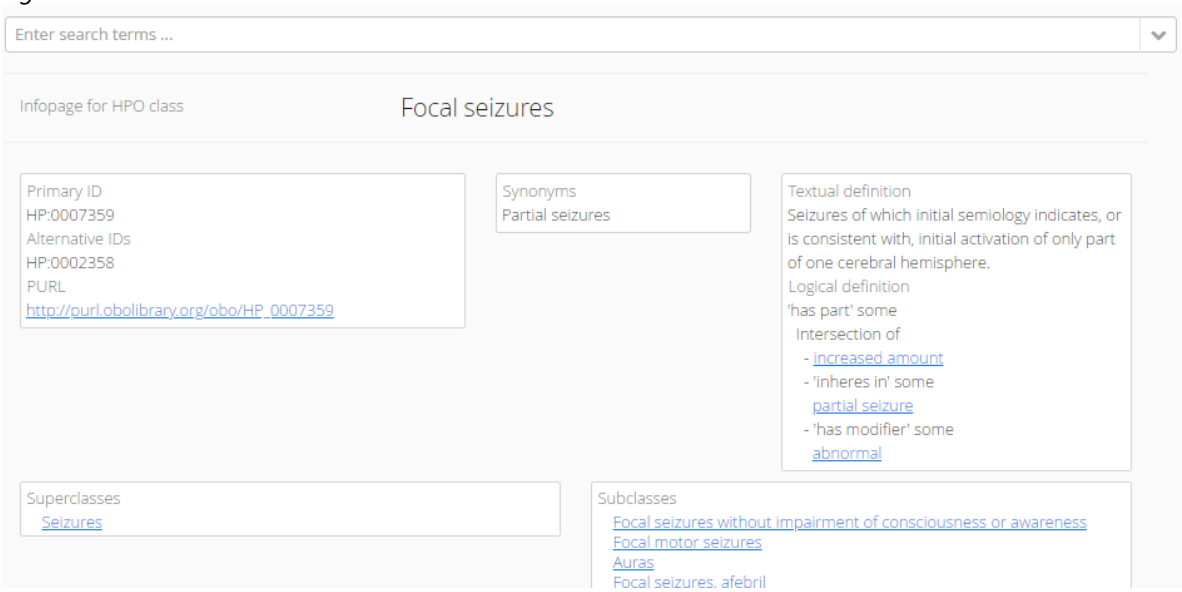
3. For this case, list all described phenotypic features (preferably in an Excel-like program).
   Example:

   > Pregnancy and delivery of the female patient were uncomplicated and at term. The patient suffered from intractable focal seizures, vomiting and loss of consciousness due to intracranial bleedings. Biochemical investigations revealed a normal level for albumin and mildly elevated values for lactate, aspartate aminotransferase and creatine kinase. Metabolic investigations revealed cholestasis and subsequent vitamin K deficiency, explaining in part her intracranial bleedings. The patient died due to brain edema at 5 weeks of age. Since the clinical phenotype of the patient was suspect for a CDG syndrome, initial CDG diagnosis was established by isoelectric focusing (IEF) of the patient's serum transferrin.
   > *Lübbehusen et al. 2010, Human Molecular Genetics*

   List: intractable focal seizures, vomiting, intracranial bleeding, elevated lactate, elevated aspartate aminotransferase, elevated creatine kinase, cholestasis, vitamin K deficiency

4. For each phenotypic feature, list the most appropriate HPO-term and the associated HPO-code via http://compbio.charite.de/hpoweb/showterm?id=HP:0000118#id=HP_0000118 (Figure 1).
   In the search bar you can type the phenotypic feature to see the possible HPO-terms. Primary ID on the left renders the HPO-code. In the middle text block, synonyms for the feature are listed and on the right a textual definition is provided. Super classes and subclasses are listed below. Always be as precise as possible, if a subclass is applicable, it is preferred over its superclass.

   Figure 1



Example:

Intractable focal seizures	Focal seizures	HP:0002358
Vomiting	Vomiting	HP:0002013
Intracranial bleeding	Intracranial hemorrhage	HP:0002170
Elevated lactate	Increased serum lactate	HP:0002151

5. Expand the list of phenotypic features by listing the super classes of each of the phenotypic features that you listed. For the patient that you started with, score each phenotypic feature and all super classes. If you do this, you will preserve the tree-like hierarchical structure of HPO.

Example:		*P1*
0. Phenotypic abnormality	*HP:0000118*	*1*
1. Abnormality of the nervous system	*HP:0000707*	*1*
2. Abnormality of nervous system physiology	*HP:0012638*	*1*
3. Seizures	*HP:0001250*	*1*
4. Focal seizures	*HP:0002358*	*1*

When more super classes are applicable, list the most appropriate one. If you cannot choose, list both.

6. When you finished this for the first patient, you can add the second:
Example: add the phenotypic feature "profound intellectual disability" to your list:

		P1	*P2*
0. Phenotypic abnormality	*HP:0000118*	*1*	*1*
1. Abnormality of the nervous system	*HP:0000707*	*1*	*1*
2. Abnormality of nervous system physiology	*HP:0012638*	*1*	*1*
3. Neurodevelopmental abnormality	*HP:0012759*	*0*	*1*
4. Intellectual disability	*HP:0001249*	*0*	*1*
5. Intellectual disability, profound	*HP:0002187*	*0*	*1*
3. Seizures	*HP:0001250*	*1*	*0*
4. Focal seizures	*HP:0002358*	*1*	*0*

7. Retake step 3-7 until you listed all phenotypic features as detailed as possible for all your patients. Be meticulous in also scoring all appropriate super classes of the phenotypic features.
Example: Supplementary Table 2

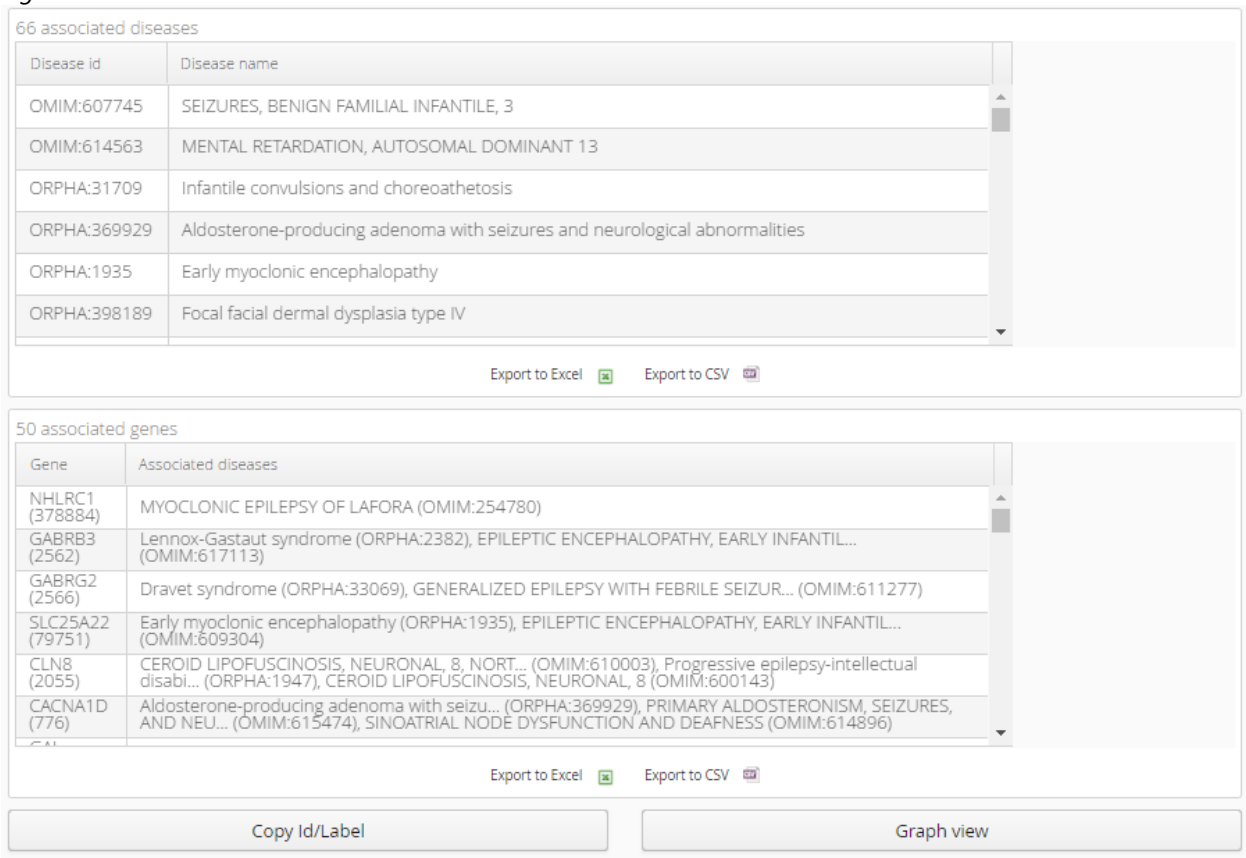
<u>Calculating gene occurrence ratios</u>

8. Sum the total number of patients for each listed phenotypic feature.
Example: Supplementary Table 2

9. Calculate the phenotypic frequency by dividing the total number of patients that present with a phenotypic feature over the total number of included patients
Example: Supplementary Table 1

10. Retrieve from HPO for each phenotypic feature the number of associated <u>genes</u> and note this number for each of the phenotypic features and all super classes.
The associated number of genes can be found <u>below</u> the number of associated diseases (Figure 2). HPO is frequently updated, so note the date you last consulted HPO and during your assessment, regularly check if nothing has changed.

Figure 2



Example: Supplementary Table 1

11. Calculate the **gene occurrence ratio** by dividing the frequency in the patient cohort over the number of associated genes.
    Example: episodic fever was noted in 20 patients (Table 1) and the feature is associated with 22 genes. Occurrence ratio is 20/22 = 0.909. For all occurrence ratios, see Supplementary Table 1.

<u>Determine the cut-off value to meet a minimum sensitivity</u>

12. Order all phenotypic features on the total number of patients that present with the phenotypic feature and calculate the number of phenotypic features that were noted in only one patient, the number of phenotypic features that were noted in only two patients, etc. and tabulate this.
    Example: Supplementary Figure 1

13. Calculate and plot the differences between these numbers. The cut-off value is the number of patients from whereon the difference in number of phenotypic features is fairly stable.
    Example: Supplementary Figure 1

14. This cut-off value can be checked by calculating the specificity ratio. This is the ratio of the median occurrence ratio of the top ten phenotypic features over the median occurrence ratio of all phenotypic features.
    - If no cut-off value is used, calculate the median occurrence ratio of the ten phenotypic features with the highest occurrence ratios. In addition, calculate the median occurrence of all other phenotypic features. The specificity ratio is calculated by dividing the median occurrence ratio of the top ten phenotypic features over the median occurrence ratio of all phenotypic features.
    - Exclude all phenotypic features that are noted in only one patient and calculate the second specificity ratio.
    - Exclude all phenotypic features that are noted in one or two patients and calculate the third specificity ratio.
    - Calculate specificity ratios for all possible phenotypic frequencies and plot this. This should support the cut-off value from step 13.

    Example: Supplementary Figure 2

Study highly specific phenotypic features

15. To study the top XX highly specific phenotypic features of a given disease (in the paper top 10 phenotypic features), retrieve the list of associated genes from HPO (Figure 2, Export to Excel).
    Example: Table 2

16. Derive the gene encoded protein functions from GeneCards.com or other resources.
    Example: for episodic fever one of the genes is ELP1. According to GeneCards this is Elongator Protein Complex 1, a scaffold protein and regulator for three different kinases involved in proinflammatory signaling.

17. Classify the protein functions into – data-driven – categories.
    Example: Table 2. ELP1 was categorized in the category "Immune system".

18. Note the number of genes per category to assess differences in distribution over the categories from the different phenotypic features.
    Example: Table 1.

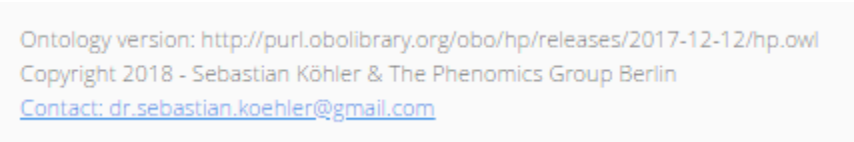
19. Based on your research question, decide what might be an interesting phenotypic feature to look into.
    Example: for episodic fever even its two super classes (fever (HP:0001945) and abnormality of temperature regulation (HP:0004370)) stood out as highly specific phenotypic features and it was not associated to any other glycosylation disorder, so this phenotypic feature was chosen to (first) look into.

20. Perform literature studies for each of the associated genes and see if you can establish one or multiple pathophysiological pathways where genes are related to. This way you can conceive a hypothesis on the possible function in this pathway of the gene you are studying.

Disclaimer

Please bear in mind that the systematic assessment of phenotypic specificity and shared pathophysiology as described here heavily relies on the extent, specificity and completeness of the initial phenotypic description, and also on the accuracy and completeness of the used open access databases: HPO and GeneCards. For HPO, be aware that it is regularly updated, not always with adjustment of the version (Figure 3).

Figure 3



Supplementary Table 1: Aligning phenotypic descriptions to assess phenotypic frequency using the Human Phenotype Ontology in COG-CDG – Summarized table

Clinical features	**HPO code**[a]	**COG1** N=3[b]	**COG2** N=1	**COG4** N=2	**COG5** N=10	**COG6** N=18	**COG7** N=8	**COG8** N=3	**Total** N=45	%
Abn. of the nervous system	HP:0000707	3	1	2	10	18	8	3	45	100
Microcephaly	HP:0000252	3	1	1	7	15	6	1	34	76
Global developmental delay	HP:0001263	3	1	2	6	16	4	1	33	73
Intellectual disability	HP:0001249	2	1		10	9		2	24	53
Seizures	HP:0001250		1	2	1	4	5	2	15	33
Cerebral atrophy	HP:0002059	2	1	2	1	4	2	1	13	29
Cerebellar atrophy	HP:0001272	2			5	1	2	2	12	27
Reduced tendon reflexes	HP:0001315				4	1	5	2	12	27
Abnormality of the abdomen	HP:0001438	1	1	2	3	12	8	3	30	67
Abnormality of the liver	HP:0001392	1	1	2	2	12	7	2	27	60
Elevated hepatic transam.	HP:0002910			2	1	12	5	2	22	49
Hepatomegaly	HP:0002240	1		1	2	6	5		15	33
Splenomegaly	HP:0001744	1		1	1	8	2		13	29
Growth abnormality	HP:0001507	3		1	6	14	8	2	34	76
Failure to thrive	HP:0001508	1		1	3	12	8	1	26	58
Short stature	HP:0004322	2		1	5	3	3	1	15	33
Muscular hypotonia	HP:0001252	1		2	7	11	8	3	32	71
Abnormality of the face	HP:0000271	3	1	1	4	15	6	2	32	71
Abnormality of the mouth	HP:0000153	2			1	6	4		13	29
Abnormality of the nose	HP:0000366	1			2	5	3	1	12	27
Abn. of the orbital region	HP:0000315	2				6	3	1	12	27
Abnormality of the skull	HP:0000929	1			1	4	5	1	12	27
Abn. of skeletal morphology	HP:0011842	2			6	11	5	1	25	56
Episodic fever	HP:0001954					11	8	1	20	44
Abnormality of blood or blood-forming tissues	HP:0001871	1	1	2	1	9	2	1	17	38
Abnormality of limbs	HP:0040064	3			2	4	4	2	15	33
Abnormality of eye physiology	HP:0012373	1		1	3	5	2	2	14	31
Abnormality of the ear	HP:0000598	2			4	1	6		13	29
Recurrent infections	HP:0002719	1		2	1	7	2		13	29
Abn. of the urinary system	HP:0000079	2			4	3	3	1	13	29
Abn.of circulating protein level	HP:0010876			1	1	5	3	2	12	27
Abn. of the cardiovasc. system	HP:0001626	2				6	4		12	27
Death in infancy	HP:0001522								15	33
Neonatal death	HP:0003811								1	

25-40%	41-55%	56-70%	71-85%	86-100%

a) HPO: Human Phenotype Ontology. b) N: number of patients.

Supplementary Table 2: Aligning phenotypic descriptions using the Human Phenotype Ontology in COG-CDG - Full table

Index	Superclass	Clinical feature	HPO code	COG1	COG2	COG4	COG5	COG6	COG7	COG8	All	Ass. genes	Occ. ratio
1	1	Abnormality of blood or blood-forming tissues	HP:0001871	1	1	2	1	9	2	1	**17**	**944**	**0.018**
2	2	Abnormal bleeding	HP:0001892	0	0	0	0	4	1	1	**6**	**340**	**0.018**
3	3	Gingival bleeding	HP:0000225	0	0	0	0	1	0	0	**1**	**27**	**0.037**
4	3	Epistaxis	HP:0000421	0	0	0	0	1	0	0	**1**	**60**	**0.017**
5	4	Spontaneous, recurrent epistaxis	HP:0004406	0	0	0	0	1	0	0	**1**	**4**	**0.250**
6	3	Internal hemorrhage	HP:0011029	0	0	0	0	4	1	0	**5**	**191**	**0.026**
7	4	Hemorrhage of the eye	HP:0011885	0	0	0	0	1	0	0	**1**	**23**	**0.043**
8	5	Retinal hemorrhage	HP:0000573	0	0	0	0	1	0	0	**1**	**10**	**0.100**
9	4	Intracranial hemorrhage	HP:0002170	0	0	0	0	2	0	0	**2**	**71**	**0.028**
10	5	Subdural hemorrhage	HP:0100309	0	0	0	0	1	0	0	**1**	**1**	**1.000**
11	4	Gastrointestinal bleeding	HP:0002239	0	0	0	0	2	0	0	**2**	**116**	**0.017**
12	3	Subcutaneous hemorrhage	HP:0001933	0	0	0	0	0	0	1	**1**	**174**	**0.006**
13	4	Spontaneous hematomas	HP:0007420	0	0	0	0	0	0	1	**1**	**23**	**0.043**
14	2	Abnormal thrombosis	HP:0001977	0	0	0	0	1	0	0	**1**	**95**	**0.011**
15	3	Disseminated intravascular coagulation	HP:0005521	0	0	0	0	1	0	0	**1**	**3**	**0.333**
16	2	Abnormality of coagulation	HP:0001928	0	1	2	1	3	1	1	**9**	**127**	**0.071**
17	3	Abnormality of the coagulation cascade	HP:0003256	0	0	2	0	1	1	1	**5**	**66**	**0.076**
18	4	Reduced antithrombin III activity	HP:0001976	0	0	1	0	0	0	1	**2**	**8**	**0.250**
19	4	Reduced protein C activity	HP:0005543	0	0	0	0	0	0	1	**1**	**2**	**0.500**
20	4	Reduced protein S activity	HP:0004855	0	0	0	0	0	0	1	**1**	**2**	**0.500**
21	4	Abnormality of the intrinsic pathway	HP:0010989	0	0	0	0	1	0	0	**1**	**23**	**0.043**
22	5	Reduced factor XI activity	HP:0001929	0	0	0	0	1	0	0	**1**	**5**	**0.200**
23	3	Prolonged partial thromboplastin time	HP:0003645	0	0	0	1	3	0	0	**4**	**14**	**0.286**
24	2	Abnormality of thrombocytes	HP:0001872	1	0	1	1	5	1	0	**9**	**260**	**0.035**
25	3	Abnormal platelet count	HP:0011873	1	0	1	1	5	1	0	**9**	**227**	**0.040**
26	4	Thrombocytopenia	HP:0001873	1	0	1	1	5	1	0	**9**	**216**	**0.042**
27	5	Macrothrombocytopenia	HP:0040185	1	0	0	0	0	0	0	**1**	**6**	**0.167**
28	2	Abnormality of bone marrow cell morphology	HP:0005561	1	0	0	0	4	1	0	**6**	**191**	**0.031**
29	3	Abnormality of cells of the erythoid lineage	HP:0012130	1	0	0	0	3	1	0	**5**	**2**	**2.500**
30	4	Abnormality of erythrocytes	HP:0001877	1	0	0	0	3	1	0	**5**	**395**	**0.013**
31	5	Anemia	HP:0001903	1	0	0	0	3	1	0	**5**	**365**	**0.014**
32	3	Abnormality of cells of the monocyte/macrophage lineage	HP:0012144	0	0	0	0	2	0	0	**2**	**33**	**0.061**
33	4	Abnormality of macrophages	HP:0004311	0	0	0	0	1	0	0	**1**	**27**	**0.037**
34	5	Hemophagocytosis	HP:0012156	0	0	0	0	1	0	0	**1**	**8**	**0.125**
35	4	Abnormal monocyte count	HP:0012310	0	0	0	0	1	0	0	**1**	**6**	**0.167**
36	5	Monocytosis	HP:0012311	0	0	0	0	1	0	0	**1**	**4**	**0.250**
37	3	Abnormality of multiple cell lineages in the bone marrow	HP:0012145	0	0	0	0	2	0	0	**2**	**88**	**0.023**
38	4	Pancytopenia	HP:0001876	0	0	0	0	2	0	0	**2**	**56**	**0.036**

39	2	Abnormality of leukocytes	HP:0001881	0	0	0	0	2	0	0	**2**	**418**	**0.005**
40	3	Abnormal leukocyte count	HP:0011893	0	0	0	0	2	0	0	**2**	**218**	**0.009**
41	4	Leukocytosis	HP:0001974	0	0	0	0	2	0	0	**2**	**57**	**0.035**
42	1	Abnormality of connective tissue	HP:0003549	0	0	0	0	1	1	0	**2**	**1019**	**0.002**
43	2	Abnormality of adipose tissue	HP:0009124	0	0	0	0	1	1	0	**2**	**157**	**0.013**
44	3	Lipodystrophy	HP:0009125	0	0	0	0	2	0	0	**2**	**84**	**0.024**
45	3	Abnormality of subcutaneous fat tissue	HP:0001001	0	0	0	0	1	1	0	**2**	**35**	**0.057**
46	4	Abnormal subcutaneous fat tissue distribution	HP:0007552	0	0	0	0	1	1	0	**2**	**7**	**0.286**
47	1	Abnormality of head or neck	HP:0000152	3	1	1	4	15	6	2	**32**	**2186**	**0.015**
48	2	Abnormality of the neck	HP:0000464	2	0	0	1	1	5	0	**9**	**414**	**0.022**
49	3	Short neck	HP:0000470	2	0	0	1	1	5	0	**9**	**238**	**0.038**
50	3	Low posterior hairline	HP:0002162	1	0	0	0	0	0	0	**1**	**69**	**0.014**
51	2	Abnormality of the head	HP:0000234	3	1	1	4	15	6	2	**32**	**2160**	**0.015**
52	3	Abnormality of the face	HP:0000271	3	1	1	4	15	6	2	**32**	**1954**	**0.016**
53	4	Abnormal facial shape	HP:0001999	0	0	0	0	0	1	0	**1**	**625**	**0.002**
54	5	Flat face	HP:0012368	0	0	0	0	0	1	0	**1**	**69**	**0.014**
55	4	Abnormality of the forehead	HP:0000290	1	0	1	1	2	3	0	**8**	**610**	**0.013**
56	5	Narrow forehead	HP:0000341	1	0	0	0	1	3	0	**5**	**61**	**0.082**
57	5	Flat forehead	HP:0004425	0	0	1	0	0	3	0	**4**	**7**	**0.571**
58	5	Abnormality of the metopic suture	HP:0005556	0	0	0	0	1	0	0	**1**	**38**	**0.026**
59	6	Metopic synostosis	HP:0011330	0	0	0	0	1	0	0	**1**	**3**	**0.333**
60	5	Abnormality of the frontal hairline	HP:0000599	0	0	0	1	1	1	0	**3**	**78**	**0.038**
61	6	Low anterior hairline	HP:0000294	0	0	0	1	1	1	0	**3**	**52**	**0.058**
62	4	Abnormality of the nose	HP:0000366	1	0	0	2	5	3	1	**12**	**1109**	**0.011**
63	5	Wide nose	HP:0000445	0	0	0	0	0	3	1	**4**	**73**	**0.055**
64	5	Abnormality of the nares	HP:0005288	1	0	0	0	1	0	0	**2**	**377**	**0.005**
65	6	Anteverted nares	HP:0000463	1	0	0	0	1	0	0	**2**	**363**	**0.006**
66	7	Broad nasal tip	HP:0000455	0	0	0	0	1	0	0	**1**	**57**	**0.018**
67	5	Abnormal nasal morphology	HP:0005105	0	0	0	2	2	3	1	**8**	**642**	**0.012**
68	6	Prominent nose	HP:0000448	0	0	0	2	2	0	1	**5**	**45**	**0.111**
69	6	Short nose	HP:0003196	0	0	0	0	0	3	0	**3**	**204**	**0.015**
70	5	Abnormality of the nasal bridge	HP:0000422	1	0	0	1	2	0	1	**5**	**657**	**0.008**
71	6	Depressed nasal bridge	HP:0005280	0	0	0	0	1	0	1	**2**	**307**	**0.007**
72	6	Wide nasal bridge	HP:0000431	1	0	0	1	1	0	0	**3**	**352**	**0.009**
73	4	Abnormality of the mouth	HP:0000153	2	0	0	1	6	4	0	**13**	**1417**	**0.009**
74	5	Abnormality of mouth size	HP:0011337	1	0	0	0	3	3	0	**7**	**237**	**0.030**
75	6	Narrow mouth	HP:0000160	1	0	0	0	0	3	0	**4**	**136**	**0.029**
76	6	Wide mouth	HP:0000154	0	0	0	0	3	0	0	**3**	**113**	**0.027**
77	5	Abnormality of the oral cavity	HP:0000163	1	0	0	1	5	3	0	**10**	**1213**	**0.008**
78	6	Abnormality of the oral mucosa	HP:0011830	0	0	0	0	1	2	0	**3**	**205**	**0.015**
79	7	Abnormality of the gingiva	HP:0000168	0	0	0	0	1	2	0	**3**	**144**	**0.021**

80	8	Gingival overgrowth	HP:0000212	0	0	0	0	1	2	0	**3**	**75**	**0.040**
81	6	Abnormality of the palate	HP:0000174	1	0	0	1	1	0	0	**3**	**677**	**0.004**
82	7	High palate	HP:0000218	1	0	0	1	1	0	0	**3**	**389**	**0.008**
83	6	Abnormality of the tongue	HP:0000157	0	0	0	0	0	1	0	**1**	**238**	**0.004**
84	7	Protruding tongue	HP:0010808	0	0	0	0	0	1	0	**1**	**19**	**0.053**
85	6	Abnormality of the teeth	HP:0000164	0	0	0	0	4	0	0	**4**	**619**	**0.006**
86	7	Abnormal number of teeth	HP:0006483	0	0	0	0	1	0	0	**1**	**200**	**0.005**
87	8	Reduced number of teeth	HP:0009804	0	0	0	0	1	0	0	**1**	**183**	**0.005**
88	7	Abnormality of dental structure	HP:0011061	0	0	0	0	3	0	0	**3**	**256**	**0.012**
89	8	Carious teeth	HP:0000670	0	0	0	0	2	0	0	**2**	**145**	**0.014**
90	8	Hypoplasia of teeth	HP:0000685	0	0	0	0	2	0	0	**2**	**60**	**0.033**
91	6	Abnormality of the oral frenula	HP:0000190	1	0	0	0	0	0	0	**1**	**29**	**0.034**
92	7	Short lingual frenulum	HP:0000200	1	0	0	0	0	0	0	**1**	**3**	**0.333**
93	5	Abnormality of the lip	HP:0000159	2	0	0	0	4	3	0	**9**	**666**	**0.014**
94	6	Abnormality of upper lip	HP:0000177	2	0	0	0	1	2	0	**5**	**517**	**0.010**
95	7	Abnormality of the philtrum	HP:0000288	2	0	0	0	1	2	0	**5**	**350**	**0.014**
96	8	Smooth philtrum	HP:0000319	1	0	0	0	0	2	0	**3**	**69**	**0.043**
97	8	Long philtrum	HP:0000343	1	0	0	0	1	0	0	**2**	**213**	**0.009**
98	8	Hypoplastic philtrum	HP:0005326	0	0	0	0	1	0	0	**1**	**1**	**1.000**
99	6	Full lips	HP:0012471	0	0	0	0	1	1	0	**2**	**155**	**0.013**
100	6	Thin vermilion border	HP:0000233	2	0	0	0	2	0	0	**4**	**192**	**0.021**
101	7	Thin upper lip vermilion	HP:0000219	2	0	0	0	0	0	0	**2**	**130**	**0.015**
102	5	Pierre-Robin sequence	HP:0000201	1	0	0	0	0	0	0	**1**	**13**	**0.077**
103	4	Abnormality of the midface	HP:0000309	1	0	0	0	0	3	0	**4**	**375**	**0.008**
104	5	Abnormality of malar bones	HP:0012369	0	0	0	0	0	3	0	**3**	**182**	**0.016**
105	6	Malar flattening	HP:0000272	0	0	0	0	0	3	0	**3**	**175**	**0.017**
106	5	Hypoplasia of midface	HP:0011800	1	0	0	0	0	0	0	**1**	**167**	**0.006**
107	4	Abnormality of the orbital region	HP:0000315	2	0	0	0	6	3	1	**12**	**1220**	**0.010**
108	5	Abnormal morphology of the ocular adnexa	HP:0030669	2	0	0	0	6	3	1	**12**	**949**	**0.013**
109	6	Abnormality of the eyelid	HP:0000492	2	0	0	0	5	3	1	**11**	**800**	**0.014**
110	7	Epicanthus	HP:0000286	0	0	0	0	3	0	1	**4**	**316**	**0.013**
111	7	Abnormality of the palpebral fissures	HP:0008050	2	0	0	0	3	3	0	**8**	**481**	**0.017**
112	8	Slanting of the palpebral fissures	HP:0200006	2	0	0	0	0	2	0	**4**	**400**	**0.010**
113	9	Upslanted palpebral fissure	HP:0000582	0	0	0	0	0	2	0	**2**	**162**	**0.012**
114	9	Downslanted palpebral fissures	HP:0000494	2	0	0	0	0	0	0	**2**	**275**	**0.007**
115	8	Abnormal size of the palpebral fissures	HP:0200007	0	0	0	0	3	3	0	**6**	**159**	**0.038**
116	9	Short palpebral fissure	HP:0012745	0	0	0	0	0	3	0	**3**	**70**	**0.043**
117	9	Long palpebral fissure	HP:0000637	0	0	0	0	3	0	0	**3**	**25**	**0.120**
118	6	Abnormality of the eyebrow	HP:0000534	0	0	0	0	1	0	0	**1**	**362**	**0.003**
119	7	Laterally extended eyebrow	HP:0011230	0	0	0	0	1	0	0	**1**	**0**	**0.000**
120	3	Abnormality of the skull	HP:0000929	1	0	0	1	4	5	1	**12**	**1454**	**0.008**

121	4	Abnormality of the calvaria	HP:0002683	0	0	0	0	1	0	0	**1**	**689**	**0.001**
122	5	Abnormality of calvarial morphology	HP:0002658	0	0	0	0	1	0	0	**1**	**31**	**0.032**
123	6	Skull asymmetry	HP:0002678	0	0	0	0	1	0	0	**1**	**5**	**0.200**
124	4	Abnormality of the facial skeleton	HP:0011821	1	0	0	1	3	5	1	**11**	**780**	**0.014**
125	5	Abnormality of the mandible	HP:0000277	1	0	0	1	2	5	1	**10**	**623**	**0.016**
126	6	Retrognathia	HP:0000278	0	0	0	1	3	3	0	**7**	**155**	**0.045**
127	6	Aplasia/hypoplasia of the mandible	HP:0009118	1	0	0	0	0	2	1	**4**	**493**	**0.008**
128	7	Micrognathia	HP:0000347	1	0	0	0	0	2	1	**4**	**489**	**0.008**
129	1	Abnormality of limbs	HP:0040064	3	0	0	2	4	4	2	**15**	**1393**	**0.011**
130	2	Abnormality of the upper limb	HP:0002817	3	0	0	0	0	3	2	**8**	**1007**	**0.008**
131	3	Abnormality of the hand	HP:0001155	3	0	0	0	0	3	2	**8**	**929**	**0.009**
132	4	Split hand	HP:0001171	0	0	0	0	0	0	1	**1**	**54**	**0.019**
133	4	Aplasia/hypoplasia involving bones of the hand	HP:0005927	1	0	0	0	0	0	1	**2**	**329**	**0.006**
134	5	Small hand	HP:0200055	1	0	0	0	0	0	1	**2**	**74**	**0.027**
135	4	Deviation of the hand or of fingers of the hand	HP:0009484	1	0	0	0	0	2	0	**3**	**318**	**0.009**
136	5	Ulnar deviation of the hand or of fingers	HP:0001193	1	0	0	0	0	2	0	**3**	**45**	**0.067**
137	6	Ulnar deviation of the hand	HP:0009487	0	0	0	0	0	2	0	**2**	**13**	**0.154**
138	6	Ulnar deviation of finger	HP:0009465	1	0	0	0	0	0	0	**1**	**33**	**0.030**
139	4	Abnormality of the palm	HP:0100871	1	0	0	0	0	3	0	**4**	**265**	**0.015**
140	5	Abnormality of the skin of the palm	HP:0040211	1	0	0	0	0	3	0	**4**	**183**	**0.022**
141	6	Abnormal palmar dermatoglyphics	HP:0001018	1	0	0	0	0	3	0	**4**	**133**	**0.030**
142	7	Abnormality of the palmar creases	HP:0010490	1	0	0	0	0	3	0	**4**	**128**	**0.031**
143	8	Simian crease	HP:0000954	1	0	0	0	0	3	0	**4**	**107**	**0.037**
144	2	Abnormality of the lower limb	HP:0002814	3	0	0	0	1	0	2	**6**	**1039**	**0.006**
145	3	Abnormality of the lower limb bone	HP:0040069	2	0	0	0	0	0	1	**3**	**474**	**0.006**
146	4	Abnormality of the femur	HP:0002823	1	0	0	0	0	0	1	**2**	**179**	**0.011**
147	5	Abnormality of the femoral neck or head region	HP:0003366	1	0	0	0	0	0	1	**2**	**112**	**0.018**
148	6	Abnormality of the femoral neck	HP:0003367	1	0	0	0	0	0	1	**2**	**96**	**0.021**
149	7	Coxa valga	HP:0003673	1	0	0	0	0	0	1	**2**	**9**	**0.222**
150	7	Broad femoral neck	HP:0006429	1	0	0	0	0	0	0	**1**	**8**	**0.125**
151	3	Abnormality of the foot	HP:0001760	1	0	0	0	1	0	2	**4**	**862**	**0.005**
152	4	Aplasia/hypoplasia involving bones of the feet	HP:0006494	1	0	0	0	0	0	1	**2**	**202**	**0.010**
153	5	Short foot	HP:0001773	1	0	0	0	0	0	1	**2**	**103**	**0.019**
154	4	Positional foot deformity	HP:0005656	1	0	0	0	1	0	1	**3**	**265**	**0.011**
155	5	Talipes	HP:0001883	1	0	0	0	1	0	1	**3**	**264**	**0.011**
156	6	Talipes equinovarus	HP:0001762	1	0	0	0	1	0	1	**3**	**211**	**0.014**
157	7	Bilateral talipes equinovarus	HP:0001776	0	0	0	0	0	0	1	**1**	**3**	**0.333**
158	2	Abnormality of limb bone	HP:0040068	0	0	0	2	4	4	1	**11**	**1025**	**0.011**
159	3	Abnormality of limb bone morphology	HP:0002813	0	0	0	2	4	4	1	**11**	**1003**	**0.011**
160	4	Abnormality of the limb diaphysis	HP:0006504	0	0	0	1	0	0	0	**1**	**182**	**0.005**
161	5	Bowing of the long bones	HP:0006487	0	0	0	1	0	0	0	**1**	**177**	**0.006**

162	6	Bowing of the legs	HP:0002979	0	0	0	1	0	0	0	**1**	**131**	**0.008**
163	7	Genu valgum	HP:0002857	0	0	0	1	0	0	0	**1**	**82**	**0.012**
164	4	Abnormality of digit	HP:0011297	0	0	0	1	4	4	1	**10**	**885**	**0.011**
165	5	Clinodactyly	HP:0030084	0	0	0	1	0	0	1	**2**	**303**	**0.007**
166	6	Finger clinodactyly	HP:0040019	0	0	0	1	0	0	0	**1**	**232**	**0.004**
167	6	Toe clinodactyly	HP:0001863	0	0	0	0	0	0	1	**1**	**13**	**0.077**
168	5	Polydactyly	HP:0010442	0	0	0	0	2	0	0	**2**	**191**	**0.010**
169	5	Abnormality of the finger	HP:0001167	0	0	0	0	2	4	0	**6**	**720**	**0.008**
170	6	Deviation of finger	HP:0004097	0	0	0	0	0	3	0	**3**	**305**	**0.010**
171	7	Overlapping fingers	HP:0010557	0	0	0	0	0	3	0	**3**	**18**	**0.167**
172	6	Long fingers	HP:0100807	0	0	0	0	1	2	0	**3**	**160**	**0.019**
173	6	Abnormality of the thumb	HP:0001172	0	0	0	0	1	4	0	**5**	**242**	**0.021**
174	7	Adducted thumbs	HP:0001181	0	0	0	0	1	4	0	**5**	**56**	**0.089**
175	6	Slender finger	HP:0001238	0	0	0	0	1	1	0	**2**	**95**	**0.021**
176	5	Abnormality of toe	HP:0001780	0	0	0	0	0	2	1	**3**	**427**	**0.007**
177	6	Overlapping toes	HP:0001845	0	0	0	0	0	1	0	**1**	**19**	**0.053**
178	6	Widely spaced toes	HP:0008094	0	0	0	0	0	0	1	**1**	**2**	**0.500**
179	6	Long toes	HP:0010511	0	0	0	0	0	2	0	**2**	**23**	**0.087**
180	7	Long hallux	HP:0001847	0	0	0	0	0	1	0	**1**	**9**	**0.111**
181	1	Abnormality of metabolism/homeostasis	HP:0001939	0	1	2	1	11	8	2	**25**	**1708**	**0.015**
182	2	Abnormality of fluid regulation	HP:0011032	0	0	0	0	0	1	0	**1**	**374**	**0.003**
183	3	Dehydration	HP:0001944	0	0	0	0	0	1	0	**1**	**70**	**0.014**
184	2	Abnormality of temperature regulation	HP:0004370	0	0	0	0	11	8	1	**20**	**244**	**0.082**
185	3	Fever	HP:0001945	0	0	0	0	11	8	1	**20**	**180**	**0.111**
186	4	Episodic fever	HP:0001954	0	0	0	0	11	8	1	**20**	**22**	**0.909**
187	2	Abnormality of cell physiology	HP:0011017	0	0	0	0	1	0	0	**1**	**326**	**0.003**
188	3	Abnormality of B cell physiology	HP:0005372	0	0	0	0	1	0	0	**1**	**142**	**0.007**
189	4	Absent specific antibody respons	HP:0005424	0	0	0	0	1	0	0	**1**	**1**	**1.000**
190	2	Abnormality of ion homeostasis	HP:0003111	0	1	0	0	1	0	0	**2**	**225**	**0.009**
191	3	Abnormality of cation homeostasis	HP:0010929	0	1	0	0	1	0	0	**2**	**203**	**0.010**
192	4	Abnormality of the transition element cation hom.	HP:0011030	0	1	0	0	1	0	0	**2**	**37**	**0.054**
193	5	Abnormality of iron homeostasis	HP:0011031	0	0	0	0	1	0	0	**1**	**28**	**0.036**
194	6	Abnormal serum ferritin	HP:0040133	0	0	0	0	1	0	0	**1**	**22**	**0.045**
195	7	Increased serum ferritin	HP:0003281	0	0	0	0	1	0	0	**1**	**22**	**0.045**
196	5	Abnormality of copper homeostasis	HP:0010836	0	1	0	0	0	0	0	**1**	**9**	**0.111**
197	6	Hypocupremia	HP:0011967	0	1	0	0	0	0	0	**1**	**3**	**0.333**
198	6	Decreased serum ceruloplasmin	HP:0010837	0	1	0	0	0	0	0	**1**	**6**	**0.167**
199	2	Abnormal enzyme/coenzyme activity	HP:0012379	0	0	1	1	2	0	0	**4**	**99**	**0.040**
200	3	Abnormality of alkaline phosphatase activity	HP:0004379	0	0	1	1	2	0	0	**4**	**61**	**0.066**
201	4	Elevated alkaline phosphatase	HP:0003155	0	0	1	1	2	0	0	**4**	**58**	**0.069**
202	2	Abnormality of circulating protein level	HP:0010876	0	0	1	1	5	3	2	**12**	**302**	**0.040**

203	3	Abnormality of circulating enzyme level	HP:0011021	0	0	0	0	3	3	2	**8**	**212**	**0.038**
204	4	Abnormal levels of creatine kinase in blood	HP:0040081	0	0	0	0	3	3	2	**8**	**211**	**0.038**
205	5	Elevated serum creatine phosphokinase	HP:0003236	0	0	0	0	3	3	2	**8**	**209**	**0.038**
206	3	Abnormal levels of alpha feto-protein	HP:0045056	0	0	1	1	2	0	0	**4**	**13**	**0.308**
207	4	Elevated alpha feto-protein	HP:0006254	0	0	1	1	2	0	0	**4**	**12**	**0.333**
208	2	Abnormality of carbohydrate metabolism/homeostasis	HP:0011013	0	0	0	0	0	1	0	**1**	**528**	**0.002**
209	3	Abnormal glucose homeostasis	HP:0011014	0	0	0	0	0	1	0	**1**	**468**	**0.002**
210	4	Abnormality of blood glucose concentration	HP:0011015	0	0	0	0	0	1	0	**1**	**178**	**0.006**
211	5	Hypoglycaemia	HP:0001943	0	0	0	0	0	1	0	**1**	**157**	**0.006**
212	6	Recurrent hypoglycaemia	HP:0001988	0	0	0	0	0	1	0	**1**	**8**	**0.125**
213	2	Abnormality of lipid metabolism	HP:0003119	0	0	1	0	0	0	0	**1**	**180**	**0.006**
214	3	Abnormality of cholesterol metabolism	HP:0003107	0	0	1	0	0	0	0	**1**	**64**	**0.016**
215	4	Hypercholesterolemia	HP:0003124	0	0	1	0	0	0	0	**1**	**33**	**0.030**
216	2	Hyperbilirubinemia	HP:0002904	0	0	0	1	1	0	0	**2**	**52**	**0.038**
217	3	Unconjugated hyperbilirubinemia	HP:0008282	0	0	0	1	0	0	0	**1**	**5**	**0.200**
218	3	Conjugated hyperbilirubinemia	HP:0002908	0	0	0	0	1	0	0	**1**	**17**	**0.059**
219	2	Abnormality of vitamin metabolism	HP:0100508	0	0	0	0	1	0	0	**1**	**58**	**0.017**
220	3	Abnormality of vitamin K metabolism	HP:0100831	0	0	0	0	1	0	0	**1**	**6**	**0.167**
221	4	Vitamin K deficiency	HP:0011892	0	0	0	0	1	0	0	**1**	**6**	**0.167**
222	1	Abnormality of prenatal development or birth	HP:0001197	0	0	0	1	6	1	0	**8**	**465**	**0.017**
223	2	Abnormality of the amniotic fluid	HP:0001560	0	0	0	1	2	1	0	**4**	**227**	**0.018**
224	3	Oligohydramnios	HP:0001562	0	0	0	1	1	1	0	**3**	**88**	**0.034**
225	3	Polyhydramnios	HP:0001561	0	0	0	0	1	0	0	**1**	**143**	**0.007**
226	2	Hydrops fetalis	HP:0001789	0	0	0	0	1	0	0	**1**	**44**	**0.023**
227	2	Premature birth	HP:0001622	0	0	0	0	4	0	0	**4**	**100**	**0.040**
228	2	Prenatal movement abnormality	HP:0001557	0	0	0	0	1	0	0	**1**	**112**	**0.009**
229	3	Decreased fetal movement	HP:0001558	0	0	0	0	1	0	0	**1**	**101**	**0.010**
230	1	Abnormality of the abdomen	HP:0001438	1	1	2	3	12	8	3	**30**	**544**	**0.055**
231	2	Abnormality of the abdominal organs	HP:0002012	1	1	2	3	12	8	3	**30**	**895**	**0.034**
232	3	Abnormality of the liver	HP:0001392	1	1	2	2	12	7	2	**27**	**769**	**0.035**
233	4	Decreased liver function	HP:0001410	0	1	0	0	2	0	0	**3**	**171**	**0.018**
234	5	Hepatic failure	HP:0001399	0	0	0	0	2	0	0	**2**	**137**	**0.015**
235	4	Hepatomegaly	HP:0002240	1	0	1	2	6	5	0	**15**	**392**	**0.038**
236	4	Elevated hepatic transaminases	HP:0002910	0	0	2	1	12	5	2	**22**	**157**	**0.140**
237	4	Cirrhosis	HP:0001394	0	0	1	1	2	0	0	**4**	**112**	**0.036**
238	4	Hepatic necrosis	HP:0002605	0	0	0	0	1	0	0	**1**	**25**	**0.040**
239	4	Abnormality of the biliary system	HP:0004297	0	0	1	1	5	3	0	**10**	**262**	**0.038**
240	5	Cholestasis	HP:0001396	0	0	1	1	5	3	0	**10**	**187**	**0.053**
241	6	Jaundice	HP:0000952	0	0	0	1	1	2	0	**4**	**145**	**0.028**
242	4	Abnormality of the hepatic vasculature	HP:0006707	0	0	0	1	0	0	0	**1**	**48**	**0.021**
243	5	Portal hypertension	HP:0001409	0	0	0	1	0	0	0	**1**	**46**	**0.022**

244	4	Hepatitis	HP:0012115	0	0	1	0	0	0	1	**2**	**64**	**0.031**
245	3	Abnormality of the spleen	HP:0001743	1	0	1	1	8	2	0	**13**	**373**	**0.035**
246	4	Splenomegaly	HP:0001744	1	0	1	1	8	2	0	**13**	**290**	**0.045**
247	3	Abnormality of the gastrointestinal tract	HP:0011024	0	0	0	2	3	5	1	**11**	**980**	**0.011**
248	4	Morphological abnormality of the GI tract	HP:0012718	0	0	0	2	3	1	1	**7**	**752**	**0.009**
249	5	Abnormality of the esophagus	HP:0002031	0	0	0	1	2	1	0	**4**	**141**	**0.028**
250	6	Gastroesophageal reflux	HP:0002020	0	0	0	1	0	1	0	**2**	**170**	**0.012**
251	6	Esophageal varix	HP:0002040	0	0	0	0	1	0	0	**1**	**18**	**0.056**
252	6	Dysphagia	HP:0002015	0	0	0	0	1	0	0	**1**	**207**	**0.005**
253	5	Abnormality of the large intestine	HP:0002250	0	0	0	0	0	0	1	**1**	**144**	**0.007**
254	6	Abnormal large intestine physiology	HP:0012700	0	0	0	0	0	0	1	**1**	**35**	**0.029**
255	7	Bowel incontinence	HP:0002607	0	0	0	0	0	0	1	**1**	**29**	**0.034**
256	5	Anorectal anomaly	HP:0012732	0	0	0	1	1	0	0	**2**	**208**	**0.010**
257	6	Abnormality of the anus	HP:0004378	0	0	0	1	1	0	0	**2**	**147**	**0.014**
258	7	Ectopic anus	HP:0004397	0	0	0	0	1	0	0	**1**	**35**	**0.029**
259	8	Anus anteposition	HP:0001545	0	0	0	0	1	0	0	**1**	**16**	**0.063**
260	7	Anal stenosis	HP:0002025	0	0	0	1	0	0	0	**1**	**27**	**0.037**
261	4	Functional abnormality of the GI tract	HP:0012719	0	0	0	0	2	4	0	**6**	**547**	**0.011**
262	5	Gastrointestinal obstruction	HP:0004796	0	0	0	0	0	3	0	**3**	**68**	**0.044**
263	5	Gastrointestinal inflammation	HP:0004386	0	0	0	0	2	1	0	**3**	**51**	**0.059**
264	6	Inflammation of the large intestine	HP:0002037	0	0	0	0	2	1	0	**3**	**28**	**0.107**
265	7	Colitis	HP:0002583	0	0	0	0	1	0	0	**1**	**22**	**0.045**
266	8	Acute colitis	HP:0100282	0	0	0	0	1	0	0	**1**	**5**	**0.200**
267	9	Enterocolitis	HP:0004387	0	0	0	0	1	0	0	**1**	**5**	**0.200**
268	2	Abdominal symptom	HP:0011458	1	0	1	2	6	6	0	**16**	**995**	**0.016**
269	3	Anorexia	HP:0002039	0	0	0	0	1	0	0	**1**	**64**	**0.016**
270	3	Nausea and vomiting	HP:0002017	0	0	0	0	2	0	0	**2**	**307**	**0.007**
271	4	Vomiting	HP:0002013	0	0	0	0	2	0	0	**2**	**161**	**0.012**
272	3	Diarrhea	HP:0002014	0	0	1	0	4	2	0	**7**	**205**	**0.034**
273	3	Feeding difficulties	HP:0011968	1	0	1	2	1	6	0	**11**	**529**	**0.021**
274	1	Abnormality of the breast	HP:0000769	1	0	0	0	3	1	0	**5**	**273**	**0.018**
275	2	Abnormality of the nipple	HP:0004404	0	0	0	0	3	1	0	**4**	**135**	**0.030**
276	3	Inverted nipples	HP:0003186	0	0	0	0	3	1	0	**4**	**14**	**0.286**
277	2	Abnormal intermamillary distance	HP:0040157	1	0	0	0	0	0	0	**1**	**72**	**0.014**
278	3	Wide intermamillary distance	HP:0006610	1	0	0	0	0	0	0	**1**	**72**	**0.014**
279	1	Abnormality of the cardiovascular system	HP:0001626	2	0	0	0	6	4	0	**12**	**1667**	**0.007**
280	2	Abnormality of cardiovascular system morphology	HP:0030680	2	0	0	0	6	3	0	**11**	**1149**	**0.010**
281	3	Abnormal heart morphology	HP:0001627	2	0	0	0	6	3	0	**11**	**910**	**0.012**
282	4	Abnormality of the heart valves	HP:0001654	1	0	0	0	1	1	0	**3**	**305**	**0.010**
283	5	Abnormality of the aortic valve	HP:0001646	1	0	0	0	1	0	0	**2**	**167**	**0.012**
284	6	Aortic regurgitation	HP:0001659	1	0	0	0	1	0	0	**2**	**59**	**0.034**

285	6	Dysplastic aortic valve	HP:0005176	0	0	0	0	1	0	0	**1**	**0**	**0.000**
286	5	Abnormality of the atrioventricular valves	HP:0006705	1	0	0	0	0	1	0	**2**	**148**	**0.014**
287	6	Abnormality of the mitral valve	HP:0001633	1	0	0	0	0	0	0	**1**	**126**	**0.008**
288	7	Mitral stenosis	HP:0001718	1	0	0	0	0	0	0	**1**	**12**	**0.083**
289	8	Parachute mitral valve	HP:0011571	1	0	0	0	0	0	0	**1**	**0**	**0.000**
290	6	Abnormality of the tricuspid valve	HP:0001702	0	0	0	0	0	1	0	**1**	**36**	**0.028**
291	7	Tricuspid insufficiency	HP:0005180	0	0	0	0	0	1	0	**1**	**17**	**0.059**
292	4	Abnormality of cardiac atrium	HP:0005120	1	0	0	0	4	2	0	**7**	**244**	**0.029**
293	5	Atrial septal defect	HP:0001631	0	0	0	0	4	2	0	**6**	**241**	**0.025**
294	6	Secundum atrial septal defect	HP:0001684	0	0	0	0	1	2	0	**3**	**7**	**0.429**
295	6	Patent foramen ovale	HP:0001655	0	0	0	0	1	0	0	**1**	**26**	**0.038**
296	4	Abnormality of cardiac ventricle	HP:0001713	1	0	0	0	2	3	0	**6**	**342**	**0.018**
297	5	Abnormality of the ventricular septum	HP:0010438	0	0	0	0	2	3	0	**5**	**245**	**0.020**
298	6	Ventricular septal defect	HP:0001629	0	0	0	0	2	3	0	**5**	**240**	**0.021**
299	5	Ventricular hypertrophy	HP:0001714	1	0	0	0	0	0	0	**1**	**74**	**0.014**
300	6	Left ventricular hypertrophy	HP:0001712	1	0	0	0	0	0	0	**1**	**54**	**0.019**
301	4	Hypoplastic heart	HP:0001961	0	0	0	0	1	0	0	**1**	**22**	**0.045**
302	3	Congenital malformation of the great arteries	HP:0011603	0	0	0	0	3	0	0	**3**	**240**	**0.013**
303	4	Persistent ductus arteriosus	HP:0001643	0	0	0	0	3	0	0	**3**	**192**	**0.016**
304	2	Abnormality of cardiovascular system physiology	HP:0011025	0	0	0	0	0	2	0	**2**	**775**	**0.003**
305	3	Arrhythmia	HP:0011675	0	0	0	0	0	1	0	**1**	**311**	**0.003**
306	4	Tachycardia	HP:0001649	0	0	0	0	0	1	0	**1**	**99**	**0.010**
307	3	Congestive heart failure	HP:0001635	0	0	0	0	0	1	0	**1**	**160**	**0.006**
308	1	Abnormality of the ear	HP:0000598	2	0	0	4	1	6	0	**13**	**1466**	**0.009**
309	2	Hearing abnormality	HP:0000364	2	0	0	2	1	2	0	**7**	**986**	**0.007**
310	3	Hearing impairment	HP:0000365	2	0	0	2	1	2	0	**7**	**969**	**0.007**
311	4	Severe hearing impairment	HP:0012714	2	0	0	2	1	2	0	**7**	**5**	**1.400**
312	5	Severe sensorineural hearing impairment	HP:0008625	0	0	0	2	1	2	0	**5**	**5**	**1.000**
313	5	Severe conductive hearing impairment	HP:0012717	1	0	0	0	0	0	0	**1**	**0**	**0.000**
314	5	Mixed hearing impairment	HP:0000410	1	0	0	0	0	0	0	**1**	**18**	**0.056**
315	2	Abnormality of the outer ear	HP:0000356	2	0	0	3	0	5	0	**10**	**760**	**0.013**
316	3	Abnormal location of ears	HP:0000357	2	0	0	3	0	5	0	**10**	**490**	**0.020**
317	4	Low set ears	HP:0000369	2	0	0	3	0	5	0	**10**	**463**	**0.022**
318	4	Posteriorly rotated ears	HP:0000358	2	0	0	3	0	2	0	**7**	**289**	**0.024**
319	3	Abnormality of the pinna	HP:0000377	0	0	0	0	0	2	0	**2**	**493**	**0.004**
320	4	Abnormality of the helix	HP:0011039	1	0	0	0	0	0	0	**1**	**93**	**0.011**
321	5	Cleft helix	HP:0009902	1	0	0	0	0	0	0	**1**	**2**	**0.500**
322	4	Abnormality of the antihelix	HP:0009738	1	0	0	0	0	0	0	**1**	**23**	**0.043**
323	4	Microtia	HP:0008551	1	0	0	0	0	0	0	**1**	**86**	**0.012**
324	3	Abnormality of the auditory canal	HP:0000372	1	0	0	0	0	0	0	**1**	**61**	**0.016**
325	4	Stenosis of the external auditory canal	HP:0000402	1	0	0	0	0	0	0	**1**	**24**	**0.042**

326	2	Abnormality of the middle ear	HP:0000370	1	0	0	0	0	0	0	**1**	**334**	**0.003**
327	3	Morphological abnormality of the middle ear	HP:0008609	1	0	0	0	0	0	0	**1**	**16**	**0.063**
328	4	Cholesteatoma	HP:0009797	1	0	0	0	0	0	0	**1**	**2**	**0.500**
329	2	Abnormality of the inner ear	HP:0000359	1	0	0	0	0	0	0	**1**	**674**	**0.001**
330	3	Morphological abnormality of the inner ear	HP:0011390	1	0	0	0	0	0	0	**1**	**55**	**0.018**
331	4	Morphological abnormality of the vestibule of the inner ear	HP:0011376	1	0	0	0	0	0	0	**1**	**20**	**0.050**
332	5	Morphological abnormality of the semicircular channel	HP:0011380	1	0	0	0	0	0	0	**1**	**17**	**0.059**
333	6	Morphological abnormality of the lateral semicircular channel	HP:0040106	1	0	0	0	0	0	0	**1**	**0**	**0.000**
334	1	Abnormality of the endocrine system	HP:0000818	0	0	0	0	2	0	0	**2**	**942**	**0.002**
335	2	Abnormality of the thymus	HP:0000777	0	0	0	0	1	0	0	**1**	**44**	**0.023**
336	3	Thymus hyperplasia	HP:0010516	0	0	0	0	1	0	0	**1**	**2**	**0.500**
337	2	Abnormality of circulating hormone level	HP:0003117	0	0	0	0	1	0	0	**1**	**293**	**0.003**
338	3	Abnormal serum insulin-like growth factor 1 level	HP:0030352	0	0	0	0	1	0	0	**1**	**9**	**0.111**
339	4	Decreased serum insuline-like growth factor 1	HP:0030353	0	0	0	0	1	0	0	**1**	**6**	**0.167**
340	2	Abnormality of the adrenal glands	HP:0000834	0	0	0	0	1	0	0	**1**	**172**	**0.006**
341	3	Abnormality of adrenal morphology	HP:0011732	0	0	0	0	1	0	0	**1**	**71**	**0.014**
342	4	Adrenal hypoplasia	HP:0000835	0	0	0	0	1	0	0	**1**	**23**	**0.043**
343	1	Abnormality of the eye	HP:0000478	2	0	1	3	6	3	2	**17**	**2050**	**0.008**
344	2	Abnormal eye morphology	HP:0012372	2	0	0	0	2	1	0	**5**	**1646**	**0.003**
345	3	Abnormality of the globe	HP:0012374	2	0	0	0	2	1	0	**5**	**1644**	**0.003**
346	4	Abnormality of globe location	HP:0100886	2	0	0	0	0	1	0	**3**	**643**	**0.005**
347	5	Hypertelorism	HP:0000316	1	0	0	0	0	1	0	**2**	**475**	**0.004**
348	5	Proptosis	HP:0000520	1	0	0	0	0	0	0	**1**	**177**	**0.006**
349	4	Abnormality of the uvea	HP:0000553	0	0	0	0	1	0	0	**1**	**402**	**0.002**
350	5	Abnormality of the iris	HP:0000525	0	0	0	0	1	0	0	**1**	**323**	**0.003**
351	6	Abnormality of the pupil	HP:0000615	0	0	0	0	1	0	0	**1**	**41**	**0.024**
352	7	Abnormal pupillary function	HP:0007686	0	0	0	0	1	0	0	**1**	**19**	**0.053**
353	8	Miosis	HP:0000616	0	0	0	0	1	0	0	**1**	**9**	**0.111**
354	4	Abnormality of the anterior segment of the globe	HP:0004328	0	0	0	0	1	0	0	**1**	**896**	**0.001**
355	5	Abnormality of the cornea	HP:0000481	0	0	0	0	1	0	0	**1**	**503**	**0.002**
356	6	Decreased corneal reflex	HP:0008000	0	0	0	0	1	0	0	**1**	**2**	**0.500**
357	4	Abnormality of the posterior segment of the globe	HP:0004329	1	0	0	0	1	0	0	**2**	**888**	**0.002**
358	5	Abnormality of the fundus	HP:0001098	1	0	0	0	1	0	0	**2**	**885**	**0.002**
359	6	Abnormality of the optic nerve	HP:0000587	0	0	0	0	1	0	0	**1**	**560**	**0.002**
360	7	Abnormality of optic disc	HP:0012795	0	0	0	0	1	0	0	**1**	**464**	**0.002**
361	8	Optic atrophy	HP:0000648	0	0	0	0	1	0	0	**1**	**415**	**0.002**
362	6	Abnormality of the retina	HP:0000479	1	0	0	0	0	0	0	**1**	**646**	**0.002**
363	7	Macular abnormality	HP:0001103	1	0	0	0	0	0	0	**1**	**145**	**0.007**
364	2	Abnormal eye physiology	HP:0012373	1	0	1	3	5	2	2	**14**	**1624**	**0.009**
365	3	Abnormality of vision	HP:0000504	0	0	0	2	0	1	0	**3**	**797**	**0.004**
366	4	Visual impairment	HP:0000505	0	0	0	2	0	1	0	**3**	**557**	**0.005**

367	5	Cortical visual impairment	HP:0100704	0	0	0	2	0	0	0	**2**	**57**	**0.035**
368	3	Abnormality of refraction	HP:0000539	0	0	0	1	0	0	0	**1**	**369**	**0.003**
369	4	Hypermetropia	HP:0000540	0	0	0	1	0	0	0	**1**	**123**	**0.008**
370	3	Abnormality of eye movement	HP:0000496	1	0	1	2	4	1	2	**11**	**1086**	**0.010**
371	4	Abnormal conjugate eye movement	HP:0000549	1	0	0	2	4	1	2	**10**	**627**	**0.016**
372	5	Strabismus	HP:0000486	1	0	0	2	4	1	2	**10**	**612**	**0.016**
373	6	Esotropia	HP:0000565	1	0	0	0	0	1	2	**4**	**59**	**0.068**
374	4	Abnormal involuntary eye movements	HP:0012547	0	0	1	0	0	0	0	**1**	**694**	**0.001**
375	5	Nystagmus	HP:0000639	0	0	1	0	0	0	0	**1**	**692**	**0.001**
376	3	Ptosis	HP:0000508	0	0	0	0	1	0	0	**1**	**441**	**0.002**
377	4	Bilateral ptosis	HP:0001488	0	0	0	0	1	0	0	**1**	**13**	**0.077**
378	3	Abnormal visual electrophysiology	HP:0030453	0	0	0	0	1	0	0	**1**	**200**	**0.005**
379	4	Abnormal electroretinogram	HP:0000512	0	0	0	0	1	0	0	**1**	**160**	**0.006**
380	5	Undetectable electroretinogram	HP:0000550	0	0	0	0	1	0	0	**1**	**20**	**0.050**
381	1	Abnormality of the genitourinary system	HP:0000119	2	0	0	4	3	3	1	**13**	**1672**	**0.008**
382	2	Abnormality of the genital system	HP:0000078	2	0	0	1	0	0	0	**3**	**1059**	**0.003**
383	3	Abnormal genital system morphology	HP:0012243	2	0	0	1	0	0	0	**3**	**962**	**0.003**
384	4	Abnormal external genitalia	HP:0000811	2	0	0	1	0	0	0	**3**	**792**	**0.004**
385	5	Abnormality of male external genitalia	HP:0000032	2	0	0	1	0	0	0	**3**	**769**	**0.004**
386	6	Abnormality of the testis	HP:0000035	2	0	0	1	0	0	0	**3**	**677**	**0.004**
387	7	Cryptorchidism	HP:0000028	2	0	0	1	0	0	0	**3**	**515**	**0.006**
388	8	Unilateral cryptorchidism	HP:0012741	1	0	0	0	0	0	0	**1**	**5**	**0.200**
389	6	Abnormality of the penis	HP:0000036	1	0	0	1	0	0	0	**2**	**507**	**0.004**
390	7	Displacement of the external urethral meatus	HP:0100627	1	0	0	0	0	0	0	**1**	**243**	**0.004**
391	8	Hypospadias	HP:0000047	1	0	0	0	0	0	0	**1**	**229**	**0.004**
392	7	Hypoplasia of penis	HP:0008736	0	0	0	1	0	0	0	**1**	**327**	**0.003**
393	8	Micropenis	HP:0000054	0	0	0	1	0	0	0	**1**	**160**	**0.006**
394	2	Abnormality of the urinary system	HP:0000079	2	0	0	4	3	3	1	**13**	**1229**	**0.011**
395	3	Abnormality of the urinary system physiology	HP:0011277	0	0	0	0	0	1	0	**1**	**700**	**0.001**
396	4	Abnormality of urine homeostasis	HP:0003110	0	0	0	0	0	1	0	**1**	**438**	**0.002**
397	5	Proteinuria	HP:0000093	0	0	0	0	0	1	0	**1**	**141**	**0.007**
398	6	Mild proteinuria	HP:0012595	0	0	0	0	0	1	0	**1**	**0**	**0.000**
399	3	Abnormality of the upper urinary tract	HP:0010935	2	0	0	0	3	1	0	**6**	**886**	**0.007**
400	4	Abnormality of the kidney	HP:0000077	2	0	0	0	3	1	0	**6**	**863**	**0.007**
401	5	Abnormal renal morphology	HP:0012210	1	0	0	0	3	1	0	**5**	**670**	**0.007**
402	6	Hyperechogenic kidneys	HP:0004719	0	0	0	0	1	0	0	**1**	**5**	**0.200**
403	6	Abnormality of the nephron	HP:0012575	0	0	0	0	1	0	0	**1**	**166**	**0.006**
404	7	Abnormality of the renal tubule	HP:0000091	0	0	0	0	1	0	0	**1**	**76**	**0.013**
405	6	Abnormality of the renal pelvis	HP:0010944	1	0	0	0	0	1	0	**2**	**142**	**0.014**
406	7	Dilatation of the renal pelvis	HP:0010946	1	0	0	0	0	1	0	**2**	**141**	**0.014**
407	8	Hydronephrosis	HP:0000126	1	0	0	0	0	1	0	**2**	**141**	**0.014**

408	6	Renal hypoplasia/aplasia	HP:0008678	0	0	0	0	1	0	0	**1**	**201**	**0.005**
409	7	Renal agenesis	HP:0000104	0	0	0	0	1	0	0	**1**	**96**	**0.010**
410	8	Unilateral kidney agenesis	HP:0000122	0	0	0	0	1	0	0	**1**	**32**	**0.031**
411	5	Abnormal renal physiology	HP:0012211	1	0	0	0	0	0	0	**1**	**454**	**0.002**
412	6	Hemolytic uremic syndrome	HP:0005575	1	0	0	0	0	0	0	**1**	**3**	**0.333**
413	6	Renal insufficiency	HP:0000083	1	0	0	0	0	0	0	**1**	**282**	**0.004**
414	3	Abnormality of the lower urinary tract	HP:0010936	0	0	0	4	1	1	1	**7**	**456**	**0.015**
415	4	Abnormality of the bladder	HP:0000014	0	0	0	4	1	1	1	**7**	**234**	**0.030**
416	5	Funcitonal abnormality of the bladder	HP:0000009	0	0	0	4	1	1	1	**7**	**210**	**0.033**
417	6	Neurogenic bladder	HP:0000011	0	0	0	2	0	1	0	**3**	**9**	**0.333**
418	6	Urinary incontinence	HP:0000020	0	0	0	2	0	0	1	**3**	**57**	**0.053**
419	1	Abnormality of the immune system	HP:0002715	1	0	2	1	8	2	0	**14**	**1251**	**0.011**
420	2	Abnormality of the lymphatic system	HP:0100763	0	0	0	0	1	0	0	**1**	**459**	**0.002**
421	3	Abnormality of the lymph nodes	HP:0002733	0	0	0	0	1	0	0	**1**	**145**	**0.007**
422	4	Lymphadenopathy	HP:0002716	0	0	0	0	1	0	0	**1**	**137**	**0.007**
423	5	Generalized lymphadenopathy	HP:0008940	0	0	0	0	1	0	0	**1**	**1**	**1.000**
424	2	Abnormality of immune system physiology	HP:0010978	1	0	2	1	7	2	0	**13**	**950**	**0.014**
425	3	Recurrent infections	HP:0002719	1	0	2	1	7	2	0	**13**	**528**	**0.025**
426	4	Recurrent respiratory infections	HP:0002205	0	0	1	0	1	0	0	**2**	**339**	**0.006**
427	4	Recurrent urinary tract infections	HP:0000010	0	0	0	1	0	0	0	**1**	**83**	**0.012**
428	3	Abnormality of humoral immunity	HP:0005368	1	0	0	0	3	0	0	**4**	**163**	**0.025**
429	4	Abnormality of complement system	HP:0005339	1	0	0	0	0	0	0	**1**	**23**	**0.043**
430	4	Abnormal immunoglobulin level	HP:0010701	0	0	0	0	3	0	0	**3**	**140**	**0.021**
431	5	Hypogammaglobulinemia	HP:0004313	0	0	0	0	2	0	0	**2**	**97**	**0.021**
432	5	Hypergammaglobulinemia	HP:0010702	0	0	0	0	1	0	0	**1**	**49**	**0.020**
433	3	Immunodeficiency	HP:0002721	0	0	0	0	2	0	0	**2**	**180**	**0.011**
434	4	Combined immunodeficiency	HP:0005387	0	0	0	0	2	0	0	**2**	**18**	**0.111**
435	1	Abnormality of the integument	HP:0001574	0	0	1	1	9	5	1	**17**	**1777**	**0.010**
436	2	Abnormality of the skin	HP:0000951	0	0	0	1	4	5	1	**11**	**1454**	**0.008**
437	3	Abnormality of skin morphology	HP:0007517	0	0	0	1	4	5	1	**11**	**1356**	**0.008**
439	4	Abnormal elasticity of skin	HP:0010647	0	0	0	1	0	5	0	**6**	**144**	**0.042**
440	5	Abnormally lax or hyperextensible skin	HP:0008067	0	0	0	1	0	5	0	**6**	**109**	**0.055**
441	6	Cutis laxa	HP:0000973	0	0	0	1	0	2	0	**3**	**73**	**0.041**
438	6	Excessive wrinkled skin	HP:0100678	0	0	0	1	0	5	0	**6**	**35**	**0.171**
442	4	Thickened skin	HP:0001072	0	0	0	0	3	0	1	**4**	**359**	**0.011**
443	5	Epidermal thickening	HP:0011368	0	0	0	0	3	0	1	**4**	**326**	**0.012**
444	6	Hyperkeratosis	HP:0000962	0	0	0	0	3	0	1	**4**	**215**	**0.019**
445	6	Hyperkeratosis pilaris	HP:0040180	0	0	0	0	0	0	1	**1**	**3**	**0.333**
446	4	Dry skin	HP:0000958	0	0	0	1	2	0	1	**4**	**114**	**0.035**
447	4	Abnormality of epidermal morphology	HP:0011124	0	0	0	1	0	0	0	**1**	**50**	**0.020**
448	5	Scaling skin	HP:0040189	0	0	0	1	0	0	0	**1**	**14**	**0.071**

449	2	Abnormality of the skin adnexa	HP:0011138	0	0	1	0	6	1	0	**8**	**945**	**0.008**
450	3	Abnormality of the hair	HP:0001595	0	0	1	0	1	1	0	**3**	**827**	**0.004**
451	4	Abnormality of hair density	HP:0011357	0	0	1	0	0	0	0	**1**	**244**	**0.004**
452	5	Thick hair	HP:0100874	0	0	1	0	0	0	0	**1**	**10**	**0.100**
453	4	Abnormal hair quantity	HP:0011362	0	0	0	0	0	1	0	**1**	**431**	**0.002**
454	5	Hypertrichosis	HP:0000998	0	0	0	0	0	1	0	**1**	**122**	**0.008**
455	4	Abnormality of hair pigmentation	HP:0009887	0	0	0	0	1	0	0	**1**	**95**	**0.011**
456	5	Hypopigmentation of hair	HP:0005599	0	0	0	0	1	0	0	**1**	**84**	**0.012**
457	3	Abnormality of the sweat gland	HP:0000971	0	0	0	0	7	0	0	**7**	**8**	**0.875**
458	4	Hypohidrosis or hyperhidrosis	HP:0007550	0	0	0	0	7	0	0	**7**	**159**	**0.044**
459	5	Hypohidrosis	HP:0000966	0	0	0	0	11	0	0	**11**	**58**	**0.190**
460	1	Abnormality of the musculature	HP:0003011	1	1	2	7	11	8	3	**33**	**1928**	**0.017**
461	2	Abnormality of muscle morphology	HP:0011805	0	0	0	1	0	2	1	**4**	**1024**	**0.004**
462	3	Decreased muscle mass	HP:0003199	0	0	0	1	0	2	1	**4**	**51**	**0.078**
463	2	Abnormality of muscle physiology	HP:0011804	1	1	2	7	11	8	3	**33**	**1633**	**0.020**
464	3	Abnormal muscle tone	HP:0003808	1	1	2	7	11	8	3	**33**	**1368**	**0.024**
465	4	Muscular hypotonia	HP:0001252	1	0	2	7	11	7	3	**31**	**1123**	**0.028**
466	5	Generalized hypotonia	HP:0001290	1	0	1	1	1	2	0	**6**	**660**	**0.009**
467	5	Axial hypotonia	HP:0008936	0	0	1	0	2	0	0	**3**	**91**	**0.033**
468	4	Hypertonia	HP:0001276	0	1	1	1	0	1	0	**4**	**607**	**0.007**
469	5	Limb hypertonia	HP:0002509	0	0	1	1	0	0	0	**2**	**19**	**0.105**
470	5	Spasticity	HP:0001257	0	1	0	1	0	1	0	**3**	**490**	**0.006**
471	6	Spastic tetraplegia	HP:0002510	0	1	0	1	0	0	0	**2**	**55**	**0.036**
472	6	Opisthotonus	HP:0002179	0	0	0	0	0	1	0	**1**	**25**	**0.040**
473	1	Abnormality of the nervous system	HP:0000707	3	1	2	10	18	8	3	**45**	**2863**	**0.016**
474	2	Abnormality of the nervous system morphology	HP:0012639	3	1	2	10	16	7	3	**42**	**2033**	**0.021**
475	3	Morphological abnormality of the CNS	HP:0002011	3	1	2	10	16	7	3	**42**	**1872**	**0.022**
476	4	Abnormality of brain morphology	HP:0012443	3	1	2	10	16	7	3	**42**	**1724**	**0.024**
477	5	Abnormality of the forebrain morphology	HP:0100547	3	1	2	7	16	7	2	**38**	**1198**	**0.032**
478	6	Abnormality of the cerebrum	HP:0002060	3	1	2	7	16	7	2	**38**	**1177**	**0.032**
479	7	Aplasia/hypoplasia of the cerebrum	HP:0007364	3	1	2	7	16	7	2	**38**	**897**	**0.042**
480	8	Microcephaly	HP:0000252	3	1	1	7	15	6	1	**34**	**736**	**0.046**
481	9	Progressive microcephaly	HP:0000253	1	0	0	2	6	5	0	**14**	**53**	**0.264**
482	9	Postnatal microcephaly	HP:0005484	0	1	0	0	0	0	0	**1**	**51**	**0.020**
483	8	Atrophy/degeneration affecting cerebrum	HP:0007369	2	1	2	1	4	2	1	**13**	**355**	**0.037**
484	9	Cerebral atrophy	HP:0002059	2	1	2	1	4	2	1	**13**	**348**	**0.037**
485	8	Aplasia/hypoplasia of the corpus callosum	HP:0007370	0	1	1	0	6	3	0	**11**	**400**	**0.028**
486	9	Hypoplasia of the corpus callosum	HP:0002079	0	1	1	0	5	3	0	**10**	**206**	**0.049**
487	9	Agenesis of the corpus callosum	HP:0001274	0	0	0	0	1	0	0	**1**	**187**	**0.005**
488	7	Abnormality of the cerebral subcortex	HP:0010993	0	0	0	1	2	2	0	**5**	**510**	**0.010**
489	8	Abnormality of the cerebral white matter	HP:0002500	0	0	0	1	2	2	0	**5**	**474**	**0.011**

490	9	Cerebral white matter atrophy	HP:0012762	0	0	0	1	2	2	0	**5**	**9**	**0.556**
491	10	Abnormality of the periventricular white matter	HP:0002518	0	0	0	0	2	0	0	**2**	**37**	**0.054**
492	11	Periventricular leukomalacia	HP:0006970	0	0	0	0	2	0	0	**2**	**8**	**0.250**
493	5	Abnormality of the hindbrain morphology	HP:0011282	3	0	1	5	3	2	2	**16**	**886**	**0.018**
494	6	Abnormality of the metencephalon	HP:0011283	3	0	1	5	3	2	2	**16**	**886**	**0.018**
495	7	Abnormality of the cerebellum	HP:0001317	3	0	1	5	3	2	2	**16**	**884**	**0.018**
496	8	Cerebellar atrophy	HP:0001272	2	0	0	5	1	2	2	**12**	**202**	**0.059**
497	9	Diffuse cerebellar atrophy	HP:0100275	0	0	0	3	0	0	1	**4**	**3**	**1.333**
498	8	Ataxia	HP:0001251	0	0	1	4	0	0	1	**6**	**602**	**0.010**
499	9	Dysmetria	HP:0001310	0	0	0	3	0	0	1	**4**	**85**	**0.047**
500	9	Dysdiadochokinesis	HP:0002075	0	0	0	3	0	0	0	**3**	**30**	**0.100**
501	9	Truncal ataxia	HP:0002078	0	0	0	1	0	0	0	**1**	**30**	**0.033**
502	9	Gait ataxia	HP:0002066	0	0	0	3	0	0	0	**3**	**78**	**0.038**
503	10	Progressive gait ataxia	HP:0007240	0	0	0	3	0	0	0	**3**	**5**	**0.600**
504	8	Aplasia/hypoplasia of the cerebellum	HP:0007360	0	0	0	0	2	0	0	**2**	**235**	**0.009**
505	9	Cerebellar hypoplasia	HP:0001321	0	0	0	0	2	0	0	**2**	**146**	**0.014**
506	8	Cerebellar malformation	HP:0002438	1	0	0	0	0	0	0	**1**	**219**	**0.005**
507	9	Abnormality of the cerebellar vermis	HP:0002334	1	0	0	0	0	0	0	**1**	**178**	**0.006**
508	10	Aplasia/hypoplasia of the cereb. vermis	HP:0006817	1	0	0	0	0	0	0	**1**	**163**	**0.006**
509	11	Hypoplastic cerebellar vermis	HP:0001320	1	0	0	0	0	0	0	**1**	**141**	**0.007**
510	5	Abnormality of brain stem morphology	HP:0002363	0	0	0	1	0	0	1	**2**	**82**	**0.024**
511	6	Atrophy/degeneration affecting the brain stem	HP:0007366	0	0	0	1	0	0	1	**2**	**11**	**0.182**
512	5	Abnormality of the cerebral ventricles	HP:0002118	2	0	0	1	2	4	1	**10**	**516**	**0.019**
513	6	Ventriculomegaly	HP:0002119	2	0	0	1	2	4	1	**10**	**381**	**0.026**
514	5	Abnormality of the pituitary gland	HP:0012503	0	1	0	0	1	0	0	**2**	**212**	**0.009**
515	6	Abnormal size of pituitary gland	HP:0012504	0	1	0	0	0	0	0	**1**	**6**	**0.167**
516	7	Small pituitary gland	HP:0012506	0	1	0	0	0	0	0	**1**	**2**	**0.500**
517	6	Abnormality of the anterior pituitary	HP:0011747	0	0	0	0	1	0	0	**1**	**194**	**0.005**
518	7	Hyperpituitarism	HP:0010514	0	0	0	0	1	0	0	**1**	**68**	**0.015**
519	8	Thyroid-stimulating hormone excess	HP:0002925	0	0	0	0	1	0	0	**1**	**11**	**0.091**
520	4	Abnormal CNS myelination	HP:0011400	0	0	0	1	2	1	0	**4**	**131**	**0.031**
521	5	Delayed CNS myelination	HP:0002188	0	0	0	1	2	1	0	**4**	**15**	**0.267**
522	3	Abnormal peripheral nervous system morphology	HP:0000759	0	0	0	1	0	1	2	**4**	**635**	**0.006**
523	4	Peripheral neuropathy	HP:0009830	0	0	0	1	0	1	2	**4**	**420**	**0.010**
524	2	Abnormality of nervous system physiology	HP:0012638	3	1	2	10	17	7	3	**43**	**2474**	**0.017**
525	3	Abnormality of higher mental function	HP:0011446	0	0	0	3	1	0	0	**4**	**1024**	**0.004**
526	4	Reduced consciousness	HP:0004372	0	0	0	0	1	0	0	**1**	**234**	**0.004**
527	5	Lethargy	HP:0001254	0	0	0	0	1	0	0	**1**	**103**	**0.010**
528	4	Neurological speech impairment	HP:0002167	0	0	0	3	1	0	0	**4**	**635**	**0.006**
529	5	Dysarthria	HP:0001260	0	0	0	3	0	0	0	**3**	**359**	**0.008**
530	3	Neurodevelopmental abnormality	HP:0012759	3	1	2	10	16	4	2	**38**	**1801**	**0.021**

531	4	Intellectual disability	HP:0001249	2	1	0	10	9	0	2	**24**	**1290**	**0.019**
532	5	Mild intellectual disability	HP:0001256	0	0	0	3	0	0	0	**3**	**137**	**0.022**
533	5	Moderate intellectual disability	HP:0002342	2	0	0	3	0	0	0	**5**	**50**	**0.100**
534	5	Severe intellectual disability	HP:0010864	0	0	0	4	0	0	0	**4**	**159**	**0.025**
535	4	Neurodevelopmental delay	HP:0012758	3	1	2	6	16	4	1	**33**	**1310**	**0.025**
536	5	Global developmental delay	HP:0001263	3	1	2	6	16	4	1	**33**	**1123**	**0.029**
537	6	Mild global developmental delay	HP:0011342	2	0	0	2	2	0	0	**6**	**22**	**0.273**
538	6	Moderate global developmental delay	HP:0011343	1	0	1	0	0	0	0	**2**	**7**	**0.286**
539	6	Severe global developmental delay	HP:0011344	0	0	0	1	1	0	0	**2**	**66**	**0.030**
540	6	Profound global developmental delay	HP:0012736	0	0	0	1	0	2	0	**3**	**24**	**0.125**
541	5	Delayed speech and language development	HP:0000750	0	0	1	4	0	0	1	**6**	**291**	**0.021**
542	6	Absent speech	HP:0001344	0	0	1	4	0	0	1	**6**	**77**	**0.078**
543	4	Developmental regression	HP:0002376	0	0	1	0	0	0	1	**2**	**197**	**0.010**
544	3	Seizures	HP:0001250	0	1	2	1	4	5	2	**15**	**1174**	**0.013**
545	4	Generalized seizures	HP:0002197	0	1	0	1	0	0	0	**2**	**136**	**0.015**
546	5	Generalized tonic seizures	HP:0010818	0	1	0	0	0	0	0	**1**	**10**	**0.100**
547	4	Focal seizures	HP:0007359	0	0	0	0	1	0	0	**1**	**50**	**0.020**
548	3	Abnormal nervous system electrophysiology	HP:0001311	0	0	0	0	0	3	0	**3**	**334**	**0.009**
549	4	Abnormality of central nervous system electrophysiology	HP:0030178	0	0	0	0	0	3	0	**3**	**257**	**0.012**
550	5	EEG abnormality	HP:0002353	0	0	0	0	0	3	0	**3**	**246**	**0.012**
551	3	Reduced tendon reflexes	HP:0001315	0	0	0	4	1	5	2	**12**	**347**	**0.035**
552	4	Areflexia	HP:0001284	0	0	0	0	1	2	0	**3**	**160**	**0.019**
553	4	Hyporeflexia	HP:0001265	0	0	0	4	0	2	2	**8**	**226**	**0.035**
554	5	Hyporeflexia of lower limbs	HP:0002600	0	0	0	1	0	0	0	**1**	**16**	**0.063**
555	3	Abnormality of central motor function	HP:0011442	0	0	1	2	0	0	0	**3**	**1294**	**0.002**
556	4	Upper motor neuron dysfunction	HP:0002493	0	0	1	1	0	0	0	**2**	**944**	**0.002**
557	5	Hyperreflexia	HP:0001347	0	0	1	1	0	0	0	**2**	**502**	**0.004**
558	4	Abnormality of coordination	HP:0011443	0	0	0	1	0	0	0	**1**	**657**	**0.002**
559	5	Slurred speech	HP:0001350	0	0	0	1	0	0	0	**1**	**25**	**0.040**
560	6	Slow slurred speech	HP:0007164	0	0	0	1	0	0	0	**1**	**2**	**0.500**
561	3	Abnormality of movement	HP:0100022	0	0	1	2	1	0	1	**5**	**1069**	**0.005**
562	4	Involuntary movements	HP:0004305	0	0	1	0	0	0	1	**2**	**558**	**0.004**
563	5	Myoclonus	HP:0001336	0	0	0	0	0	0	1	**1**	**143**	**0.007**
564	4	Gait disturbance	HP:0001288	0	0	0	2	1	0	0	**3**	**536**	**0.006**
565	5	Broad-based gait	HP:0002136	0	0	0	1	0	0	0	**1**	**33**	**0.030**
566	5	Inability to walk	HP:0002540	0	0	0	1	1	0	0	**2**	**53**	**0.038**
567	3	Behavioral abnormality	HP:0000708	0	0	0	3	2	1	0	**6**	**965**	**0.006**
568	4	Short attention span	HP:0000736	0	0	0	1	0	0	0	**1**	**144**	**0.007**
569	5	Attention deficit hyperactive disorder	HP:0007018	0	0	0	1	0	0	0	**1**	**146**	**0.007**
570	4	Abnormal emotion/affect behavior	HP:0100851	0	0	0	0	1	1	0	**2**	**246**	**0.008**
571	5	Irritability	HP:0000737	0	0	0	0	0	1	0	**1**	**104**	**0.010**

572	5	Abnormal aggressive, impulsive or violent behav.	HP:0006919	0	0	0	0	1	1	0	**2**	**146**	**0.014**
573	6	Aggressive behaviour	HP:0000718	0	0	0	0	1	1	0	**2**	**121**	**0.017**
574	4	Autistic behaviour	HP:0000729	0	0	0	2	1	0	0	**3**	**205**	**0.015**
575	4	Stereotypic behaviours	HP:0000733	0	0	0	0	1	0	0	**1**	**66**	**0.015**
576	3	Encephalopathy	HP:0001298	0	0	0	0	0	0	1	**1**	**170**	**0.006**
577	4	Acute encephalopathy	HP:0006846	0	0	0	0	0	0	1	**1**	**25**	**0.040**
578	1	Abnormality of the respiratory system	HP:0002086	1	0	0	0	3	5	0	**9**	**1215**	**0.007**
579	2	Abnormal respiratory system morphology	HP:0012252	1	0	0	0	2	3	0	**6**	**818**	**0.007**
580	3	Abnormality of the upper respiratory tract	HP:0002087	0	0	0	0	1	1	0	**2**	**340**	**0.006**
581	4	Abnormality of the larynx	HP:0001600	0	0	0	0	0	1	0	**1**	**146**	**0.007**
582	5	Laryngomalacia	HP:0001601	0	0	0	0	0	1	0	**1**	**46**	**0.022**
583	4	Abnormality of the tracheobronchial system	HP:0005607	0	0	0	0	1	0	0	**1**	**226**	**0.004**
584	5	Abnormality of the bronchi	HP:0002109	0	0	0	0	1	0	0	**1**	**108**	**0.009**
585	6	Bronchiectasis	HP:0002110	0	0	0	0	1	0	0	**1**	**84**	**0.012**
586	3	Abnormality of the lung	HP:0002088	1	0	0	0	1	2	0	**4**	**676**	**0.006**
587	4	Pneumothorax	HP:0002107	0	0	0	0	1	0	0	**1**	**24**	**0.042**
588	4	Abnormality of the pulmonary vasculature	HP:0004930	1	0	0	0	0	2	0	**3**	**99**	**0.030**
589	5	Abnormality of the pulmonary artery	HP:0004414	1	0	0	0	0	2	0	**3**	**81**	**0.037**
590	6	Elevated pulmonary artery pressure	HP:0004890	1	0	0	0	0	2	0	**3**	**94**	**0.032**
591	7	Pulmonary hypertension	HP:0002092	1	0	0	0	0	2	0	**3**	**94**	**0.032**
592	2	Functional respiratory abnormality	HP:0002795	0	0	0	0	2	3	0	**5**	**816**	**0.006**
593	3	Respiratory insufficiency	HP:0002093	0	0	0	0	2	3	0	**5**	**408**	**0.012**
594	4	Neonatal respiratory distress	HP:0002643	0	0	0	0	2	2	0	**4**	**23**	**0.174**
595	5	Neonatal asphyxia	HP:0012768	0	0	0	0	0	2	0	**2**	**1**	**2.000**
596	1	Abnormality of the skeletal system	HP:0000924	2	0	0	6	11	5	1	**25**	**2129**	**0.012**
597	2	Abnormality of skeletal morphology	HP:0011842	2	0	0	6	11	5	1	**25**	**2049**	**0.012**
598	3	Skeletal dysplasia	HP:0002652	0	0	0	0	7	0	0	**7**	**149**	**0.047**
599	3	Abnormal axial skeleton morphology	HP:0009121	2	0	0	4	2	0	1	**9**	**1706**	**0.005**
600	4	Abnormality of the vertebral column	HP:0000925	2	0	0	4	1	0	1	**8**	**922**	**0.009**
601	5	Abnormality of the vertebrae	HP:0003468	2	0	0	0	0	0	0	**2**	**316**	**0.006**
602	6	Abnormal form of the vertebral bodies	HP:0003312	1	0	0	0	0	0	0	**1**	**235**	**0.004**
603	7	Vertebral clefting	HP:0008428	1	0	0	0	0	0	0	**1**	**22**	**0.045**
604	8	Butterfly vertebrae	HP:0003316	1	0	0	0	0	0	0	**1**	**10**	**0.100**
605	6	Vertebral segmentation defect	HP:0003422	1	0	0	0	0	0	0	**1**	**103**	**0.010**
606	5	Abnormality of the curvature of the vert. column	HP:0010674	1	0	0	4	1	0	1	**7**	**705**	**0.010**
607	6	Scoliosis	HP:0002650	1	0	0	4	1	0	1	**7**	**649**	**0.011**
608	7	Thoracic scoliosis	HP:0002943	1	0	0	0	0	0	0	**1**	**14**	**0.071**
609	7	Kyphoscoliosis	HP:0002751	1	0	0	0	0	0	0	**1**	**94**	**0.011**
610	4	Abnormality of the thorax	HP:0000765	2	0	0	0	1	0	0	**3**	**541**	**0.006**
611	5	Abnormality of the rib cage	HP:0001547	2	0	0	0	1	0	0	**3**	**254**	**0.012**
612	6	Asymmetry of the thorax	HP:0001555	0	0	0	0	1	0	0	**1**	**8**	**0.125**

613	6	Abnormality of the ribs	HP:0000772	2	0	0	0	0	0	0	**2**	**199**	**0.010**
614	7	Rib fusion	HP:0000902	2	0	0	0	0	0	0	**2**	**27**	**0.074**
615	7	Rib gap	HP:0030280	1	0	0	0	0	0	0	**1**	**1**	**1.000**
616	8	Posterior rib gap	HP:0030282	1	0	0	0	0	0	0	**1**	**1**	**1.000**
617	3	Abnormal appendicular skeleton morphology	HP:0011844	1	0	0	0	1	2	0	**4**	**1084**	**0.004**
618	4	Abnormality of long bone morphology	HP:0011314	1	0	0	0	0	2	0	**3**	**470**	**0.006**
619	5	Short long bones	HP:0003026	1	0	0	0	0	1	0	**2**	**153**	**0.013**
620	5	Abnormality of epiphysis morphology	HP:0005930	0	0	0	0	0	1	0	**1**	**205**	**0.005**
621	6	Absent epiphyses	HP:0010577	0	0	0	0	0	1	0	**1**	**13**	**0.077**
622	7	Absent knee epiphyses	HP:0006400	0	0	0	0	0	1	0	**1**	**1**	**1.000**
623	4	Abnormality of pelvic girdle bone morphology	HP:0002644	1	0	0	0	1	0	0	**2**	**388**	**0.005**
624	5	Abnormality of the hip bone	HP:0003272	1	0	0	0	1	0	0	**2**	**329**	**0.006**
625	6	Hip dysplasia	HP:0001385	0	0	0	0	1	0	0	**1**	**91**	**0.011**
626	6	Abnormality of the ilium	HP:0002867	1	0	0	0	0	0	0	**1**	**72**	**0.014**
627	7	Squared iliac bones	HP:0003177	1	0	0	0	0	0	0	**1**	**4**	**0.250**
628	6	Abnormality of the hip joint	HP:0001384	1	0	0	0	0	0	0	**1**	**200**	**0.005**
629	7	Abnormality of the acetabulum	HP:0003170	1	0	0	0	0	0	0	**1**	**49**	**0.020**
630	8	Flat acetabular roof	HP:0003180	1	0	0	0	0	0	0	**1**	**16**	**0.063**
631	3	Abnormal joint morphology	HP:0001367	1	0	0	2	9	3	1	**16**	**999**	**0.016**
632	4	Abnormality of joint mobility	HP:0011729	1	0	0	2	9	3	1	**16**	**795**	**0.020**
633	5	Joint hypermobility	HP:0001382	0	0	0	2	9	0	0	**11**	**229**	**0.048**
634	5	Flexion contracture	HP:0001371	1	0	0	2	1	3	1	**8**	**492**	**0.016**
635	6	Congenital contracture	HP:0002803	0	0	0	0	1	1	0	**2**	**120**	**0.017**
636	7	Arthrogryposis multiplex congenita	HP:0002804	0	0	0	0	1	0	0	**1**	**102**	**0.010**
637	7	Distal arthrogryposis	HP:0005684	0	0	0	0	0	1	0	**1**	**12**	**0.083**
638	6	Multiple joint contractures	HP:0002828	0	0	0	0	7	2	0	**9**	**11**	**0.818**
639	6	Flexion contracture of digit	HP:0030044	1	0	0	2	0	2	0	**5**	**216**	**0.023**
640	7	Camptodactyly	HP:0012385	1	0	0	2	0	2	0	**5**	**212**	**0.024**
641	8	Camptodactyly of finger	HP:0100490	1	0	0	2	0	2	0	**5**	**146**	**0.034**
642	9	Camptodactyly of 2nd - 5th fingers	HP:0001215	1	0	0	1	0	0	0	**2**	**1**	**2.000**
643	6	Limb joint contracture	HP:0003121	0	0	0	1	0	0	1	**2**	**256**	**0.008**
644	7	Contractures of the joints of upper limbs	HP:0100360	0	0	0	1	0	0	1	**2**	**208**	**0.010**
645	8	Elbow flexion contracture	HP:0002987	0	0	0	1	0	0	0	**1**	**46**	**0.022**
646	7	Contractures of the joints of lower limbs	HP:0005750	0	0	0	1	0	0	1	**2**	**94**	**0.021**
647	8	Knee flexion contracture	HP:0006380	0	0	0	1	0	0	0	**1**	**39**	**0.026**
648	3	Abnormal bone structure	HP:0003330	1	0	0	0	1	1	0	**3**	**565**	**0.005**
649	4	Abnormal bone ossification	HP:0011849	0	0	0	0	0	1	0	**1**	**497**	**0.002**
650	5	Abnormal enchondral ossification	HP:0003336	0	0	0	0	0	1	0	**1**	**100**	**0.010**
651	6	Abnormal humeral ossification	HP:0012791	0	0	0	0	0	1	0	**1**	**1**	**1.000**
652	7	Absent humeral epiphyseal ossification	HP:0003892	0	0	0	0	0	1	0	**1**	**0**	**0.000**
653	4	Abnormality of bone mineral density	HP:0004348	1	0	0	0	1	0	0	**2**	**419**	**0.005**

654	5	Reduced bone mineral density	HP:0004349	1	0	0	0	1	0	0	**2**	**360**	**0.006**
655	6	Osteopenia	HP:0000938	1	0	0	0	1	0	0	**2**	**176**	**0.011**
656	1	Growth abnormality	HP:0001507	3	0	1	6	14	8	2	**34**	**1708**	**0.020**
657	2	Growth delay	HP:0001510	3	0	1	6	3	7	1	**21**	**1206**	**0.017**
658	3	Short stature	HP:0004322	2	0	1	5	3	3	1	**15**	**919**	**0.016**
659	3	Intrauterine growth retardation	HP:0001511	2	0	0	2	1	5	0	**10**	**319**	**0.031**
660	2	Abnormality of body weight	HP:0004323	1	0	1	3	12	8	1	**26**	**1060**	**0.025**
661	3	Decreased body weight	HP:0004325	1	0	1	3	12	8	1	**26**	**870**	**0.030**
662	4	Failure to thrive	HP:0001508	1	0	1	3	12	8	1	**26**	**637**	**0.041**
663	1	Mortality/Aging	HP:0040006	0	0	1	0	8	7	0	**16**	**231**	**0.069**
664	2	Age of death	HP:0011420	0	0	1	0	8	7	0	**16**	**231**	**0.069**
665	3	Death in infancy	HP:0001522	0	0	1	0	7	7	0	**15**	**132**	**0.114**
666	3	Neonatal death	HP:0003811	0	0	0	0	1	0	0	**1**	**17**	**0.059**

Supplementary Table 3: Genes associated to phenotypic features discussed throughout the manuscript

Gene	Gene name (GeneCards)
Abnormality of temperature regulation (HP:0004370), 244 genes	
ABCA1	ATP Binding Cassette Subfamily A Member 1
ABCA12	ATP Binding Cassette Subfamily A Member 12
ABCC2	ATP Binding Cassette Subfamily C Member 2
ABL1	ABL Proto-Oncogene 1, Non-Receptor Tyrosine Kinase
ACADSB	Acyl-CoA Dehydrogenase Short/Branched Chain
ADA	Adenosine Deaminase
ADA2	Adenosine Deaminase 2
ADAMTS13	ADAM Metallopeptidase With Thrombospondin Type 1 Motif 13
AK2	Adenylate Kinase 2
ALG11	ALG11 Alpha-1,2-Mannosyltransferase
ALPL	Alkaline Phosphatase, Biomineralization Associated
AQP2	Aquaporin 2
ASCL1	Achaete-Scute Family BHLH Transcription Factor 1
ATM	ATM Serine/Threonine Kinase
ATP1A2	ATPase Na+/K+ Transporting Subunit Alpha 2
ATP7A	ATPase Copper Transporting Alpha
AVP	Arginine Vasopressin
AVPR2	Arginine Vasopressin Receptor 2
BCL10	BCL10 Immune Signaling Adaptor
BCL2	BCL2 Apoptosis Regulator
BCL6	BCL6 Transcription Repressor
BCR	BCR Activator Of RhoGEF And GTPase
BDNF	Brain Derived Neurotrophic Factor
BIRC3	Baculoviral IAP Repeat Containing 3
BLNK	B Cell Linker
BRCA2	BRCA2, DNA Repair Associated
BTK	Bruton Tyrosine Kinase
BTNL2	Butyrophilin Like 2
C4A	Complement C4A (Rodgers Blood Group)
CACNA1A	Calcium Voltage-Gated Channel Subunit Alpha1 A
CCND1	Cyclin D1
CCR1	C-C Motif Chemokine Receptor 1
CD27	CD27 Molecule
CD79A	CD79a Molecule
CD79B	CD79b Molecule
CHD7	Chromodomain Helicase DNA Binding Protein 7
CHEK2	Checkpoint Kinase 2
CHRNA1	Cholinergic Receptor Nicotinic Alpha 1 Subunit
CHRND	Cholinergic Receptor Nicotinic Delta Subunit
CHRNG	Cholinergic Receptor Nicotinic Gamma Subunit
CLCF1	Cardiotrophin Like Cytokine Factor 1
CLCN7	Chloride Voltage-Gated Channel 7
CLEC7A	C-Type Lectin Domain Containing 7A
COL1A1	Collagen Type I Alpha 1 Chain
COX1	Mitochondrially Encoded Cytochrome C Oxidase I
COX2	Mitochondrially Encoded Cytochrome C Oxidase II
COX3	Mitochondrially Encoded Cytochrome C Oxidase III
CRLF1	Cytokine Receptor Like Factor 1
CTLA4	Cytotoxic T-Lymphocyte Associated Protein 4
CYBA	Cytochrome B-245 Alpha Chain
CYBB	Cytochrome B-245 Beta Chain

Gene	Gene name (GeneCards)
Abnormally lax or hyperextensible skin (HP:0008067), 109 genes	
ABCC6	ATP Binding Cassette Subfamily C Member 6
ACD	ACD Shelterin Complex Subunit And Telomerase Recruitment Factor
ADAMTS2	ADAM Metallopeptidase With Thrombospondin Type 1 Motif 2
AKT3	AKT Serine/Threonine Kinase 3
ALDH18A1	Aldehyde Dehydrogenase 18 Family Member A1
ANTXR1	ANTXR Cell Adhesion Molecule 1
ARID1B	AT-Rich Interaction Domain 1B
ATP6V0A2	ATPase H+ Transporting V0 Subunit A2
ATP6V1A	ATPase H+ Transporting V1 Subunit A
ATP6V1E1	ATPase H+ Transporting V1 Subunit E1
ATP7A	ATPase Copper Transporting Alpha
B3GALT6	Beta-1,3-Galactosyltransferase 6
B3GAT3	Beta-1,3-Glucuronyltransferase 3
B4GALT7	Beta-1,4-Galactosyltransferase 7
BAZ1B	Bromodomain Adjacent To Zinc Finger Domain 1B
BRAF	B-Raf Proto-Oncogene, Serine/Threonine Kinase
C1R	Complement C1r
C1S	Complement C1s
CD96	CD96 Molecule
CHST14	Carbohydrate Sulfotransferase 14
CHST3	Carbohydrate Sulfotransferase 3
CLIP2	CAP-Gly Domain Containing Linker Protein 2
COL1A1	Collagen Type I Alpha 1 Chain
COL1A2	Collagen Type I Alpha 2 Chain
COL3A1	Collagen Type III Alpha 1 Chain
COL5A1	Collagen Type V Alpha 1 Chain
COL5A2	Collagen Type V Alpha 2 Chain
CSTA	Cystatin A
DKC1	Dyskerin Pseudouridine Synthase 1
EED	Embryonic Ectoderm Development
EFEMP2	EGF Containing Fibulin Like Extracellular Matrix Protein 2
ELN	Elastin
ENPP1	Ectonucleotide Pyrophosphatase/Phosphodiesterase 1
EXT1	Exostosin Glycosyltransferase 1
EZH2	Enhancer Of Zeste 2 Polycomb Repressive Complex 2 Subunit
FBLN5	Fibulin 5
FBN1	Fibrillin 1
FGD1	FYVE, RhoGEF And PH Domain Containing 1
FGF10	Fibroblast Growth Factor 10
FGF20	Fibroblast Growth Factor 20
FGFR2	Fibroblast Growth Factor Receptor 2
FGFR3	Fibroblast Growth Factor Receptor 3
FIG4	FIG4 Phosphoinositide 5-Phosphatase
FKBP14	FKBP Prolyl Isomerase 14
FOXC1	Forkhead Box C1
GGCX	Gamma-Glutamyl Carboxylase
GORAB	Golgin, RAB6 Interacting
GPX4	Glutathione Peroxidase 4
GSN	Gelsolin
GTF2I	General Transcription Factor IIi
GTF2IRD1	GTF2I Repeat Domain Containing 1

CYP11B2	Cytochrome P450 Family 11 Subfamily B Member 2
CYP21A2	Cytochrome P450 Family 21 Subfamily A Member 2
DBH	Dopamine Beta-Hydroxylase
DCLRE1C	DNA Cross-Link Repair 1C
DDB2	Damage Specific DNA Binding Protein 2
DDC	Dopa Decarboxylase
DGUOK	Deoxyguanosine Kinase
DIS3L2	DIS3 Like 3'-5' Exoribonuclease 2
DST	Dystonin
EDA	Ectodysplasin A
EDAR	Ectodysplasin A Receptor
EDARADD	EDAR Associated Death Domain
EDN3	Endothelin 3
ELANE	Elastase, Neutrophil Expressed
ELP1	Elongator Complex Protein 1
ERAP1	Endoplasmic Reticulum Aminopeptidase 1
ERCC2	ERCC Excision Repair 2, TFIIH Core Complex Helicase Subunit
ERCC3	ERCC Excision Repair 3, TFIIH Core Complex Helicase Subunit
ERCC4	ERCC Excision Repair 4, Endonuclease Catalytic Subunit
ERCC5	ERCC Excision Repair 5, Endonuclease
F5	Coagulation Factor V
FAS	Fas Cell Surface Death Receptor
FBP1	Fibrillin 1
FOXP1	Forkhead Box P1
GAA	Glucosidase Alpha, Acid
GALC	Galactosylceramidase
GATA2	GATA Binding Protein 2
GCDH	Glutaryl-CoA Dehydrogenase
GCH1	GTP Cyclohydrolase 1
GDNF	Glial Cell Derived Neurotrophic Factor
GLA	Galactosidase Alpha
GPC3	Glypican 3
GPR35	G Protein-Coupled Receptor 35
H19	H19 Imprinted Maternally Expressed Transcript
HBB	Hemoglobin Subunit Beta
HERC2	HECT And RLD Domain Containing E3 Ubiquitin Protein Ligase 2
HEXB	Hexosaminidase Subunit Beta
HLA-B	Major Histocompatibility Complex, Class I, B
HLA-DPB1	Major Histocompatibility Complex, Class II, DP Beta 1
HLA-DRB1	Major Histocompatibility Complex, Class II, DR Beta 1
HMGCL	3-Hydroxy-3-Methylglutaryl-CoA Lyase
HSPG2	Heparan Sulfate Proteoglycan 2
HTR1A	5-Hydroxytryptamine Receptor 1A
IGH	Immunoglobulin Heavy Locus
IGHM	Immunoglobulin Heavy Constant Mu
IGLL1	Immunoglobulin Lambda Like Polypeptide 1
IKZF1	IKAROS Family Zinc Finger 1
IL10	Interleukin 10
IL12A	Interleukin 12A
IL12A-AS1	IL12A Antisense RNA 1
IL12B	Interleukin 12B
IL17F	Interleukin 17F
HPGD	15-Hydroxyprostaglandin Dehydrogenase
HRAS	HRas Proto-Oncogene, GTPase
IFT43	Intraflagellar Transport 43
KRAS	KRAS Proto-Oncogene, GTPase
LIMK1	LIM Domain Kinase 1
LTBP4	Latent Transforming Growth Factor Beta Binding Protein 4
MAP2K1	Mitogen-Activated Protein Kinase Kinase 1
MAP2K2	Mitogen-Activated Protein Kinase Kinase 1
MAPRE2	Microtubule Associated Protein RP/EB Family Member 2
MEGF8	Multiple EGF Like Domains 8
MLXIPL	MLX Interacting Protein Like
MRPS16	Mitochondrial Ribosomal Protein S16
MRPS22	Mitochondrial Ribosomal Protein S22
NAA10	N(Alpha)-Acetyltransferase 10, NatA Catalytic Subunit
NBAS	Neuroblastoma Amplified Sequence
NPR2	Natriuretic Peptide Receptor 2
NSD1	Nuclear Receptor Binding SET Domain Protein 1
OSMR	Oncostatin M Receptor
PARN	Poly(A)-Specific Ribonuclease
PAX2	Paired Box 2
PDGFRB	Platelet Derived Growth Factor Receptor Beta
PEX1	Peroxisomal Biogenesis Factor 1
PIK3R1	Phosphoinositide-3-Kinase Regulatory Subunit 1
PITX2	Paired Like Homeodomain 2
PLOD1	Procollagen-Lysine,2-Oxoglutarate 5-Dioxygenase 1
PRDM5	PR/SET Domain 5
PTDSS1	Phosphatidylserine Synthase 1
PTEN	Phosphatase And Tensin Homolog
PTPN11	Protein Tyrosine Phosphatase, Non-Receptor Type 11
PYCR1	Pyrroline-5-Carboxylate Reductase 1
RAF1	Raf-1 Proto-Oncogene, Serine/Threonine Kinase
RFC2	Replication Factor C Subunit 2
RIN2	Ras And Rab Interactor 2
RIT1	Ras Like Without CAAX 1
RPS6KA3	Ribosomal Protein S6 Kinase A3
RTEL1	Regulator Of Telomere Elongation Helicase 1
SKI	SKI Proto-Oncogene
SLC26A2	Solute Carrier Family 26 Member 2
SLC2A10	Solute Carrier Family 2 Member 10
SLC39A13	Solute Carrier Family 39 Member 13
SLC6A8	Solute Carrier Family 6 Member 8
SLC7A7	Solute Carrier Family 7 Member 7
SMARCA2	SWI/SNF Related, Matrix Associated, Actin Dependent Regulator Of Chromatin, Subfamily A, Member 2
SPINT2	Serine Peptidase Inhibitor, Kunitz Type 2
SRD5A3	Steroid 5 Alpha-Reductase 3
SUZ12	SUZ12 Polycomb Repressive Complex 2 Subunit
TBL2	Transducin Beta Like 2
TBX15	T-Box 15
TERT	Telomerase Reverse Transcriptase
TGM5	Transglutaminase 5
TINF2	TERF1 Interacting Nuclear Factor 2
TNXB	Tenascin XB

IL17RA	Interleukin 17 Receptor A
IL17RC	Interleukin 17 Receptor C
IL23R	Interleukin 23 Receptor
IL2RG	Interleukin 2 Receptor Subunit Gamma
IL36RN	Interleukin 36 Receptor Antagonist
IL6	Interleukin 6
IL7R	Interleukin 7 Receptor
IPW	Imprinted In Prader-Willi Syndrome
IRF8	Interferon Regulatory Factor 8
ITPR2	Inositol 1,4,5-Trisphosphate Receptor Type 2
JAK2	Janus Kinase 2
KCNJ1	Potassium Voltage-Gated Channel Subfamily J Member 1
KLRC4	Killer Cell Lectin Like Receptor C4
KRT14	Keratin 14
LACC1	Laccase Domain Containing 1
LBR	Lamin B Receptor
LIFR	LIF Receptor Alpha
LIG4	DNA Ligase 4
LIPA	Lipase A, Lysosomal Acid Type
LPIN1	Lipin 1
LPIN2	Lipin 2
LRRC8A	Leucine Rich Repeat Containing 8 VRAC Subunit A
LYST	Lysosomal Trafficking Regulator
MAGEL2	MAGE Family Member L2
MALT1	MALT1 Paracaspase
MAP2K1	Mitogen-Activated Protein Kinase Kinase 1
MAP2K2	Mitogen-Activated Protein Kinase Kinase 1
MBTPS2	Membrane Bound Transcription Factor Peptidase, Site 2
MEFV	MEFV, Pyrin Innate Immunity Regulator
MIF	Macrophage Migration Inhibitory Factor
MKRN3	Makorin Ring Finger Protein 3
MKRN3-AS1	MKRN3 Antisense RNA 1
MLX	MAX Dimerization Protein MLX
MST1	Macrophage Stimulating 1
MVK	Mevalonate Kinase
MYD88	Myeloid Differentiation Primary Response 88
MYH3	Myosin Heavy Chain 3
NALCN	Sodium Leak Channel, Non-Selective
NCF1	Neutrophil Cytosolic Factor 1
NCF2	Neutrophil Cytosolic Factor 2
NCF4	Neutrophil Cytosolic Factor 4
ND1	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 1
ND4	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 4
ND5	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 5
ND6	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 6
NDN	Necdin, MAGE Family Member
NECTIN4	Nectin Cell Adhesion Molecule 4
NFKBIA	NFKB Inhibitor Alpha
NGF	Nerve Growth Factor
NGLY1	N-Glycanase 1
NLRC4	NLR Family CARD Domain Containing 4
NLRP12	NLR Family Pyrin Domain Containing 12
TRPS1	Transcriptional Repressor GATA Binding 1
TUBB	Tubulin Beta Class I
TWIST2	Twist Family BHLH Transcription Factor 2
WDR19	WD Repeat Domain 19
XYLT2	Xylosyltransferase 2
ZNF469	Zinc Finger Protein 469

Excessive wrinkled skin (HP:0007392), 35 genes

ABCC6	ATP Binding Cassette Subfamily C Member 6
ACD	ACD Shelterin Complex Subunit And Telomerase Recruitment Factor
ADAMTS2	ADAM Metallopeptidase With Thrombospondin Type 1 Motif 2
ARID1B	AT-Rich Interaction Domain 1B
ATP6V0A2	ATPase H+ Transporting V0 Subunit A2
ATP6V1A	ATPase H+ Transporting V1 Subunit A
ATP6V1E1	ATPase H+ Transporting V1 Subunit E1
BRAF	B-Raf Proto-Oncogene, Serine/Threonine Kinase
C1R	Complement C1r
COL1A2	Collagen Type I Alpha 2 Chain
COL3A1	Collagen Type III Alpha 1 Chain
COL5A1	Collagen Type V Alpha 1 Chain
CSTA	Cystatin A
DKC1	Dyskerin Pseudouridine Synthase 1
ENPP1	Ectonucleotide Pyrophosphatase/Phosphodiesterase 1
FGFR2	Fibroblast Growth Factor Receptor 2
FGFR3	Fibroblast Growth Factor Receptor 3
KRAS	KRAS Proto-Oncogene, GTPase
MAP2K1	Mitogen-Activated Protein Kinase Kinase 1
MAP2K2	Mitogen-Activated Protein Kinase Kinase 1
PARN	Poly(A)-Specific Ribonuclease
PIK3R1	Phosphoinositide-3-Kinase Regulatory Subunit 1
PLOD1	Procollagen-Lysine,2-Oxoglutarate 5-Dioxygenase 1
PTEN	Phosphatase And Tensin Homolog
PTPN11	Protein Tyrosine Phosphatase, Non-Receptor Type 11
PYCR1	Pyrroline-5-Carboxylate Reductase 1
RAF1	Raf-1 Proto-Oncogene, Serine/Threonine Kinase
RIT1	Ras Like Without CAAX 1
RTEL1	Regulator Of Telomere Elongation Helicase 1
SMARCA2	SWI/SNF Related, Matrix Associated, Actin Dependent Regulator Of Chromatin, Subfamily A, Member 2
TERT	Telomerase Reverse Transcriptase
TGM5	Transglutaminase 5
TINF2	TERF1 Interacting Nuclear Factor 2
TWIST2	Twist Family BHLH Transcription Factor 2
ZNF469	Zinc Finger Protein 469

Elevated hepatic transaminases (HP:0002910), 157 genes

ABCB4	ATP Binding Cassette Subfamily B Member 4
ABCD3	ATP Binding Cassette Subfamily D Member 3
ACAD9	Acyl-CoA Dehydrogenase Family Member 9
ACADM	Acyl-CoA Dehydrogenase Medium Chain
ACOX1	Acyl-CoA Oxidase 1
ADA2	Adenosine Deaminase 2
ADK	Adenosine Kinase

NLRP3	NLR Family Pyrin Domain Containing 3
NME1	NME/NM23 Nucleoside Diphosphate Kinase 1
NOD2	Nucleotide Binding Oligomerization Domain Containing 2
NOTCH3	Notch Receptor 3
NPAP1	Nuclear Pore Associated Protein 1
NTRK1	Neurotrophic Receptor Tyrosine Kinase 1
ORAI1	ORAI Calcium Release-Activated Calcium Modulator 1
PAX8	Paired Box 8
PHOX2B	Paired Like Homeobox 2b
PIK3R1	Phosphoinositide-3-Kinase Regulatory Subunit 1
POU6F2	POU Class 6 Homeobox 2
PRKAR1A	Protein Kinase CAMP-Dependent Type I Regulatory Subunit Alpha
PRNP	Prion Protein
PRSS1	Serine Protease 1
PRTN3	Proteinase 3
PSMB8	Proteasome Subunit Beta 8
PSTPIP1	Proline-Serine-Threonine Phosphatase Interacting Protein 1
PTPN22	Protein Tyrosine Phosphatase Non-Receptor Type 22
PTPN3	Protein Tyrosine Phosphatase Non-Receptor Type 3
PTS	6-Pyruvoyltetrahydropterin Synthase
PWAR1	Prader Willi/Angelman Region RNA 1
PWRN1	Prader-Willi Region Non-Protein Coding RNA 1
QDPR	Quinoid Dihydropteridine Reductase
RAB27A	RAB27A, Member RAS Oncogene Family
RAG1	Recombination Activating 1
RAG2	Recombination Activating 2
RB1	RB Transcriptional Corepressor 1
REST	RE1 Silencing Transcription Factor
RET	Ret Proto-Oncogene
RMRP	RNA Component Of Mitochondrial RNA Processing Endoribonuclease
RUNX1	RUNX Family Transcription Factor 1
RYR1	Ryanodine Receptor 1
SCN4A	Sodium Voltage-Gated Channel Alpha Subunit 4
SCN9A	Sodium Voltage-Gated Channel Alpha Subunit 9
SHANK3	SH3 And Multiple Ankyrin Repeat Domains 3
SLC11A1	Solute Carrier Family 11 Member 1
SLC12A1	Solute Carrier Family 12 Member 1
SLC19A3	Solute Carrier Family 19 Member 3
SLC25A19	Solute Carrier Family 25 Member 19
SLC25A20	Solute Carrier Family 25 Member 20
SLC29A3	Solute Carrier Family 29 Member 3
SLCO1B1	Solute Carrier Organic Anion Transporter Family Member 1B1
SLCO1B3	Solute Carrier Organic Anion Transporter Family Member 1B3
SNORD115-1	Small Nucleolar RNA, C/D Box 115-1
SNORD116-1	Small Nucleolar RNA, C/D Box 116-1
SNRPN	Small Nuclear Ribonucleoprotein Polypeptide N
SNX10	Sorting Nexin 10
SPINK1	Serine Peptidase Inhibitor, Kazal Type 1
SPR	Sepiapterin Reductase
STAC3	SH3 And Cysteine Rich Domain 3
AGL	Amylo-Alpha-1, 6-Glucosidase, 4-Alpha-Glucanotransferase
AGPAT2	1-Acylglycerol-3-Phosphate O-Acyltransferase 2
AKR1D1	Aldo-Keto Reductase Family 1 Member D1
ALAS2	5'-Aminolevulinate Synthase 2
ALDOB	Aldolase, Fructose-Bisphosphate B
ALG6	ALG6 Alpha-1,3-Glucosyltransferase
ALMS1	ALMS1 Centrosome And Basal Body Associated Protein
AMACR	Alpha-Methylacyl-CoA Racemase
ATM	ATM Serine/Threonine Kinase
ATP6AP1	ATPase H+ Transporting Accessory Protein 1
ATP7B	ATPase Copper Transporting Beta
ATP8B1	ATPase Phospholipid Transporting 8B1
ATRX	ATRX, Chromatin Remodeler
BCS1L	BCS1 Homolog, Ubiquinol-Cytochrome C Reductase Complex Chaperone
BRCA1	BRCA1, DNA Repair Associated
BRCA2	BRCA2, DNA Repair Associated
BSCL2	BSCL2, Seipin Lipid Droplet Biogenesis Associated
CAVIN1	Caveolae Associated Protein 1
CBS	Cystathionine-Beta-Synthase
CC2D2A	Coiled-Coil And C2 Domain Containing 2A
CCDC115	Coiled-Coil Domain Containing 115
CD19	CD19 Molecule
CD81	CD81 Molecule
CDKN2A	Cyclin Dependent Kinase Inhibitor 2A
COG2	COG complex subunit 2
COG4	COG complex subunit 4
COG6	COG complex subunit 6
COG8	COG complex subunit 8
CPT1A	Carnitine Palmitoyltransferase 1A
CPT2	Carnitine Palmitoyltransferase 2
CR2	Complement C3d Receptor 2
CYC1	Cytochrome C1
CYP7B1	Cytochrome P450 Family 7 Subfamily B Member 1
DAXX	Death Domain Associated Protein
DCDC2	Doublecortin Domain Containing 2
DDOST	Dolichyl-Diphosphooligosaccharide--Protein Glycosyltransferase Non-Catalytic Subunit
DGUOK	Deoxyguanosine Kinase
DLD	Dihydrolipoamide Dehydrogenase
DMD	Dystrophin
DOLK	Dolichol Kinase
DPAGT1	Dolichyl-Phosphate N-Acetylglucosaminephosphotransferase 2
DPM1	Dolichyl-Phosphate Mannosyltransferase Subunit 1, Catalytic
DPM3	Dolichyl-Phosphate Mannosyltransferase Subunit 3
EIF2AK3	Eukaryotic Translation Initiation Factor 2 Alpha Kinase 3
ERCC4	ERCC Excision Repair 4, Endonuclease Catalytic Subunit
F5	Coagulation Factor V
FAH	Fumarylacetoacetate Hydrolase
FAN1	FANCD2 And FANCI Associated Nuclease 1
FBXL4	F-Box And Leucine Rich Repeat Protein 4
G6PC	Glucose-6-Phosphatase Catalytic Subunit

STAT3	Signal Transducer And Activator Of Transcription 3
STAT4	Signal Transducer And Activator Of Transcription 4
STIM1	Stromal Interaction Molecule 1
STX11	Syntaxin 11
STXBP2	Syntaxin Binding Protein 2
TBX3	T-Box 3
TCF3	Transcription Factor 3
TCF4	Transcription Factor 4
TCIRG1	T Cell Immune Regulator 1, ATPase H+ Transporting V0 Subunit A3
TLR4	Toll Like Receptor 4
TMEM165	Transmembrane Protein 165
TMEM173	Transmembrane Protein 173
TNFAIP3	TNF Alpha Induced Protein 3
TNFRSF1A	TNF Receptor Superfamily Member 1A
TNFSF11	TNF Superfamily Member 11
TP53	Tumor Protein P53
TRAF3IP2	TRAF3 Interacting Protein 2
TRAPPC9	Trafficking Protein Particle Complex 9
TREX1	Three Prime Repair Exonuclease 1
TRIP13	Thyroid Hormone Receptor Interactor 13
TRNF	Mitochondrially Encoded TRNA-Phe (UUU/C)
TRNH	Mitochondrially Encoded TRNA-His (CAU/C)
TRNL1	Mitochondrially Encoded TRNA-Leu (UUA/G) 1
TRNQ	Mitochondrially Encoded TRNA-Gln (CAA/G)
TRNS1	Mitochondrially Encoded TRNA-Ser (UCN) 1
TRNS2	Mitochondrially Encoded TRNA-Ser (AGU/C) 2
TRNW	Mitochondrially Encoded TRNA-Trp (UGA/G)
TSC1	TSC Complex Subunit 1
TSC2	TSC Complex Subunit 2
TSPYL1	TSPY Like 1
UBAC2	UBA Domain Containing 2
UNC13D	Unc-13 Homolog D
VPS11	VPS11, CORVET/HOPS Core Subunit
WAS	WASP Actin Nucleation Promoting Factor
WIPF1	WAS/WASL Interacting Protein Family Member 1
WT1	WT1 Transcription Factor
XIAP	X-Linked Inhibitor Of Apoptosis
XPA	XPA, DNA Damage Recognition And Repair Factor
XPC	XPC Complex Subunit, DNA Damage Recognition And Repair Factor

Fever (HP:0001945), 180 genes

ABCC2	ATP Binding Cassette Subfamily C Member 2
ABL1	ABL Proto-Oncogene 1, Non-Receptor Tyrosine Kinase
ADA	Adenosine Deaminase
ADA2	Adenosine Deaminase 2
ADAMTS13	ADAM Metallopeptidase With Thrombospondin Type 1 Motif 13
AK2	Adenylate Kinase 2
ALPL	Alkaline Phosphatase, Biomineralization Associated
AQP2	Aquaporin 2
ATM	ATM Serine/Threonine Kinase
ATP1A2	ATPase Na+/K+ Transporting Subunit Alpha 2
AVP	Arginine Vasopressin
GDF2	Growth Differentiation Factor 2
GLRX5	Glutaredoxin 5
GNAS	GNAS Complex Locus
GNE	Glucosamine (UDP-N-Acetyl)-2-Epimerase/N-Acetylmannosamine Kinase
GNMT	Glycine N-Methyltransferase
GPD1	Glycerol-3-Phosphate Dehydrogenase 1
GPR35	G Protein-Coupled Receptor 35
GYS2	Glycogen Synthase 2
HADH	Hydroxyacyl-CoA Dehydrogenase
HADHA	Hydroxyacyl-CoA Dehydrogenase/3-Ketoacyl-CoA Thiolase/Enoyl-CoA Hydratase (Trifunctional Protein), Alpha Subunit
HADHB	Hydroxyacyl-CoA Dehydrogenase/3-Ketoacyl-CoA Thiolase/Enoyl-CoA Hydratase (Trifunctional Protein), Beta Subunit
HAMP	Hepcidin Antimicrobial Peptide
HBB	Hemoglobin Subunit Beta
HFE	Hemochromatosis
HFE2	Hemochromatosis Type 2 (Juvenile)
HLA-B	Major Histocompatibility Complex, Class I, B
HNF1A	HNF1 Homeobox A
HNF1B	HNF1 Homeobox B
HNF4A	Hepatocyte Nuclear Factor 4 Alpha
HSD17B4	Hydroxysteroid 17-Beta Dehydrogenase 4
HSD3B7	Hydroxy-Delta-5-Steroid Dehydrogenase, 3 Beta- And Steroid Delta-Isomerase 7
IARS	Isoleucyl-TRNA Synthetase
ICOS	Inducible T-Cell Costimulator
IER3IP1	Immediate Early Response 3 Interacting Protein 1
IFT172	Intraflagellar Transport 172
IKZF1	IKAROS Family Zinc Finger 1
INPP5E	Inositol Polyphosphate-5-Phosphatase E
JAG1	Jagged 1
JAK2	Janus Kinase 2
KIF23	Kinesin Family Member 23
KRAS	KRAS Proto-Oncogene, GTPase
LARS	Leucyl-TRNA Synthetase
LBR	Lamin B Receptor
LHX1	LIM Homeobox 1
LIPT1	Lipoyltransferase 1
MARS	Methionyl-TRNA Synthetase
MPV17	MPV17, Mitochondrial Inner Membrane Protein
MRPL3	Mitochondrial Ribosomal Protein L3
MRPL44	Mitochondrial Ribosomal Protein L44
MRPS16	Mitochondrial Ribosomal Protein S16
MS4A1	Membrane Spanning 4-Domains A1
MST1	Macrophage Stimulating 1
MVK	Mevalonate Kinase
NFKB1	Nuclear Factor Kappa B Subunit 1
NFKB2	Nuclear Factor Kappa B Subunit 2
NGLY1	N-Glycanase 1
NR1H4	Nuclear Receptor Subfamily 1 Group H Member 4
OCLN	Occludin
OFD1	OFD1, Centriole And Centriolar Satellite Protein
PALB2	Partner And Localizer Of BRCA2
PALLD	Palladin, Cytoskeletal Associated Protein
PEX13	Peroxisomal Biogenesis Factor 13

Mitochond
Membrane

AVPR2	Arginine Vasopressin Receptor 2
BCL10	BCL10 Immune Signaling Adaptor
BCL2	BCL2 Apoptosis Regulator
BCL6	BCL6 Transcription Repressor
BCR	BCR Activator Of RhoGEF And GTPase
BIRC3	Baculoviral IAP Repeat Containing 3
BLNK	B Cell Linker
BRCA2	BRCA2, DNA Repair Associated
BTK	Bruton Tyrosine Kinase
BTNL2	Butyrophilin Like 2
C4A	Complement C4A (Rodgers Blood Group)
CACNA1A	Calcium Voltage-Gated Channel Subunit Alpha1 A
CCND1	Cyclin D1
CCR1	C-C Motif Chemokine Receptor 1
CD27	CD27 Molecule
CD79A	CD79a Molecule
CD79B	CD79b Molecule
CHD7	Chromodomain Helicase DNA Binding Protein 7
CHEK2	Checkpoint Kinase 2
COL1A1	Collagen Type I Alpha 1 Chain
COX1	Mitochondrially Encoded Cytochrome C Oxidase I
COX2	Mitochondrially Encoded Cytochrome C Oxidase II
COX3	Mitochondrially Encoded Cytochrome C Oxidase III
CRLF1	Cytokine Receptor Like Factor 1
CTLA4	Cytotoxic T-Lymphocyte Associated Protein 4
CYBA	Cytochrome B-245 Alpha Chain
CYBB	Cytochrome B-245 Beta Chain
CYP11B2	Cytochrome P450 Family 11 Subfamily B Member 2
CYP21A2	Cytochrome P450 Family 21 Subfamily A Member 2
DCLRE1C	DNA Cross-Link Repair 1C
DDB2	Damage Specific DNA Binding Protein 2
DIS3L2	DIS3 Like 3'-5' Exoribonuclease 2
DST	Dystonin
EDA	Ectodysplasin A
ELANE	Elastase, Neutrophil Expressed
ELP1	Elongator Complex Protein 1
ERAP1	Endoplasmic Reticulum Aminopeptidase 1
ERCC2	ERCC Excision Repair 2, TFIIH Core Complex Helicase Subunit
ERCC3	ERCC Excision Repair 3, TFIIH Core Complex Helicase Subunit
ERCC4	ERCC Excision Repair 4, Endonuclease Catalytic Subunit
ERCC5	ERCC Excision Repair 5, Endonuclease
F5	Coagulation Factor V
FAS	Fas Cell Surface Death Receptor
FBP1	Fibrillin 1
FOXP1	Forkhead Box P1
GAA	Glucosidase Alpha, Acid
GALC	Galactosylceramidase
GATA2	GATA Binding Protein 2
GCH1	GTP Cyclohydrolase 1
GLA	Galactosidase Alpha
GPC3	Glypican 3
GPR35	G Protein-Coupled Receptor 35
H19	H19 Imprinted Maternally Expressed Transcript
HBB	Hemoglobin Subunit Beta
PEX19	Peroxisomal Biogenesis Factor 19
PHKA2	Phosphorylase Kinase Regulatory Subunit Alpha 2
PHKG2	Phosphorylase Kinase Catalytic Subunit Gamma 2
PMM2	Phosphomannomutase 2
PNPLA2	Patatin Like Phospholipase Domain Containing 2
POLD1	DNA Polymerase Delta 1, Catalytic Subunit
POLG	DNA Polymerase Gamma, Catalytic Subunit
POLG2	DNA Polymerase Gamma 2, Accessory Subunit
PRKCD	Protein Kinase C Delta
PSMB8	Proteasome Subunit Beta 8
RBCK1	RANBP2-Type And C3HC4-Type Zinc Finger Containing 1
RNASEH2A	Ribonuclease H2 Subunit A
RNASEH2C	Ribonuclease H2 Subunit C
RPGRIP1L	RPGRIP1 Like
SAR1B	Secretion Associated Ras Related GTPase 1B
SBDS	SBDS, Ribosome Maturation Factor
SC5D	Sterol-C5-Desaturase
SDHD	Succinate Dehydrogenase Complex Subunit D
SERPINA1	Serpin Family A Member 1
SLC22A5	Solute Carrier Family 22 Member 5
SLC25A13	Solute Carrier Family 25 Member 13
SLC25A20	Solute Carrier Family 25 Member 20
SLC30A10	Solute Carrier Family 30 Member 10
SLC37A4	Solute Carrier Family 37 Member 4
SMAD4	SMAD Family Member 4
SRD5A3	Steroid 5 Alpha-Reductase 3
STEAP3	STEAP3 Metalloreductase
TANGO2	Transport And Golgi Organization 2 Homolog
TCF4	Transcription Factor 4
TFAM	Transcription Factor A, Mitochondrial
TFR2	Transferrin Receptor 2
TMEM165	Transmembrane Protein 165
TMEM199	Transmembrane Protein 199
TMEM67	Transmembrane Protein 67
TNFRSF13B	TNF Receptor Superfamily Member 13B
TNFRSF13C	TNF Receptor Superfamily Member 13C
TNFSF12	TNF Superfamily Member 12
TP53	Tumor Protein P53
TRAPPC11	Trafficking Protein Particle Complex 11
TREX1	Three Prime Repair Exonuclease 1
TRMT10C	TRNA Methyltransferase 10C, Mitochondrial RNase P Subunit
TRMU	TRNA 5-Methylaminomethyl-2-Thiouridylate Methyltransferase
TWNK	Twinkle MtDNA Helicase
USP18	Ubiquitin Specific Peptidase 18
VIPAS39	VPS33B Interacting Protein, Apical-Basolateral Polarity Regulator, Spe-39 Homolog
VPS13A	Vacuolar Protein Sorting 13 Homolog A
VPS33B	VPS33B, Late Endosome And Lysosome Associated
YARS2	Tyrosyl-TRNA Synthetase 2

Abnormality of coagulation (HP:0001928), 127 genes

A2ML1	Alpha-2-Macroglobulin Like 1
ABCC2	ATP Binding Cassette Subfamily C Member 2
ACD	ACD Shelterin Complex Subunit And Telomerase Recruitment Factor
AGGF1	Angiogenic Factor With G-Patch And FHA Domains

HLA-B	Major Histocompatibility Complex, Class I, B
HLA-DPB1	Major Histocompatibility Complex, Class II, DP Beta 1
HLA-DRB1	Major Histocompatibility Complex, Class II, DR Beta 1
HMGCL	3-Hydroxy-3-Methylglutaryl-CoA Lyase
HTR1A	5-Hydroxytryptamine Receptor 1A
IGH	Immunoglobulin Heavy Locus
IGHM	Immunoglobulin Heavy Constant Mu
IGLL1	Immunoglobulin Lambda Like Polypeptide 1
IKZF1	IKAROS Family Zinc Finger 1
IL10	Interleukin 10
IL12A	Interleukin 12A
IL12A-AS1	IL12A Antisense RNA 1
IL12B	Interleukin 12 B
IL23R	Interleukin 23 Receptor
IL2RG	Interleukin 2 Receptor Subunit Gamma
IL36RN	Interleukin 36 Receptor Antagonist
IL6	Interleukin 6
IL7R	Interleukin 7 Receptor
IRF8	Interferon Regulatory Factor 8
JAK2	Janus Kinase 2
KCNJ1	Potassium Voltage-Gated Channel Subfamily J Member 1
KLRC4	Killer Cell Lectin Like Receptor C4
LACC1	Laccase Domain Containing 1
LBR	Lamin B Receptor
LIFR	LIF Receptor Alpha
LIG4	DNA Ligase 4
LIPA	Lipase A, Lysosomal Acid Type
LPIN1	Lipin 1
LPIN2	Lipin 2
LRRC8A	Leucine Rich Repeat Containing 8 VRAC Subunit A
LYST	Lysosomal Trafficking Regulator
MALT1	MALT1 Paracaspase
MEFV	MEFV, Pyrin Innate Immunity Regulator
MIF	Macrophage Migration Inhibitory Factor
MLX	MAX Dimerization Protein MLX
MST1	Macrophage Stimulating 1
MVK	Mevalonate Kinase
MYD88	Myeloid Differentiation Primary Response 88
MYH3	Myosin Heavy Chain 3
NCF1	Neutrophil Cytosolic Factor 1
NCF2	Neutrophil Cytosolic Factor 2
NCF4	Neutrophil Cytosolic Factor 4
ND1	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 1
ND4	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 4
ND5	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 5
ND6	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 6
NGF	Nerve Growth Factor
NGLY1	N-Glycanase 1
NLRC4	NLR Family CARD Domain Containing 4
NLRP12	NLR Family Pyrin Domain Containing 12
AKR1D1	Aldo-Keto Reductase Family 1 Member D1
ALG12	ALG12 Alpha-1,6-Mannosyltransferase
ALG2	ALG2 Alpha-1,3/1,6-Mannosyltransferase
ALG6	ALG6 Alpha-1,3-Glucosyltransferase
AMACR	Alpha-Methylacyl-CoA Racemase
ANO6	Anoctamin 6
ATP6V0A2	ATPase H+ Transporting V0 Subunit A2
ATP6V1A	ATPase H+ Transporting V1 Subunit A
ATP6V1E1	ATPase H+ Transporting V1 Subunit E1
B4GALT1	Beta-1,4-Galactosyltransferase 1
BCS1L	BCS1 Homolog, Ubiquinol-Cytochrome C Reductase Complex Chaperone
BRAF	B-Raf Proto-Oncogene, Serine/Threonine Kinase
COG4	COG complex subunit 4
CTC1	CST Telomere Replication Complex Component 1
CYP7B1	Cytochrome P450 Family 7 Subfamily B Member 1
DDOST	Dolichyl-Diphosphooligosaccharide--Protein Glycosyltransferase Non-Catalytic Subunit
DKC1	Dyskerin Pseudouridine Synthase 1
DLD	Dihydrolipoamide Dehydrogenase
DOLK	Dolichol Kinase
DPAGT1	Dolichyl-Phosphate N-Acetylglucosaminephosphotransferase 2
DPM1	Dolichyl-Phosphate Mannosyltransferase Subunit 1, Catalytic
F10	Coagulation Factor X
F11	Coagulation Factor XI
F12	Coagulation Factor XII
F13A1	Coagulation Factor XIII A Chain
F13B	Coagulation Factor XIII B Chain
F2	Coagulation Factor II, Thrombin
F5	Coagulation Factor V
F7	Coagulation Factor VII
F8	Coagulation Factor VIII
F9	Coagulation Factor IX
FAH	Fumarylacetoacetate Hydrolase
FGA	Fibrinogen Alpha Chain
FGB	Fibrinogen Beta Chain
FGG	Fibrinogen Gamma Chain
FLNA	Filamin A
GATA2	GATA Binding Protein 2
GATA6	GATA Binding Protein 6
GFI1B	Growth Factor Independent 1B Transcriptional Repressor
GGCX	Gamma-Glutamyl Carboxylase
GNE	Glucosamine (UDP-N-Acetyl)-2-Epimerase/N-Acetylmannosamine Kinase
GPR35	G Protein-Coupled Receptor 35
HADH	Hydroxyacyl-CoA Dehydrogenase
HRG	Histidine Rich Glycoprotein
HSD3B7	Hydroxy-Delta-5-Steroid Dehydrogenase, 3 Beta- And Steroid Delta-Isomerase 7
IDH1	Isocitrate Dehydrogenase (NADP(+)) 1, Cytosolic
IDH2	Isocitrate Dehydrogenase (NADP(+)) 2, Mitochondrial
KAT6B	Lysine Acetyltransferase 6B
KLKB1	Kallikrein B1
KRAS	KRAS Proto-Oncogene, GTPase

NLRP3	NLR Family Pyrin Domain Containing 3
NME1	NME/NM23 Nucleoside Diphosphate Kinase 1
NOD2	Nucleotide Binding Oligomerization Domain Containing 2
NOTCH3	Notch Receptor 3
NTRK1	Neurotrophic Receptor Tyrosine Kinase 1
ORAI1	ORAI Calcium Release-Activated Calcium Modulator 1
PIK3R1	Phosphoinositide-3-Kinase Regulatory Subunit 1
POU6F2	POU Class 6 Homeobox 2
PRKAR1A	Protein Kinase CAMP-Dependent Type I Regulatory Subunit Alpha
PRNP	Prion Protein
PRSS1	Serine Protease 1
PRTN3	Proteinase 3
PSMB8	Proteasome Subunit Beta 8
PSTPIP1	Proline-Serine-Threonine Phosphatase Interacting Protein 1
PTPN22	Protein Tyrosine Phosphatase Non-Receptor Type 22
PTPN3	Protein Tyrosine Phosphatase Non-Receptor Type 3
PTS	6-Pyruvoyltetrahydropterin Synthase
QDPR	Quinoid Dihydropteridine Reductase
RAB27A	RAB27A, Member RAS Oncogene Family
RAG1	Recombination Activating 1
RAG2	Recombination Activating 2
RB1	RB Transcriptional Corepressor 1
REST	RE1 Silencing Transcription Factor
RMRP	RNA Component Of Mitochondrial RNA Processing Endoribonuclease
RUNX1	RUNX Family Transcription Factor 1
RYR1	Ryanodine Receptor 1
SLC11A1	Solute Carrier Family 11 Member 1
SLC12A1	Solute Carrier Family 12 Member 1
SLC19A3	Solute Carrier Family 19 Member 3
SLC29A3	Solute Carrier Family 29 Member 3
SLCO1B1	Solute Carrier Organic Anion Transporter Family Member 1B1
SLCO1B3	Solute Carrier Organic Anion Transporter Family Member 1B3
SPINK1	Serine Peptidase Inhibitor, Kazal Type 1
STAT3	Signal Transducer And Activator Of Transcription 3
STAT4	Signal Transducer And Activator Of Transcription 4
STIM1	Stromal Interaction Molecule 1
STX11	Syntaxin 11
STXBP2	Syntaxin Binding Protein 2
TCF3	Transcription Factor 3
TCF4	Transcription Factor 4
TLR4	Toll Like Receptor 4
TMEM165	Transmembrane Protein 165
TMEM173	Transmembrane Protein 173
TNFAIP3	TNF Alpha Induced Protein 3
TNFRSF1A	TNF Receptor Superfamily Member 1A
TP53	Tumor Protein P53
TREX1	Three Prime Repair Exonuclease 1
TRIP13	Thyroid Hormone Receptor Interactor 13
TRNF	Mitochondrially Encoded TRNA-Phe (UUU/C)
TRNH	Mitochondrially Encoded TRNA-His (CAU/C)
TRNL1	Mitochondrially Encoded TRNA-Leu (UUA/G) 1
TRNQ	Mitochondrially Encoded TRNA-Gln (CAA/G)
LARS	Leucyl-TRNA Synthetase
LMAN1	Lectin, Mannose Binding 1
LMNA	Lamin A/C
LYST	Lysosomal Trafficking Regulator
LZTR1	Leucine Zipper Like Transcription Regulator 1
MAP2K1	Mitogen-Activated Protein Kinase Kinase 1
MCFD2	Multiple Coagulation Factor Deficiency 2
MGAT2	Mannosyl (Alpha-1,6-)-Glycoprotein Beta-1,2-N-Acetylglucosaminyltransferase
MPDU1	Mannose-P-Dolichol Utilization Defect 1
MPI	Mannose Phosphate Isomerase
MST1	Macrophage Stimulating 1
MYD88	Myeloid Differentiation Primary Response 88
NBEAL2	Neurobeachin Like 2
NHP2	NHP2 Ribonucleoprotein
NOP10	NOP10 Ribonucleoprotein
NR1H4	Nuclear Receptor Subfamily 1 Group H Member 4
NRAS	NRAS Proto-Oncogene, GTPase
ORAI1	ORAI Calcium Release-Activated Calcium Modulator 1
PARN	Poly(A)-Specific Ribonuclease
PEX1	Peroxisomal Biogenesis Factor 1
PEX10	Peroxisomal Biogenesis Factor 10
PEX11B	Peroxisomal Biogenesis Factor 11B
PEX12	Peroxisomal Biogenesis Factor 12
PEX13	Peroxisomal Biogenesis Factor 13
PEX14	Peroxisomal Biogenesis Factor 14
PEX16	Peroxisomal Biogenesis Factor 16
PEX19	Peroxisomal Biogenesis Factor 19
PEX2	Peroxisomal Biogenesis Factor 2
PEX26	Peroxisomal Biogenesis Factor 26
PEX3	Peroxisomal Biogenesis Factor 3
PEX5	Peroxisomal Biogenesis Factor 5
PEX6	Peroxisomal Biogenesis Factor 6
PIGA	Phosphatidylinositol Glycan Anchor Biosynthesis Class A
PLAT	Plasminogen Activator, Tissue Type
PLG	Plasminogen
PMM2	Phosphomannomutase 2
PRF1	Perforin 1
PROC	Protein C, Inactivator Of Coagulation Factors Va And VIIIa
PROS1	Protein S
PTH1R	Parathyroid Hormone 1 Receptor
PTPN11	Protein Tyrosine Phosphatase, Non-Receptor Type 11
RAF1	Raf-1 Proto-Oncogene, Serine/Threonine Kinase
RASA2	RAS P21 Protein Activator 2
RBM8A	RNA Binding Motif Protein 8A
RFT1	RFT1 Homolog
RIT1	Ras Like Without CAAX 1
RRAS	RAS Related
RTEL1	Regulator Of Telomere Elongation Helicase 1
RYR1	Ryanodine Receptor 1
SERAC1	Serine Active Site Containing 1
SERPINC1	Serpin Family C Member 1
SERPINF2	Serpin Family H Member 1

TRNS1	Mitochondrially Encoded TRNA-Ser (UCN) 1
TRNS2	Mitochondrially Encoded TRNA-Ser (AGU/C) 2
TRNW	Mitochondrially Encoded TRNA-Trp (UGA/G)
TSC1	TSC Complex Subunit 1
TSC2	TSC Complex Subunit 2
UBAC2	UBA Domain Containing 2
UNC13D	Unc-13 Homolog D
WAS	WASP Actin Nucleation Promoting Factor
WIPF1	WAS/WASL Interacting Protein Family Member 1
WT1	WT1 Transcription Factor
XIAP	X-Linked Inhibitor Of Apoptosis
XPA	XPA, DNA Damage Recognition And Repair Factor
XPC	XPC Complex Subunit, DNA Damage Recognition And Repair Factor

Episodic fever (HP:0001954), 22 genes

CRLF1	Cytokine Receptor Like Factor 1
CYP11B2	Cytochrome P450 Family 11 Subfamily B Member 2
ELP1	Elongator Complex Protein 1
GALC	Galactosylceramidase
GCH1	GTP Cyclohydrolase 1
LIFR	LIF Receptor Alpha
LPIN2	Lipin 2
MEFV	MEFV, Pyrin Innate Immunity Regulator
MVK	Mevalonate Kinase
NGF	Nerve Growth Factor
NLRP12	NLR Family Pyrin Domain Containing 12
NLRP3	NLR Family Pyrin Domain Containing 3
NTRK1	Neurotrophic Receptor Tyrosine Kinase 1
ORAI1	ORAI Calcium Release-Activated Calcium Modulator 1
PSMB8	Proteasome Subunit Beta 8
PTS	6-Pyruvoyltetrahydropterin Synthase
QDPR	Quinoid Dihydropteridine Reductase
SLC29A3	Solute Carrier Family 29 Member 3
STIM1	Stromal Interaction Molecule 1
STXBP2	Syntaxin Binding Protein 2
TNFAIP3	TNF Alpha Induced Protein 3
TNFRSF1A	TNF Receptor Superfamily Member 1A

Hearing impairment (HP:0000365), 969 genes

A2ML1	Alpha-2-Macroglobulin Like 1
AAAS	Aladin WD Repeat Nucleoporin
AARS	Alanyl-TRNA Synthetase
ABCA12	ATP Binding Cassette Subfamily A Member 12
ABCA4	ATP Binding Cassette Subfamily A Member 4
ABCB6	ATP Binding Cassette Subfamily B Member 6
ABCC8	ATP Binding Cassette Subfamily C Member 8
ABCC9	ATP Binding Cassette Subfamily C Member 9
ABCD1	ATP Binding Cassette Subfamily D Member 1
ABHD12	Abhydrolase Domain Containing 12
ABHD5	Abhydrolase Domain Containing 5
ACO2	Aconitase 2
ACOX1	Acyl-CoA Oxidase 1
ACSL4	Acyl-CoA Synthetase Long Chain Family Member 4
SOS1	SOS Ras/Rac Guanine Nucleotide Exchange Factor 1
SOS2	SOS Ras/Rac Guanine Nucleotide Exchange Factor 2
SRD5A3	Steroid 5 Alpha-Reductase 3
SSR4	Signal Sequence Receptor Subunit 4
STIM1	Stromal Interaction Molecule 1
STX11	Syntaxin 11
TCF4	Transcription Factor 4
TEK	TEK Receptor Tyrosine Kinase
TERC	Telomerase RNA Component
TERT	Telomerase Reverse Transcriptase
TFAM	Transcription Factor A, Mitochondrial
THBD	Thrombomodulin
TINF2	TERF1 Interacting Nuclear Factor 2
TRMU	TRNA 5-Methylaminomethyl-2-Thiouridylate Methyltransferase
UNC13D	Unc-13 Homolog D
USB1	U6 SnRNA Biogenesis Phosphodiesterase 1
VKORC1	Vitamin K Epoxide Reductase Complex Subunit 1
VWF	Von Willebrand Factor
WRAP53	WD Repeat Containing Antisense To TP53
XIAP	X-Linked Inhibitor Of Apoptosis
ZMPSTE24	Zinc Metallopeptidase STE24

Absent speech (HP:0001344), 77 genes

ADSL	Adenylosuccinate Lyase
ALG11	ALG11 Alpha-1,2-Mannosyltransferase
AP3B2	Adaptor Related Protein Complex 3 Subunit Beta 2
BCL11B	B-Cell CLL/Lymphoma 11B
BMP4	Bone Morphogenetic Protein 4
C2CD3	C2 Calcium Dependent Domain Containing 3
CASK	Calcium/Calmodulin Dependent Serine Protein Kinase
CHMP1A	Charged Multivesicular Body Protein 1A
CLIC2	Chloride Intracellular Channel 2
CLP1	Cleavage And Polyadenylation Factor I Subunit 3
COG4	COG complex subunit 4
CUL4B	Cullin 4B
DAG1	Dystroglycan 1
DEAF1	DEAF1, Transcription Factor
DNM1	Dynamin 1 Like
EARS2	Glutamyl-TRNA Synthetase 2, Mitochondrial
ERLIN2	ER Lipid Raft Associated 2
EXOSC3	Exosome Component 3
FGF12	Fibroblast Growth Factor 12
FOXG1	Forkhead Box G1
FRRS1L	Ferric Chelate Reductase 1 Like
GABRD	Gamma-Aminobutyric Acid Type A Receptor Delta Subunit
GMPPB	GDP-Mannose Pyrophosphorylase B
GNAO1	G Protein Subunit Alpha O1
GNS	Glucosamine (N-Acetyl)-6-Sulfatase
GPT2	Glutamic--Pyruvic Transaminase 2
GRIN2B	Glutamate Ionotropic Receptor NMDA Type Subunit 2B
IFIH1	Interferon Induced With Helicase C Domain 1
ISCA2	Iron-Sulfur Cluster Assembly 2
KCNAB2	Potassium Voltage-Gated Channel Subfamily A Regulatory Beta Subunit 2

ACTB	Actin Beta
ACTC1	Actin Alpha Cardiac Muscle 1
ACTG1	Actin Gamma 1
ACTN2	Actinin Alpha 2
ACVR1	Activin A Receptor Type 1
ACY1	Aminoacylase 1
ADCY1	Adenylate Cyclase 1
ADGRV1	Adhesion G Protein-Coupled Receptor V1
ADK	Adenosine Kinase
AFF4	AF4/FMR2 Family Member 4
AGBL5	ATP/GTP Binding Protein Like 5
AHSG	Alpha 2-HS Glycoprotein
AIFM1	Apoptosis Inducing Factor Mitochondria Associated 1
AIPL1	Aryl Hydrocarbon Receptor Interacting Protein Like 1
AK2	Adenylate Kinase 2
AKT1	AKT Serine/Threonine Kinase 1
ALG11	ALG11 Alpha-1,2-Mannosyltransferase
ALMS1	ALMS1 Centrosome And Basal Body Associated Protein
ALOX12B	Arachidonate 12-Lipoxygenase, 12R Type
ALOXE3	Arachidonate Lipoxygenase 3
ALX3	ALX Homeobox 3
AMER1	APC Membrane Recruitment Protein 1
AMMECR1	Alport Syndrome, Mental Retardation, Midface Hypoplasia And Elliptocytosis Chromosomal Region Gene 1
ANKH	ANKH Inorganic Pyrophosphate Transport Regulator
ANKRD1	Ankyrin Repeat Domain 1
ANKRD11	Ankyrin Repeat Domain 11
ANOS1	Anosmin 1
ANTXR1	ANTXR Cell Adhesion Molecule 1
AP1S1	Adaptor Related Protein Complex 1 Subunit Sigma 1
AP1S2	Adaptor Related Protein Complex 1 Subunit Sigma 2
APC	APC Regulator Of WNT Signaling Pathway
APC2	APC Regulator Of WNT Signaling Pathway 2
APOPT1	Cytochrome C Oxidase Assembly Factor 8
ARHGDIA	Rho GDP Dissociation Inhibitor Alpha
ARHGEF18	Rho/Rac Guanine Nucleotide Exchange Factor 18
ARHGEF6	Rac/Cdc42 Guanine Nucleotide Exchange Factor 6
ARID1A	AT-Rich Interaction Domain 1A
ARID1B	AT-Rich Interaction Domain 1B
ARID2	AT-Rich Interaction Domain 2
ARL2BP	ADP Ribosylation Factor Like GTPase 2 Binding Protein
ARL6	ADP Ribosylation Factor Like GTPase 6
ARNT2	Aryl Hydrocarbon Receptor Nuclear Translocator 2
ARSB	Arylsulfatase B
ARSE	Arylsulfatase E
ARVCF	ARVCF Delta Catenin Family Member
ASCL1	Achaete-Scute Family BHLH Transcription Factor 1
ASPA	Aspartoacylase
ASPM	Abnormal Spindle Microtubule Assembly
ASXL1	Additional Sex Combs Like 1, Transcriptional Regulator
ATP1A2	ATPase Na+/K+ Transporting Subunit Alpha 2
KIF5C	Kinesin Family Member 5C
KLHL15	Kelch Like Family Member 15
MCOLN1	Mucolipin 1
MDH2	Malate Dehydrogenase 2
MECP2	Methyl-CpG Binding Protein 2
MFF	Mitochondrial Fission Factor
MFSD2A	Major Facilitator Superfamily Domain Containing 2A
MID2	Midline 2
MPDU1	Mannose-P-Dolichol Utilization Defect 1
NACC1	Nucleus Accumbens Associated 1
NEXMIF	Neurite Extension And Migration Factor
PIGA	Phosphatidylinositol Glycan Anchor Biosynthesis Class A
PIGN	Phosphatidylinositol Glycan Anchor Biosynthesis Class N
PIGV	Phosphatidylinositol Glycan Anchor Biosynthesis Class V
PNPT1	Polyribonucleotide Nucleotidyltransferase 1
POMT1	Protein O-Mannosyltransferase 1
PRDM16	PR/SET Domain 16
PRPS1	Phosphoribosyl Pyrophosphate Synthetase 1
PURA	Purine Rich Element Binding Protein A
PYCR2	Pyrroline-5-Carboxylate Reductase 2
RARS2	Arginyl-TRNA Synthetase 2, Mitochondrial
RERE	Arginine-Glutamic Acid Dipeptide Repeats
SETBP1	SET Binding Protein 1
SIK1	Salt Inducible Kinase 1
SKI	SKI Proto-Oncogene
SLC16A2	Solute Carrier Family 16 Member 2
SLC25A12	Solute Carrier Family 25 Member 12
SLC2A1	Solute Carrier Family 2 Member 1
SLC33A1	Solute Carrier Family 33 Member 1
SLC9A6	Solute Carrier Family 9 Member 6
SMARCA2	SWI/SNF Related, Matrix Associated, Actin Dependent Regulator Of Chromatin, Subfamily A, Member 2
SMARCE1	SWI/SNF Related, Matrix Associated, Actin Dependent Regulator Of Chromatin, Subfamily E, Member 1
SPATA5	Spermatogenesis Associated 5
ST3GAL5	ST3 Beta-Galactoside Alpha-2,3-Sialyltransferase 5
STXBP1	Syntaxin Binding Protein 1
TCF4	Transcription Factor 4
TCTN2	Tectonic Family Member 2
TELO2	Telomere Maintenance 2
TMEM231	Transmembrane Protein 231
UBA5	Ubiquitin Like Modifier Activating Enzyme 5
UBE3A	Ubiquitin Protein Ligase E3A
UNC80	Unc-80 Homolog, NALCN Channel Complex Subunit
UQCRQ	Ubiquinol-Cytochrome C Reductase Complex III Subunit VII
VPS11	VPS11, CORVET/HOPS Core Subunit
WDR45	WD Repeat Domain 45
ZDHHC15	Zinc Finger DHHC-Type Containing 15
ZEB2	Zinc Finger E-Box Binding Homeobox 2

Multiple joint contractures (HP:0002828), 11 genes

ALG14	ALG14 UDP-N-Acetylglucosaminyltransferase Subunit

ATP1A3	ATPase Na+/K+ Transporting Subunit Alpha 3
ATP6AP1	ATPase H+ Transporting Accessory Protein 1
ATP6V0A4	ATPase H+ Transporting V0 Subunit A4
ATP6V1B1	ATPase H+ Transporting V1 Subunit B1
ATP6V1B2	ATPase H+ Transporting V1 Subunit B2
ATP8	Mitochondrially Encoded ATP Synthase Membrane Subunit 8
ATP8B1	ATPase Phospholipid Transporting 8B1
ATRX	ATRX, Chromatin Remodeler
B3GLCT	Beta 3-Glucosyltransferase
BAG3	BCL2 Associated Athanogene 3
BAZ1B	Bromodomain Adjacent To Zinc Finger Domain 1B
BBIP1	BBSome Interacting Protein 1
BBS1	Bardet-Biedl Syndrome 1
BBS10	Bardet-Biedl Syndrome 10
BBS12	Bardet-Biedl Syndrome 12
BBS2	Bardet-Biedl Syndrome 2
BBS4	Bardet-Biedl Syndrome 4
BBS5	Bardet-Biedl Syndrome 5
BBS7	Bardet-Biedl Syndrome 7
BBS9	Bardet-Biedl Syndrome 9
BCAP31	B Cell Receptor Associated Protein 31
BCOR	BCL6 Corepressor
BCR	BCR Activator Of RhoGEF And GTPase
BCS1L	BCS1 Homolog, Ubiquinol-Cytochrome C Reductase Complex Chaperone
BEAN1	Brain Expressed Associated With NEDD4 1
BEST1	Bestrophin 1
BMP15	Bone Morphogenetic Protein 15
BMP4	Bone Morphogenetic Protein 4
BRAF	B-Raf Proto-Oncogene, Serine/Threonine Kinase
BRCA2	BRCA2, DNA Repair Associated
BRIP1	BRCA1 Interacting Protein C-Terminal Helicase 1
BSND	Barttin CLCNK Type Accessory Beta Subunit
BTD	Biotinidase
BTK	Bruton Tyrosine Kinase
BTRC	Beta-Transducin Repeat Containing E3 Ubiquitin Protein Ligase
BUB1	BUB1 Mitotic Checkpoint Serine/Threonine Kinase
BUB1B	BUB1 Mitotic Checkpoint Serine/Threonine Kinase B
BUB3	BUB3 Mitotic Checkpoint Protein
C2ORF71	Photoreceptor Cilium Actin Regulator
C5ORF42	Ciliogenesis And Planar Polarity Effector 1
C8ORF37	Chromosome 8 Open Reading Frame 37
CA4	Carbonic Anhydrase 4
CABP2	Calcium Binding Protein 2
CACNA1A	Calcium Voltage-Gated Channel Subunit Alpha1 A
CACNA1D	Calcium Voltage-Gated Channel Subunit Alpha1 D
CASK	Calcium/Calmodulin Dependent Serine Protein Kinase
CATSPER2	Cation Channel Sperm Associated 2
CCBE1	Collagen And Calcium Binding EGF Domains 1
CCDC141	Coiled-Coil Domain Containing 141
CCDC50	Coiled-Coil Domain Containing 50
CD151	CD151 Molecule (Raph Blood Group)
CD164	CD164 Molecule
CDC42	Cell Division Cycle 42
CDC45	Cell Division Cycle 45
DOK7	Docking Protein 7
FLNA	Filamin A
LMNA	Lamin A/C
MED12	Mediator Complex Subunit 12
MUSK	Muscle Associated Receptor Tyrosine Kinase
MYOD1	Myogenic Differentiation 1
NDE1	NudE Neurodevelopment Protein 1
RAPSN	Receptor Associated Protein Of The Synapse
UBA1	Ubiquitin Like Modifier Activating Enzyme 1
ZMPSTE24	Zinc Metallopeptidase STE24

Progressive microcephaly (HP:0000253), 53 genes

ACO2	Aconitase 2
ALG12	ALG12 Alpha-1,6-Mannosyltransferase
AMPD2	Adenosine Monophosphate Deaminase 2
ARFGEF2	ADP Ribosylation Factor Guanine Nucleotide Exchange Factor 2
ASNS	Asparagine Synthetase (Glutamine-Hydrolyzing)
ATP6V0A2	ATPase H+ Transporting V0 Subunit A2
ATP6V1A	ATPase H+ Transporting V1 Subunit A
ATP6V1E1	ATPase H+ Transporting V1 Subunit E1
BRAT1	BRCA1 Associated ATM Activator 1
CASK	Calcium/Calmodulin Dependent Serine Protein Kinase
CCDC88A	Coiled-Coil Domain Containing 88A
CDC42	Cell Division Cycle 42
CDC45	Cell Division Cycle 45
CDKL5	Cyclin Dependent Kinase Like 5
CLP1	Cleavage And Polyadenylation Factor I Subunit 3
COG1	COG complex subunit 1
EFTUD2	Elongation Factor Tu GTP Binding Domain Containing 2
EXOSC3	Exosome Component 3
EXTL3	Exostosin Like Glycosyltransferase 3
FOXG1	Forkhead Box G1
MECP2	Methyl-CpG Binding Protein 2
MED17	Mediator Complex Subunit 17
MFSD2A	Major Facilitator Superfamily Domain Containing 2A
MPC1	Mitochondrial Pyruvate Carrier 1
PCLO	Piccolo Presynaptic Cytomatrix Protein
PNKP	Polynucleotide Kinase 3'-Phosphatase
PNPO	Pyridoxamine 5'-Phosphate Oxidase
POMK	Protein-O-Mannose Kinase
PPT1	Palmitoyl-Protein Thioesterase 1
PTRH2	Peptidyl-TRNA Hydrolase 2
PYCR2	Pyrroline-5-Carboxylate Reductase 2
QARS	Glutaminyl-TRNA Synthetase
RARS2	Arginyl-TRNA Synthetase 2, Mitochondrial
RNASEH2A	Ribonuclease H2 Subunit A
RNASEH2C	Ribonuclease H2 Subunit C
RNF113A	Ring Finger Protein 113A
SCN8A	Sodium Voltage-Gated Channel Alpha Subunit 8
SEPSECS	Sep (O-Phosphoserine) TRNA:Sec (Selenocysteine) TRNA Synthase
SLC1A4	Solute Carrier Family 1 Member 4
SLC25A19	Solute Carrier Family 25 Member 19
SLC25A22	Solute Carrier Family 25 Member 22
SLC2A1	Solute Carrier Family 2 Member 1

CDC6	Cell Division Cycle 6
CDCA7	Cell Division Cycle Associated 7
CDH23	Cadherin Related 23
CDHR1	Cadherin Related Family Member 1
CDK5RAP2	CDK5 Regulatory Subunit Associated Protein 2
CDKL5	Cyclin Dependent Kinase Like 5
CDT1	Chromatin Licensing And DNA Replication Factor 1
CEACAM16	Carcinoembryonic Antigen Related Cell Adhesion Molecule 16
CEP290	Centrosomal Protein 290
CEP57	Centrosomal Protein 57
CEP78	Centrosomal Protein 78
CERKL	Ceramide Kinase Like
CERS3	Ceramide Synthase 3
CHCHD10	Coiled-Coil-Helix-Coiled-Coil-Helix Domain Containing 10
CHD4	Chromodomain Helicase DNA Binding Protein 4
CHD7	Chromodomain Helicase DNA Binding Protein 7
CHN1	Chimerin 1
CHRNG	Cholinergic Receptor Nicotinic Gamma Subunit
CHST14	Carbohydrate Sulfotransferase 14
CHST3	Carbohydrate Sulfotransferase 3
CHSY1	Chondroitin Sulfate Synthase 1
CIB2	Calcium And Integrin Binding Family Member 2
CISD2	CDGSH Iron Sulfur Domain 2
CLCN7	Chloride Voltage-Gated Channel 7
CLCNKA	Chloride Voltage-Gated Channel Ka
CLCNKB	Chloride Voltage-Gated Channel Kb
CLDN14	Claudin 14
CLIC5	Chloride Intracellular Channel 5
CLIP2	CAP-Gly Domain Containing Linker Protein 2
CLPP	Caseinolytic Mitochondrial Matrix Peptidase Proteolytic Subunit
CLRN1	Clarin 1
CNGA1	Cyclic Nucleotide Gated Channel Alpha 1
CNGB1	Cyclic Nucleotide Gated Channel Beta 1
COA5	Cytochrome C Oxidase Assembly Factor 5
COA7	Cytochrome C Oxidase Assembly Factor 7
COCH	Cochlin
COL10A1	Collagen Type X Alpha 1 Chain
COL11A1	Collagen Type XI Alpha 1 Chain
COL11A2	Collagen Type XI Alpha 2 Chain
COL1A1	Collagen Type I Alpha 1 Chain
COL1A2	Collagen Type I Alpha 2 Chain
COL25A1	Collagen Type XXV Alpha 1 Chain
COL2A1	Collagen Type II Alpha 1 Chain
COL4A3	Collagen Type IV Alpha 3 Chain
COL4A3BP	Collagen Type IV Alpha 3 Binding Protein
COL4A4	Collagen Type IV Alpha 4 Chain
COL4A5	Collagen Type IV Alpha 5 Chain
COL4A6	Collagen Type IV Alpha 6 Chain
COL7A1	Collagen Type VII Alpha 1 Chain
COL9A1	Collagen Type IX Alpha 1 Chain
COL9A2	Collagen Type IX Alpha 2 Chain
COL9A3	Collagen Type IX Alpha 3 Chain
COLEC10	Collectin Subfamily Member 10
COLEC11	Collectin Subfamily Member 11
COMT	Catechol-O-Methyltransferase
COQ2	Coenzyme Q2, Polyprenyltransferase
SNAP29	Synaptosome Associated Protein 29
SPTAN1	Spectrin Alpha, Non-Erythrocytic 1
STAMBP	STAM Binding Protein
TOE1	Target Of EGR1, Member 1 (Nuclear)
TREX1	Three Prime Repair Exonuclease 1
TSEN15	TRNA Splicing Endonuclease Subunit 15
TSEN2	TRNA Splicing Endonuclease Subunit 2
TSEN54	TRNA Splicing Endonuclease Subunit 54
VPS53	VPS53, GARP Complex Subunit
WWOX	WW Domain Containing Oxidoreductase
ZNHIT3	Zinc Finger HIT-Type Containing 3

Hypohidrosis (HP:0000966), 58 genes

ABCA12	ATP Binding Cassette Subfamily A Member 12
ALOX12B	Arachidonate 12-Lipoxygenase, 12R Type
ALOXE3	Arachidonate Lipoxygenase 3
ALX4	ALX Homeobox 4
ARNT2	Aryl Hydrocarbon Receptor Nuclear Translocator 2
ARX	Aristaless Related Homeobox
CASK	Calcium/Calmodulin Dependent Serine Protein Kinase
CERS3	Ceramide Synthase 3
CLCF1	Cardiotrophin Like Cytokine Factor 1
COG6	COG complex subunit 6
COL11A1	Collagen Type XI Alpha 1 Chain
CRLF1	Cytokine Receptor Like Factor 1
CTNS	Cystinosin, Lysosomal Cystine Transporter
EDA	Ectodysplasin A
EDA2R	Ectodysplasin A2 Receptor
EDAR	Ectodysplasin A Receptor
EDARADD	EDAR Associated Death Domain
ELOVL4	ELOVL Fatty Acid Elongase 4
ELP1	Elongator Complex Protein 1
FAM111B	Family With Sequence Similarity 111 Member B
FGFR1	Fibroblast Growth Factor Receptor 1
GHR	Growth Hormone Receptor
GJB2	Gap Junction Protein Beta 2
GJB6	Gap Junction Protein Beta 6
GLA	Galactosidase Alpha
GMPPA	GDP-Mannose Pyrophosphorylase A
HESX1	HESX Homeobox 1
HEXB	Hexosaminidase Subunit Beta
KCTD1	Potassium Channel Tetramerization Domain Containing 1
KDF1	Keratinocyte Differentiation Factor 1
KRT14	Keratin 14
LIFR	LIF Receptor Alpha
LMNA	Lamin A/C
LMNB1	Lamin B1
MBTPS2	Membrane Bound Transcription Factor Peptidase, Site 2
NECTIN1	Nectin Cell Adhesion Molecule 1
NFKBIA	NFKB Inhibitor Alpha
NIPAL4	NIPA Like Domain Containing 4
OTX2	Orthodenticle Homeobox 2
PNPLA1	Patatin Like Phospholipase Domain Containing 1
POLA1	DNA Polymerase Alpha 1, Catalytic Subunit
PRDM12	PR/SET Domain 12
PROKR2	Prokineticin Receptor 2

COQ6	Coenzyme Q6, Monooxygenase
COQ7	Coenzyme Q7, Hydroxylase
COX1	Mitochondrially Encoded Cytochrome C Oxidase I
COX10	Cytochrome C Oxidase Assembly Factor Heme A:Farnesyltransferase COX10
COX14	Cytochrome C Oxidase Assembly Factor COX14
COX15	Cytochrome C Oxidase Assembly Homolog COX15
COX2	Mitochondrially Encoded Cytochrome C Oxidase II
COX20	Cytochrome C Oxidase Assembly Factor COX20
COX3	Mitochondrially Encoded Cytochrome C Oxidase III
COX6B1	Cytochrome C Oxidase Subunit 6B1
COX7B	Cytochrome C Oxidase Subunit 7B
CPLX1	Complexin 1
CRB1	Crumbs Cell Polarity Complex Component 1
CREBBP	CREB Binding Protein
CRKL	CRK Like Proto-Oncogene, Adaptor Protein
CRX	Cone-Rod Homeobox
CRYAB	Crystallin Alpha B
CRYM	Crystallin Mu
CSPP1	Centrosome And Spindle Pole Associated Protein 1
CSRP3	Cysteine And Glycine Rich Protein 3
CTBP1	C-Terminal Binding Protein 1
CTC1	CST Telomere Replication Complex Component 1
CTLA4	Cytotoxic T-Lymphocyte Associated Protein 4
CTNNB1	Catenin Beta 1
CTSA	Cathepsin A
CYP7B1	Cytochrome P450 Family 7 Subfamily B Member 1
CYTB	Mitochondrially Encoded Cytochrome B
DAB1	DAB Adaptor Protein 1
DACT1	Dishevelled Binding Antagonist Of Beta Catenin 1
DCAF17	DDB1 And CUL4 Associated Factor 17
DCDC2	Doublecortin Domain Containing 2
DCHS1	Dachsous Cadherin-Related 1
DDB2	Damage Specific DNA Binding Protein 2
DDX11	DEAD/H-Box Helicase 11
DDX3X	DEAD-Box Helicase 3 X-Linked
DEAF1	DEAF1, Transcription Factor
DES	Desmin
DGUOK	Deoxyguanosine Kinase
DHCR7	7-Dehydrocholesterol Reductase
DHDDS	Dehydrodolichyl Diphosphate Synthase Subunit
DHODH	Dihydroorotate Dehydrogenase (Quinone)
DIABLO	Diablo IAP-Binding Mitochondrial Protein
DIAPH1	Diaphanous Related Formin 1
DIAPH3	Diaphanous Related Formin 3
DKC1	Dyskerin Pseudouridine Synthase 1
DKK1	Dickkopf WNT Signaling Pathway Inhibitor 1
DLX5	Distal-Less Homeobox 5
DLX6	Distal-Less Homeobox 6
DMD	Dystrophin
DMP1	Dentin Matrix Acidic Phosphoprotein 1
DMXL2	Dmx Like 2
DNAAF3	Dynein Axonemal Assembly Factor 3
ROGDI	Rogdi Homolog
SCN9A	Sodium Voltage-Gated Channel Alpha Subunit 9
SHANK3	SH3 And Multiple Ankyrin Repeat Domains 3
SLC13A5	Solute Carrier Family 13 Member 5
SMARCAD1	SWI/SNF-Related, Matrix-Associated Actin-Dependent Regulator Of Chromatin, Subfamily A, Containing DEAD/H Box 1
SOX10	SRY-Box 10
SOX2	SRY-Box 2
SOX3	SRY-Box 3
STIM1	Stromal Interaction Molecule 1
STS	Steroid Sulfatase
TGM1	Transglutaminase 1
TP63	Tumor Protein P63
TRIP4	Thyroid Hormone Receptor Interactor 4
WNT10A	Wnt Family Member 10A
ZMPSTE24	Zinc Metallopeptidase STE24

Death in infancy (HP:0001522), 132 genes

AARS2	Alanyl-TRNA Synthetase 2, Mitochondrial
ACOX1	Acyl-CoA Oxidase 1
ACTG2	Actin Gamma 2, Smooth Muscle
AIMP1	Aminoacyl TRNA Synthetase Complex Interacting Multifunctional Protein 1
ALG1	ALG1 Chitobiosyldiphosphodolichol Beta-Mannosyltransferase
ALPL	Alkaline Phosphatase, Biomineralization Associated
AMT	Aminomethyltransferase
ANTXR2	ANTXR Cell Adhesion Molecule 2
ARX	Aristaless Related Homeobox
ASCL1	Achaete-Scute Family BHLH Transcription Factor 1
ASXL1	Additional Sex Combs Like 1, Transcriptional Regulator
ATRX	ATRX, Chromatin Remodeler
BMPR1B	Bone Morphogenetic Protein Receptor Type 1B
BOLA3	BolA Family Member 3
BRAT1	BRCA1 Associated ATM Activator 1
CCDC22	Coiled-Coil Domain Containing 22
CD96	CD96 Molecule
CDON	Cell Adhesion Associated, Oncogene Regulated
CLCF1	Cardiotrophin Like Cytokine Factor 1
CNTN1	Contactin 1
COG6	COG complex subunit 6
CPT2	Carnitine Palmitoyltransferase 2
CRLF1	Cytokine Receptor Like Factor 1
CRTAP	Cartilage Associated Protein
DCX	Doublecortin
DLL3	Delta Like Canonical Notch Ligand 3
DNM1L	Dynamin 1 Like
DNM2	Dynamin 2
DOLK	Dolichol Kinase
DPM2	Dolichyl-Phosphate Mannosyltransferase Subunit 2, Regulatory
EIF2AK3	Eukaryotic Translation Initiation Factor 2 Alpha Kinase 3
EMG1	N1-Specific Pseudouridine Methyltransferase
EPCAM	Epithelial Cell Adhesion Molecule
EPG5	Ectopic P-Granules Autophagy Protein 5 Homolog
ERCC1	ERCC Excision Repair 1, Endonuclease Non-Catalytic Subunit

DNAI1	Dynein Axonemal Intermediate Chain 1
DNAJC3	DnaJ Heat Shock Protein Family (Hsp40) Member C3
DNASE1L3	Deoxyribonuclease 1 Like 3
DNMT1	DNA Methyltransferase 1
DOLK	Dolichol Kinase
DSG2	Desmoglein 2
DSPP	Dentin Sialophosphoprotein
DUSP6	Dual Specificity Phosphatase 6
DUX4	Double Homeobox 4
DVL1	Dishevelled Segment Polarity Protein 1
DVL3	Dishevelled Segment Polarity Protein 3
EBP	EBP Cholestenol Delta-Isomerase
ECE1	Endothelin Converting Enzyme 1
ECHS1	Enoyl-CoA Hydratase, Short Chain 1
EDC3	Enhancer Of MRNA Decapping 3
EDN1	Endothelin 1
EDN3	Endothelin 3
EDNRA	Endothelin Receptor Type A
EDNRB	Endothelin Receptor Type B
EFNB1	Ephrin B1
EFTUD2	Elongation Factor Tu GTP Binding Domain Containing 2
EHMT1	Euchromatic Histone Lysine Methyltransferase 1
ELAC2	ElaC Ribonuclease Z 2
ELMOD3	ELMO Domain Containing 3
ELN	Elastin
ENPP1	Ectonucleotide Pyrophosphatase/Phosphodiesterase 1
EP300	E1A Binding Protein P300
EPG5	Ectopic P-Granules Autophagy Protein 5 Homolog
EPS8	Epidermal Growth Factor Receptor Pathway Substrate 8
ERCC1	ERCC Excision Repair 1, Endonuclease Non-Catalytic Subunit
ERCC2	ERCC Excision Repair 2, TFIIH Core Complex Helicase Subunit
ERCC3	ERCC Excision Repair 3, TFIIH Core Complex Helicase Subunit
ERCC4	ERCC Excision Repair 4, Endonuclease Catalytic Subunit
ERCC5	ERCC Excision Repair 5, Endonuclease
ERCC6	ERCC Excision Repair 6, Chromatin Remodeling Factor
ERCC8	ERCC Excision Repair 8, CSA Ubiquitin Ligase Complex Subunit
ERF	ETS2 Repressor Factor
ESPN	Espin
ESRRB	Estrogen Related Receptor Beta
EXOSC8	Exosome Component 8
EXT1	Exostosin Glycosyltransferase 1
EYA1	EYA Transcriptional Coactivator And Phosphatase 1
EYA4	EYA Transcriptional Coactivator And Phosphatase 4
EYS	Eyes Shut Homolog
FAM161A	FAM161 Centrosomal Protein A
FAM20C	FAM20C Golgi Associated Secretory Pathway Kinase
FANCA	FA Complementation Group A
FANCB	FA Complementation Group B
FANCC	FA Complementation Group C
FANCD2	FA Complementation Group D2
FANCE	FA Complementation Group E
FANCF	FA Complementation Group F
FANCG	FA Complementation Group G
FANCI	FA Complementation Group I
FANCL	FA Complementation Group L
ERCC2	ERCC Excision Repair 2, TFIIH Core Complex Helicase Subunit
ERCC5	ERCC Excision Repair 5, Endonuclease
ERCC6	ERCC Excision Repair 6, Chromatin Remodeling Factor
FAN1	FANCD2 And FANCI Associated Nuclease 1
FARS2	Phenylalanyl-TRNA Synthetase 2, Mitochondrial
FGFR3	Fibroblast Growth Factor Receptor 3
FLI1	Fli-1 Proto-Oncogene, ETS Transcription Factor
FRAS1	Fraser Extracellular Matrix Complex Subunit 1
FREM2	FRAS1 Related Extracellular Matrix Protein 2
GBA	Glucosylceramidase Beta
GCSH	Glycine Cleavage System Protein H
GDF5	Growth Differentiation Factor 5
GLB1	Galactosidase Beta 1
GLDC	Glycine Decarboxylase
GLUL	Glutamine synthetase family
GPC3	Glypican 3
GPC4	Glypican 4
GPR161	G Protein-Coupled Receptor 161
GRIP1	Glutamate Receptor Interacting Protein 1
HDAC6	Histone Deacetylase 6
HESX1	HESX Homeobox 1
HSPG2	Heparan Sulfate Proteoglycan 2
HTRA2	HtrA Serine Peptidase 2
IDUA	Iduronidase, Alpha-L-
ITGA6	Integrin Subunit Alpha 6
ITGB4	Integrin Subunit Beta 4
ITPA	Inosine Triphosphatase
KRAS	KRAS Proto-Oncogene, GTPase
LAMA3	Laminin Subunit Alpha 3
LAMB3	Laminin Subunit Beta 3
LAMC2	Laminin Subunit Gamma 2
LHX4	LIM Homeobox 4
LIPA	Lipase A, Lysosomal Acid Type
LIPT1	Lipoyltransferase 1
LMNA	Lamin A/C
LMOD1	Leiomodin 1
LTC4S	Leukotriene C4 Synthase
MESP2	Mesoderm Posterior BHLH Transcription Factor 2
MLH1	MutL Homolog 1
MLH3	MutL Homolog 3
MRPS22	Mitochondrial Ribosomal Protein S22
MSH2	MutS Homolog 2
MSH6	MutS Homolog 6
MYH11	Myosin Heavy Chain 11
MYLK	Myosin Light Chain Kinase
MYO5B	Myosin VB
NADK2	NAD Kinase 2, Mitochondrial
NAXE	NAD(P)HX Epimerase
NRAS	NRAS Proto-Oncogene, GTPase
OCRL	OCRL, Inositol Polyphosphate-5-Phosphatase
PEX1	Peroxisomal Biogenesis Factor 1
PEX10	Peroxisomal Biogenesis Factor 10
PEX11B	Peroxisomal Biogenesis Factor 11B
PEX12	Peroxisomal Biogenesis Factor 12
PEX13	Peroxisomal Biogenesis Factor 13

FANCM	FA Complementation Group M
FARS2	Phenylalanyl-TRNA Synthetase 2, Mitochondrial
FAS	Fas Cell Surface Death Receptor
FASTKD2	FAST Kinase Domains 2
FAT4	FAT Atypical Cadherin 4
FBN1	Fibrillin 1
FERMT1	Fermitin Family Member 1
FEZF1	FEZ Family Zinc Finger 1
FGF10	Fibroblast Growth Factor 10
FGF17	Fibroblast Growth Factor 17
FGF3	Fibroblast Growth Factor 3
FGF8	Fibroblast Growth Factor 8
FGF9	Fibroblast Growth Factor 9
FGFR1	Fibroblast Growth Factor Receptor 1
FGFR2	Fibroblast Growth Factor Receptor 2
FGFR3	Fibroblast Growth Factor Receptor 3
FGFRL1	Fibroblast Growth Factor Receptor Like 1
FHL2	Four And A Half LIM Domains 2
FIBP	FGF1 Intracellular Binding Protein
FIG4	FIG4 Phosphoinositide 5-Phosphatase
FKBP14	FKBP Prolyl Isomerase 14
FKTN	Fukutin
FLCN	Folliculin
FLII	FLII Actin Remodeling Protein
FLNA	Filamin A
FLNB	Filamin B
FLRT3	Fibronectin Leucine Rich Transmembrane Protein 3
FMR1	Fragile X Mental Retardation 1
FOXC1	Forkhead Box C1
FOXG1	Forkhead Box G1
FOXI1	Forkhead Box I1
FOXRED1	FAD Dependent Oxidoreductase Domain Containing 1
FRAS1	Fraser Extracellular Matrix Complex Subunit 1
FREM2	FRAS1 Related Extracellular Matrix Protein 2
FRG1	FSHD Region Gene 1
FSCN2	Fascin Actin-Bundling Protein 2, Retinal
FSHR	Follicle Stimulating Hormone Receptor
FTO	FTO Alpha-Ketoglutarate Dependent Dioxygenase
FUCA1	Alpha-L-Fucosidase 1
FXN	Frataxin
G6PC3	Glucose-6-Phosphatase Catalytic Subunit 3
GAA	Glucosidase Alpha, Acid
GABBR2	Gamma-Aminobutyric Acid Type B Receptor Subunit 2
GABRD	Gamma-Aminobutyric Acid Type A Receptor Delta Subunit
GALC	Galactosylceramidase
GALE	UDP-Galactose-4-Epimerase
GALNS	Galactosamine (N-Acetyl)-6-Sulfatase
GAS8	Growth Arrest Specific 8
GATA1	GATA Binding Protein 1
GATA2	GATA Binding Protein 2
GATA3	GATA Binding Protein 3
GATAD1	GATA Zinc Finger Domain Containing 1
GBA	Glucosylceramidase Beta
PEX14	Peroxisomal Biogenesis Factor 14
PEX16	Peroxisomal Biogenesis Factor 16
PEX19	Peroxisomal Biogenesis Factor 19
PEX2	Peroxisomal Biogenesis Factor 2
PEX26	Peroxisomal Biogenesis Factor 26
PEX3	Peroxisomal Biogenesis Factor 3
PEX5	Peroxisomal Biogenesis Factor 5
PEX6	Peroxisomal Biogenesis Factor 6
PHOX2B	Paired Like Homeobox 2b
PIGA	Phosphatidylinositol Glycan Anchor Biosynthesis Class A
PIK3CA	Phosphatidylinositol-4,5-Bisphosphate 3-Kinase Catalytic Subunit Alpha
PLEC	Plectin
PLXND1	Plexin D1
PMS1	PMS1 Homolog 1, Mismatch Repair System Component
PMS2	PMS1 Homolog 2, Mismatch Repair System Component
PQBP1	Polyglutamine Binding Protein 1
PROKR2	Prokineticin Receptor 2
PRPS1	Phosphoribosyl Pyrophosphate Synthetase 1
PSAP	Prosaposin
PTF1A	Pancreas Specific Transcription Factor, 1a
RBM8A	RNA Binding Motif Protein 8A
RET	Ret Proto-Oncogene
REV3L	REV3 Like, DNA Directed Polymerase Zeta Catalytic Subunit
RMND1	Required For Meiotic Nuclear Division 1 Homolog
ROBO1	Roundabout Guidance Receptor 1
ROR2	Receptor Tyrosine Kinase Like Orphan Receptor 2
SCN4A	Sodium Voltage-Gated Channel Alpha Subunit 4
SCO2	SCO2, Cytochrome C Oxidase Assembly Protein
SLC25A19	Solute Carrier Family 25 Member 19
SMOC1	SPARC Related Modular Calcium Binding 1
SNRPB	Small Nuclear Ribonucleoprotein Polypeptides B And B1
SUOX	Sulfite Oxidase
TFAM	Transcription Factor A, Mitochondrial
TGFBR2	Transforming Growth Factor Beta Receptor 2
TMEM70	Transmembrane Protein 70
TSEN54	TRNA Splicing Endonuclease Subunit 54
TSPYL1	TSPY Like 1
TUFM	Tu Translation Elongation Factor, Mitochondrial
UBR1	Ubiquitin Protein Ligase E3 Component N-Recognin 1
VPS33B	VPS33B, Late Endosome And Lysosome Associated
WASHC5	WASH Complex Subunit 5
WDR11	WD Repeat Domain 11

GBA2	Glucosylceramidase Beta 2
GCK	Glucokinase
GDF3	Growth Differentiation Factor 3
GDF5	Growth Differentiation Factor 5
GDF6	Growth Differentiation Factor 6
GDNF	Glial Cell Derived Neurotrophic Factor
GFER	Growth Factor, Augmenter Of Liver Regeneration
GIPC3	GIPC PDZ Domain Containing Family Member 3
GJA1	Gap Junction Protein Alpha 1
GJB1	Gap Junction Protein Beta 1
GJB2	Gap Junction Protein Beta 2
GJB3	Gap Junction Protein Beta 3
GJB4	Gap Junction Protein Beta 4
GJB6	Gap Junction Protein Beta 6
GJC2	Gap Junction Protein Gamma 2
GLA	Galactosidase Alpha
GLB1	Galactosidase Beta 1
GLI3	GLI Family Zinc Finger 3
GLIS3	GLIS Family Zinc Finger 3
GLYCTK	Glycerate Kinase
GMNN	Geminin DNA Replication Inhibitor
GMPPA	GDP-Mannose Pyrophosphorylase A
GMPPB	GDP-Mannose Pyrophosphorylase B
GNAS	GNAS Complex Locus
GNRH1	Gonadotropin Releasing Hormone 1
GNRHR	Gonadotropin Releasing Hormone 1
GNS	Glucosamine (N-Acetyl)-6-Sulfatase
GP1BB	Glycoprotein Ib Platelet Subunit Beta
GPC3	Glypican 3
GPC4	Glypican 4
GPSM2	G Protein Signaling Modulator 2
GRHL2	Grainyhead Like Transcription Factor 2
GRIP1	Glutamate Receptor Interacting Protein 1
GRXCR1	Glutaredoxin And Cysteine Rich Domain Containing 1
GRXCR2	Glutaredoxin And Cysteine Rich Domain Containing 2
GSC	Goosecoid Homeobox
GSDME	Gasdermin E
GTF2E2	General Transcription Factor IIE Subunit 2
GTF2I	General Transcription Factor IIi
GTF2IRD1	GTF2I Repeat Domain Containing 1
GUCA1B	Guanylate Cyclase Activator 1B
GUCY2D	Guanylate Cyclase 2D, Retinal
GUSB	Glucuronidase Beta
HACE1	HECT Domain And Ankyrin Repeat Containing E3 Ubiquitin Protein Ligase 1
HARS	Histidyl-TRNA Synthetase
HARS2	Histidyl-TRNA Synthetase 2, Mitochondrial
HBB	Hemoglobin Subunit Beta
HCCS	Holocytochrome C Synthase
HDAC4	Histone Deacetylase 4
HDAC8	Histone Deacetylase 8
HESX1	HESX Homeobox 1
HGF	Hepatocyte Growth Factor
HGSNAT	Heparan-Alpha-Glucosaminide N-Acetyltransferase
HIRA	Histone Cell Cycle Regulator
HLA-DPB1	Major Histocompatibility Complex, Class II, DP Beta 1
HNF1B	HNF1 Homeobox B
HNF4A	Hepatocyte Nuclear Factor 4 Alpha
HOXA1	Homeobox A1
HOXA11	Homeobox A11

HOXA2	Homeobox A2
HOXB1	Homeobox B1
HS6ST1	Heparan Sulfate 6-O-Sulfotransferase 1
HSD17B10	Hydroxysteroid 17-Beta Dehydrogenase 10
HSD17B4	Hydroxysteroid 17-Beta Dehydrogenase 4
HTRA2	HtrA Serine Peptidase 2
HYMAI	Hydatidiform Mole Associated And Imprinted
IARS	Isoleucyl-TRNA Synthetase
IARS2	Isoleucyl-TRNA Synthetase 2, Mitochondrial
IDH3B	Isocitrate Dehydrogenase (NAD(+)) 3 Beta
IDS	Iduronate 2-Sulfatase
IDUA	Iduronidase, Alpha-L-
IFT140	Intraflagellar Transport 140
IFT172	Intraflagellar Transport 172
IFT27	Intraflagellar Transport 27
IGBP1	Immunoglobulin Binding Protein 1
IGF1	Insulin Like Growth Factor 1
IL17RD	Interleukin 17 Receptor D
ILDR1	Immunoglobulin Like Domain Containing Receptor 1
IMPAD1	Inositol Monophosphatase Domain Containing 1
IMPDH1	Inosine Monophosphate Dehydrogenase 1
IMPG2	Interphotoreceptor Matrix Proteoglycan 2
INS	Insulin
IQCB1	IQ Motif Containing B1
IQSEC2	IQ Motif And Sec7 Domain 2
IRX5	Iroquois Homeobox 5
ITGB6	Integrin Subunit Beta 6
ITM2B	Integral Membrane Protein 2B
JMJD1C	Jumonji Domain Containing 1C
KARS	Lysyl-TRNA Synthetase
KAT6B	Lysine Acetyltransferase 6B
KCNAB2	Potassium Voltage-Gated Channel Subfamily A Regulatory Beta Subunit 2
KCNC3	Potassium Voltage-Gated Channel Subfamily C Member 3
KCNE1	Potassium Voltage-Gated Channel Subfamily E Regulatory Subunit 1
KCNE5	Potassium Voltage-Gated Channel Subfamily E Regulatory Subunit 5
KCNH1	Potassium Voltage-Gated Channel Subfamily H Regulatory Subunit 1
KCNJ10	Potassium Voltage-Gated Channel Subfamily J Member 10
KCNJ11	Potassium Voltage-Gated Channel Subfamily J Member 11
KCNJ13	Potassium Voltage-Gated Channel Subfamily J Member 13
KCNQ1	Potassium Voltage-Gated Channel Subfamily Q Member 1
KCNQ4	Potassium Voltage-Gated Channel Subfamily Q Member 4
KDM6A	Lysine Demethylase 6A
KDSR	3-Ketodihydrosphingosine Reductase
KIF7	Kinesin Family Member 7
KISS1	KiSS-1 Metastasis Suppressor
KISS1R	KISS1 Receptor
KIT	KIT Proto-Oncogene, Receptor Tyrosine Kinase
KITLG	KIT Ligand
KIZ	Kizuna Centrosomal Protein
KLHL7	Kelch Like Family Member 7
KLLN	Killin, P53 Regulated DNA Replication Inhibitor
KMT2A	Lysine Methyltransferase 2A
KMT2D	Lysine Methyltransferase 2D
KRAS	KRAS Proto-Oncogene, GTPase

KYNU	Kynureninase
L2HGDH	L-2-Hydroxyglutarate Dehydrogenase
LAMA4	Laminin Subunit Alpha 4
LAMB1	Laminin Subunit Beta 1
LARS	Leucyl-TRNA Synthetase
LARS2	Leucyl-TRNA Synthetase 2, Mitochondrial
LCA5	Lebercilin LCA5
LDB3	LIM Domain Binding 3
LEMD3	LEM Domain Containing 3
LETM1	Leucine Zipper And EF-Hand Containing Transmembrane Protein 1
LHFPL5	LHFPL Tetraspan Subfamily Member 5
LHX1	LIM Homeobox 1
LHX3	LIM Homeobox 3
LIG4	DNA Ligase 4
LIMK1	LIM Domain Kinase 1
LMNA	Lamin A/C
LMNB1	Lamin B1
LMNB2	Lamin B2
LMX1B	LIM Homeobox Transcription Factor 1 Beta
LONP1	Lon Peptidase 1, Mitochondrial
LOR	Loricrin
LOXHD1	Lipoxygenase Homology Domains 1
LRAT	Lecithin Retinol Acyltransferase
LRP2	LDL Receptor Related Protein 2
LRP4	LDL Receptor Related Protein 4
LRP5	LDL Receptor Related Protein 5
LRTOMT	Leucine Rich Transmembrane And O-Methyltransferase Domain Containing
LZTFL1	Leucine Zipper Transcription Factor Like 1
LZTR1	Leucine Zipper Like Transcription Regulator 1
MAD2L2	Mitotic Arrest Deficient 2 Like 2
MAF	MAF BZIP Transcription Factor
MAFB	MAF BZIP Transcription Factor B
MAK	Male Germ Cell Associated Kinase
MAN2B1	Mannosidase Alpha Class 2B Member 1
MANBA	Mannosidase Beta
MAP2K1	Mitogen-Activated Protein Kinase Kinase 1
MAP3K20	Mitogen-Activated Protein Kinase Kinase Kinase 20
MAP3K7	Mitogen-Activated Protein Kinase Kinase Kinase 7
MAPK1	Mitogen-Activated Protein Kinase 1
MARS2	Methionyl-TRNA Synthetase 2, Mitochondrial
MARVELD2	MARVEL Domain Containing 2
MASP1	Mannan Binding Lectin Serine Peptidase 1
MBTPS2	Membrane Bound Transcription Factor Peptidase, Site 2
MCM2	Minichromosome Maintenance Complex Component 2
MCTP2	Multiple C2 And Transmembrane Domain Containing 2
MECOM	MDS1 And EVI1 Complex Locus
MECP2	Methyl-CpG Binding Protein 2
MED12	Mediator Complex Subunit 12
MEGF8	Multiple EGF Like Domains 8
MEOX1	Mesenchyme Homeobox 1
MERTK	MER Proto-Oncogene, Tyrosine Kinase
MFN2	Mitofusin 2
MGAT2	Mannosyl (Alpha-1,6-)-Glycoprotein Beta-1,2-N-Acetylglucosaminyltransferase
MGP	Matrix Gla Protein
MIR96	MicroRNA 96
MITF	Melanocyte Inducing Transcription Factor
MKKS	McKusick-Kaufman Syndrome

MKS1	MKS Transition Zone Complex Subunit 1
MLXIPL	MLX Interacting Protein Like
MOGS	Mannosyl-Oligosaccharide Glucosidase
MORC2	MORC Family CW-Type Zinc Finger 2
MPZ	Myelin Protein Zero
MSRB3	Methionine Sulfoxide Reductase B3
MYBPC3	Myosin Binding Protein C, Cardiac
MYCN	MYCN Proto-Oncogene, BHLH Transcription Factor
MYD88	Myeloid Differentiation Primary Response 88
MYH14	Myosin Heavy Chain 14
MYH3	Myosin Heavy Chain 3
MYH6	Myosin Heavy Chain 6
MYH7	Myosin Heavy Chain 7
MYH9	Myosin Heavy Chain 9
MYO15A	Myosin XVA
MYO1A	Myosin IA
MYO3A	Myosin IIIA
MYO6	Myosin VI
MYO7A	Myosin VIIA
MYPN	Myopalladin
NAA10	N(Alpha)-Acetyltransferase 10, NatA Catalytic Subunit
NAGA	Alpha-N-Acetylgalactosaminidase
NAGLU	N-Acetyl-Alpha-Glucosaminidase
NALCN	Sodium Leak Channel, Non-Selective
ND1	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 1
ND4	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 4
ND5	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 5
ND6	Mitochondrially Encoded NADH:Ubiquinone Oxidoreductase Core Subunit 6
NDP	Norrin Cystine Knot Growth Factor NDP
NDRG1	N-Myc Downstream Regulated 1
NDUFA1	NADH:Ubiquinone Oxidoreductase Subunit A1
NDUFA10	NADH:Ubiquinone Oxidoreductase Subunit A10
NDUFA11	NADH:Ubiquinone Oxidoreductase Subunit A11
NDUFA12	NADH:Ubiquinone Oxidoreductase Subunit A12
NDUFA2	NADH:Ubiquinone Oxidoreductase Subunit A2
NDUFA9	NADH:Ubiquinone Oxidoreductase Subunit A9
NDUFAF1	NADH:Ubiquinone Oxidoreductase Complex Assembly Factor 1
NDUFAF2	NADH:Ubiquinone Oxidoreductase Complex Assembly Factor 2
NDUFAF3	NADH:Ubiquinone Oxidoreductase Complex Assembly Factor 3
NDUFAF4	NADH:Ubiquinone Oxidoreductase Complex Assembly Factor 4
NDUFAF5	NADH:Ubiquinone Oxidoreductase Complex Assembly Factor 5
NDUFAF6	NADH:Ubiquinone Oxidoreductase Complex Assembly Factor 6
NDUFB11	NADH:Ubiquinone Oxidoreductase Subunit B11
NDUFB3	NADH:Ubiquinone Oxidoreductase Subunit B3
NDUFB9	NADH:Ubiquinone Oxidoreductase Subunit B9
NDUFS1	NADH:Ubiquinone Oxidoreductase Core Subunit S1
NDUFS2	NADH:Ubiquinone Oxidoreductase Core Subunit S2
NDUFS3	NADH:Ubiquinone Oxidoreductase Core Subunit S3
NDUFS4	NADH:Ubiquinone Oxidoreductase Core Subunit S4
NDUFS6	NADH:Ubiquinone Oxidoreductase Core Subunit S6
NDUFS7	NADH:Ubiquinone Oxidoreductase Core Subunit S7
NDUFS8	NADH:Ubiquinone Oxidoreductase Core Subunit S8

NDUFV1	NADH:Ubiquinone Oxidoreductase Core Subunit V1
NDUFV2	NADH:Ubiquinone Oxidoreductase Core Subunit V2
NEBL	Nebulette
NEDD4L	NEDD4 Like E3 Ubiquitin Protein Ligase
NEK2	NIMA Related Kinase 2
NELFA	Negative Elongation Factor Complex Member A
NEU1	Neuraminidase 1
NEXN	Nexilin F-Actin Binding Protein
NF2	Neurofibromin 2
NFIX	Nuclear Factor I X
NHP2	NHP2 Ribonucleoprotein
NIPAL4	NIPA Like Domain Containing 4
NIPBL	NIPBL Cohesin Loading Factor
NLRC4	NLR Family CARD Domain Containing 4
NLRP12	NLR Family Pyrin Domain Containing 12
NLRP3	NLR Family Pyrin Domain Containing 3
NMNAT1	Nicotinamide Nucleotide Adenylyltransferase 1
NOG	Noggin
NOP10	NOP10 Ribonucleoprotein
NOTCH2	Notch Receptor 2
NOTCH3	Notch Receptor 3
NPHP1	Nephrocystin 1
NR2E3	Nuclear Receptor Subfamily 2 Group E Member 3
NR5A1	Nuclear Receptor Subfamily 5 Group A Member 1
NRAS	NRAS Proto-Oncogene, GTPase
NRL	Neural Retina Leucine Zipper
NRTN	Neurturin
NSD1	Nuclear Receptor Binding SET Domain Protein 1
NSD2	Nuclear Receptor Binding SET Domain Protein 2
NSDHL	NAD(P) Dependent Steroid Dehydrogenase-Like
NSMF	NMDA Receptor Synaptonuclear Signaling And Neuronal Migration Factor
NSUN2	NOP2/Sun RNA Methyltransferase 2
NTNG1	Netrin G1
NUBPL	Nucleotide Binding Protein Like
NUP107	Nucleoporin 107
NUS1	NUS1 Dehydrodolichyl Diphosphate Synthase Subunit
OFD1	OFD1, Centriole And Centriolar Satellite Protein
OPA1	OPA1 Mitochondrial Dynamin Like GTPase
ORC1	Origin Recognition Complex Subunit 1
ORC4	Origin Recognition Complex Subunit 4
ORC6	Origin Recognition Complex Subunit 6
OSBPL2	Oxysterol Binding Protein Like 2
OTOA	Otoancorin
OTOF	Otoferlin
OTOG	Otogelin
OTOGL	Otogelin Like
OTUD6B	OTU Domain Containing 6B
OTX2	Orthodenticle Homeobox 2
P2RX2	Purinergic Receptor P2X 2
PALB2	Partner And Localizer Of BRCA2
PARN	Poly(A)-Specific Ribonuclease
PAX1	Paired Box 1
PAX2	Paired Box 2
PAX3	Paired Box 3
PCDH15	Protocadherin Related 15
PCLO	Piccolo Presynaptic Cytomatrix Protein
PCNT	Pericentrin
PCYT1A	Phosphate Cytidylyltransferase 1, Choline, Alpha
PDE4D	Phosphodiesterase 4D

PDE6A	Phosphodiesterase 6A
PDE6B	Phosphodiesterase 6B
PDE6G	Phosphodiesterase 6G
PDGFRB	Platelet Derived Growth Factor Receptor Beta
PDX1	Pancreatic And Duodenal Homeobox 1
PDZD7	PDZ Domain Containing 7
PEPD	Peptidase D
PET100	PET100 Cytochrome C Oxidase Chaperone
PEX1	Peroxisomal Biogenesis Factor 1
PEX10	Peroxisomal Biogenesis Factor 10
PEX11B	Peroxisomal Biogenesis Factor 11B
PEX12	Peroxisomal Biogenesis Factor 12
PEX13	Peroxisomal Biogenesis Factor 13
PEX14	Peroxisomal Biogenesis Factor 14
PEX16	Peroxisomal Biogenesis Factor 16
PEX19	Peroxisomal Biogenesis Factor 19
PEX2	Peroxisomal Biogenesis Factor 2
PEX26	Peroxisomal Biogenesis Factor 26
PEX3	Peroxisomal Biogenesis Factor 3
PEX5	Peroxisomal Biogenesis Factor 5
PEX6	Peroxisomal Biogenesis Factor 6
PEX7	Peroxisomal Biogenesis Factor 7
PGM3	Phosphoglucomutase 3
PHEX	Phosphate Regulating Endopeptidase Homolog X-Linked
PHF6	PHD Finger Protein 6
PHOX2B	Paired Like Homeobox 2b
PHYH	Phytanoyl-CoA 2-Hydroxylase
PIEZO1	Piezo Type Mechanosensitive Ion Channel Component 1
PIEZO2	Piezo Type Mechanosensitive Ion Channel Component 2
PIGA	Phosphatidylinositol Glycan Anchor Biosynthesis Class A
PIGL	Phosphatidylinositol Glycan Anchor Biosynthesis Class L
PIGV	Phosphatidylinositol Glycan Anchor Biosynthesis Class V
PIK3CA	Phosphatidylinositol-4,5-Bisphosphate 3-Kinase Catalytic Subunit Alpha
PIK3R1	Phosphoinositide-3-Kinase Regulatory Subunit 1
PITX2	Paired Like Homeodomain 2
PJVK	Pejvakin
PLA2G6	Phospholipase A2 Group VI
PLAGL1	PLAG1 Like Zinc Finger 1
PLN	Phospholamban
PLOD3	Procollagen-Lysine,2-Oxoglutarate 5-Dioxygenase 3
PLP1	Proteolipid Protein 1
PLXND1	Plexin D1
PMP22	Peripheral Myelin Protein 22
PNPLA1	Patatin Like Phospholipase Domain Containing 1
PNPLA2	Patatin Like Phospholipase Domain Containing 2
PNPLA6	Patatin Like Phospholipase Domain Containing 6
PNPLA8	Patatin Like Phospholipase Domain Containing 8
PNPT1	Polyribonucleotide Nucleotidyltransferase 1
POGZ	Pogo Transposable Element Derived With ZNF Domain
POLA1	DNA Polymerase Alpha 1, Catalytic Subunit
POLD1	DNA Polymerase Delta 1, Catalytic Subunit
POLG	DNA Polymerase Gamma, Catalytic Subunit
POLR1C	RNA Polymerase I And III Subunit C
POLR1D	RNA Polymerase I And III Subunit D
POMGNT1	Protein O-Linked Mannose N-Acetylglucosaminyltransferase 1 (Beta 1,2-)
POMK	Protein-O-Mannose Kinase
POR	Cytochrome P450 Oxidoreductase
PORCN	Porcupine O-Acyltransferase

POU3F4	POU Class 3 Homeobox 4
POU4F3	POU Class 4 Homeobox 3
PPP1R15B	Protein Phosphatase 1 Regulatory Subunit 15B
PQBP1	Polyglutamine Binding Protein 1
PRCD	Photoreceptor Disc Component
PRDM16	PR/SET Domain 16
PRDM5	PR/SET Domain 5
PRKAR1A	Protein Kinase CAMP-Dependent Type I Regulatory Subunit Alpha
PRKDC	Protein Kinase, DNA-Activated, Catalytic Subunit
PROK2	Prokineticin 2
PROKR2	Prokineticin Receptor 2
PROM1	Prominin 1
PRPF3	Pre-MRNA Processing Factor 3
PRPF31	Pre-MRNA Processing Factor 31
PRPF4	Pre-MRNA Processing Factor 4
PRPF6	Pre-MRNA Processing Factor 6
PRPF8	Pre-MRNA Processing Factor 8
PRPH2	Peripherin 2
PRPS1	Phosphoribosyl Pyrophosphate Synthetase 1
PRRT2	Proline Rich Transmembrane Protein 2
PRRX1	Paired Related Homeobox 1
PRTN3	Proteinase 3
PSEN1	Presenilin 1
PSEN2	Presenilin 2
PSMC3IP	PSMC3 Interacting Protein
PTDSS1	Phosphatidylserine Synthase 1
PTEN	Phosphatase And Tensin Homolog
PTH1R	Parathyroid Hormone 1 Receptor
PTPN11	Protein Tyrosine Phosphatase, Non-Receptor Type 11
PTPN22	Protein Tyrosine Phosphatase Non-Receptor Type 22
PTPRQ	Protein Tyrosine Phosphatase Receptor Type Q
PTRH2	Peptidyl-TRNA Hydrolase 2
PYCR2	Pyrroline-5-Carboxylate Reductase 2
RAB23	RAB23, Member RAS Oncogene Family
RAD21	RAD21 Cohesin Complex Component
RAD51	RAD51 Recombinase
RAD51C	RAD51 Paralog C
RAF1	Raf-1 Proto-Oncogene, Serine/Threonine Kinase
RAI1	Retinoic Acid Induced 1
RAP1A	RAP1A, Member Of RAS Oncogene Family
RAP1B	RAP1B, Member Of RAS Oncogene Family
RASA2	RAS P21 Protein Activator 2
RBM20	RNA Binding Motif Protein 20
RBM8A	RNA Binding Motif Protein 8A
RBP3	Retinol Binding Protein 3
RD3	Retinal Degeneration 3, GUCY2D Regulator
RDH12	Retinol Dehydrogenase 12
RDX	Radixin
RECQL4	RecQ Like Helicase 4
REEP6	Receptor Accessory Protein 6
RERE	Arginine-Glutamic Acid Dipeptide Repeats
RET	Ret Proto-Oncogene
REV3L	REV3 Like, DNA Directed Polymerase Zeta Catalytic Subunit
RFC2	Replication Factor C Subunit 2
RFT1	RFT1 Homolog
RGR	Retinal G Protein Coupled Receptor
RHO	Rhodopsin
RIPOR2	RHO Family Interacting Cell Polarization Regulator 2

RIT1	Ras Like Without CAAX 1
RLBP1	Retinaldehyde Binding Protein 1
RMND1	Required For Meiotic Nuclear Division 1 Homolog
RNASET2	Ribonuclease T2
RNF135	Ring Finger Protein 135
RNR1	RNA, Ribosomal 45S Cluster 1
ROBO3	Roundabout Guidance Receptor 3
ROM1	Retinal Outer Segment Membrane Protein 1
ROR2	Receptor Tyrosine Kinase Like Orphan Receptor 2
RP1	RP1 Axonemal Microtubule Associated
RP2	RP2 Activator Of ARL3 GTPase
RP9	RP9 Pre-MRNA Splicing Factor
RPE65	Retinoid Isomerohydrolase RPE65
RPGR	Retinitis Pigmentosa GTPase Regulator
RPGRIP1	RPGR Interacting Protein 1
RPL10	Ribosomal Protein L10
RPL11	Ribosomal Protein L11
RPS23	Ribosomal Protein S23
RPS26	Ribosomal Protein S26
RPS28	Ribosomal Protein S28
RPS6KA3	Ribosomal Protein S6 Kinase A3
RRAS	RAS Related
RREB1	Ras Responsive Element Binding Protein 1
RRM2B	Ribonucleotide Reductase Regulatory TP53 Inducible Subunit M2B
RTEL1	Regulator Of Telomere Elongation Helicase 1
RUNX2	RUNX Family Transcription Factor 2
S1PR2	Sphingosine-1-Phosphate Receptor 2
SAG	S-Antigen Visual Arrestin
SALL1	Spalt Like Transcription Factor 1
SALL4	Spalt Like Transcription Factor 4
SBF2	SET Binding Factor 2
SC5D	Sterol-C5-Desaturase
SCN1A	Sodium Voltage-Gated Channel Alpha Subunit 1
SCN5A	Sodium Voltage-Gated Channel Alpha Subunit 5
SCO1	SCO Cytochrome C Oxidase Assembly Protein 1
SDCCAG8	Serologically Defined Colon Cancer Antigen 8
SDHA	Succinate Dehydrogenase Complex Flavoprotein Subunit A
SDHB	Succinate Dehydrogenase Complex Iron Sulfur Subunit B
SDHC	Succinate Dehydrogenase Complex Subunit C
SDHD	Succinate Dehydrogenase Complex Subunit D
SEC23B	SEC23 Homolog B, Coat Complex II Component
SEC24C	SEC24 Homolog C, COPII Coat Complex Component
SEM1	SEM1 26S Proteasome Complex Subunit
SEMA3A	Semaphorin 3A
SEMA3C	Semaphorin 3C
SEMA3D	Semaphorin 3D
SEMA3E	Semaphorin 3E
SEMA4A	Semaphorin 4A
SERAC1	Serine Active Site Containing 1
SERPINB6	Serpin Family B Member 6
SETD2	SET Domain Containing 2
SETD5	SET Domain Containing 5
SF3B4	Splicing Factor 3b Subunit 4
SGCD	Sarcoglycan Delta
SGSH	N-Sulfoglucosamine Sulfohydrolase
SH3TC2	SH3 Domain And Tetratricopeptide Repeats 2
SHANK3	SH3 And Multiple Ankyrin Repeat Domains 3
SHOC2	SHOC2 Leucine Rich Repeat Scaffold Protein

SIN3A	SIN3 Transcription Regulator Family Member A
SIX1	SIX Homeobox 1
SIX5	SIX Homeobox 5
SIX6	SIX Homeobox 6
SKI	SKI Proto-Oncogene
SLC17A8	Solute Carrier Family 17 Member 8
SLC19A2	Solute Carrier Family 19 Member 2
SLC25A4	Solute Carrier Family 25 Member 4
SLC26A2	Solute Carrier Family 26 Member 2
SLC26A4	Solute Carrier Family 26 Member 4
SLC26A5	Solute Carrier Family 26 Member 5
SLC29A3	Solute Carrier Family 29 Member 3
SLC33A1	Solute Carrier Family 33 Member 1
SLC39A8	Solute Carrier Family 39 Member 8
SLC4A11	Solute Carrier Family 4 Member 11
SLC52A2	Solute Carrier Family 52 Member 2
SLC52A3	Solute Carrier Family 52 Member 3
SLC7A14	Solute Carrier Family 7 Member 14
SLITRK6	SLIT And NTRK Like Family Member 6
SLX4	SLX4 Structure-Specific Endonuclease Subunit
SMAD4	SMAD Family Member 4
SMARCA4	SWI/SNF Related, Matrix Associated, Actin Dependent Regulator Of Chromatin, Subfamily A, Member 4
SMARCB1	SWI/SNF Related, Matrix Associated, Actin Dependent Regulator Of Chromatin, Subfamily B, Member 1
SMARCE1	SWI/SNF Related, Matrix Associated, Actin Dependent Regulator Of Chromatin, Subfamily E, Member 1
SMC1A	Structural Maintenance Of Chromosomes 1A
SMC3	Structural Maintenance Of Chromosomes 3
SMCHD1	Structural Maintenance Of Chromosomes Flexible Hinge Domain Containing 1
SNAI2	Snail Family Transcriptional Repressor 2
SNAP29	Synaptosome Associated Protein 29
SNRNP200	Small Nuclear Ribonucleoprotein U5 Subunit 200
SNRPB	Small Nuclear Ribonucleoprotein Polypeptides B And B1
SNX10	Sorting Nexin 10
SNX14	Sorting Nexin 14
SOS1	SOS Ras/Rac Guanine Nucleotide Exchange Factor 1
SOS2	SOS Ras/Rac Guanine Nucleotide Exchange Factor 2
SOST	Sclerostin
SOX10	SRY-Box 10
SOX11	SRY-Box 11
SOX2	SRY-Box 2
SOX3	SRY-Box 3
SOX9	SRY-Box 9
SPATA5	Spermatogenesis Associated 5
SPATA7	Spermatogenesis Associated 7
SPECC1L	Sperm Antigen With Calponin Homology And Coiled-Coil Domains 1 Like
SPIDR	Scaffold Protein Involved In DNA Repair
SPRY4	Sprouty RTK Signaling Antagonist 4
SPTLC1	Serine Palmitoyltransferase Long Chain Base Subunit 1
SQSTM1	Sequestosome 1
SRCAP	Snf2 Related CREBBP Activator Protein
SRP72	Signal Recognition Particle 72
SRY	Sex Determining Region Y
ST3GAL5	ST3 Beta-Galactoside Alpha-2,3-Sialyltransferase 5
STAMBP	STAM Binding Protein
STAT3	Signal Transducer And Activator Of Transcription 3
STRC	Stereocilin
SUCLA2	Succinate-CoA Ligase ADP-Forming Beta Subunit

SUCLG1	Succinate-CoA Ligase Alpha Subunit
SUMF1	Sulfatase Modifying Factor 1
SURF1	SURF1 Cytochrome C Oxidase Assembly Factor
SYNE4	Spectrin Repeat Containing Nuclear Envelope Family Member 4
SYT2	Synaptotagmin 2
TAC3	Tachykinin 3
TACO1	Translational Activator Of Cytochrome C Oxidase I
TACR3	Tachykinin Receptor 3
TAF1	TATA-Box Binding Protein Associated Factor 1
TANGO2	Transport And Golgi Organization 2 Homolog
TAZ	Tafazzin
TBC1D24	TBC1 Domain Family Member 24
TBL2	Transducin Beta Like 2
TBX1	T-Box 1
TBX15	T-Box 15
TBX22	T-Box 22
TBX4	T-Box 4
TCAP	Titin-Cap
TCF12	Transcription Factor 12
TCIRG1	T Cell Immune Regulator 1, ATPase H+ Transporting V0 Subunit A3
TCOF1	Treacle Ribosome Biogenesis Factor 1
TCTN3	Tectonic Family Member 3
TECTA	Tectorin Alpha
TELO2	Telomere Maintenance 2
TERC	Telomerase RNA Component
TERT	Telomerase Reverse Transcriptase
TFAP2A	Transcription Factor AP-2 Alpha
TFAP2B	Transcription Factor AP-2 Beta
TGFB1	Transforming Growth Factor Beta 1
TGM1	Transglutaminase 1
THRB	Thyroid Hormone Receptor Beta
TIMM8A	Translocase Of Inner Mitochondrial Membrane 8A
TIMMDC1	Translocase Of Inner Mitochondrial Membrane Domain Containing 1
TINF2	TERF1 Interacting Nuclear Factor 2
TK2	Thymidine Kinase 2
TMC1	Transmembrane Channel Like 1
TMEM126A	Transmembrane Protein 126A
TMEM126B	Transmembrane Protein 126B
TMIE	Transmembrane Inner Ear
TMPO	Thymopoietin
TNC	Tenascin C
TNFRSF11A	TNF Receptor Superfamily Member 11a
TNFRSF11B	TNF Receptor Superfamily Member 11b
TNFSF11	TNF Superfamily Member 11
TNNC1	Troponin C1, Slow Skeletal And Cardiac Type
TNNI3	Troponin I3, Cardiac Type
TNNT2	Troponin T2, Cardiac Type
TOPORS	TOP1 Binding Arginine/Serine Rich Protein
TP63	Tumor Protein P63
TPM1	Tropomyosin 1
TPRN	Taperin
TRAPPC11	Trafficking Protein Particle Complex 11
TRIM32	Tripartite Motif Containing 32
TRIOBP	TRIO And F-Actin Binding Protein
TRIP13	Thyroid Hormone Receptor Interactor 13
TRMU	TRNA 5-Methylaminomethyl-2-Thiouridylate Methyltransferase

TRNC	Mitochondrially Encoded TRNA-Cys (UGU/C)
TRNE	Mitochondrially Encoded TRNA-Glu (GAA/G)
TRNF	Mitochondrially Encoded TRNA-Phe (UUU/C)
TRNH	Mitochondrially Encoded TRNA-His (CAU/C)
TRNI	Mitochondrially Encoded TRNA-Ile (AUU/C)
TRNK	Mitochondrially Encoded TRNA-Lys (AAA/G)
TRNL1	Mitochondrially Encoded TRNA-Leu (UUA/G) 1
TRNN	Mitochondrially Encoded TRNA-Asn (AAU/C)
TRNP	Mitochondrially Encoded TRNA-Pro (CCN)
TRNQ	Mitochondrially Encoded TRNA-Gln (CAA/G)
TRNS1	Mitochondrially Encoded TRNA-Ser (UCN) 1
TRNS2	Mitochondrially Encoded TRNA-Ser (AGU/C) 2
TRNT1	TRNA Nucleotidyl Transferase 1
TRNV	Mitochondrially Encoded TRNA-Val (GUN)
TRNW	Mitochondrially Encoded TRNA-Trp (UGA/G)
TRPS1	Transcriptional Repressor GATA Binding 1
TRPV4	Transient Receptor Potential Cation Channel Subfamily V Member 4
TSHZ1	Teashirt Zinc Finger Homeobox 1
TSPEAR	Thrombospondin Type Laminin G Domain And EAR Repeats
TSR2	TSR2 Ribosome Maturation Factor
TTC19	Tetratricopeptide Repeat Domain 19
TTC8	Tetratricopeptide Repeat Domain 8
TTN	Titin
TTR	Transthyretin
TUB	TUB Bipartite Transcription Factor
TULP1	TUB Like Protein 1
TWIST1	Twist Family BHLH Transcription Factor 1
TWIST2	Twist Family BHLH Transcription Factor 2
TWNK	Twinkle MtDNA Helicase
TXNL4A	Thioredoxin Like 4A
TXNRD2	Thioredoxin Reductase 2
TYMP	Thymidine Phosphorylase
TYR	Tyrosinase
UBE2T	Ubiquitin Conjugating Enzyme E2 T
UBR1	Ubiquitin Protein Ligase E3 Component N-Recognin 1
UFD1	Ubiquitin Recognition Factor In ER Associated Degradation 1
UGT1A1	UDP Glucuronosyltransferase Family 1 Member A1
USB1	U6 SnRNA Biogenesis Phosphodiesterase 1
USH1C	USH1 Protein Network Component Harmonin
USH1G	USH1 Protein Network Component Sans
USH2A	Usherin
USP9X	Ubiquitin Specific Peptidase 9 X-Linked
VCL	Vinculin
VHL	Von Hippel-Lindau Tumor Suppressor
VPS11	VPS11, CORVET/HOPS Core Subunit
VPS13B	Vacuolar Protein Sorting 13 Homolog B
WAC	WW Domain Containing Adaptor With Coiled-Coil
WDPCP	WD Repeat Containing Planar Cell Polarity Effector
WDR11	WD Repeat Domain 11
WFS1	Wolframin ER Transmembrane Glycoprotein
WHRN	Whirlin
WNT10B	Wnt Family Member 10B
WNT5A	Wnt Family Member 5A
WRAP53	WD Repeat Containing Antisense To TP53
XPA	XPA, DNA Damage Recognition And Repair Factor
XPC	XPC Complex Subunit, DNA Damage Recognition And Repair Factor

XPNPEP3	X-Prolyl Aminopeptidase 3
XRCC2	X-Ray Repair Cross Complementing 2
XRCC4	X-Ray Repair Cross Complementing 4
XYLT2	Xylosyltransferase 2
YAP1	Yes Associated Protein 1
ZBTB20	Zinc Finger And BTB Domain Containing 20
ZFP57	ZFP57 Zinc Finger Protein
ZIC1	Zic Family Member 1
ZMPSTE24	Zinc Metallopeptidase STE24
ZNF408	Zinc Finger Protein 408
ZNF469	Zinc Finger Protein 469
ZNF513	Zinc Finger Protein 513

Severe hearing impairment (HP:0012714), 5 genes

BRAF	B-Raf Proto-Oncogene, Serine/Threonine Kinase
GPSM2	G Protein Signaling Modulator 2
PTPN11	Protein Tyrosine Phosphatase, Non-Receptor Type 11
RAF1	Raf-1 Proto-Oncogene, Serine/Threonine Kinase
TRIOBP	TRIO And F-Actin Binding Protein