

## dbCMMS.v13

Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild (844st)

AARS2	ALG11	BCAT2	COG8
AASS	ALG12	BCKDHA	COQ2
ABAT	ALG13	BCKDHB	COQ4
ABCC8	ALG14	BCKDK	COQ5
ABCD1	ALG2	BCS1L	COQ6
ABCD4	ALG3	BLOC1S3	COQ7
ABHD12	ALG6	BOLA3	COQ8A
ABHD5	ALG8	BSCL2	COQ9
ACAD8	ALG9	BTD	COX10
ACAD9	ALKBH1	C12ORF65	COX14
ACADM	ALPL	C19ORF12	COX15
ACADS	AMACR	C19ORF70	COX20
ACADSB	AMN	C1QBP	COX4I1
ACADVL	AMPD1	CA5A	COX4I2
ACAT1	AMPD2	CACNA1A	COX5A
ACER3	AMT	CACNA1S	COX6B1
ACO2	ANTXR2	CAD	COX8A
ACOX1	AP1S1	CARS2	CP
ACSF3	AP3B1	CAT	CPOX
ACY1	AP3D1	CBS	CPS1
ADA	AP5Z1	CCDC115	CPT1A
ADAR	APOPT1	CCDC174	CPT2
ADCK2	APRT	CCDC88C	CRAT
ADCY5	ARG1	CD320	CTH
ADK	ARSA	CEP89	CTNS
ADPRHL2	ARSB	CERS1	CTSA
ADSL	ASAH1	CHCHD10	CTSD
ADSSL1	ASL	CHKB	CTSF
AFG3L2	ASNS	CISD2	CTSK
AGA	ASPA	CLDN16	CUBN
AGK	ASS1	CLN3	CWF19L1
AGL	ATAD3A	CLN5	CYP27A1
AGPS	ATAD3B	CLN6	CYP7B1
AGXT	ATAD3C	CLN8	D2HGDH
AHCY	ATIC	CLPB	DARS
AIFM1	ATP13A2	CLPP	DARS2
AIMP1	ATP5E	CLTC	DBH
AKR1D1	ATP5F1D	CMPK2	DBT
ALAD	ATP5MD	CNKSR2	DCAF17
ALAS2	ATP6AP1	COA5	DDC
ALDH18A1	ATP6V0A2	COA6	DGUOK
ALDH3A2	ATP7A	COA7	DHCR24
ALDH4A1	ATP7B	COASY	DHCR7
ALDH5A1	ATPAF2	COG1	DHDDS
ALDH6A1	AUH	COG2	DHFR
ALDH7A1	B4GALNT1	COG4	DHODH
ALDOA	B4GALT1	COG5	DIAPH1
ALDOB	B4GALT7	COG6	DLAT
ALG1	BAAT	COG7	DLD

**dbCMMS.v13**

DMGDH	FDX2	GLUL	HSD17B10
DNA2	FDXR	GLYCTK	HSD17B4
DNAJC12	FECH	GM2A	HSD3B7
DNAJC19	FH	GMPPA	HSPD1
DNAJC3	FIG4	GMPPB	HTRA2
DNAJC5	FLAD1	GNAO1	HYAL1
DNM1L	FMO3	GNE	HYOU1
DOLK	FOLR1	GNMT	IARS
DPAGT1	FOXI1	GNPAT	IARS2
DPM1	FOXRED1	GNPTAB	IBA57
DPM2	FTCD	GNPTG	IDH2
DPM3	FTL	GNS	IDH3A
DPYD	FUCA1	GOT2	IDS
DPYS	FUT8	GPD1	IDUA
DTNBP1	FXN	GPHN	IER3IP1
EARS2	G6PC	GPR161	IFIH1
EBP	G6PD	GRHPR	IGSF1
ECHS1	GAA	GRN	INSR
ECSIT	GABBR2	GSS	IRF2BPL
EIF2B1	GABRG2	GTPBP3	ISCA1
EIF2B2	GALC	GUSB	ISCA2
EIF2B3	GALE	GYG1	ISCU
EIF2B4	GALK1	GYG2	ISPD
EIF2B5	GALNS	GYS1	IVD
ELAC2	GALNT3	GYS2	JAG1
ELOVL4	GALT	HAAO	KARS
ELOVL5	GAMT	HACE1	KCNA4
ENO3	GARS	HADH	KCNJ11
EPG5	GATB	HADHA	KIF5A
EPHX1	GATC	HADHB	KYNU
EPM2A	GATM	HARS	L2HGDH
EPRS	GBA	HCFC1	LAMA1
ERAL1	GBE1	HEPACAM	LAMP2
ERCC6	GCDH	HEXA	LARS
ETFA	GCH1	HEXB	LARS2
ETFB	GCK	HGD	LBR
ETFDH	GCLC	HGSNAT	LDHA
ETHE1	GCSH	HIBCH	LHX4
EXOSC3	GFER	HIKESHI	LIAS
EXOSC8	GFM1	HLCS	LIPA
EXT1	GGT1	HMBS	LIPE
EXT2	GIF	HMGCL	LIPT1
FA2H	GJC2	HMGCS2	LIPT2
FAH	GK	HOGA1	LMBRD1
FAM126A	GLA	HPD	LNPK
FAR1	GLB1	HPRT1	LPIN1
FARS2	GLDC	HPS1	LRP2
FASTKD2	GLRA1	HPS3	LRPPRC
FBP1	GLRX5	HPS4	LYRM4
FBXL4	GLS	HPS5	LYRM7
FDFT1	GLUD1	HPS6	MAG

**dbCMMS.v13**

MAGED2	MSTO1	NDUFS4	PDSS1
MAGEL2	MTFMT	NDUFS5	PDSS2
MAGT1	MTHFD1	NDUFS6	PEPD
MAN1B1	MTHFR	NDUFS7	PET100
MAN2B1	MTO1	NDUFS8	PET117
MANBA	MTPAP	NDUFV1	PEX1
MAOA	MTR	NDUFV2	PEX10
MARS	MTRR	NDUFV3	PEX11B
MARS2	MUT	NEU1	PEX12
MAT1A	MVK	NFE2L2	PEX13
MBD5	MYO5A	NFS1	PEX14
MCAT	NAA10	NFU1	PEX16
MCCC1	NADK2	NGLY1	PEX19
MCCC2	NAGA	NKX6-2	PEX2
MCEE	NAGLU	NOTCH2	PEX26
MCOLN1	NAGS	NPC1	PEX3
MECR	NALCN	NPC2	PEX5
MFF	NANS	NSDHL	PEX6
MFN2	NARS2	NSUN3	PEX7
MFSD8	NAXD	NT5C3A	PFKM
MGAT2	NAXE	NUBPL	PGAM2
MGME1	NBAS	NUS1	PGAP2
MICU1	NDST1	OAT	PGK1
MICU2	NDUFA1	OCRL	PGM1
MIEF2	NDUFA10	OGDH	PGM3
MIPEP	NDUFA11	OPA1	PHGDH
MLC1	NDUFA12	OPA3	PHKA1
MLPH	NDUFA13	OPLAH	PHKA2
MLYCD	NDUFA2	OTC	PHKB
MMAA	NDUFA4	OXA1L	PHKG2
MMAB	NDUFA6	OXCT1	PHYH
MMACHC	NDUFA8	PAH	PIGA
MMADHC	NDUFA9	PANK2	PIGM
MOCS1	NDUFAF1	PARK7	PIGN
MOCS2	NDUFAF2	PARS2	PIGO
MOCS3	NDUFAF3	PAX7	PIGQ
MOGS	NDUFAF4	PAXBP1	PIGT
MPC1	NDUFAF5	PC	PIGV
MPDU1	NDUFAF6	PCBD1	PIGW
MPI	NDUFAF8	PCCA	PIK3R4
MPV17	NDUFB1	PCCB	PITRM1
MRM2	NDUFB10	PCK1	PKLR
MRPL12	NDUFB11	PCLO	PLA2G6
MRPL44	NDUFB3	PCYT1A	PLAA
MRPL57	NDUFB6	PDE10A	PLIN5
MRPS16	NDUFB8	PDE12	PMM2
MRPS2	NDUFB9	PDHA1	PMPCA
MRPS22	NDUFC2	PDHB	PMPCB
MRPS23	NDUFS1	PDHX	PNP
MRPS34	NDUFS2	PDK3	PNPLA2
MRPS7	NDUFS3	PDP1	PNPLA4

**dbCMMS.v13**

PNPLA6	RPIA	SLC35A1	TARS2
PNPO	RRM2B	SLC35A2	TAT
PNPT1	RTN4IP1	SLC35C1	TAZ
POLG	RYR1	SLC37A4	TBCD
POLG2	SARS	SLC39A14	TBCK
POLR1A	SARS2	SLC39A4	TBL1X
POLR1C	SC5D	SLC39A8	TCN1
POLR3A	SCN8A	SLC3A1	TCN2
POLR3B	SCO1	SLC46A1	TDO2
POLRMT	SCO2	SLC4A10	TFAM
POMGNT1	SCP2	SLC52A1	TH
POMT1	SCYL1	SLC52A2	THAP11
PPA2	SDHA	SLC52A3	TIMM50
PPCS	SDHAF1	SLC5A1	TIMM8A
PPM1K	SDHAF2	SLC5A2	TIMMDC1
PPOX	SDHB	SLC6A17	TJP2
PPP1R15B	SDHC	SLC6A19	TK2
PPP2R1A	SDHD	SLC6A3	TKT
PPP2R5D	SELENOI	SLC6A8	TMEM106B
PPT1	SERAC1	SLC6A9	TMEM126A
PRKAG2	SFXN4	SLC7A1	TMEM126B
PRMT7	SGSH	SLC7A2	TMEM165
PRODH	SLC13A3	SLC7A7	TMEM199
PRODH2	SLC16A1	SLC7A9	TMEM240
PRPS1	SLC16A2	SLC9A1	TMEM260
PRUNE1	SLC17A5	SMPD1	TMEM70
PSAP	SLC19A2	SMS	TMLHE
PSPH	SLC19A3	SNORD118	TMTC3
PTCD1	SLC1A1	SNX14	TNNI3
PTS	SLC1A4	SPG7	TPK1
PURA	SLC22A5	SPR	TPP1
PUS1	SLC25A1	SPTBN4	TRAPPC11
PYCR1	SLC25A10	SQSTM1	TRAPPC12
PYCR2	SLC25A12	SRD5A3	TRIAP1
PYGL	SLC25A13	SSR4	TRIM37
PYGM	SLC25A15	ST3GAL5	TRIT1
PYROXD1	SLC25A19	STAT2	TRMT10C
QARS	SLC25A20	STX3	TRMT5
QDPR	SLC25A22	STXBP5L	TRMU
QRSL1	SLC25A24	SUCLA2	TRNT1
RAB11B	SLC25A26	SUCLG1	TRPM6
RAB27A	SLC25A3	SUGCT	TSMF
RAB3GAP1	SLC25A32	SUMF1	TTC19
RANBP2	SLC25A4	SUOX	TTPA
RARS	SLC25A42	SURF1	TUBB2A
RARS2	SLC25A46	SYT1	TUBB3
REPS1	SLC27A4	SZT2	TUBB4A
RFT1	SLC2A1	TACO1	TUBB4B
RMND1	SLC2A2	TAF1	TUFM
RNASEH1	SLC30A10	TALDO1	TWNK
RNF213	SLC30A9	TANGO2	TXN2

## dbCMMS.v13

TYMP  
UBQLN2  
UBTF  
UFM1  
UGT1A1  
UMPS  
UNC80  
UPB1  
UQCC2  
UQCC3  
UQCRB  
UQCRC2  
UQCRFS1  
UQCRH  
UQCRQ  
UROD  
UROS  
VAC14  
VARS  
VARS2  
WARS2  
WDR45  
WDR45B  
WFS1  
VMA21  
VPS11  
VPS13D  
VPS33A  
VPS53  
WVOX  
XDH  
XPR1  
YARS  
YARS2  
ZBTB42  
ZNHIT3

## dbCMMS.v13

Gener för epileptisk encefalopati (308st), 102 ingår i listan för medfödda metabola sjukdomar

AARS	CLN5	GABRA3	KDM5C
ABAT	CLN6	GABRA5	KIAA2022
ADAM22	CLN8	GABRB1	KIF1A
ADAR	CLTC	GABRB2	KIF5A
ADCY5	CNKSR2	GABRB3	KIF5C
ADRA2B	CNNM2	GABRG2	KMT2A
ADSL	CNPY3	GAL	LGI1
AFG3L2	CNTNAP2	GAMT	LIAS
AGTR2	COL4A1	GATM	LMNB2
AKT3	CPA6	GCSH	LNPB
ALDH5A1	CPLX1	GLDC	MAGEL2
ALDH7A1	CRADD	GNAO1	MAGI2
ALG1	CSNK2B	GNB1	MAPK10
ALG13	CSTB	GNB5	MBD5
ALG14	CTSD	GOSR2	MBOAT7
ALG3	CTSF	GPAA1	MDH2
ALG6	CUX2	GPHN	MECP2
AMT	CYFIP2	GRIA3	MED13L
ANKRD11	DCX	GRIA4	MED17
AP3B2	DDX3X	GRIN1	MED23
AP4S1	DEAF1	GRIN2A	MEF2C
APP	DENND5A	GRIN2B	MFF
ARHGEF9	DEPDC5	GRIN2D	MFSD8
ARID1B	DHDDS	GRN	MID2
ARV1	DNAJC5	GUF1	MOCS1
ARX	DNM1	HCN1	MOCS2
ASAH1	DNM1L	HECW2	MTOR
ASNS	DNMT3A	HIVEP2	NAA10
ATP13A2	DOCK7	HNRNPH2	NACC1
ATP1A2	DOLK	HNRNPU	NAPB
ATP1A3	DPM1	HSD17B10	NECAP1
ATP6AP2	DPM2	IQSEC2	NF1
ATP6V1A	DPYD	IRF2BPL	NGLY1
ATP7A	DYNC1H1	ITPA	NHLRC1
BRAT1	DYRK1A	KCNA1	NPRL2
BRWD3	EEF1A2	KCNA2	NPRL3
BSCL2	EHMT1	KCNB1	NR2F1
CACNA1A	EIF2S3	KCNC1	NRXN1
CACNA1D	EPM2A	KCNH1	NTRK2
CACNA2D1	FGF12	KCNH5	NUS1
CAD	FGF13	KCNJ10	OPHN1
CASK	FLNA	KCNMA1	OTUD6B
CDKL5	FOLR1	KCNQ1	PAFAH1B1
CERS1	FOXG1	KCNQ2	PAK3
CHD2	FOXP1	KCNQ3	PAX7
CHRNA2	FRRS1L	KCNQ5	PCDH12
CHRNA4	GABBR2	KCNT1	PCDH19
CHRN2	GABRA1	KCNT2	PIGA
CLN3	GABRA2	KCTD7	PIGC

**dbCMMS.v13**

PIGG	SLC12A5	UBE3A
PIGN	SLC13A5	UNC80
PIGO	SLC1A2	WASF1
PIGP	SLC1A4	WDR45
PIGQ	SLC25A12	WDR45B
PIGT	SLC25A22	WWOX
PIGW	SLC2A1	YWHAG
PLCB1	SLC35A2	ZBTB18
PLPBP	SLC35A3	ZEB2
PNKP	SLC39A8	ZNHIT3
PNPO	SLC45A1	
POLG	SLC6A1	
PPP2R1A	SLC6A8	
PPP3CA	SLC9A6	
PPT1	SMARCA2	
PRDM8	SMC1A	
PRICKLE1	SNAP25	
PRICKLE2	SPATA5	
PRRT2	SPTAN1	
PRUNE1	SRPX2	
PTPN23	ST3GAL3	
PURA	ST3GAL5	
QARS	STAMBP	
RAB11A	STRADA	
RAB11B	STX1B	
RAB39B	STXBP1	
RARS2	STYXL1	
RBSN	SUMF1	
RELN	SV2A	
RFT1	SYN1	
RHOBTB2	SYNGAP1	
RNASEH2A	SYNJ1	
RNASEH2B	SYP	
RNASEH2C	SZT2	
ROGDI	TBC1D24	
RORB	TBCD	
RUSC2	TCF4	
SAMD12	TDP2	
SAMHD1	TMTTC3	
SCARB2	TPP1	
SCN1A	TRAK1	
SCN1B	TRAPPC12	
SCN2A	TRAPPC6B	
SCN3A	TREX1	
SCN8A	TRIM8	
SCN9A	TRPM6	
SEPSECS	TSC1	
SERPINI1	TSC2	
SETD1B	TUBA1A	
SHANK3	TUBB2A	
SIK1	UBA5	

## dbCMMS.v13

Gener för mitokondriella sjukdomar (241st), alla ingår i listan för medfödda metabola sjukdomar

AARS2	COX8A	MPV17	NDUFS7
ACAD9	CRAT	MRM2	NDUFS8
ADCK2	DARS2	MRPL12	NDUFV1
AFG3L2	DCAF17	MRPL44	NDUFV2
AGK	DGUOK	MRPL57	NDUFV3
AIFM1	DNA2	MRPS16	NFS1
ALKBH1	DNAJC19	MRPS2	NFU1
ANTXR2	DNM1L	MRPS22	NSUN3
APOPT1	EARS2	MRPS23	NUBPL
ATAD3A	ECHS1	MRPS34	OPA1
ATAD3B	ECSIT	MRPS7	OPA3
ATAD3C	ELAC2	MSTO1	OXA1L
ATP5E	EPG5	MTO1	PARK7
ATP5F1D	ERAL1	MTPAP	PARS2
ATP5MD	ETHE1	NADK2	PDE12
ATP6AP1	FARS2	NARS2	PDK3
ATPAF2	FASTKD2	NAXD	PDSS1
BCS1L	FDX2	NAXE	PDSS2
BOLA3	FDXR	NDUFA1	PET100
C12ORF65	FOXRED1	NDUFA10	PET117
C19ORF70	FTL	NDUFA11	PITRM1
C1QBP	FXN	NDUFA12	PMPCB
CA5A	GARS	NDUFA13	PNPLA4
CARS2	GATB	NDUFA2	PNPT1
CEP89	GATC	NDUFA4	POLG
CHCHD10	GFER	NDUFA6	POLG2
CISD2	GFM1	NDUFA8	POLRMT
CLPB	GTPBP3	NDUFA9	PPA2
CLPP	HSPD1	NDUFAF1	PTCD1
CMPK2	IARS2	NDUFAF2	PUS1
COA5	IBA57	NDUFAF3	QRSL1
COA6	ISCA1	NDUFAF4	RARS2
COA7	ISCA2	NDUFAF5	REPS1
COASY	ISCU	NDUFAF6	RMND1
COQ2	KARS	NDUFAF8	RNASEH1
COQ4	KIF5A	NDUFB1	RRM2B
COQ5	LARS2	NDUFB10	RTN4IP1
COQ6	LRPPRC	NDUFB11	SARS2
COQ7	LYRM7	NDUFB3	SCO1
COQ8A	MCAT	NDUFB6	SCO2
COQ9	MECR	NDUFB8	SDHA
COX10	MFF	NDUFB9	SDHAF1
COX14	MFN2	NDUFC2	SDHAF2
COX15	MGME1	NDUFS1	SDHB
COX20	MICU1	NDUFS2	SDHC
COX4I1	MICU2	NDUFS3	SDHD
COX4I2	MIEF2	NDUFS4	SFXN4
COX5A	MIPEP	NDUFS5	SLC25A1
COX6B1	MPC1	NDUFS6	SLC25A10



## dbCMMS.v13

SLC25A19  
SLC25A24  
SLC25A3  
SLC25A32  
SLC25A4  
SLC25A42  
SLC25A46  
SPG7  
SUCLA2  
SUCLG1  
SURF1  
TACO1  
TARS2  
TAZ  
TFAM  
TIMM50  
TIMM8A  
TIMMDC1  
TK2  
TMEM126A  
TMEM126B  
TMEM70  
TNNI3  
TRIAP1  
TRIT1  
TRMT10C  
TRMT5  
TRMU  
TSFM  
TTC19  
TUFM  
TWNK  
TYMP  
UQCC2  
UQCC3  
UQCRB  
UQCRC2  
UQCRFS1  
UQCRH  
UQCRQ  
VARS2  
WARS2  
VMA21  
VPS13D  
YARS2

## dbCMMS.v13

Gener för leversjukdomar hos barn (89st), 48 ingår i listan för medfödda metabola sjukdomar

ABCB11	MYO5B
ABCB4	NEK8
ABCC2	NOTCH2
ABCD3	NPC1
ACOX2	NPC2
AGL	NPHP1
AKR1D1	NPHP3
ALAS2	NPHP4
ALDOB	NR1H4
ALG3	PHKA2
ALG8	PHKB
AMACR	PHKG2
ANKS6	PKD1
ARG1	PKD2
ASAH1	PKHD1
ASS1	POLG
ATP6AP1	PYGL
ATP7B	RPGRIP1L
ATP8B1	SDCCAG8
BAAT	SERPINA1
BCS1L	SLC25A13
CCDC115	SLC27A5
CEP164	SLC2A2
CEP290	SLC37A4
CLDN1	SLCO1B1
CYP27A1	SLCO1B3
CYP7B1	TALDO1
DCDC2	TJP2
DGUOK	TMEM199
EIF2AK3	TMEM67
FAH	TRMU
FBP1	TTC21B
FECH	TWNK
G6PC	UGT1A1
GALE	UNC45
GALT	WDR19
GATA6	VIPAS39
GBE1	VPS33B
GLIS2	XPNPEP3
GPD1	ZNF423
GYS2	
HSD17B4	
HSD3B7	
INVS	
IQCB1	
JAG1	
LIPA	
MPI	
MPV17	