

## dbCMMS.v14

Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild (870st)

AARS2	ALG11	B4GALT7	COG5
AASS	ALG12	BAAT	COG6
ABAT	ALG13	BCAT2	COG7
ABCC8	ALG14	BCKDHA	COG8
ABCD1	ALG2	BCKDHB	COLGALT1
ABCD4	ALG3	BCKDK	COQ2
ABHD12	ALG6	BCS1L	COQ4
ABHD5	ALG8	BLOC1S3	COQ5
ACAD8	ALG9	BOLA3	COQ6
ACAD9	ALKBH1	BSCL2	COQ7
ACADM	ALPL	BTD	COQ8A
ACADS	AMACR	C12ORF65	COQ9
ACADSB	AMN	C19ORF12	COX10
ACADVL	AMPD1	C1QBP	COX14
ACAT1	AMPD2	CA5A	COX15
ACER3	AMT	CACNA1A	COX20
ACO2	ANTXR2	CACNA1E	COX4I1
ACOX1	AP1S1	CACNA1S	COX4I2
ACSF3	AP3B1	CAD	COX5A
ACY1	AP3D1	CARS	COX6B1
ADA	AP5Z1	CARS2	COX8A
ADAR	APOPT1	CAT	CP
ADCK2	APRT	CBS	CPOX
ADCY5	ARG1	CCDC115	CPS1
ADK	ARL3	CCDC174	CPT1A
ADPRHL2	ARSA	CCDC88C	CPT2
ADSL	ARSB	CD320	CRAT
ADSSL1	ASAH1	CEP89	CTH
AFG3L2	ASL	CERS1	CTNS
AGA	ASNS	CHCHD10	CTSA
AGK	ASPA	CHKB	CTSD
AGL	ASS1	CISD2	CTSF
AGPS	ATAD3A	CLDN16	CTSK
AGXT	ATAD3B	CLN3	CUBN
AHCY	ATAD3C	CLN5	CWF19L1
AIFM1	ATIC	CLN6	CYP27A1
AIMP1	ATP13A2	CLN8	CYP7B1
AKR1D1	ATP1A1	CLPB	D2HGDH
ALAD	ATP5F1D	CLPP	DARS
ALAS2	ATP5F1E	CLTC	DARS2
ALDH18A1	ATP5MD	CMPK2	DBH
ALDH3A2	ATP6AP1	CNKSR2	DBT
ALDH4A1	ATP6V0A2	COA5	DCAF17
ALDH5A1	ATP7A	COA6	DDC
ALDH6A1	ATP7B	COA7	DEGS1
ALDH7A1	ATPAF2	COASY	DGAT1
ALDOA	AUH	COG1	DGUOK
ALDOB	B4GALNT1	COG2	DHCR24
ALG1	B4GALT1	COG4	DHCR7

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DHDDS	FAH	GIF	HMGCL
DHFR	FAM126A	GJC2	HMGCS2
DHODH	FAM149B1	GK	HOGA1
DHPS	FAR1	GLA	HPD
DIAPH1	FARS2	GLB1	HPRT1
DLAT	FARSB	GLDC	HPS1
DLD	FASTKD2	GLRA1	HPS3
DMGDH	FBP1	GLRX5	HPS4
DNA2	FBXL4	GLS	HPS5
DNAJC12	FDFT1	GLUD1	HPS6
DNAJC19	FDX2	GLUL	HSD17B10
DNAJC3	FDXR	GLYCTK	HSD17B4
DNAJC5	FECH	GM2A	HSD3B7
DNM1L	FH	GMPPA	HSPD1
DOLK	FIG4	GMPPB	HTRA2
DPAGT1	FLAD1	GNAO1	HYAL1
DPM1	FMO3	GNE	HYOU1
DPM2	FOLR1	GNMT	IARS
DPM3	FOX11	GNPAT	IARS2
DPYD	FOXRED1	GNPTAB	IBA57
DPYS	FTCD	GNPTG	IDH2
DTNBP1	FTL	GNS	IDH3A
EARS2	FUCA1	GOT2	IDS
EBP	FUT8	GPD1	IDUA
ECHS1	FXN	GPHN	IER3IP1
ECM1	G6PC	GPR161	IFIH1
ECSIT	G6PD	GRHPR	IGSF1
EIF2B1	GAA	GRN	INSR
EIF2B2	GABBR2	GSS	IRF2BPL
EIF2B3	GABRG2	GTPBP3	ISCA1
EIF2B4	GALC	GUSB	ISCA2
EIF2B5	GALE	GYG1	ISCU
ELAC2	GALK1	GYG2	ISPD
ELOVL4	GALNS	GYS1	IVD
ELOVL5	GALNT3	GYS2	JAG1
ENO3	GALT	HAAO	KARS
EPG5	GAMT	HACE1	KCNA4
EPHX1	GARS	HADH	KCNJ11
EPM2A	GATB	HADHA	KIF5A
EPRS	GATC	HADHB	KYNU
ERAL1	GATM	HARS	L2HGDH
ERCC6	GBA	HCFC1	LAMA1
ETFA	GBE1	HEPACAM	LAMP2
ETFB	GCDH	HEXA	LARS
ETFDH	GCH1	HEXB	LARS2
ETHE1	GCK	HGD	LBR
EXOSC3	GCLC	HGSNAT	LDHA
EXOSC8	GCSH	HIBCH	LHX4
EXT1	GFER	HIKESHI	LIAS
EXT2	GFM1	HLCS	LIPA
FA2H	GGT1	HMBS	LIPE

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LIPT1	MPI	NDUFAF4	PAX7
LIPT2	MPV17	NDUFAF5	PAXBP1
LMBRD1	MRM2	NDUFAF6	PC
LNPK	MRPL12	NDUFAF8	PCBD1
LONP1	MRPL44	NDUFB1	PCCA
LPIN1	MRPL57	NDUFB10	PCCB
LRP2	MRPS14	NDUFB11	PCK1
LRPPRC	MRPS16	NDUFB3	PCLO
LYRM4	MRPS2	NDUFB6	PCYT1A
LYRM7	MRPS22	NDUFB8	PDE10A
MAG	MRPS23	NDUFB9	PDE12
MAGED2	MRPS34	NDUFC2	PDHA1
MAGEL2	MRPS7	NDUFS1	PDHB
MAGT1	MSL3	NDUFS2	PDHX
MAN1B1	MSTO1	NDUFS3	PDK3
MAN2B1	MTFMT	NDUFS4	PDP1
MANBA	MTHFD1	NDUFS5	PDSS1
MAOA	MTHFR	NDUFS6	PDSS2
MARS	MTO1	NDUFS7	PEPD
MARS2	MTPAP	NDUFS8	PET100
MAT1A	MTR	NDUFV1	PET117
MBD5	MTRR	NDUFV2	PEX1
MCAT	MUT	NDUFV3	PEX10
MCCC1	MVK	NEU1	PEX11B
MCCC2	MYO5A	NFE2L2	PEX12
MCEE	MYORG	NFS1	PEX13
MCOLN1	NAA10	NFU1	PEX14
MECR	NADK2	NGLY1	PEX16
MFF	NAGA	NKX6-2	PEX19
MFN2	NAGLU	NOTCH2	PEX2
MFSD8	NAGS	NPC1	PEX26
MGAT2	NALCN	NPC2	PEX3
MGME1	NANS	NSDHL	PEX5
MICOS13	NARS2	NSUN3	PEX6
MICU1	NAXD	NT5C3A	PEX7
MICU2	NAXE	NUBPL	PFKM
MIEF2	NBAS	NUS1	PGAM2
MIPEP	NDST1	OAT	PGAP2
MLC1	NDUFA1	OCRL	PGK1
MLPH	NDUFA10	OGDH	PGM1
MLYCD	NDUFA11	OPA1	PGM3
MMAA	NDUFA12	OPA3	PHGDH
MMAB	NDUFA13	OPLAH	PHKA1
MMACHC	NDUFA2	OTC	PHKA2
MMADHC	NDUFA4	OXA1L	PHKB
MOCS1	NDUFA6	OXCT1	PHKG2
MOCS2	NDUFA8	PAH	PHYH
MOCS3	NDUFA9	PAK1	PIGA
MOGS	NDUFAF1	PANK2	PIGM
MPC1	NDUFAF2	PARK7	PIGN
MPDU1	NDUFAF3	PARS2	PIGO

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PIGQ	PYCR1	SLC22A5	SNX14
PIGS	PYCR2	SLC25A1	SPG7
PIGT	PYGL	SLC25A10	SPR
PIGV	PYGM	SLC25A12	SPTBN4
PIGW	PYROXD1	SLC25A13	SQSTM1
PIK3R4	QARS	SLC25A15	SRD5A3
PITRM1	QDPR	SLC25A19	SSR4
PKLR	QRSL1	SLC25A20	ST3GAL5
PLA2G6	RAB11B	SLC25A22	STAT2
PLAA	RAB27A	SLC25A24	STX3
PLIN5	RAB3GAP1	SLC25A26	STXBP5L
PMM2	RANBP2	SLC25A3	SUCLA2
PMPCA	RARS	SLC25A32	SUCLG1
PMPCB	RARS2	SLC25A4	SUGCT
PNP	REPS1	SLC25A42	SUMF1
PNPLA2	RFT1	SLC25A46	SUOX
PNPLA4	RMND1	SLC27A4	SURF1
PNPLA6	RNASEH1	SLC2A1	SYT1
PNPO	RNF13	SLC2A2	SZT2
PNPT1	RNF213	SLC30A10	TACO1
POLG	RPIA	SLC30A9	TAF1
POLG2	RRM2B	SLC35A1	TALDO1
POLR1A	RTN4IP1	SLC35A2	TANGO2
POLR1C	RYR1	SLC35C1	TARS2
POLR3A	SARS	SLC37A4	TAT
POLR3B	SARS2	SLC39A14	TAZ
POLRMT	SC5D	SLC39A4	TBCD
POMGNT1	SCN8A	SLC39A8	TBCK
POMT1	SCO1	SLC3A1	TBL1X
PPA2	SCO2	SLC46A1	TCN1
PPCS	SCP2	SLC4A10	TCN2
PPM1K	SCYL1	SLC52A1	TDO2
PPOX	SDHA	SLC52A2	TFAM
PPP1R15B	SDHAF1	SLC52A3	TH
PPP1R21	SDHAF2	SLC5A1	THAP11
PPP2R1A	SDHB	SLC5A2	TIMM50
PPP2R5D	SDHC	SLC6A17	TIMM8A
PPT1	SDHD	SLC6A19	TIMMDC1
PRKAG2	SELENOI	SLC6A3	TJP2
PRMT7	SERAC1	SLC6A8	TK2
PRODH	SFXN4	SLC6A9	TKT
PRODH2	SGSH	SLC7A1	TMEM106B
PRPS1	SLC13A3	SLC7A2	TMEM126A
PRUNE1	SLC16A1	SLC7A7	TMEM126B
PSAP	SLC16A2	SLC7A9	TMEM165
PSPH	SLC17A5	SLC9A1	TMEM199
PTCD1	SLC19A2	SLC9A7	TMEM240
PTS	SLC19A3	SMARCD1	TMEM260
PURA	SLC1A1	SMPD1	TMEM70
PUS1	SLC1A4	SMS	TMLHE
PUS7	SLC20A2	SNORD118	TMTC3

## dbCMMS.v14

TNNI3	VPS11
TPK1	VPS13D
TPP1	VPS33A
TRAPPC11	VPS53
TRAPPC12	WVOX
TRAPPC2L	XDH
TRIAP1	XPR1
TRIM37	YARS
TRIT1	YARS2
TRMT1	ZBTB42
TRMT10C	ZNHIT3
TRMT5	
TRMU	
TRNT1	
TRPM6	
TSFM	
TTC19	
TTPA	
TUBB2A	
TUBB3	
TUBB4A	
TUBB4B	
TUFM	
TWNK	
TXN2	
TYMP	
UBQLN2	
UBTF	
UFM1	
UGT1A1	
UMPS	
UNC80	
UPB1	
UQCC2	
UQCC3	
UQCRB	
UQCRC2	
UQCRFS1	
UQCRH	
UQCRQ	
UROD	
UROS	
VAC14	
VAMP2	
VARS	
VARS2	
WARS2	
WDR45	
WDR45B	
WFS1	
VMA21	

## dbCMMS.v14

Gener för epileptisk encefalopati (353st), 110 ingår i listan för medfödda metabola sjukdomar

AARS	CAMK2A	EEF1A2	IQSEC2
ABAT	CAMK2B	EHMT1	IRF2BPL
ACTL6B	CASK	EIF2S3	ITPA
ADAM22	CDKL5	EPM2A	KANSL1
ADAR	CERS1	FGF12	KCNA1
ADCY5	CERT1	FGF13	KCNA2
ADRA2B	CHD2	FGFR3	KCNB1
ADSL	CHRNA2	FLNA	KCNC1
AFG3L2	CHRNA4	FOLR1	KCND2
AGTR2	CHRNA7	FOXG1	KCNH1
AIMP1	CHRN2	FOXP1	KCNH5
AKT3	CLCN4	FRRS1L	KCNJ10
ALDH5A1	CLN3	GABBR2	KCNMA1
ALDH7A1	CLN5	GABRA1	KCNQ1
ALG1	CLN6	GABRA2	KCNQ2
ALG11	CLN8	GABRA3	KCNQ3
ALG13	CLTC	GABRA5	KCNQ5
ALG14	CNKSR2	GABRB1	KCNT1
ALG3	CNNM2	GABRB2	KCNT2
ALG6	CNPY3	GABRB3	KCTD7
AMT	CNTN2	GABRG2	KDM5C
ANKRD11	CNTNAP2	GAL	KIAA2022
AP2M1	COL4A1	GAMT	KIF1A
AP3B2	CPA6	GATM	KIF5A
AP4S1	CPLX1	GCSH	KIF5C
APP	CRADD	GLDC	KMT2A
ARHGEF9	CSNK2B	GLRA1	LGI1
ARID1B	CSTB	GLS	LIAS
ARV1	CTSD	GNAO1	LMNB2
ARX	CTSF	GNB1	LNPX
ASAH1	CUX2	GNB5	MAGEL2
ASNS	CYFIP2	GOSR2	MAGI2
ASXL3	DCX	GPAA1	MAPK10
ATP13A2	DDX3X	GPHN	MBD5
ATP1A2	DEAF1	GRIA3	MBOAT7
ATP1A3	DENND5A	GRIA4	MDH2
ATP6AP2	DEPDC5	GRIN1	MECP2
ATP6V1A	DHDDS	GRIN2A	MED13L
ATP7A	DNAJC5	GRIN2B	MED17
ATXN2	DNM1	GRIN2D	MED23
BRAT1	DNM1L	GRN	MEF2C
BRWD3	DNMT3A	GUF1	MFF
BSCL2	DOCK7	HACE1	MFSD8
CACNA1A	DOLK	HCN1	MID2
CACNA1D	DPM1	HECW2	MOCS1
CACNA1E	DPM2	HIVEP2	MOCS2
CACNA2D1	DPYD	HNRNPH2	MTHFS
CACNA2D2	DYNC1H1	HNRNPU	MTOR
CAD	DYRK1A	HSD17B10	NAA10

**dbCMMS.v14**

NACC1	PTPN23	SNAP25	ZBTB18
NAPB	PURA	SORCS3	ZEB2
NECAP1	QARS	SPATA5	ZNHIT3
NEDD4	RAB11A	SPTAN1	
NEUROD2	RAB11B	SRPX2	
NF1	RAB39B	ST3GAL3	
NGLY1	RAPGEF2	ST3GAL5	
NHLRC1	RARS2	STAMBP	
NPRL2	RBSN	STRADA	
NPRL3	RELN	STT3B	
NR2F1	RFT1	STX1B	
NRXN1	RHOBTB2	STXBP1	
NTRK2	RNASEH2A	STYXL1	
NUS1	RNASEH2B	SUMF1	
OPHN1	RNASEH2C	SV2A	
OTUD6B	RNF13	SYN1	
PACS2	ROGDI	SYNGAP1	
PAFAH1B1	RORA	SYNJ1	
PAK1	RORB	SYP	
PAK3	RUSC2	SZT2	
PAX7	SAMD12	TBC1D24	
PCDH12	SAMHD1	TBCD	
PCDH19	SCARB2	TBL1XR1	
PHACTR1	SCN1A	TCF4	
PIGA	SCN1B	TDP2	
PIGC	SCN2A	TMTC3	
PIGG	SCN3A	TNRC6A	
PIGH	SCN8A	TPP1	
PIGL	SCN9A	TRAK1	
PIGN	SEPSECS	TRAPPC12	
PIGO	SERPINI1	TRAPPC6B	
PIGP	SETBP1	TREX1	
PIGQ	SETD1B	TRIM8	
PIGS	SGCE	TRPM6	
PIGT	SHANK3	TSC1	
PIGW	SIK1	TSC2	
PLCB1	SLC12A5	TUBA1A	
PLP1	SLC13A5	TUBB2A	
PLPBP	SLC1A2	TUBB2B	
PNKP	SLC1A4	UBA5	
PNPO	SLC25A12	UBE3A	
POLG	SLC25A22	UFC1	
PPP1CB	SLC2A1	UNC80	
PPP2R1A	SLC35A2	VARS	
PPP3CA	SLC35A3	WASF1	
PPT1	SLC39A8	WDR26	
PRDM8	SLC45A1	WDR45	
PRICKLE1	SLC6A1	WDR45B	
PRICKLE2	SLC6A8	WDR62	
PRRT2	SLC9A6	WWOX	
PRUNE1	SMARCA2	YWHAG	

## dbCMMS.v14

Gener för mitokondriella sjukdomar (243st), alla ingår i listan för medfödda metabola sjukdomar

AARS2	COX8A	MPC1	NDUFS6
ACAD9	CRAT	MPV17	NDUFS7
ADCK2	DARS2	MRM2	NDUFS8
AFG3L2	DCAF17	MRPL12	NDUFV1
AGK	DGUOK	MRPL44	NDUFV2
AIFM1	DNA2	MRPL57	NDUFV3
ALKBH1	DNAJC19	MRPS14	NFS1
ANTXR2	DNM1L	MRPS16	NFU1
APOPT1	EARS2	MRPS2	NSUN3
ATAD3A	ECHS1	MRPS22	NUBPL
ATAD3B	ECSIT	MRPS23	OPA1
ATAD3C	ELAC2	MRPS34	OPA3
ATP5E	EPG5	MRPS7	OXA1L
ATP5F1D	ERAL1	MSTO1	PARK7
ATP5MD	ETHE1	MTO1	PARS2
ATP6AP1	FARS2	MTPAP	PDE12
ATPAF2	FASTKD2	NADK2	PDK3
BCS1L	FDX2	NARS2	PDSS1
BOLA3	FDXR	NAXD	PDSS2
C12ORF65	FOXRED1	NAXE	PET100
C19ORF70	FTL	NDUFA1	PET117
C1QBP	FXN	NDUFA10	PITRM1
CA5A	GARS	NDUFA11	PMPCB
CARS2	GATB	NDUFA12	PNPLA4
CEP89	GATC	NDUFA13	PNPT1
CHCHD10	GFER	NDUFA2	POLG
CISD2	GFM1	NDUFA4	POLG2
CLPB	GTPBP3	NDUFA6	POLRMT
CLPP	HSPD1	NDUFA8	PPA2
CMPK2	IARS2	NDUFA9	PTCD1
COA5	IBA57	NDUF AF1	PUS1
COA6	ISCA1	NDUF AF2	QRSL1
COA7	ISCA2	NDUF AF3	RARS2
COASY	ISCU	NDUF AF4	REPS1
COQ2	KARS	NDUF AF5	RMND1
COQ4	KIF5A	NDUF AF6	RNASEH1
COQ5	LARS2	NDUF AF8	RRM2B
COQ6	LONP1	NDUFB1	RTN4IP1
COQ7	LRPPRC	NDUFB10	SARS2
COQ8A	LYRM7	NDUFB11	SCO1
COQ9	MCAT	NDUFB3	SCO2
COX10	MECR	NDUFB6	SDHA
COX14	MFF	NDUFB8	SDHAF1
COX15	MFN2	NDUFB9	SDHAF2
COX20	MGME1	NDUFC2	SDHB
COX4I1	MICU1	NDUFS1	SDHC
COX4I2	MICU2	NDUFS2	SDHD
COX5A	MIEF2	NDUFS3	SFXN4
COX6B1	MIPEP	NDUFS4	SLC25A1



## dbCMMS.v14

SLC25A10  
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.v14

Gener för leversjukdomar hos barn (124st), 52 ingår i listan för medfödda metabola sjukdomar

ABCA1	GBE1	SLC37A4
ABCB11	GDF1	SLCO1B1
ABCB4	GPD1	SLCO1B3
ABCC2	GUCY2D	SMPD1
ABCD3	GYS2	TALDO1
ACOX2	HAMP	TCTN2
AGL	HFE	TFR2
AKR1D1	HJV	TJP2
ALAS2	HSD17B4	TMEM107
ALDOB	HSD3B7	TMEM199
ALG3	IFT140	TMEM216
ALG8	IFT172	TMEM67
ALMS1	IFT43	TRAF3IP1
AMACR	IFT52	TRMU
ANKS6	IFT81	TTC21B
ARG1	INPP5E	TTC37
ASAH1	INTU	TWNK
ASS1	JAG1	UGT1A1
ATP6AP1	KRT18	UNC45
ATP7B	KRT8	WDR19
ATP8B1	LIPA	WDR34
BAAT	LRP5	WDR35
BBS1	MPI	WDR60
BCS1L	MPV17	VIPAS39
CC2D2A	MYO5B	VPS33B
CCDC115	NEK1	XPNPEP3
CEP164	NOTCH2	
CEP290	NPC1	
CEP41	NPC2	
CEP83	NPHP3	
CLDN1	NR1H4	
CSPP1	OFD1	
CYP27A1	PHKA2	
CYP7A1	PHKB	
CYP7B1	PHKG2	
DCDC2	PKD1	
DGUOK	PKD2	
DNAJB11	PKHD1	
DYNC2H1	POLG	
DYNC2LI1	PRKCSH	
EIF2AK3	PYGL	
FAH	RPGRIP1L	
FBP1	SC5D	
FECH	SEC63	
G6PC	SERPINA1	
GALE	SKIV2L	
GALT	SLC25A13	
GANAB	SLC27A5	
GATA6	SLC2A2	