

## dbCMMS.v10.1

Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild (732st)

AARS2	AMPD2	CLPP	DNA2
AASS	ANTXR2	COA5	DNAJC19
ABCC8	AP1S1	COA6	DNAJC3
ABCD1	AP3B1	COASY	DNM1L
ABCD4	AP3D1	COG1	DPAGT1
ABHD12	AP5Z1	COG2	DPM3
ABHD5	APOPT1	COG4	DPYS
ACAD8	APRT	COG5	DTNBP1
ACAD9	ARG1	COG6	DUOX2
ACADM	ARSA	COG7	DUOXA2
ACADS	ARSB	COG8	EARS2
ACADSB	ASL	COQ2	EBP
ACADVL	ASPA	COQ4	ECHS1
ACAT1	ASS1	COQ7	ECSIT
ACER3	ATAD3A	COQ8A	EIF2B1
ACO2	ATIC	COQ9	EIF2B2
ACOX1	ATP5E	COX10	EIF2B3
ACSF3	ATP6AP1	COX15	EIF2B4
ADA	ATP6V0A2	COX4I2	EIF2B5
ADCK2	ATP7B	COX6B1	ELAC2
ADK	ATPAF2	COX8A	ELOVL4
ADSSL1	AUH	CP	ELOVL5
AFG3L2	B4GALNT1	CPOX	ENO3
AGA	B4GALT1	CPS1	EPG5
AGK	B4GALT7	CPT1A	EPHX1
AGL	BAAT	CPT2	ERCC6
AGPS	BCAT2	CTH	ETFA
AGXT	BCKDHA	CTNS	ETFB
AHCY	BCKDHB	CTSA	ETFDH
AIFM1	BCKDK	CTSK	ETHE1
AIMP1	BCS1L	CUBN	EXOSC3
AKR1D1	BLOC1S3	CWF19L1	EXOSC8
ALAD	BOLA3	CYP27A1	EXT1
ALAS2	BTD	CYP7B1	EXT2
ALDH18A1	C12ORF65	D2HGDH	FA2H
ALDH3A2	C19ORF12	DARS2	FAH
ALDH4A1	CA5A	DBH	FAM126A
ALDH6A1	CARS2	DBT	FAR1
ALDOA	CAT	DCAF17	FARS2
ALDOB	CBS	DDC	FASTKD2
ALG11	CCDC115	DGUOK	FBP1
ALG12	CCDC174	DHCR24	FBXL4
ALG2	CCDC88C	DHCR7	FDX1L
ALG8	CD320	DHFR	FECH
ALG9	CEP89	DHODH	FH
ALPL	CHCHD10	DIAPH1	FIG4
AMACR	CHKB	DLAT	FLAD1
AMN	CLDN16	DLD	FMO3
AMPD1	CLPB	DMGDH	FOXE1

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FOXRED1	HADHA	LHX4	MTFMT
FTCD	HADHB	LIPA	MTHFD1
FTL	HARS	LIPE	MTHFR
FUCA1	HCFC1	LIPT1	MTO1
FXN	HEPACAM	LIPT2	MTPAP
G6PC	HEXA	LMBRD1	MTR
G6PD	HEXB	LPIN1	MTRR
GAA	HGD	LRP2	MUT
GALC	HGSNAT	LRPPRC	MVK
GALE	HIBCH	LYRM4	MYO5A
GALK1	HIKESHI	LYRM7	NAA10
GALNS	HLCS	MAG	NADK2
GALNT3	HMBS	MAGED2	NAGA
GALT	HMGCL	MAGT1	NAGLU
GARS	HMGCS2	MAN1B1	NAGS
GBA	HOGA1	MAN2B1	NALCN
GBE1	HPD	MANBA	NANS
GCDH	HPRT1	MAOA	NARS2
GCH1	HPS1	MARS2	NAXE
GCK	HPS3	MAT1A	NBAS
GCLC	HPS4	MCAT	NDST1
GFER	HPS5	MCCC1	NDUFA1
GFM1	HPS6	MCCC2	NDUFA10
GIF	HSD17B4	MCEE	NDUFA11
GJC2	HSD3B7	MCOLN1	NDUFA12
GK	HSPD1	MFF	NDUFA13
GLA	HTRA2	MFN2	NDUFA2
GLB1	HYAL1	MGAT2	NDUFA4
GLRA1	IARS	MGME1	NDUFA6
GLRX5	IARS2	MICU1	NDUFA8
GLS	IBA57	MIEF2	NDUFA9
GLUD1	IDH2	MLC1	NDUFAF1
GLUL	IDS	MLPH	NDUFAF2
GM2A	IDUA	MLYCD	NDUFAF3
GNE	INSR	MMAA	NDUFAF4
GNMT	ISCA2	MMAB	NDUFAF5
GNPAT	ISCU	MMACHC	NDUFAF6
GNPTAB	ISPD	MMADHC	NDUFB1
GNPTG	IVD	MOGS	NDUFB11
GNS	IYD	MPC1	NDUFB3
GPR161	JAG1	MPDU1	NDUFB6
GRHPR	KARS	MPI	NDUFB8
GSS	KCNA4	MPV17	NDUFB9
GTPBP3	KCNJ11	MRPL12	NDUFC2
GUSB	KIF5A	MRPL44	NDUFS1
GYG1	L2HGDH	MRPL57	NDUFS2
GYG2	LAMA1	MRPS16	NDUFS3
GYS1	LAMP2	MRPS2	NDUFS4
GYS2	LARS2	MRPS22	NDUFS5
HACE1	LBR	MRPS23	NDUFS6
HADH	LDHA	MRPS7	NDUFS7

**dbCMMS.v10.1**

NDUFS8	PEX14	PUS1	SLC26A4
NDUFV1	PEX16	PYCR1	SLC27A4
NDUFV2	PEX19	PYCR2	SLC2A2
NDUFV3	PEX2	PYGL	SLC30A10
NEU1	PEX26	PYGM	SLC35A1
NFS1	PEX3	QDPR	SLC35C1
NFU1	PEX5	QRSL1	SLC37A4
NKX2-1	PEX6	RAB27A	SLC39A14
NKX2-5	PEX7	RAB3GAP1	SLC39A4
NOTCH2	PFKM	RANBP2	SLC3A1
NPC1	PGAM2	RARS	SLC46A1
NPC2	PGAP2	RARS2	SLC4A10
NSDHL	PGK1	RMND1	SLC52A1
NT5C3A	PGM1	RNASEH1	SLC52A2
NUBPL	PGM3	RPIA	SLC52A3
NUS1	PHGDH	RRM2B	SLC5A1
OAT	PHKA1	RTN4IP1	SLC5A2
OCRL	PHKA2	RYR1	SLC5A5
OGDH	PHKB	SARS2	SLC6A17
OPA1	PHKG2	SC5D	SLC6A19
OPA3	PHYH	SCO1	SLC6A3
OPLAH	PIGM	SCO2	SLC7A1
OTC	PLA2G6	SCP2	SLC7A2
OXCT1	PMM2	SCYL1	SLC7A7
PAH	PMPCA	SDHA	SLC7A9
PANK2	PNP	SDHAF1	SLC9A1
PARK7	PNPLA2	SDHAF2	SMPD1
PARS2	PNPLA4	SDHB	SMS
PAX8	PNPLA6	SDHC	SNORD118
PC	PNPT1	SDHD	SNX14
PCBD1	POLG	SERAC1	SPG7
PCCA	POLG2	SGSH	SPR
PCCB	POLR1C	SLC16A1	SQSTM1
PCK1	POLR3A	SLC16A2	SRD5A3
PCLO	POLR3B	SLC17A5	SSR4
PCYT1A	POMGNT1	SLC19A2	STAT2
PDE10A	POMT1	SLC19A3	STX3
PDHA1	PPA2	SLC1A1	STXBP5L
PDHB	PPOX	SLC1A4	SUCLA2
PDHX	PPP1R15B	SLC22A5	SUCLG1
PDK3	PPP2R1A	SLC25A1	SUGCT
PDP1	PPP2R5D	SLC25A13	SUOX
PDSS1	PRKAG2	SLC25A15	SURF1
PDSS2	PRMT7	SLC25A19	TACO1
PEPD	PRODH	SLC25A20	TAF1
PET100	PRODH2	SLC25A26	TALDO1
PEX1	PRPS1	SLC25A3	TANGO2
PEX10	PSAP	SLC25A32	TARS2
PEX11B	PSPH	SLC25A4	TAT
PEX12	PTCD1	SLC25A42	TAZ
PEX13	PTS	SLC25A46	TBCD

## dbCMMS.v10.1

TBCK	VPS11
TCN1	VPS53
TCN2	XDH
TFAM	XPR1
TG	YARS2
TH	ZBTB42
TIMM8A	
TJP2	
TK2	
TKT	
TMEM126A	
TMEM126B	
TMEM165	
TMEM199	
TMEM240	
TMEM70	
TMLHE	
TNNI3	
TPK1	
TPO	
TRIM37	
TRIT1	
TRMT10C	
TRMT5	
TRMU	
TRNT1	
TRPM6	
TSFM	
TSHR	
TTC19	
TTPA	
TUBB4A	
TUFM	
TWNK	
TXN2	
TYMP	
UBQLN2	
UGT1A1	
UMPS	
UPB1	
UQCC2	
UQCC3	
UQCRB	
UQCRC2	
UQCRQ	
UROD	
UROS	
VAC14	
VARS2	
WFS1	
VMA21	

## dbCMMS.v10.1

Gener för epileptisk encefalopati (222st), 78 ingår i listan för medfödda metabola sjukdomar

AARS	CTSF	KCNA1
ABAT	DCX	KCNA2
ADAR	DEAF1	KCNB1
ADCY5	DENND5A	KCNC1
ADRA2B	DEPDC5	KCNH1
ADSL	DNAJC5	KCNJ10
AFG3L2	DNM1	KCNMA1
AGTR2	DNM1L	KCNQ2
ALDH5A1	DOCK7	KCNQ3
ALDH7A1	DOLK	KCNT1
ALG1	DPM1	KCTD7
ALG13	DPM2	KDM5C
ALG3	DPYD	KIAA2022
ALG6	DYRK1A	KIF1A
AMT	EEF1A2	KIF5A
AP3B2	EHMT1	KMT2A
AP4S1	EPM2A	LGI1
ARHGEF9	FGF12	LIAS
ARV1	FLNA	LMNB2
ARX	FOLR1	MAGEL2
ASAH1	FOXP1	MAGI2
ASNS	FRRS1L	MAPK10
ATP13A2	GABRA1	MBD5
ATP1A2	GABRB1	MBOAT7
ATP1A3	GABRB3	MECP2
ATP6AP2	GABRG2	MEF2C
ATP7A	GAL	MFF
BRAT1	GAMT	MFSD8
BRWD3	GATM	MOCS1
CACNA1A	GCSH	MOCS2
CACNA2D1	GLDC	NAPB
CAD	GNAO1	NECAP1
CASK	GNB1	NGLY1
CDKL5	GNB5	NHLRC1
CERS1	GOSR2	NPRL2
CHD2	GPHN	NPRL3
CHRNA2	GRIA3	NR2F1
CHRNA4	GRIN1	NRXN1
CHRN2	GRIN2A	PAFAH1B1
CLN3	GRIN2B	PAK3
CLN5	GRIN2D	PCDH19
CLN6	GRN	PIGA
CLN8	GUF1	PIGG
CNKS2	HCN1	PIGN
CNNM2	HECW2	PIGO
CNTNAP2	HNRNPU	PIGQ
CPA6	HSD17B10	PIGT
CSTB	IQSEC2	PIGW
CTSD	ITPA	PLCB1

## dbCMMS.v10.1

PNKP	STX1B
PNPO	STXBP1
POLG	STYXL1
PPT1	SUMF1
PRDM8	SV2A
PRICKLE1	SYN1
PRICKLE2	SYNGAP1
PRRT2	SYNJ1
PRUNE1	SYP
PURA	SZT2
QARS	TBC1D24
RAB39B	TCF4
RARS2	TDP2
RBSN	TPP1
RELN	TREX1
RFT1	TRPM6
RNASEH2A	TSC1
RNASEH2B	TSC2
RNASEH2C	TUBA1A
ROGDI	UBA5
RORB	UBE3A
SAMHD1	UNC80
SCARB2	WDR45
SCN1A	WWOX
SCN1B	ZBTB18
SCN2A	ZEB2
SCN3A	
SCN8A	
SCN9A	
SERPINI1	
SHANK3	
SIK1	
SLC12A5	
SLC13A5	
SLC1A2	
SLC25A12	
SLC25A22	
SLC2A1	
SLC35A2	
SLC39A8	
SLC6A1	
SLC6A8	
SLC9A6	
SPATA5	
SPTAN1	
SRPX2	
ST3GAL3	
ST3GAL5	
STAMBP	

## dbCMMS.v10.1

Gener för mitokondriella sjukdomar (192st), alla ingår i listan för medfödda metabola sjukdomar

AARS2	FOXRED1	NDUFAF2	SCO2
ACAD9	FTL	NDUFAF3	SDHA
ADCK2	FXN	NDUFAF4	SDHAF1
AFG3L2	GARS	NDUFAF5	SDHAF2
AGK	GFER	NDUFAF6	SDHB
AIFM1	GFM1	NDUFB1	SDHC
ANTXR2	GTPBP3	NDUFB11	SDHD
APOPT1	HSPD1	NDUFB3	SLC25A1
ATAD3A	IARS2	NDUFB6	SLC25A19
ATP5E	IBA57	NDUFB8	SLC25A3
ATP6AP1	ISCA2	NDUFB9	SLC25A32
ATPAF2	ISCU	NDUFC2	SLC25A4
BCS1L	KARS	NDUFS1	SLC25A42
BOLA3	KIF5A	NDUFS2	SLC25A46
C12ORF65	LARS2	NDUFS3	SPG7
CA5A	LRPPRC	NDUFS4	SUCLA2
CARS2	LYRM7	NDUFS5	SUCLG1
CEP89	MCAT	NDUFS6	SURF1
CHCHD10	MFF	NDUFS7	TACO1
CLPB	MFN2	NDUFS8	TARS2
CLPP	MGME1	NDUFV1	TAZ
COA5	MICU1	NDUFV2	TFAM
COA6	MIEF2	NDUFV3	TIMM8A
COASY	MPC1	NFS1	TK2
COQ2	MPV17	NFU1	TMEM126A
COQ4	MRPL12	NUBPL	TMEM126B
COQ7	MRPL44	OPA1	TMEM70
COQ8A	MRPL57	OPA3	TNNI3
COQ9	MRPS16	PARK7	TRIT1
COX10	MRPS2	PARS2	TRMT10C
COX15	MRPS22	PDK3	TRMT5
COX4I2	MRPS23	PDSS1	TRMU
COX6B1	MRPS7	PDSS2	TSFM
COX8A	MTO1	PET100	TTC19
DARS2	MTPAP	PNPLA4	TUFM
DCAF17	NADK2	PNPT1	TWNK
DGUOK	NARS2	POLG	TYMP
DNA2	NAXE	POLG2	UQCC2
DNAJC19	NDUFA1	PPA2	UQCC3
DNM1L	NDUFA10	PTCD1	UQCRB
EARS2	NDUFA11	PUS1	UQCRC2
ECHS1	NDUFA12	QRSL1	UQCRCQ
ECSIT	NDUFA13	RARS2	VARS2
ELAC2	NDUFA2	RMND1	VMA21
EPG5	NDUFA4	RNASEH1	YARS2
ETHE1	NDUFA6	RRM2B	
FARS2	NDUFA8	RTN4IP1	
FASTKD2	NDUFA9	SARS2	
FDX1L	NDUFAF1	SCO1	

## dbCMMS.v10.1

Gener för leversjukdomar hos barnrn (58st), 29 ingår i listan för medfödda metabola sjukdomar

ABCB11	TMEM67
ABCB4	TRMU
AGL	TTC21B
AKR1D1	TWNK
ALDOB	WDR19
AMACR	VIPAS39
ANKS6	VPS33B
ATP7B	XPNPEP3
ATP8B1	ZNF423
BCS1L	
CEP164	
CEP290	
CLDN1	
CYP7B1	
DCDC2	
DGUOK	
G6PC	
GBE1	
GLIS2	
GYS2	
HSD3B7	
INVS	
IQCB1	
JAG1	
MPV17	
MYO5B	
NEK8	
NOTCH2	
NPC1	
NPC2	
NPHP1	
NPHP3	
NPHP4	
NR1H4	
PHKA2	
PHKB	
PHKG2	
PKD1	
PKD2	
PKHD1	
POLG	
PYGL	
RPGRIP1L	
SDCCAG8	
SLC25A13	
SLC2A2	
SLC37A4	
TALDO1	
TJP2	