

dbCMMS.v12

Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild (798st)

AARS2	ALG12	BCKDHB	COQ4
AASS	ALG13	BCKDK	COQ6
ABAT	ALG14	BCS1L	COQ7
ABCC8	ALG2	BLOC1S3	COQ8A
ABCD1	ALG3	BOLA3	COQ9
ABCD4	ALG6	BSCL2	COX10
ABHD12	ALG8	BTD	COX14
ABHD5	ALG9	C12ORF65	COX15
ACAD8	ALPL	C19ORF12	COX20
ACAD9	AMACR	C19ORF70	COX4I1
ACADM	AMN	C1QBP	COX4I2
ACADS	AMPD1	CA5A	COX5A
ACADSB	AMPD2	CACNA1A	COX6B1
ACADVL	AMT	CACNA1S	COX8A
ACAT1	ANTXR2	CAD	CP
ACER3	AP1S1	CARS2	CPOX
ACO2	AP3B1	CAT	CPS1
ACOX1	AP3D1	CBS	CPT1A
ACSF3	AP5Z1	CCDC115	CPT2
ACY1	APOPT1	CCDC174	CRAT
ADA	APRT	CCDC88C	CTH
ADAR	ARG1	CD320	CTNS
ADCK2	ARSA	CEP89	CTSA
ADCY5	ARSB	CERS1	CTSD
ADK	ASAH1	CHCHD10	CTSF
ADSL	ASL	CHKB	CTSK
ADSSL1	ASNS	CISD2	CUBN
AFG3L2	ASPA	CLDN16	CWF19L1
AGA	ASS1	CLN3	CYP27A1
AGK	ATAD3A	CLN5	CYP7B1
AGL	ATAD3B	CLN6	D2HGDH
AGPS	ATAD3C	CLN8	DARS2
AGXT	ATIC	CLPB	DBH
AHCY	ATP13A2	CLPP	DBT
AIFM1	ATP5E	CLTC	DCAF17
AIMP1	ATP5F1D	CMPK2	DDC
AKR1D1	ATP5MD	CNKSR2	DGUOK
ALAD	ATP6AP1	COA5	DHCR24
ALAS2	ATP6V0A2	COA6	DHCR7
ALDH18A1	ATP7A	COA7	DHDDS
ALDH3A2	ATP7B	COASY	DHFR
ALDH4A1	ATPAF2	COG1	DHODH
ALDH5A1	AUH	COG2	DIAPH1
ALDH6A1	B4GALNT1	COG4	DLAT
ALDH7A1	B4GALT1	COG5	DLD
ALDOA	B4GALT7	COG6	DMGDH
ALDOB	BAAT	COG7	DNA2
ALG1	BCAT2	COG8	DNAJC12
ALG11	BCKDHA	COQ2	DNAJC19

dbCMMS.v12

DNAJC3	FMO3	GPR161	IGSF1
DNAJC5	FOLR1	GRHPR	INSR
DNM1L	FOXRED1	GRN	ISCA1
DOLK	FTCD	GSS	ISCA2
DPAGT1	FTL	GTPBP3	ISCU
DPM1	FUCA1	GUSB	ISPD
DPM2	FXN	GYG1	IVD
DPM3	G6PC	GYG2	JAG1
DPYD	G6PD	GYS1	KARS
DPYS	GAA	GYS2	KCNA4
DTNBP1	GABBR2	HAAO	KCNJ11
EARS2	GABRG2	HACE1	KIF5A
EBP	GALC	HADH	KYNU
ECHS1	GALE	HADHA	L2HGDH
ECSIT	GALK1	HADHB	LAMA1
EIF2B1	GALNS	HARS	LAMP2
EIF2B2	GALNT3	HCFC1	LARS2
EIF2B3	GALT	HEPACAM	LBR
EIF2B4	GAMT	HEXA	LDHA
EIF2B5	GARS	HEXB	LHX4
ELAC2	GATM	HGD	LIAS
ELOVL4	GBA	HGSNAT	LIPA
ELOVL5	GBE1	HIBCH	LIPE
ENO3	GCDH	HIKESHI	LIPT1
EPG5	GCH1	HLCS	LIPT2
EPHX1	GCK	HMBS	LMBRD1
EPM2A	GCLC	HMGCL	LPIN1
ERAL1	GCSH	HMGCS2	LRP2
ERCC6	GFER	HOGA1	LRPPRC
ETFA	GFM1	HPD	LYRM4
ETFB	GIF	HPRT1	LYRM7
ETFDH	GJC2	HPS1	MAG
ETHE1	GK	HPS3	MAGED2
EXOSC3	GLA	HPS4	MAGEL2
EXOSC8	GLB1	HPS5	MAGT1
EXT1	GLDC	HPS6	MAN1B1
EXT2	GLRA1	HSD17B10	MAN2B1
FA2H	GLRX5	HSD17B4	MANBA
FAH	GLS	HSD3B7	MAOA
FAM126A	GLUD1	HSPD1	MARS2
FAR1	GLUL	HTRA2	MAT1A
FARS2	GM2A	HYAL1	MBD5
FASTKD2	GMPPA	HYOU1	MCAT
FBP1	GNAO1	IARS	MCCC1
FBXL4	GNE	IARS2	MCCC2
FDX2	GNMT	IBA57	MCEE
FDXR	GNPAT	IDH2	MCOLN1
FECH	GNPTAB	IDS	MECR
FH	GNPTG	IDUA	MFF
FIG4	GNS	IER3IP1	MFN2
FLAD1	GPHN	IFIH1	MFSD8

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MGAT2	NAXE	OCRL	PGM3
MGME1	NBAS	OGDH	PHGDH
MICU1	NDST1	OPA1	PHKA1
MICU2	NDUFA1	OPA3	PHKA2
MIEF2	NDUFA10	OPLAH	PHKB
MIPEP	NDUFA11	OTC	PHKG2
MLC1	NDUFA12	OXA1L	PHYH
MLPH	NDUFA13	OXCT1	PIGA
MLYCD	NDUFA2	PAH	PIGM
MMAA	NDUFA4	PANK2	PIGN
MMAB	NDUFA6	PARK7	PIGO
MMACHC	NDUFA8	PARS2	PIGQ
MMADHC	NDUFA9	PAX7	PIGT
MOCS1	NDUFAF1	PC	PIGV
MOCS2	NDUFAF2	PCBD1	PIGW
MOCS3	NDUFAF3	PCCA	PITRM1
MOGS	NDUFAF4	PCCB	PLA2G6
MPC1	NDUFAF5	PCK1	PLAA
MPDU1	NDUFAF6	PCLO	PMM2
MPI	NDUFB1	PCYT1A	PMPCA
MPV17	NDUFB10	PDE10A	PNP
MRM2	NDUFB11	PDE12	PNPLA2
MRPL12	NDUFB3	PDHA1	PNPLA4
MRPL44	NDUFB6	PDHB	PNPLA6
MRPL57	NDUFB8	PDHX	PNPO
MRPS16	NDUFB9	PDK3	PNPT1
MRPS2	NDUFC2	PDP1	POLG
MRPS22	NDUFS1	PDSS1	POLG2
MRPS23	NDUFS2	PDSS2	POLR1A
MRPS34	NDUFS3	PEPD	POLR1C
MRPS7	NDUFS4	PET100	POLR3A
MSTO1	NDUFS5	PET117	POLR3B
MTFMT	NDUFS6	PEX1	POMGNT1
MTHFD1	NDUFS7	PEX10	POMT1
MTHFR	NDUFS8	PEX11B	PPA2
MTO1	NDUFV1	PEX12	PPM1K
MTPAP	NDUFV2	PEX13	PPOX
MTR	NDUFV3	PEX14	PPP1R15B
MTRR	NEU1	PEX16	PPP2R1A
MUT	NFE2L2	PEX19	PPP2R5D
MVK	NFS1	PEX2	PPT1
MYO5A	NFU1	PEX26	PRKAG2
NAA10	NKX6-2	PEX3	PRMT7
NADK2	NOTCH2	PEX5	PRODH
NAGA	NPC1	PEX6	PRODH2
NAGLU	NPC2	PEX7	PRPS1
NAGS	NSDHL	PFKM	PRUNE1
NALCN	NT5C3A	PGAM2	PSAP
NANS	NUBPL	PGAP2	PSPH
NARS2	NUS1	PGK1	PTCD1
NAXD	OAT	PGM1	PTS

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PURA	SLC25A19	STX3	TRNT1
PUS1	SLC25A20	STXBP5L	TRPM6
PYCR1	SLC25A22	SUCLA2	TSFM
PYCR2	SLC25A24	SUCLG1	TTC19
PYGL	SLC25A26	SUGCT	TTPA
PYGM	SLC25A3	SUMF1	TUBB3
PYROXD1	SLC25A32	SUOX	TUBB4A
QARS	SLC25A4	SURF1	TUFM
QDPR	SLC25A42	SZT2	TWNK
QRSL1	SLC25A46	TACO1	TXN2
RAB11B	SLC27A4	TAF1	TYMP
RAB27A	SLC2A1	TALDO1	UBQLN2
RAB3GAP1	SLC2A2	TANGO2	UBTF
RANBP2	SLC30A10	TARS2	UGT1A1
RARS	SLC30A9	TAT	UMPS
RARS2	SLC35A1	TAZ	UNC80
REPS1	SLC35A2	TBCD	UPB1
RFT1	SLC35C1	TBCK	UQCC2
RMND1	SLC37A4	TBL1X	UQCC3
RNASEH1	SLC39A14	TCN1	UQCRB
RPIA	SLC39A4	TCN2	UQCRC2
RRM2B	SLC39A8	TDO2	UQCRFS1
RTN4IP1	SLC3A1	TFAM	UQCRQ
RYR1	SLC46A1	TH	UROD
SARS2	SLC4A10	THAP11	UROS
SC5D	SLC52A1	TIMM50	VAC14
SCO1	SLC52A2	TIMM8A	VARS2
SCO2	SLC52A3	TIMMDC1	WARS2
SCP2	SLC5A1	TJP2	WDR45
SCYL1	SLC5A2	TK2	WFS1
SDHA	SLC6A17	TKT	VMA21
SDHAF1	SLC6A19	TMEM126A	VPS11
SDHAF2	SLC6A3	TMEM126B	VPS33A
SDHB	SLC6A8	TMEM165	VPS53
SDHC	SLC6A9	TMEM199	WWOX
SDHD	SLC7A1	TMEM240	XDH
SELENOI	SLC7A2	TMEM260	XPR1
SERAC1	SLC7A7	TMEM70	YARS
SGSH	SLC7A9	TMLHE	YARS2
SLC16A1	SLC9A1	TMTC3	ZBTB42
SLC16A2	SMPD1	TNNI3	ZNHIT3
SLC17A5	SMS	TPK1	
SLC19A2	SNORD118	TPP1	
SLC19A3	SNX14	TRAPPC11	
SLC1A1	SPG7	TRAPPC12	
SLC1A4	SPR	TRIAP1	
SLC22A5	SQSTM1	TRIM37	
SLC25A1	SRD5A3	TRIT1	
SLC25A12	SSR4	TRMT10C	
SLC25A13	ST3GAL5	TRMT5	
SLC25A15	STAT2	TRMU	

dbCMMS.v12

Gener för epileptisk encefalopati (290st), 94 ingår i listan för medfödda metabola sjukdomar

AARS	CLTC	GAMT	MAGI2
ABAT	CNKSR2	GATM	MAPK10
ADAR	CNNM2	GCSH	MBD5
ADCY5	CNTNAP2	GLDC	MBOAT7
ADRA2B	COL4A1	GNAO1	MDH2
ADSL	CPA6	GNB1	MECP2
AFG3L2	CPLX1	GNB5	MED13L
AGTR2	CRADD	GOSR2	MED17
AKT3	CSNK2B	GPAA1	MED23
ALDH5A1	CSTB	GPHN	MEF2C
ALDH7A1	CTSD	GRIA3	MFF
ALG1	CTSF	GRIN1	MFSD8
ALG13	CUX2	GRIN2A	MOCS1
ALG14	DCX	GRIN2B	MOCS2
ALG3	DDX3X	GRIN2D	MTOR
ALG6	DEAF1	GRN	NAA10
AMT	DENND5A	GUF1	NACC1
ANKRD11	DEPDC5	HCN1	NAPB
AP3B2	DHDDS	HECW2	NECAP1
AP4S1	DNAJC5	HIVEP2	NF1
APP	DNM1	HNRNPH2	NGLY1
ARHGEF9	DNM1L	HNRNPU	NHLRC1
ARID1B	DNMT3A	HSD17B10	NPRL2
ARV1	DOCK7	IQSEC2	NPRL3
ARX	DOLK	ITPA	NR2F1
ASAH1	DPM1	KCNA1	NRXN1
ASNS	DPM2	KCNA2	NTRK2
ATP13A2	DPYD	KCNB1	NUS1
ATP1A2	DYNC1H1	KCNC1	OPHN1
ATP1A3	DYRK1A	KCNH1	OTUD6B
ATP6AP2	EEF1A2	KCNH5	PAFAH1B1
ATP7A	EHMT1	KCNJ10	PAK3
BRAT1	EIF2S3	KCNMA1	PAX7
BRWD3	EPM2A	KCNQ1	PCDH12
BSCL2	FGF12	KCNQ2	PCDH19
CACNA1A	FGF13	KCNQ3	PIGA
CACNA2D1	FLNA	KCNQ5	PIGC
CAD	FOLR1	KCNT1	PIGG
CASK	FOXP1	KCNT2	PIGN
CDKL5	FOXP1	KCTD7	PIGO
CERS1	FRRS1L	KDM5C	PIGP
CHD2	GABBR2	KIAA2022	PIGQ
CHRNA2	GABRA1	KIF1A	PIGT
CHRNA4	GABRA3	KIF5A	PIGW
CHRNB2	GABRB1	KMT2A	PLCB1
CLN3	GABRB2	LGI1	PLPBP
CLN5	GABRB3	LIAS	PNKP
CLN6	GABRG2	LMNB2	PNPO
CLN8	GAL	MAGEL2	POLG

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PPP2R1A	SMC1A
PPP3CA	SNAP25
PPT1	SPATA5
PRDM8	SPTAN1
PRICKLE1	SRPX2
PRICKLE2	ST3GAL3
PRRT2	ST3GAL5
PRUNE1	STAMBP
PURA	STRADA
QARS	STX1B
RAB11A	STXBP1
RAB11B	STYXL1
RAB39B	SUMF1
RARS2	SV2A
RBSN	SYN1
RELN	SYNGAP1
RFT1	SYNJ1
RNASEH2A	SYP
RNASEH2B	SZT2
RNASEH2C	TBC1D24
ROGDI	TBCD
RORB	TCF4
RUSC2	TDP2
SAMHD1	TMTC3
SCARB2	TPP1
SCN1A	TRAK1
SCN1B	TRAPPC12
SCN2A	TREX1
SCN3A	TRIM8
SCN8A	TRPM6
SCN9A	TSC1
SEPSECS	TSC2
SERPINI1	TUBA1A
SETD1B	TUBB2A
SHANK3	UBA5
SIK1	UBE3A
SLC12A5	UNC80
SLC13A5	WDR45
SLC1A2	WWOX
SLC1A4	YWHAG
SLC25A12	ZBTB18
SLC25A22	ZEB2
SLC2A1	ZNHIT3
SLC35A2	
SLC35A3	
SLC39A8	
SLC45A1	
SLC6A1	
SLC6A8	
SLC9A6	
SMARCA2	

dbCMMS.v12

Gener för mitokondriella sjukdomar (229st), alla ingår i listan för medfödda metabola sjukdomar

AARS2	DARS2	MRPL57	NFU1
ACAD9	DCAF17	MRPS16	NUBPL
ADCK2	DGUOK	MRPS2	OPA1
AFG3L2	DNA2	MRPS22	OPA3
AGK	DNAJC19	MRPS23	OXA1L
AIFM1	DNM1L	MRPS34	PARK7
ANTXR2	EARS2	MRPS7	PARS2
APOPT1	ECHS1	MSTO1	PDE12
ATAD3A	ECSIT	MTO1	PKD3
ATAD3B	ELAC2	MTPAP	PDSS1
ATAD3C	EPG5	NADK2	PDSS2
ATP5E	ERAL1	NARS2	PET100
ATP5F1D	ETHE1	NAXD	PET117
ATP5MD	FARS2	NAXE	PITRM1
ATP6AP1	FASTKD2	NDUFA1	PNPLA4
ATPAF2	FDX1L	NDUFA10	PNPT1
BCS1L	FDXR	NDUFA11	POLG
BOLA3	FOXRED1	NDUFA12	POLG2
C12ORF65	FTL	NDUFA13	PPA2
C19ORF70	FXN	NDUFA2	PTCD1
C1QBP	GARS	NDUFA4	PUS1
CA5A	GFER	NDUFA6	QRSL1
CARS2	GFM1	NDUFA8	RARS2
CEP89	GTPBP3	NDUFA9	REPS1
CHCHD10	HSPD1	NDUFAF1	RMND1
CISD2	IARS2	NDUFAF2	RNASEH1
CLPB	IBA57	NDUFAF3	RRM2B
CLPP	ISCA1	NDUFAF4	RTN4IP1
CMPK2	ISCA2	NDUFAF5	SARS2
COA5	ISCU	NDUFAF6	SCO1
COA6	KARS	NDUFB1	SCO2
COA7	KIF5A	NDUFB10	SDHA
COASY	LARS2	NDUFB11	SDHAF1
COQ2	LRPPRC	NDUFB3	SDHAF2
COQ4	LYRM7	NDUFB6	SDHB
COQ6	MCAT	NDUFB8	SDHC
COQ7	MECR	NDUFB9	SDHD
COQ8A	MFF	NDUFC2	SLC25A1
COQ9	MFN2	NDUFS1	SLC25A19
COX10	MGME1	NDUFS2	SLC25A24
COX14	MICU1	NDUFS3	SLC25A3
COX15	MICU2	NDUFS4	SLC25A32
COX20	MIEF2	NDUFS5	SLC25A4
COX4I1	MIPEP	NDUFS6	SLC25A42
COX4I2	MPC1	NDUFS7	SLC25A46
COX5A	MPV17	NDUFS8	SPG7
COX6B1	MRM2	NDUFV1	SUCLA2
COX8A	MRPL12	NDUFV2	SUCLG1
CRAT	MRPL44	NDUFV3	SURF1

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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
UQCRQ
VARS2
WARS2
VMA21
YARS2

dbCMMS.v12

Gener för leversjukdomar hos barn (60st), 29 ingår i listan för medfödda metabola sjukdomar

ABCB11	TALDO1
ABCB4	TJP2
ABCD3	TMEM67
ACOX2	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	