

dbCMMS.v11

Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild (757st)

AARS2	ALG13	C12ORF65	CPOX
AASS	ALG2	C19ORF12	CPS1
ABAT	ALG3	C19ORF70	CPT1A
ABCC8	ALG6	CA5A	CPT2
ABCD1	ALG8	CACNA1A	CTH
ABCD4	ALG9	CACNA1S	CTNS
ABHD12	ALPL	CAD	CTSA
ABHD5	AMACR	CARS2	CTSD
ACAD8	AMN	CAT	CTSF
ACAD9	AMPD1	CBS	CTSK
ACADM	AMPD2	CCDC115	CUBN
ACADS	AMT	CCDC174	CWF19L1
ACADSB	ANTXR2	CCDC88C	CYP27A1
ACADVL	AP1S1	CD320	CYP7B1
ACAT1	AP3B1	CEP89	D2HGDH
ACER3	AP3D1	CERS1	DARS2
ACO2	AP5Z1	CHCHD10	DBH
ACOX1	APOPT1	CHKB	DBT
ACSF3	APRT	CLDN16	DCAF17
ADA	ARG1	CLN3	DDC
ADAR	ARSA	CLN5	DGUOK
ADCK2	ARSB	CLN6	DHCR24
ADCY5	ASAH1	CLN8	DHCR7
ADK	ASL	CLPB	DHFR
ADSL	ASNS	CLPP	DHODH
ADSSL1	ASPA	CNKSR2	DIAPH1
AFG3L2	ASS1	COA5	DLAT
AGA	ATAD3A	COA6	DLD
AGK	ATIC	COASY	DMGDH
AGL	ATP13A2	COG1	DNA2
AGPS	ATP5E	COG2	DNAJC12
AGXT	ATP6AP1	COG4	DNAJC19
AHCY	ATP6V0A2	COG5	DNAJC3
AIFM1	ATP7A	COG6	DNAJC5
AIMP1	ATP7B	COG7	DNM1L
AKR1D1	ATPAF2	COG8	DOLK
ALAD	AUH	COQ2	DPAGT1
ALAS2	B4GALNT1	COQ4	DPM1
ALDH18A1	B4GALT1	COQ7	DPM2
ALDH3A2	B4GALT7	COQ8A	DPM3
ALDH4A1	BAAT	COQ9	DPYD
ALDH5A1	BCAT2	COX10	DPYS
ALDH6A1	BCKDHA	COX14	DTNBP1
ALDH7A1	BCKDHB	COX15	DUOX2
ALDOA	BCKDK	COX20	DUOXA2
ALDOB	BCS1L	COX4I2	EARS2
ALG1	BLOC1S3	COX6B1	EBP
ALG11	BOLA3	COX8A	ECHS1
ALG12	BTD	CP	ECSIT

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EIF2B1	GALT	HEXB	LIPE
EIF2B2	GAMT	HGD	LIPT1
EIF2B3	GARS	HGSNAT	LIPT2
EIF2B4	GATM	HIBCH	LMBRD1
EIF2B5	GBA	HIKESHI	LPIN1
ELAC2	GBE1	HLCS	LRP2
ELOVL4	GCDH	HMBS	LRPPRC
ELOVL5	GCH1	HMGCL	LYRM4
ENO3	GCK	HMGCS2	LYRM7
EPG5	GCLC	HOGA1	MAG
EPHX1	GCSH	HPD	MAGED2
EPM2A	GFER	HPRT1	MAGEL2
ERCC6	GFM1	HPS1	MAGT1
ETFA	GIF	HPS3	MAN1B1
ETFB	GJC2	HPS4	MAN2B1
ETFDH	GK	HPS5	MANBA
ETHE1	GLA	HPS6	MAOA
EXOSC3	GLB1	HSD17B10	MARS2
EXOSC8	GLDC	HSD17B4	MAT1A
EXT1	GLRA1	HSD3B7	MBD5
EXT2	GLRX5	HSPD1	MCAT
FA2H	GLS	HTRA2	MCCC1
FAH	GLUD1	HYAL1	MCCC2
FAM126A	GLUL	HYOU1	MCEE
FAR1	GM2A	IARS	MCOLN1
FARS2	GNAO1	IARS2	MECR
FASTKD2	GNE	IBA57	MFF
FBP1	GNMT	IDH2	MFN2
FBXL4	GNPAT	IDS	MFSD8
FDX1L	GNPTAB	IDUA	MGAT2
FECH	GNPTG	IGSF1	MGME1
FH	GNS	INSR	MICU1
FIG4	GPHN	ISCA2	MIEF2
FLAD1	GPR161	ISCU	MIPEP
FMO3	GRHPR	ISPD	MLC1
FOLR1	GRN	IVD	MLPH
FOXE1	GSS	IYD	MLYCD
FOXRED1	GTPBP3	JAG1	MMAA
FTCD	GUSB	KARS	MMAB
FTL	GYG1	KCNA4	MMACHC
FUCA1	GYG2	KCNJ11	MMADHC
FXN	GYS1	KIF5A	MOCS1
G6PC	GYS2	L2HGDH	MOCS2
G6PD	HACE1	LAMA1	MOGS
GAA	HADH	LAMP2	MPC1
GABRG2	HADHA	LARS2	MPDU1
GALC	HADHB	LBR	MPI
GALE	HARS	LDHA	MPV17
GALK1	HCFC1	LHX4	MRPL12
GALNS	HEPACAM	LIAS	MRPL44
GALNT3	HEXA	LIPA	MRPL57

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MRPS16	NDUFS3	PEX1	PPA2
MRPS2	NDUFS4	PEX10	PPOX
MRPS22	NDUFS5	PEX11B	PPP1R15B
MRPS23	NDUFS6	PEX12	PPP2R1A
MRPS7	NDUFS7	PEX13	PPP2R5D
MTFMT	NDUFS8	PEX14	PPT1
MTHFD1	NDUFV1	PEX16	PRKAG2
MTHFR	NDUFV2	PEX19	PRMT7
MTO1	NDUFV3	PEX2	PRODH
MTPAP	NEU1	PEX26	PRODH2
MTR	NFS1	PEX3	PRPS1
MTRR	NFU1	PEX5	PRUNE1
MUT	NKX2-1	PEX6	PSAP
MVK	NKX2-5	PEX7	PSPH
MYO5A	NOTCH2	PFKM	PTCD1
NAA10	NPC1	PGAM2	PTS
NADK2	NPC2	PGAP2	PURA
NAGA	NSDHL	PGK1	PUS1
NAGLU	NT5C3A	PGM1	PYCR1
NAGS	NUBPL	PGM3	PYCR2
NALCN	NUS1	PHGDH	PYGL
NANS	OAT	PHKA1	PYGM
NARS2	OCRL	PHKA2	PYROXD1
NAXE	OGDH	PHKB	QARS
NBAS	OPA1	PHKG2	QDPR
NDST1	OPA3	PHYH	QRSL1
NDUFA1	OPLAH	PIGA	RAB27A
NDUFA10	OTC	PIGM	RAB3GAP1
NDUFA11	OXCT1	PIGN	RANBP2
NDUFA12	PAH	PIGO	RARS
NDUFA13	PANK2	PIGQ	RARS2
NDUFA2	PARK7	PIGT	RFT1
NDUFA4	PARS2	PIGW	RMND1
NDUFA6	PAX8	PLA2G6	RNASEH1
NDUFA8	PC	PLAA	RPIA
NDUFA9	PCBD1	PMM2	RRM2B
NDUFAF1	PCCA	PMPCA	RTN4IP1
NDUFAF2	PCCB	PNP	RYR1
NDUFAF3	PCK1	PNPLA2	SARS2
NDUFAF4	PCLO	PNPLA4	SC5D
NDUFAF5	PCYT1A	PNPLA6	SCO1
NDUFAF6	PDE10A	PNPO	SCO2
NDUFB1	PDHA1	PNPT1	SCP2
NDUFB11	PDHB	POLG	SCYL1
NDUFB3	PDHX	POLG2	SDHA
NDUFB6	PDK3	POLR1A	SDHAF1
NDUFB8	PDP1	POLR1C	SDHAF2
NDUFB9	PDSS1	POLR3A	SDHB
NDUFC2	PDSS2	POLR3B	SDHC
NDUFS1	PEPD	POMGNT1	SDHD
NDUFS2	PET100	POMT1	SELENOI

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SERAC1	SLC7A2	TMEM70
SGSH	SLC7A7	TMLHE
SLC16A1	SLC7A9	TMTC3
SLC16A2	SLC9A1	TNNI3
SLC17A5	SMPD1	TPK1
SLC19A2	SMS	TPO
SLC19A3	SNORD118	TPP1
SLC1A1	SNX14	TRAPPC11
SLC1A4	SPG7	TRIM37
SLC22A5	SPR	TRIT1
SLC25A1	SQSTM1	TRMT10C
SLC25A12	SRD5A3	TRMT5
SLC25A13	SSR4	TRMU
SLC25A15	ST3GAL5	TRNT1
SLC25A19	STAT2	TRPM6
SLC25A20	STX3	TSFM
SLC25A22	STXBP5L	TSHR
SLC25A26	SUCLA2	TTC19
SLC25A3	SUCLG1	TTPA
SLC25A32	SUGCT	TUBB4A
SLC25A4	SUMF1	TUFM
SLC25A42	SUOX	TWNK
SLC25A46	SURF1	TXN2
SLC26A4	SZT2	TYMP
SLC27A4	TACO1	UBQLN2
SLC2A1	TAF1	UGT1A1
SLC2A2	TALDO1	UMPS
SLC30A10	TANGO2	UNC80
SLC30A9	TARS2	UPB1
SLC35A1	TAT	UQCC2
SLC35A2	TAZ	UQCC3
SLC35C1	TBCD	UQCRB
SLC37A4	TBCK	UQCRC2
SLC39A14	TBL1X	UQCRQ
SLC39A4	TCN1	UROD
SLC39A8	TCN2	UROS
SLC3A1	TDO2	VAC14
SLC46A1	TFAM	VARS2
SLC4A10	TG	WARS2
SLC52A1	TH	WDR45
SLC52A2	TIMM50	WFS1
SLC52A3	TIMM8A	VMA21
SLC5A1	TJP2	VPS11
SLC5A2	TK2	VPS53
SLC5A5	TKT	WVOX
SLC6A17	TMEM126A	XDH
SLC6A19	TMEM126B	XPR1
SLC6A3	TMEM165	YARS
SLC6A8	TMEM199	YARS2
SLC6A9	TMEM240	ZBTB42
SLC7A1	TMEM260	ZNHIT3

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Gener för epileptisk encefalopati (242st), 82 ingår i listan för medfödda metabola sjukdomar

AARS	CRADD	HNRNPH2	PCDH12
ABAT	CSTB	HNRNPU	PCDH19
ADAR	CTSD	HSD17B10	PIGA
ADCY5	CTSF	IQSEC2	PIGG
ADRA2B	DCX	ITPA	PIGN
ADSL	DEAF1	KCNA1	PIGO
AFG3L2	DENND5A	KCNA2	PIGQ
AGTR2	DEPDC5	KCNB1	PIGT
ALDH5A1	DNAJC5	KCNC1	PIGW
ALDH7A1	DNM1	KCNH1	PLCB1
ALG1	DNM1L	KCNJ10	PLPBP
ALG13	DOCK7	KCNMA1	PNKP
ALG3	DOLK	KCNQ2	PNPO
ALG6	DPM1	KCNQ3	POLG
AMT	DPM2	KCNT1	PPT1
AP3B2	DPYD	KCTD7	PRDM8
AP4S1	DYRK1A	KDM5C	PRICKLE1
APP	EEF1A2	KIAA2022	PRICKLE2
ARHGEF9	EHMT1	KIF1A	PRRT2
ARV1	EIF2S3	KIF5A	PRUNE1
ARX	EPM2A	KMT2A	PURA
ASAH1	FGF12	LGI1	QARS
ASNS	FLNA	LIAS	RAB39B
ATP13A2	FOLR1	LMNB2	RARS2
ATP1A2	FOXP1	MAGEL2	RBSN
ATP1A3	FRRS1L	MAGI2	RELN
ATP6AP2	GABRA1	MAPK10	RFT1
ATP7A	GABRB1	MBD5	RNASEH2A
BRAT1	GABRB3	MBOAT7	RNASEH2B
BRWD3	GABRG2	MDH2	RNASEH2C
BSCL2	GAL	MECP2	ROGDI
CACNA1A	GAMT	MED17	RORB
CACNA2D1	GATM	MEF2C	RUSC2
CAD	GCSH	MFF	SAMHD1
CASK	GLDC	MFSD8	SCARB2
CDKL5	GNAO1	MOCS1	SCN1A
CERS1	GNB1	MOCS2	SCN1B
CHD2	GNB5	NACC1	SCN2A
CHRNA2	GOSR2	NAPB	SCN3A
CHRNA4	GPHN	NECAP1	SCN8A
CHRN2	GRIA3	NGLY1	SCN9A
CLN3	GRIN1	NHLRC1	SEPSECS
CLN5	GRIN2A	NPRL2	SERPINI1
CLN6	GRIN2B	NPRL3	SHANK3
CLN8	GRIN2D	NR2F1	SIK1
CNKS2R2	GRN	NRXN1	SLC12A5
CNNM2	GUF1	OTUD6B	SLC13A5
CNTNAP2	HCN1	PAFAH1B1	SLC1A2
CPA6	HECW2	PAK3	SLC1A4

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SLC25A12
SLC25A22
SLC2A1
SLC35A2
SLC39A8
SLC6A1
SLC6A8
SLC9A6
SMC1A
SPATA5
SPTAN1
SRPX2
ST3GAL3
ST3GAL5
STAMBP
STRADA
STX1B
STXBP1
STYXL1
SUMF1
SV2A
SYN1
SYNGAP1
SYNJ1
SYP
SZT2
TBC1D24
TBCD
TCF4
TDP2
TPP1
TRAK1
TRES1
TRIM8
TRPM6
TSC1
TSC2
TUBA1A
UBA5
UBE3A
UNC80
WDR45
WVOX
ZBTB18
ZEB2
ZNHIT3

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Gener för mitokondriella sjukdomar (199st), alla ingår i listan för medfödda metabola sjukdomar

AARS2	FASTKD2	NDUFA8	SARS2
ACAD9	FDX1L	NDUFA9	SCO1
ADCK2	FOXRED1	NDUFAF1	SCO2
AFG3L2	FTL	NDUFAF2	SDHA
AGK	FXN	NDUFAF3	SDHAF1
AIFM1	GARS	NDUFAF4	SDHAF2
ANTXR2	GFER	NDUFAF5	SDHB
APOPT1	GFM1	NDUFAF6	SDHC
ATAD3A	GTPBP3	NDUFB1	SDHD
ATP5E	HSPD1	NDUFB11	SLC25A1
ATP6AP1	IARS2	NDUFB3	SLC25A19
ATPAF2	IBA57	NDUFB6	SLC25A3
BCS1L	ISCA2	NDUFB8	SLC25A32
BOLA3	ISCU	NDUFB9	SLC25A4
C12ORF65	KARS	NDUFC2	SLC25A42
C19ORF70	KIF5A	NDUFS1	SLC25A46
CA5A	LARS2	NDUFS2	SPG7
CARS2	LRPPRC	NDUFS3	SUCLA2
CEP89	LYRM7	NDUFS4	SUCLG1
CHCHD10	MCAT	NDUFS5	SURF1
CLPB	MECR	NDUFS6	TACO1
CLPP	MFF	NDUFS7	TARS2
COA5	MFN2	NDUFS8	TAZ
COA6	MGME1	NDUFV1	TFAM
COASY	MICU1	NDUFV2	TIMM50
COQ2	MIEF2	NDUFV3	TIMM8A
COQ4	MIPEP	NFS1	TK2
COQ7	MPC1	NFU1	TMEM126A
COQ8A	MPV17	NUBPL	TMEM126B
COQ9	MRPL12	OPA1	TMEM70
COX10	MRPL44	OPA3	TNNI3
COX14	MRPL57	PARK7	TRIT1
COX15	MRPS16	PARS2	TRMT10C
COX20	MRPS2	PDK3	TRMT5
COX4I2	MRPS22	PDSS1	TRMU
COX6B1	MRPS23	PDSS2	TSFM
COX8A	MRPS7	PET100	TTC19
DARS2	MTO1	PNPLA4	TUFM
DCAF17	MTPAP	PNPT1	TWNK
DGUOK	NADK2	POLG	TYMP
DNA2	NARS2	POLG2	UQCC2
DNAJC19	NAXE	PPA2	UQCC3
DNM1L	NDUFA1	PTCD1	UQCRB
EARS2	NDUFA10	PUS1	UQCRC2
ECHS1	NDUFA11	QRSL1	UQCRQ
ECSIT	NDUFA12	RARS2	VARS2
ELAC2	NDUFA13	RMND1	WARS2
EPG5	NDUFA2	RNASEH1	VMA21
ETHE1	NDUFA4	RRM2B	YARS2
FARS2	NDUFA6	RTN4IP1	

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Gener för leversjukdomar hos barn (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	