

dbCMMS.v9

Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild (681st)

AARS2	ALG13	C12ORF65	CPT1A
AASS	ALG2	C19ORF12	CPT2
ABAT	ALG3	CA5A	CTH
ABCC8	ALG6	CACNA1A	CTNS
ABCD1	ALG8	CARS2	CTSA
ABCD4	ALG9	CAT	CTSD
ABHD12	ALPL	CBS	CTSF
ABHD5	AMACR	CCDC115	CTSK
ACAD8	AMN	CCDC174	CUBN
ACAD9	AMPD1	CCDC88C	CWF19L1
ACADM	AMPD2	CD320	CYP27A1
ACADS	AMT	CEP89	CYP7B1
ACADSB	ANTXR2	CERS1	D2HGDH
ACADVL	AP1S1	CHCHD10	DARS2
ACAT1	AP3B1	CHKB	DBH
ACO2	AP5Z1	CLDN16	DBT
ACOX1	APOPT1	CLN3	DCAF17
ACSF3	APRT	CLN5	DDC
ADA	ARG1	CLN6	DGUOK
ADAR	ARSA	CLN8	DHCR24
ADCK2	ARSB	CLPB	DHCR7
ADK	ASAH1	CLPP	DHFR
ADSL	ASL	CNKSR2	DHODH
AFG3L2	ASNS	COA5	DIAPH1
AGA	ASPA	COA6	DLAT
AGK	ASS1	COASY	DLD
AGL	ATIC	COG1	DMGDH
AGPS	ATP13A2	COG2	DNA2
AGXT	ATP5E	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	BCKDHA	COX10	DPYS
ALDH6A1	BCKDHB	COX15	DTNBP1
ALDH7A1	BCKDK	COX4I2	DUOX2
ALDOA	BCS1L	COX6B1	DUOXA2
ALDOB	BLOC1S3	COX8A	EARS2
ALG1	BOLA3	CP	EBP
ALG11	BTD	CPOX	ECHS1
ALG12	C10ORF2	CPS1	ECSIT

EIF2B1	GAMT	HIKESHI	MAOA
EIF2B2	GARS	HLCS	MARS2
EIF2B3	GATM	HMBS	MAT1A
EIF2B4	GBA	HMGCL	MBD5
EIF2B5	GBE1	HMGCS2	MCAT
ELAC2	GCDH	HOGA1	MCCC1
ELOVL4	GCH1	HPD	MCCC2
ELOVL5	GCK	HPRT1	MCEE
ENO3	GCLC	HPS1	MCOLN1
EPG5	GCSH	HPS3	MFF
EPHX1	GFER	HPS4	MFN2
EPM2A	GFM1	HPS5	MFSD8
ERCC6	GIF	HPS6	MGAT2
ETFA	GJC2	HSD17B10	MGME1
ETFB	GK	HSD17B4	MIEF2
ETFDH	GLA	HSD3B7	MLC1
ETHE1	GLB1	HSPD1	MLPH
EXOSC8	GLDC	HYAL1	MLYCD
EXT1	GLRA1	IARS2	MMAA
EXT2	GLRX5	IBA57	MMAB
FA2H	GLUD1	IDH2	MMACHC
FAH	GLUL	IDS	MMADHC
FAM126A	GM2A	IDUA	MOGS
FAR1	GNE	INSR	MPC1
FARS2	GNMT	ISCA2	MPDU1
FASTKD2	GNPAT	ISCU	MPI
FBP1	GNPTAB	ISPD	MPV17
FBXL4	GNPTG	IVD	MRPL12
FDX1L	GNS	IYD	MRPL44
FECH	GPR161	JAG1	MRPS16
FH	GRHPR	KCNJ11	MRPS22
FIG4	GRN	L2HGDH	MRPS23
FLAD1	GSS	LAMA1	MRPS7
FMO3	GTPBP3	LAMP2	MTFMT
FOLR1	GUSB	LARS2	MTHFD1
FOXE1	GYG1	LBR	MTHFR
FOXRED1	GYG2	LDHA	MTO1
FTCD	GYS1	LHX4	MTPAP
FTL	GYS2	LIAS	MTR
FUCA1	HACE1	LIPA	MTRR
FXN	HADH	LIPE	MUT
G6PC	HADHA	LIPT1	MVK
G6PD	HADHB	LIPT2	MYO5A
GAA	HARS	LMBRD1	NAA10
GABRG2	HCFC1	LPIN1	NADK2
GALC	HEPACAM	LRPPRC	NAGA
GALE	HEXA	LYRM4	NAGLU
GALK1	HEXB	MAG	NAGS
GALNS	HGD	MAGEL2	NALCN
GALNT3	HGSNAT	MAN2B1	NARS2
GALT	HIBCH	MANBA	NAXE

NBAS	OPLAH	PIGQ	RTN4IP1
NDST1	OTC	PIGT	SARS2
NDUFA1	OXCT1	PIGW	SC5D
NDUFA10	PAH	PLA2G6	SCO1
NDUFA11	PANK2	PMM2	SCO2
NDUFA12	PARK7	PMPCA	SCP2
NDUFA13	PARS2	PNP	SCYL1
NDUFA2	PAX8	PNPLA2	SDHA
NDUFA4	PC	PNPLA4	SDHAF1
NDUFA6	PCBD1	PNPLA6	SDHAF2
NDUFA8	PCCA	PNPO	SDHB
NDUFA9	PCCB	PNPT1	SDHC
NDUFAF1	PCLO	POLG	SDHD
NDUFAF2	PCYT1A	POLG2	SERAC1
NDUFAF3	PDHA1	POLR1C	SGSH
NDUFAF4	PDHB	POLR3A	SLC16A1
NDUFAF5	PDHX	POLR3B	SLC16A2
NDUFAF6	PDK3	POMGNT1	SLC17A5
NDUFB1	PDP1	POMT1	SLC19A2
NDUFB11	PDSS1	PPOX	SLC19A3
NDUFB3	PDSS2	PPP1R15B	SLC1A1
NDUFB6	PEPD	PPP2R1A	SLC1A4
NDUFB9	PET100	PPP2R5D	SLC22A5
NDUFC2	PEX1	PPT1	SLC25A1
NDUFS1	PEX10	PRKAG2	SLC25A12
NDUFS2	PEX11B	PRMT7	SLC25A13
NDUFS3	PEX12	PRODH	SLC25A15
NDUFS4	PEX13	PRPS1	SLC25A19
NDUFS5	PEX14	PSAP	SLC25A20
NDUFS6	PEX16	PSPH	SLC25A22
NDUFS7	PEX19	PTCD1	SLC25A26
NDUFS8	PEX2	PTS	SLC25A3
NDUFV1	PEX26	PURA	SLC25A32
NDUFV2	PEX3	PUS1	SLC25A4
NDUFV3	PEX5	PYCR1	SLC26A4
NEU1	PEX6	PYCR2	SLC27A4
NFS1	PEX7	PYGL	SLC2A1
NFU1	PFKM	PYGM	SLC2A2
NKX2-1	PGAM2	QARS	SLC30A10
NKX2-5	PGAP2	QDPR	SLC35A1
NOTCH2	PGK1	QRSL1	SLC35A2
NPC1	PGM1	RAB27A	SLC35C1
NPC2	PHGDH	RAB3GAP1	SLC37A4
NSDHL	PHKA1	RANBP2	SLC39A4
NT5C3A	PHKA2	RARS	SLC39A8
NUBPL	PHKB	RARS2	SLC3A1
NUS1	PHKG2	RFT1	SLC46A1
OAT	PHYH	RMND1	SLC52A1
OGDH	PIGA	RNASEH1	SLC52A2
OPA1	PIGN	RPIA	SLC52A3
OPA3	PIGO	RRM2B	SLC5A1

SLC5A2	TPK1
SLC5A5	TPO
SLC6A17	TPP1
SLC6A19	TRIM37
SLC6A3	TRIT1
SLC6A8	TRMT10C
SLC7A7	TRMT5
SLC7A9	TRMU
SLC9A1	TRNT1
SMPD1	TRPM6
SMS	TSFM
SNX14	TSHR
SPG7	TTC19
SPR	TTPA
SQSTM1	TUBB4A
SRD5A3	TUFM
SSR4	TXN2
ST3GAL5	TYMP
STAT2	UBQLN2
STX3	UGT1A1
STXBP5L	UMPS
SUCLA2	UPB1
SUCLG1	UQCC2
SUGCT	UQCC3
SUMF1	UQCRB
SUOX	UQCRQ
SURF1	UROD
SZT2	UROS
TACO1	VARS2
TAF1	WDR45
TALDO1	WFS1
TANGO2	VPS11
TARS2	VPS53
TAT	WVOX
TAZ	XDH
TBCK	XPR1
TCN1	YARS2
TCN2	ZBTB42
TG	
TH	
TIMM8A	
TJP2	
TK2	
TKT	
TMEM126A	
TMEM126B	
TMEM165	
TMEM199	
TMEM240	
TMEM70	
TNNI3	

Gener för epileptisk encefalopati (183st)

AARS	DEPDC5	KIF1A	SCN1A
ABAT	DNAJC5	KMT2A	SCN1B
ADAR	DNM1	LGI1	SCN2A
ADRA2B	DOCK7	LIAS	SCN3A
ADSL	DOLK	LMNB2	SCN8A
AFG3L2	DPM1	MAGEL2	SCN9A
AGTR2	DPM2	MAGI2	SERPINI1
ALDH7A1	DPYD	MAPK10	SIK1
ALG1	DYRK1A	MBD5	SLC12A5
ALG13	EEF1A2	MECP2	SLC13A5
ALG3	EHMT1	MEF2C	SLC25A22
ALG6	EPM2A	MFSB8	SLC2A1
AMT	FOLR1	NAPB	SLC35A2
AP4S1	FOXG1	NECAP1	SLC39A8
ARHGEF9	GABRA1	NHLRC1	SLC6A1
ARX	GABRB3	NPRL3	SLC6A8
ASAH1	GABRG2	NR2F1	SLC9A6
ASNS	GAL	NRXN1	SPATA5
ATP13A2	GAMT	PAK3	SPTAN1
ATP1A2	GATM	PCDH19	SRPX2
ATP1A3	GCSH	PIGA	ST3GAL3
ATP6AP2	GLDC	PIGN	ST3GAL5
ATP7A	GNAO1	PIGO	STX1B
BRAT1	GOSR2	PIGQ	STXBP1
BRWD3	GRIA3	PIGT	STYXL1
CACNA1A	GRIN1	PIGW	SUMF1
CACNA2D1	GRIN2A	PLCB1	SV2A
CASK	GRIN2B	PNKP	SYN1
CDKL5	GRN	PNPO	SYNGAP1
CERS1	HCN1	POLG	SYNJ1
CHD2	HNRNPU	PPT1	SYP
CHRNA2	HSD17B10	PRDM8	SZT2
CHRNA4	IQSEC2	PRICKLE1	TBC1D24
CHRN2	ITPA	PRICKLE2	TCF4
CLN3	KCNA1	PRRT2	TDP2
CLN5	KCNA2	PURA	TPP1
CLN6	KCNB1	QARS	TREX1
CLN8	KCNC1	RAB39B	TRPM6
CNKSR2	KCNH1	RBSN	TSC1
CNNM2	KCNJ10	RELN	TSC2
CNTNAP2	KCNMA1	RNASEH2A	UBE3A
CPA6	KCNQ2	RNASEH2B	UNC80
CSTB	KCNQ3	RNASEH2C	WDR45
CTSD	KCNT1	ROGDI	WVVOX
CTSF	KCTD7	SAMHD1	ZEB2
DEAF1	KDM5C	SCARB2	

Gener för mitokondriella sjukdomar (177st), alla ingår i listan för medfödda metabola sjukdomar

AARS2	FOXRED1	NDUFB1	SDHD
ACAD9	FTL	NDUFB11	SLC25A1
ADCK2	FXN	NDUFB3	SLC25A19
AFG3L2	GARS	NDUFB6	SLC25A3
AGK	GFER	NDUFB9	SLC25A32
AIFM1	GFM1	NDUFC2	SLC25A4
ANTXR2	GTPBP3	NDUFS1	SPG7
APOPT1	HSPD1	NDUFS2	SUCLA2
ATP5E	IARS2	NDUFS3	SUCLG1
ATPAF2	IBA57	NDUFS4	SURF1
BCS1L	ISCA2	NDUFS5	TACO1
BOLA3	ISCU	NDUFS6	TARS2
C10ORF2	LARS2	NDUFS7	TAZ
C12ORF65	LRPPRC	NDUFS8	TIMM8A
CA5A	MCAT	NDUFV1	TK2
CARS2	MFF	NDUFV2	TMEM126A
CEP89	MFN2	NDUFV3	TMEM126B
CHCHD10	MGME1	NFS1	TMEM70
CLPB	MIEF2	NFU1	TNNI3
CLPP	MPC1	NUBPL	TRIT1
COA5	MPV17	OPA1	TRMT10C
COA6	MRPL12	OPA3	TRMT5
COASY	MRPL44	PARK7	TRMU
COQ2	MRPS16	PARS2	TSFM
COQ4	MRPS22	PDK3	TTC19
COQ7	MRPS23	PDSS1	TUFM
COQ8A	MRPS7	PDSS2	TYMP
COQ9	MTO1	PET100	UQCC2
COX10	MTPAP	PNPLA4	UQCC3
COX15	NADK2	PNPT1	UQCRB
COX4I2	NARS2	POLG	UQCRQ
COX6B1	NAXE	POLG2	VAR52
COX8A	NDUFA1	PTCD1	YARS2
DARS2	NDUFA10	PUS1	
DCAF17	NDUFA11	QRSL1	
DGUOK	NDUFA12	RARS2	
DNA2	NDUFA13	RMND1	
DNAJC19	NDUFA2	RNASEH1	
DNM1L	NDUFA4	RRM2B	
EARS2	NDUFA6	RTN4IP1	
ECHS1	NDUFA8	SARS2	
ECSIT	NDUFA9	SCO1	
ELAC2	NDUFAF1	SCO2	
EPG5	NDUFAF2	SDHA	
ETHE1	NDUFAF3	SDHAF1	
FARS2	NDUFAF4	SDHAF2	
FASTKD2	NDUFAF5	SDHB	
FDX1L	NDUFAF6	SDHC	