

dbCMMS.v7

Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild (631st)

AARS2	ALG3	CARS2	CYP7B1
AASS	ALG6	CBS	D2HGDH
ABAT	ALG8	CCDC88C	DARS2
ABCC8	ALG9	CD320	DBH
ABCD1	ALPL	CEP89	DBT
ABCD4	AMACR	CERS1	DCAF17
ABHD12	AMN	CHCHD10	DDC
ABHD5	AMPD1	CHKB	DGUOK
ACAD8	AMPD2	CLDN16	DHCR24
ACAD9	AMT	CLN3	DHCR7
ACADM	ANTXR2	CLN5	DHFR
ACADS	AP1S1	CLN6	DHODH
ACADSB	AP5Z1	CLN8	DIAPH1
ACADVL	APOPT1	CLPB	DLAT
ACAT1	APRT	CLPP	DLD
ACO2	ARG1	CNKSR2	DMGDH
ACOX1	ARSA	COA5	DNA2
ACSF3	ARSB	COA6	DNAJC19
ADA	ASAH1	COASY	DNAJC3
ADAR	ASL	COG1	DNAJC5
ADCK3	ASNS	COG2	DNM1L
ADK	ASPA	COG4	DOLK
ADSL	ASS1	COG5	DPAGT1
AFG3L2	ATIC	COG6	DPM1
AGA	ATP13A2	COG7	DPM3
AGK	ATP5E	COG8	DPYD
AGL	ATP6V0A2	COQ2	DPYS
AGPS	ATP7A	COQ4	DUOX2
AGXT	ATP7B	COQ9	DUOXA2
AHCY	ATPAF2	COX10	EARS2
AIFM1	AUH	COX15	EBP
AKR1D1	B4GALNT1	COX4I2	ECHS1
ALAD	B4GALT1	COX6B1	ECSIT
ALAS2	B4GALT7	CP	EIF2B1
ALDH18A1	BAAT	CPOX	EIF2B2
ALDH3A2	BCKDHA	CPS1	EIF2B3
ALDH4A1	BCKDHB	CPT1A	EIF2B4
ALDH5A1	BCKDK	CPT2	EIF2B5
ALDH6A1	BCS1L	CTH	ELAC2
ALDH7A1	BOLA3	CTNS	ELOVL4
ALDOA	BTD	CTSA	ELOVL5
ALDOB	C10orf2	CTSD	ENO3
ALG1	C11orf83	CTSF	EPG5
ALG11	C12orf65	CTSK	EPHX1
ALG12	C19orf12	CUBN	EPM2A
ALG13	CA5A	CWF19L1	ERCC6
ALG2	CACNA1A	CYP27A1	ETFA

ETFB	GLDC	ISCA2	MRPL12
ETFDH	GLRA1	ISCU	MRPL44
ETHE1	GLRX5	ISPD	MRPS16
EXOSC8	GLUD1	IVD	MRPS22
EXT1	GLUL	IYD	MRPS7
EXT2	GM2A	JAG1	MTFMT
FA2H	GNE	KCNJ11	MTHFD1
FAH	GNMT	L2HGDH	MTHFR
FAR1	GNPAT	LAMA1	MTO1
FARS2	GNPTAB	LAMP2	MTPAP
FASTKD2	GNPTG	LARS2	MTR
FBP1	GNS	LBR	MTRR
FBXL4	GPR161	LDHA	MUT
FDX1L	GRHPR	LHX4	MVK
FECH	GRN	LIAS	MYO5A
FH	GSS	LIPA	NAA10
FIG4	GTPBP3	LIPE	NADK2
FLAD1	GUSB	LIPT1	NAGA
FMO3	GYG1	LIPT2	NAGLU
FOLR1	GYG2	LMBRD1	NAGS
FOXE1	GYS1	LPIN1	NALCN
FOXRED1	GYS2	LRPPRC	NARS2
FTCD	HADH	LYRM4	NBAS
FTL	HADHA	MAN2B1	NDST1
FUCA1	HADHB	MANBA	NDUFA1
FXN	HARS	MAOA	NDUFA10
G6PC	HCFC1	MARS2	NDUFA11
G6PD	HEPACAM	MAT1A	NDUFA12
GAA	HEXA	MBD5	NDUFA13
GABRG2	HEXB	MCAT	NDUFA2
GALC	HGD	MCCC1	NDUFA4
GALE	HGSNAT	MCCC2	NDUFA6
GALK1	HIBCH	MCEE	NDUFA8
GALNS	HLCS	MCOLN1	NDUFA9
GALNT3	HMBS	MFF	NDUFAF1
GALT	HMGCL	MFN2	NDUFAF2
GAMT	HMGCS2	MFSD8	NDUFAF3
GARS	HOGA1	MGAT2	NDUFAF4
GATM	HPD	MGME1	NDUFAF5
GBA	HPRT1	MLC1	NDUFAF6
GBE1	HSD17B10	MLPH	NDUFB1
GCDH	HSD17B4	MLYCD	NDUFB11
GCH1	HSD3B7	MMAA	NDUFB3
GCK	HSPD1	MMAB	NDUFB6
GCLC	HYAL1	MMACHC	NDUFB9
GCSH	IARS2	MMADHC	NDUFC2
GFER	IBA57	MOGS	NDUFS1
GFM1	IDH2	MPC1	NDUFS2
GIF	IDS	MPDU1	NDUFS3
GLA	IDUA	MPI	NDUFS4
GLB1	INSR	MPV17	NDUFS5

NDUFS6	PEX16	PYCR2	SLC35A1
NDUFS7	PEX19	PYGL	SLC35A2
NDUFS8	PEX2	PYGM	SLC35C1
NDUFV1	PEX26	QARS	SLC37A4
NDUFV2	PEX3	QDPR	SLC39A4
NDUFV3	PEX5	QRSL1	SLC3A1
NEU1	PEX6	RAB27A	SLC46A1
NFS1	PEX7	RAB3GAP1	SLC52A1
NFU1	PFKM	RANBP2	SLC52A2
NKX2-1	PGAM2	RARS	SLC52A3
NKX2-5	PGAP2	RARS2	SLC5A1
NOTCH2	PGK1	RFT1	SLC5A2
NPC1	PGM1	RMND1	SLC5A5
NPC2	PHGDH	RNASEH1	SLC6A17
NSDHL	PHKA1	RPIA	SLC6A19
NT5C3A	PHKA2	RRM2B	SLC6A3
NUBPL	PHKB	SARS2	SLC6A8
NUS1	PHKG2	SC5D	SLC7A7
OAT	PHYH	SCO1	SLC7A9
OGDH	PIGA	SCO2	SLC9A1
OPA1	PIGN	SCP2	SMPD1
OPA3	PIGO	SDHA	SMS
OPLAH	PIGQ	SDHAF1	SNX14
OTC	PIGT	SDHAF2	SPG7
OXCT1	PIGW	SDHB	SPR
PAH	PLA2G6	SDHC	SRD5A3
PANK2	PMM2	SDHD	SSR4
PARK7	PMPCA	SERAC1	ST3GAL5
PARS2	PNP	SGSH	STAT2
PAX8	PNPLA2	SLC16A1	STX3
PC	PNPLA6	SLC16A2	STXBP5L
PCBD1	PNPO	SLC17A5	SUCLA2
PCCA	PNPT1	SLC19A2	SUCLG1
PCCB	POLG	SLC19A3	SUMF1
PCLO	POLG2	SLC1A4	SUOX
PCYT1A	POMGNT1	SLC22A5	SURF1
PDHA1	POMT1	SLC25A1	SZT2
PDHB	PPOX	SLC25A12	TACO1
PDHX	PPP2R1A	SLC25A13	TALDO1
PDK3	PPP2R5D	SLC25A15	TARS2
PDP1	PPT1	SLC25A19	TAT
PDSS1	PRKAG2	SLC25A20	TAZ
PDSS2	PRODH	SLC25A22	TCN1
PEPD	PRPS1	SLC25A3	TCN2
PET100	PSAP	SLC25A32	TG
PEX1	PSPH	SLC25A4	TH
PEX10	PTCD1	SLC26A4	TIMM8A
PEX11B	PTS	SLC27A4	TJP2
PEX12	PURA	SLC2A1	TK2
PEX13	PUS1	SLC2A2	TKT
PEX14	PYCR1	SLC30A10	TMEM126A

TMEM165
TMEM240
TMEM70
TPK1
TPO
TPP1
TRIT1
TRMT5
TRMU
TRNT1
TRPM6
TSFM
TSHR
TTC19
TTPA
TUFM
TXN2
TYMP
UBQLN2
UGT1A1
UMPS
UPB1
UQCC2
UQCRB
UQCRQ
UROD
UROS
VARS2
VPS53
WDR45
WVOX
XDH
XPR1
YARS2
ZBTB42

Gener för epileptisk encefalopati (161st)

AARS	FOLR1	PIGN	SYNGAP1
ABAT	FOXG1	PIGO	SYP
ADAR	GABRA1	PIGQ	SZT2
ADRA2B	GABRB3	PIGT	TBC1D24
ADSL	GABRG2	PIGW	TCF4
AFG3L2	GAL	PLCB1	TPP1
AGTR2	GAMT	PNKP	TREX1
ALDH7A1	GATM	PNPO	TRPM6
ALG13	GCSH	POLG	TSC1
AMT	GLDC	PPT1	TSC2
ARHGEF9	GNAO1	PRICKLE1	UBE3A
ARX	GOSR2	PRICKLE2	WDR45
ASAH1	GRIA3	PRRT2	WWOX
ASNS	GRIN1	PURA	ZFYVE20
ATP13A2	GRIN2A	QARS	
ATP1A2	GRIN2B	RAB39B	
ATP1A3	GRN	RELN	
ATP6AP2	HCN1	RNASEH2A	
ATP7A	HNRNPU	RNASEH2B	
BRWD3	HSD17B10	RNASEH2C	
CACNA2D1	KCNA1	ROGDI	
CDKL5	KCNA2	SAMHD1	
CERS1	KCNB1	SCARB2	
CHD2	KCNC1	SCN1A	
CHRNA2	KCNH1	SCN1B	
CHRNA4	KCNJ10	SCN2A	
CHRNB2	KCNMA1	SCN3A	
CLN3	KCNQ2	SCN8A	
CLN5	KCNQ3	SCN9A	
CLN6	KCNT1	SERPIN1	
CLN8	KCTD7	SIK1	
CNKS2	KDM5C	SLC12A5	
CNNM2	KIF1A	SLC13A5	
CNTNAP2	LGI1	SLC1A4	
CPA6	LIAS	SLC25A22	
CSTB	LMNB2	SLC2A1	
CTSD	MAGI2	SLC35A2	
CTSF	MAPK10	SLC6A1	
DEAF1	MBD5	SLC6A8	
DEPDC5	MECP2	SLC9A6	
DNAJC5	MEF2C	SPTAN1	
DNM1	MFSD8	SRPX2	
DOCK7	NECAP1	ST3GAL3	
DOLK	NHLRC1	ST3GAL5	
DPM1	NR2F1	STX1B	
DPYD	NRXN1	STXBP1	
DYRK1A	PAK3	STYXL1	
EEF1A2	PCDH19	SUMF1	
EPM2A	PIGA	SYN1	

Gener för mitokondriella sjukdomar (166st), alla ingår i listan för medfödda metabola sjukdomar

AARS2	GARS	NDUFS2	TACO1
ACAD9	GFER	NDUFS3	TARS2
ADCK3	GFM1	NDUFS4	TAZ
AFG3L2	GTPBP3	NDUFS5	TIMM8A
AGK	HSPD1	NDUFS6	TK2
AIFM1	IARS2	NDUFS7	TMEM126A
ANTXR2	IBA57	NDUFS8	TMEM70
APOPT1	ISCA2	NDUFV1	TRIT1
ATP5E	ISCU	NDUFV2	TRMT5
ATPAF2	LARS2	NDUFV3	TRMU
BCS1L	LRPPRC	NFS1	TSFM
BOLA3	MCAT	NFU1	TTC19
C10orf2	MFF	NUBPL	TUFM
C11orf83	MFN2	OPA1	TYMP
C12orf65	MGME1	OPA3	UQCC2
CA5A	MPC1	PARK7	UQCRB
CARS2	MPV17	PARS2	UQCRQ
CEP89	MRPL12	PDK3	VARS2
CHCHD10	MRPL44	PDSS1	YARS2
CLPB	MRPS16	PDSS2	
CLPP	MRPS22	PET100	
COA5	MRPS7	PNPT1	
COA6	MTO1	POLG	
COASY	MTPAP	POLG2	
COQ2	NADK2	PTCD1	
COQ4	NARS2	PUS1	
COQ9	NDUFA1	QRSL1	
COX10	NDUFA10	RARS2	
COX15	NDUFA11	RMND1	
COX4I2	NDUFA12	RNASEH1	
COX6B1	NDUFA13	RRM2B	
DARS2	NDUFA2	SARS2	
DCAF17	NDUFA4	SCO1	
DGUOK	NDUFA6	SCO2	
DNA2	NDUFA8	SDHA	
DNAJC19	NDUFA9	SDHAF1	
DNM1L	NDUFAF1	SDHAF2	
EARS2	NDUFAF2	SDHB	
ECHS1	NDUFAF3	SDHC	
ECSIT	NDUFAF4	SDHD	
ELAC2	NDUFAF5	SLC25A1	
EPG5	NDUFAF6	SLC25A19	
ETHE1	NDUFB1	SLC25A3	
FARS2	NDUFB11	SLC25A32	
FASTKD2	NDUFB3	SLC25A4	
FDX1L	NDUFB6	SPG7	
FOXRED1	NDUFB9	SUCLA2	
FTL	NDUFC2	SUCLG1	
FXN	NDUFS1	SURF1	