

dbCMMS.v6

Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild (610st)

AARS2	ALG2	CBS	DBH
AASS	ALG3	CCDC88C	DBT
ABAT	ALG6	CD320	DCAF17
ABCC8	ALG8	CEP89	DDC
ABCD1	ALG9	CERS1	DECR1
ABCD4	AMACR	CHCHD10	DGUOK
ABHD12	AMN	CHKB	DHCR24
ABHD5	AMPD1	CLDN16	DHCR7
ACAD8	AMPD2	CLN3	DHFR
ACAD9	AMT	CLN5	DIAPH1
ACADM	ANTXR2	CLN6	DLAT
ACADS	AP1S1	CLN8	DLD
ACADSB	APOPT1	CLPB	DMGDH
ACADVL	APRT	CLPP	DNA2
ACAT1	ARG1	CNKSR2	DNAJC19
ACO2	ARSA	COA5	DNAJC3
ACOX1	ARSB	COA6	DNAJC5
ADA	ASAH1	COASY	DNM1L
ADAR	ASL	COG1	DOLK
ADCK3	ASNS	COG2	DPAGT1
ADK	ASPA	COG4	DPM1
ADSL	ASS1	COG5	DPM3
AFG3L2	ATIC	COG7	DPYD
AGA	ATP13A2	COG8	DPYS
AGK	ATP5E	COQ2	DUOX2
AGL	ATP5L	COQ4	DUOXA2
AGPS	ATP6V0A2	COQ9	EARS2
AGXT	ATP7A	COX10	EBP
AHCY	ATP7B	COX15	ECHS1
AIFM1	ATPAF2	COX4I2	ECSIT
AIFM2	AUH	COX6B1	EIF2B1
AIFM3	B4GALNT1	CP	EIF2B2
AKR1D1	B4GALT1	CPOX	EIF2B3
ALAD	B4GALT7	CPS1	EIF2B4
ALAS2	BAAT	CPT1A	EIF2B5
ALDH18A1	BCKDHA	CPT2	ELAC2
ALDH3A2	BCKDHB	CTH	ELOVL4
ALDH4A1	BCKDK	CTNS	ELOVL5
ALDH5A1	BCS1L	CTSA	ENO3
ALDH6A1	BOLA3	CTSD	EPG5
ALDH7A1	BTD	CTSF	EPHX1
ALDOA	C10orf2	CTSK	EPM2A
ALDOB	C12orf65	CUBN	ERCC6
ALG1	C19orf12	CYP27A1	ETFA
ALG11	CA5A	CYP7B1	ETFB
ALG12	CACNA1A	D2HGDH	ETFDH
ALG13	CARS2	DARS2	ETHE1

EXOSC8	GM2A	JAG1	MTRR
EXT1	GNE	KCNJ11	MUT
EXT2	GNMT	L2HGDH	MVK
FA2H	GNPAT	LAMA1	MYO5A
FAH	GNPTAB	LAMP2	NAA10
FAR1	GNPTG	LARS2	NADK2
FARS2	GNS	LBR	NAGA
FASTKD2	GRHPR	LDHA	NAGLU
FBP1	GRN	LIAS	NAGS
FBXL4	GSS	LIPE	NALCN
FDX1L	GTPBP3	LIPT1	NARS2
FECH	GUSB	LMBRD1	NDST1
FH	GYG1	LPIN1	NDUFA1
FIG4	GYS1	LRPPRC	NDUFA10
FMO3	GYS2	LYRM4	NDUFA11
FOLR1	HADH	MAN2B1	NDUFA12
FOXE1	HADHA	MANBA	NDUFA13
FOXRED1	HADHB	MAOA	NDUFA2
FTCD	HARS	MAT1A	NDUFA3
FTL	HCFC1	MBD5	NDUFA4
FUCA1	HEPACAM	MCAT	NDUFA5
FXN	HEXA	MCCC1	NDUFA6
G6PC	HEXB	MCCC2	NDUFA7
G6PD	HGD	MCEE	NDUFA8
GAA	HGSNAT	MCOLN1	NDUFA9
GABRG2	HIBADH	MFN2	NDUFAB1
GALC	HIBCH	MFSD8	NDUFAF1
GALE	HLCS	MGAT2	NDUFAF2
GALK1	HMBS	MGME1	NDUFAF3
GALNS	HMGCL	MLC1	NDUFAF4
GALNT3	HMGCS2	MLPH	NDUFAF5
GALT	HOGA1	MLYCD	NDUFAF6
GAMT	HPD	MMAA	NDUFB1
GARS	HPRT1	MMAB	NDUFB10
GATM	HSD17B10	MMACHC	NDUFB11
GBA	HSD17B4	MMADHC	NDUFB2
GBE1	HSD3B7	MOGS	NDUFB3
GCDH	HSPD1	MPDU1	NDUFB4
GCH1	HSPE1	MPI	NDUFB5
GCK	HYAL1	MPV17	NDUFB6
GCLC	IARS2	MRPL12	NDUFB7
GCSH	IBA57	MRPL44	NDUFB8
GFER	IDH2	MRPS16	NDUFB9
GFM1	IDS	MRPS22	NDUFC1
GIF	IDUA	MRPS7	NDUFC2
GLA	INSR	MTFMT	NDUFS1
GLB1	ISCA2	MTHFD1	NDUFS2
GLDC	ISCU	MTHFR	NDUFS3
GLRA1	ISPD	MTO1	NDUFS4
GLUD1	IVD	MTPAP	NDUFS5
GLUL	IYD	MTR	NDUFS6

NDUFS7	PEX3	RARS	SLC6A19
NDUFS8	PEX5	RARS2	SLC6A3
NDUFV1	PEX6	RFT1	SLC6A8
NDUFV2	PEX7	RMND1	SLC7A7
NDUFV3	PFKM	RPIA	SLC7A9
NEU1	PGAM2	RRM2B	SMPD1
NFS1	PGAP2	SARS2	SMS
NFU1	PGK1	SC5D	SNX14
NKX2-1	PGM1	SCO1	SPG7
NKX2-5	PHGDH	SCO2	SPR
NOTCH2	PHKA1	SCP2	SRD5A3
NPC1	PHKA2	SDHA	ST3GAL5
NPC2	PHKB	SDHAF1	SUCLA2
NSDHL	PHKG2	SDHAF2	SUCLG1
NT5C3A	PHYH	SDHB	SUMF1
NUBPL	PIGA	SDHC	SUOX
OAT	PIGN	SDHD	SURF1
OGDH	PIGO	SERAC1	SZT2
OPA1	PIGQ	SGSH	TACO1
OPA3	PIGT	SLC16A1	TALDO1
OPLAH	PIGW	SLC16A2	TARS2
OTC	PLA2G6	SLC17A5	TAT
OXCT1	PMM2	SLC19A2	TAZ
PAH	PNP	SLC19A3	TCN1
PANK2	PNPLA2	SLC22A5	TCN2
PARK7	PNPLA6	SLC25A12	TG
PARS2	PNPO	SLC25A13	TH
PAX8	PNPT1	SLC25A15	TIMM8A
PC	POLG	SLC25A19	TJP2
PCBD1	POLG2	SLC25A20	TK2
PCCA	POMGNT1	SLC25A22	TMEM126A
PCCB	POMT1	SLC25A3	TMEM165
PCYT1A	PPOX	SLC25A4	TMEM240
PDHA1	PPT1	SLC26A4	TMEM70
PDHB	PRKAG2	SLC27A4	TPK1
PDHX	PRODH	SLC2A1	TPO
PDP1	PRPS1	SLC2A2	TPP1
PDSS1	PSAP	SLC30A10	TRIT1
PDSS2	PSPH	SLC35A1	TRMU
PEPD	PTCD1	SLC35A2	TRNT1
PET100	PTS	SLC35C1	TRPM6
PEX1	PURA	SLC37A4	TSFM
PEX10	PUS1	SLC39A4	TSHR
PEX11B	PYCR1	SLC3A1	TTC19
PEX12	PYGL	SLC46A1	TTPA
PEX13	PYGM	SLC52A1	TUFM
PEX14	QARS	SLC52A2	TYMP
PEX16	QDPR	SLC52A3	UBQLN2
PEX19	RAB27A	SLC5A1	UGT1A1
PEX2	RAB3GAP1	SLC5A2	UMPS
PEX26	RANBP2	SLC5A5	UPB1

UQCC2
UQCC3
UQCRB
UQCRQ
UROD
UROS
VARS2
WDR45
VPS53
VTA1
WVOX
XDH
YARS2
ZBTB42

Gener för epileptisk encefalopati (138st)

ABAT	GOSR2	RBSN
ADAR	GRIA3	RNASEH2A
ADSL	GRIN2A	RNASEH2B
AGTR2	GRIN2B	RNASEH2C
ALDH7A1	GRN	SAMHD1
ALG13	HCN1	SCARB2
AMT	HNRNPU	SCN1A
ARHGEF9	HSD17B10	SCN1B
ARX	KCNA1	SCN2A
ASNS	KCNA2	SCN3A
ATP13A2	KCNB1	SCN8A
ATP1A2	KCNC1	SCN9A
ATP1A3	KCNH1	SERPINI1
ATP6AP2	KCNJ10	SLC12A5
ATP7A	KCNMA1	SLC13A5
CDKL5	KCNQ2	SLC25A22
CERS1	KCNQ3	SLC2A1
CHD2	KCNT1	SLC35A2
CHRNA2	KCTD7	SLC6A8
CHRNA4	KDM5C	SLC9A6
CHRNA4	LGI1	SPTAN1
CLN3	LIAS	SRPX2
CLN5	MAGI2	ST3GAL3
CLN6	MAPK10	ST3GAL5
CLN8	MBD5	STX1B
CNKSR2	MECP2	STXBP1
CNNM2	MEF2C	SUMF1
CNTNAP2	MFSD8	SYN1
CPA6	NECAP1	SYNGAP1
CSTB	NHLRC1	SYP
CTSD	NRXN1	SZT2
CTSF	PAK3	TBC1D24
DEPDC5	PCDH19	TCF4
DNAJC5	PIGA	TPP1
DNM1	PIGN	TREX1
DOCK7	PIGO	TRPM6
DOLK	PIGQ	TSC1
DPYD	PIGW	TSC2
EPM2A	PLCB1	UBE3A
FOLR1	PNKP	WWOX
FOXG1	PNPO	
GABRA1	POLG	
GABRB3	PPT1	
GABRG2	PRICKLE1	
GAMT	PRICKLE2	
GATM	PRRT2	
GCSH	PURA	
GLDC	QARS	
GNAO1	RAB39B	

Gener för mitokondriella sjukdomar (173st), alla ingår i listan för medfödda metabola sjukdomar

AARS2	FTL	NDUFB1	SLC25A19
ACAD9	FXN	NDUFB2	SLC25A3
ADCK3	GARS	NDUFB3	SLC25A4
AFG3L2	GFER	NDUFB4	SPG7
AGK	GFM1	NDUFB5	SUCLA2
AIFM1	HSPD1	NDUFB6	SUCLG1
AIFM2	HSPE1	NDUFB7	SURF1
AIFM3	IARS2	NDUFB8	TACO1
ANTXR2	IBA57	NDUFB9	TARS2
APOPT1	CLPB	NDUFC1	TAZ
ATP5E	ISCA2	NDUFC2	TIMM8A
ATP5L	GTPBP3	NDUFS1	TK2
ATPAF2	COQ4	NDUFS2	TMEM126A
BCS1L	ISCU	NDUFS3	TMEM70
BOLA3	LARS2	NDUFS4	TRIT1
C10orf2	LRPPRC	NDUFS5	TRMU
C12orf65	MCAT	NDUFS6	TSMF
NDUFAF5	MFN2	NDUFS7	TTC19
NDUFAF6	MGME1	NDUFS8	TUFM
CA5A	MPV17	NDUFV1	TYMP
CARS2	MRPL12	NDUFV2	UQCC2
CEP89	MRPL44	NDUFV3	UQCC3
CHCHD10	MRPS16	NFS1	UQCRB
CLPP	MRPS22	NFU1	UQCRQ
COA5	MRPS7	NUBPL	VARA2
COA6	MTO1	OPA1	YARS2
COASY	MTPAP	OPA3	
COQ2	NADK2	PARK7	
COQ9	NARS2	PARS2	
COX10	NDUFA10	PDSS1	
COX15	NDUFA11	PDSS2	
COX4I2	NDUFA12	PET100	
COX6B1	NDUFA13	PNPT1	
DARS2	NDUFA1	POLG2	
DCAF17	NDUFA2	POLG	
DGUOK	NDUFA3	PTCD1	
DNA2	NDUFA4	PUS1	
DNAJC19	NDUFA5	RARS2	
DNM1L	NDUFA6	RMND1	
EARS2	NDUFA7	RRM2B	
ECHS1	NDUFA8	SARS2	
ECSIT	NDUFA9	SCO1	
ELAC2	NDUFAB1	SCO2	
EPG5	NDUFAF1	SDHAF1	
ETHE1	NDUFAF2	SDHAF2	
FARS2	NDUFAF3	SDHA	
FASTKD2	NDUFAF4	SDHB	
FDX1L	NDUFB10	SDHC	
FOXRED1	NDUFB11	SDHD	