

## dbCMMS.v1.1

Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild (526st)

AARS2	ALG3	CLDN16	DNAJC5
AASS	ALG6	CLN3	DNM1L
ABAT	ALG8	CLN5	DOLK
ABCC8	ALG9	CLN6	DPAGT1
ABCD1	AMACR	CLN8	DPM1
ABCD4	AMN	COA5	DPM3
ABHD12	AMPD1	COG1	DPYD
ABHD5	AMT	COG4	DPYS
ACAD8	AP1S1	COG5	EBP
ACAD9	APRT	COG7	ECSIT
ACADM	ARG1	COG8	EIF2B1
ACADS	ARSA	COQ2	EIF2B2
ACADSB	ARSB	COQ9	EIF2B3
ACADVL	ASAH1	COX10	EIF2B4
ACAT1	ASL	COX15	EIF2B5
ACO2	ASPA	COX4I2	ELAC2
ACOX1	ASS1	COX6B1	ELOVL4
ADA	ATIC	CP	ENO3
ADCK3	ATP13A2	CPOX	EPHX1
ADK	ATP5E	CPS1	EPM2A
ADSL	ATP5L	CPT1A	ETFA
AFG3L2	ATP6V0A2	CPT2	ETFB
AGA	ATP7A	CTH	ETFDH
AGK	ATP7B	CTNS	ETHE1
AGL	ATPAF2	CTSA	EXT1
AGPS	AUH	CTSD	EXT2
AGXT	B4GALT1	CTSF	FA2H
AHCY	B4GALT7	CTSK	FAH
AIFM1	BAAT	CUBN	FASTKD2
AIFM2	BCKDHA	CYP27A1	FBP1
AIFM3	BCKDHB	CYP7B1	FBXL4
AKR1D1	BCKDK	D2HGDH	FECH
ALAD	BCS1L	DARS2	FH
ALAS2	BOLA3	DBH	FIG4
ALDH18A1	BTD	DBT	FMO3
ALDH3A2	C10orf2	DDC	FOLR1
ALDH4A1	C12orf65	DECR1	FOXRED1
ALDH5A1	C19orf12	DGUOK	FTCD
ALDH6A1	C20orf7	DHCR24	FUCA1
ALDH7A1	C8orf38	DHCR7	FXN
ALDOA	CA5A	DHFR	G6PC
ALDOB	CACNA1A	DLAT	G6PD
ALG1	CBS	DLD	GAA
ALG11	CD320	DMGDH	GABRG2
ALG12	CEP89	DNA2	GALC
ALG2	CHKB	DNAJC19	GALE

GALK1	HMGCL	MOGS	NDUFC1
GALNS	HMGCS2	MPDU1	NDUFC2
GALNT3	HOGA1	MPI	NDUFS1
GALT	HPD	MPV17	NDUFS2
GAMT	HPRT1	MRPL12	NDUFS3
GATM	HSD17B10	MRPL44	NDUFS4
GBA	HSD17B4	MRPS16	NDUFS5
GBE1	HSD3B7	MRPS22	NDUFS6
GCDH	HSPD1	MTFMT	NDUFS7
GCH1	HYAL1	MTHFD1	NDUFS8
GCK	IBA57	MTHFR	NDUFV1
GCLC	IDH2	MTO1	NDUFV2
GCSH	IDS	MTPAP	NDUFV3
GFER	IDUA	MTR	NEU1
GFM1	INSR	MTRR	NFU1
GIF	ISCU	MUT	NOTCH2
GLA	ISPD	MVK	NPC1
GLB1	IVD	NAGA	NPC2
GLDC	JAG1	NAGLU	NSDHL
GLRA1	KCNJ11	NAGS	NT5C3
GLUD1	L2HGDH	NALCN	NUBPL
GLUL	LAMP2	NDUFA1	OAT
GM2A	LARS2	NDUFA10	OGDH
GNE	LBR	NDUFA11	OPA1
GNMT	LDHA	NDUFA12	OPA3
GNPAT	LIAS	NDUFA13	OPLAH
GNPTAB	LIPT1	NDUFA2	OTC
GNPTG	LMBRD1	NDUFA3	OXCT1
GNS	LPIN1	NDUFA4	PAH
GRHPR	LRPPRC	NDUFA5	PANK2
GRN	LYRM4	NDUFA6	PARK7
GSS	MAN2B1	NDUFA7	PC
GUSB	MANBA	NDUFA8	PCBD1
GYG1	MAOA	NDUFA9	PCCA
GYS1	MAT1A	NDUFAB1	PCCB
GYS2	MCAT	NDUFAF1	PDHA1
HADH	MCCC1	NDUFAF2	PDHB
HADHA	MCCC2	NDUFAF3	PDHX
HADHB	MCEE	NDUFAF4	PDP1
HARS	MCOLN1	NDUFB1	PDSS1
HCFC1	MFN2	NDUFB10	PDSS2
HEPACAM	MFSD8	NDUFB11	PEPD
HEXA	MGAT2	NDUFB2	PET100
HEXB	MGME1	NDUFB3	PEX1
HGD	MLC1	NDUFB4	PEX10
HGSNAT	MLYCD	NDUFB5	PEX12
HIBADH	MMAA	NDUFB6	PEX13
HIBCH	MMAB	NDUFB7	PEX14
HLCS	MMACHC	NDUFB8	PEX16
HMBS	MMADHC	NDUFB9	PEX19

<i>PEX2</i>	<i>SCO1</i>	<i>ST3GAL5</i>
<i>PEX26</i>	<i>SCO2</i>	<i>SUCLA2</i>
<i>PEX3</i>	<i>SCP2</i>	<i>SUCLG1</i>
<i>PEX5</i>	<i>SDHA</i>	<i>SUOX</i>
<i>PEX6</i>	<i>SDHAF1</i>	<i>SURF1</i>
<i>PEX7</i>	<i>SDHAF2</i>	<i>SZT2</i>
<i>PFKM</i>	<i>SDHB</i>	<i>TACO1</i>
<i>PGAM2</i>	<i>SDHC</i>	<i>TALDO1</i>
<i>PGAP2</i>	<i>SDHD</i>	<i>TAT</i>
<i>PGK1</i>	<i>SERAC1</i>	<i>TAZ</i>
<i>PGM1</i>	<i>SGSH</i>	<i>TCN1</i>
<i>PHGDH</i>	<i>SLC16A1</i>	<i>TCN2</i>
<i>PHKA1</i>	<i>SLC16A2</i>	<i>TH</i>
<i>PHKA2</i>	<i>SLC17A5</i>	<i>TIMM8A</i>
<i>PHKB</i>	<i>SLC19A2</i>	<i>TJP2</i>
<i>PHKG2</i>	<i>SLC19A3</i>	<i>TK2</i>
<i>PHYH</i>	<i>SLC22A5</i>	<i>TMEM126A</i>
<i>PIGT</i>	<i>SLC25A12</i>	<i>TMEM165</i>
<i>PLA2G6</i>	<i>SLC25A13</i>	<i>TMEM70</i>
<i>PMM2</i>	<i>SLC25A15</i>	<i>TPK1</i>
<i>PNP</i>	<i>SLC25A19</i>	<i>TPP1</i>
<i>PNPLA6</i>	<i>SLC25A20</i>	<i>TRMU</i>
<i>PNPO</i>	<i>SLC25A22</i>	<i>TRPM6</i>
<i>PNPT1</i>	<i>SLC25A3</i>	<i>TSFM</i>
<i>POLG</i>	<i>SLC25A4</i>	<i>TTC19</i>
<i>POLG2</i>	<i>SLC27A4</i>	<i>TTPA</i>
<i>POMGNT1</i>	<i>SLC2A1</i>	<i>TUFM</i>
<i>POMT1</i>	<i>SLC2A2</i>	<i>TYMP</i>
<i>PPOX</i>	<i>SLC30A10</i>	<i>UGT1A1</i>
<i>PPT1</i>	<i>SLC35A1</i>	<i>UGT1A1</i>
<i>PRKAG2</i>	<i>SLC35C1</i>	<i>UMPS</i>
<i>PRODH</i>	<i>SLC37A4</i>	<i>UPB1</i>
<i>PRPS1</i>	<i>SLC39A4</i>	<i>UQCC2</i>
<i>PSAP</i>	<i>SLC3A1</i>	<i>UQCRB</i>
<i>PSPH</i>	<i>SLC46A1</i>	<i>UQCRQ</i>
<i>PTS</i>	<i>SLC52A2</i>	<i>UROD</i>
<i>PUS1</i>	<i>SLC52A3</i>	<i>UROS</i>
<i>PYCR1</i>	<i>SLC5A1</i>	<i>WDR45</i>
<i>PYGL</i>	<i>SLC5A2</i>	<i>VTA1</i>
<i>PYGM</i>	<i>SLC6A19</i>	<i>WVOX</i>
<i>QDPR</i>	<i>SLC6A3</i>	<i>XDH</i>
<i>RAB3GAP1</i>	<i>SLC6A8</i>	<i>YARS2</i>
<i>RANBP2</i>	<i>SLC7A7</i>	
<i>RARS2</i>	<i>SLC7A9</i>	
<i>RFT1</i>	<i>SMPD1</i>	
<i>RMND1</i>	<i>SMS</i>	
<i>RPIA</i>	<i>SPG7</i>	
<i>RRM2B</i>	<i>SPR</i>	
<i>SARS2</i>	<i>SRD5A3</i>	
<i>SC5DL</i>	<i>SSR3</i>	

**Gener för epileptisk encefalopati (120st)**

ABAT	KCNMA1	SCN2A
ADAR	KCNQ2	SCN3A
ADSL	KCNQ3	SCN8A
AGTR2	KCNT1	SCN9A
ALDH7A1	KCTD7	SERPINI1
AMT	KDM5C	SLC12A5
ARHGEF9	LGI1	SLC25A22
ARX	LIAS	SLC2A1
ATP13A2	MAGI2	SLC6A8
ATP1A2	MAPK10	SLC9A6
ATP6AP2	MECP2	SPTAN1
ATP7A	MEF2C	SRPX2
CDKL5	MFSD8	ST3GAL3
CHD2	NHLRC1	ST3GAL5
CHRNA2	NRXN1	STXBP1
CHRNA4	PAK3	SYN1
CHRN2	PCDH19	SYNGAP1
CLN3	PEX1	SYP
CLN5	PEX10	SZT2
CLN6	PEX12	TBC1D24
CLN8	PEX13	TCF4
CNTNAP2	PEX14	TPP1
CPA6	PEX16	TREX1
CSTB	PEX19	TRPM6
CTSD	PEX2	TSC1
CTSF	PEX26	TSC2
DNAJC5	PEX3	UBE3A
DOLK	PEX5	WWOX
DPYD	PEX6	
EPM2A	PEX7	
FOLR1	PLCB1	
FOXP1	PNKP	
GABRB3	PNPO	
GABRG2	POLG	
GAMT	PPT1	
GATM	PRICKLE1	
GCSH	PRICKLE2	
GLDC	PRRT2	
GNAO1	RAB39B	
GOSR2	RNASEH2A	
GRIA3	RNASEH2B	
GRIN2A	RNASEH2C	
GRN	SAMHD1	
HSD17B10	SCARB2	
KCNA1	SCN1A	
KCNJ10	SCN1B	

49 gener ingår i båda listorna