

**IEM-panel (910 st):****Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild**

AARS2	ALDH7A1	ATP5MD	CLN5	CTNS
AASS	ALDOA	ATP6AP1	CLN6	CTSA
ABAT	ALDOB	ATP6V0A2	CLN8	CTSD
ABCC8	ALG1	ATP7A	CLPB	CTSF
ABCD1	ALG11	ATP7B	CLPP	CTSK
ABCD4	ALG12	ATPAF2	CLTC	CUBN
ABHD12	ALG13	AUH	CMPK2	CWF19L1
ABHD5	ALG14	B4GALNT1	CNKSR2	CYP27A1
ACAD8	ALG2	B4GALT1	COA5	CYP7B1
ACAD9	ALG3	B4GALT7	COA6	D2HGDH
ACADM	ALG6	BAAT	COA7	DARS
ACADS	ALG8	BBOX1	COASY	DARS2
ACADSB	ALG9	BCAT2	COG1	DBH
ACADVL	ALKBH1	BCKDHA	COG2	DBT
ACAT1	ALPL	BCKDHB	COG4	DCAF17
ACER3	AMACR	BCKDK	COG5	DDC
ACO2	AMN	BCS1L	COG6	DEGS1
ACOX1	AMPD1	BLOC1S3	COG7	DGAT1
ACSF3	AMPD2	BOLA3	COG8	DGUOK
ACY1	AMT	BSCL2	COLGALT1	DHCR24
ADA	ANO10	BTD	COQ2	DHCR7
ADAR	ANTXR2	C12orf65	COQ4	DHDDS
ADCK2	AP1S1	C19orf12	COQ5	DHFR
ADCY5	AP3B1	C1QBP	COQ6	DHODH
ADK	AP3D1	CA5A	COQ7	DHPS
ADPRHL2	AP5Z1	CACNA1A	COQ8A	DHTKD1
ADSL	APOPT1	CACNA1E	COQ9	DIAPH1
ADSSL1	APRT	CACNA1S	COX10	DLAT
AFG3L2	ARG1	CAD	COX14	DLD
AGA	ARL3	CARS	COX15	DMGDH
AGK	ARNT2	CARS2	COX20	DNA2
AGL	ARSA	CAT	COX4I1	DNAJC12
AGPS	ARSB	CBLIF	COX4I2	DNAJC19
AGTPBP1	ASAH1	CBS	COX5A	DNAJC3
AGXT	ASL	CCDC115	COX6A2	DNAJC5
AHCY	ASNS	CCDC174	COX6B1	DNM1L
AIFM1	ASPA	CCDC88C	COX8A	DOLK
AIMP1	ASS1	CD320	CP	DPAGT1
AKR1D1	ATAD3A	CEP89	CPOX	DPM1
ALAD	ATAD3B	CERS1	CPS1	DPM2
ALAS2	ATAD3C	CHCHD10	CPT1A	DPM3
ALDH18A1	ATIC	CHKB	CPT2	DPYD
ALDH3A2	ATP13A2	CISD2	CRAT	DPYS
ALDH4A1	ATP1A1	CLCN2	CSF1R	DTNBP1
ALDH5A1	ATP5F1D	CLDN16	CTCF	DTYMK
ALDH6A1	ATP5F1E	CLN3	CTH	EARS2

EBP	FTCD	GNE	HSD17B10	LPIN1
ECHS1	FTL	GNMT	HSD17B4	LRP2
ECM1	FUCA1	GNPAT	HSD3B7	LRPPRC
ECSIT	FUT8	GNPTAB	HSPD1	LYRM4
EIF2B1	FXN	GNPTG	HTRA2	LYRM7
EIF2B2	G6PC	GNS	HYAL1	MAG
EIF2B3	G6PD	GOT2	HYOU1	MAGED2
EIF2B4	GAA	GPD1	IARS	MAGEL2
EIF2B5	GABBR2	GPHN	IARS2	MAGT1
ELAC2	GABRG2	GPR161	IBA57	MAN1B1
ELOVL4	GALC	GRHPR	IDH2	MAN2B1
ELOVL5	GALE	GRN	IDH3A	MANBA
ENO3	GALK1	GSS	IDI1	MAOA
EPG5	GALM	GSX2	IDS	MARS
EPHX1	GALNS	GTPBP3	IDUA	MARS2
EPM2A	GALNT3	GUSB	IER3IP1	MAT1A
EPRS	GALT	GYG1	IFIH1	MBD5
ERAL1	GAMT	GYG2	IGSF1	MCAT
ERCC6	GARS	GYS1	INSR	MCCC1
ETFA	GATB	GYS2	IRF2BPL	MCCC2
ETFB	GATC	HAAO	ISCA1	MCEE
ETFDH	GATM	HACE1	ISCA2	MCOLN1
ETHE1	GBA	HADH	ISCU	MDH1
EXOSC3	GBA2	HADHA	ISPD	MECR
EXOSC8	GBE1	HADHB	ITPA	MFF
EXT1	GCDH	HAL	IVD	MFN2
EXT2	GCH1	HARS	JAG1	MFSD8
FA2H	GCK	HCFC1	KARS	MGAT2
FAH	GCLC	HEPACAM	KCNA4	MGME1
FAM126A	GCSH	HEXA	KCNJ11	MICOS13
FAM149B1	GFER	HEXB	KIF5A	MICU1
FAR1	GFM1	HGD	KYNU	MICU2
FARS2	GGT1	HGSNAT	L2HGDH	MIEF2
FARSB	GJC2	HIBCH	LAMA1	MIPEP
FASTKD2	GK	HIKESHI	LAMP2	MLC1
FBP1	GLA	HK1	LARS	MLPH
FBXL4	GLB1	HLCS	LARS2	MLYCD
FDFT1	GLDC	HMBS	LBR	MMAA
FDX2	GLRA1	HMGCL	LDHA	MMAB
FDXR	GLRX5	HMGCS2	LHX4	MMACHC
FECH	GLS	HOGA1	LIAS	MMADHC
FH	GLUD1	HPD	LIPA	MMUT
FIG4	GLUL	HPRT1	LIPE	MOCS1
FLAD1	GLYCTK	HPS1	LIPT1	MOCS2
FMO3	GM2A	HPS3	LIPT2	MOCS3
FOLR1	GMPPA	HPS4	LMBRD1	MOGS
FOXI1	GMPPB	HPS5	LNPK	MPC1
FOXRED1	GNAO1	HPS6	LONP1	MPDU1

MPI	NDUFA9	OPLAH	PGAP2	PPP1R15B
MPV17	NDUFAF1	OTC	PGK1	PPP1R21
MRM2	NDUFAF2	OXA1L	PGM1	PPP2R1A
MRPL12	NDUFAF3	OXCT1	PGM3	PPP2R5D
MRPL44	NDUFAF4	PAH	PHGDH	PPT1
MRPL57	NDUFAF5	PAK1	PHKA1	PRKAG2
MRPS14	NDUFAF6	PANK2	PHKA2	PRKN
MRPS16	NDUFAF8	PARK7	PHKB	PRMT7
MRPS2	NDUFB1	PARS2	PHKG2	PRODH
MRPS22	NDUFB10	PAX7	PHYH	PRODH2
MRPS23	NDUFB11	PAXBP1	PIGA	PRPS1
MRPS25	NDUFB3	PC	PIGM	PRUNE1
MRPS34	NDUFB6	PCBD1	PIGN	PSAP
MRPS36	NDUFB8	PCCA	PIGO	PSAT1
MRPS7	NDUFB9	PCCB	PIGQ	PSPH
MSL3	NDUFC2	PCK1	PIGS	PTCD1
MSTO1	NDUFS1	PCLO	PIGT	PTCD3
MTFMT	NDUFS2	PCYT1A	PIGV	PTS
MTHFD1	NDUFS3	PDE10A	PIGW	PURA
MTHFR	NDUFS4	PDE12	PIK3R4	PUS1
MTO1	NDUFS5	PDHA1	PITRM1	PUS7
MTPAP	NDUFS6	PDHB	PKLR	PYCR1
MTR	NDUFS7	PDHX	PLA2G6	PYCR2
MTRR	NDUFS8	PDK3	PLAA	PYGL
MVK	NDUFV1	PDP1	PLIN5	PYGM
MYO5A	NDUFV2	PDPR	PMM2	PYROXD1
MYORG	NDUFV3	PDSS1	PMPCA	QARS
NAA10	NEU1	PDSS2	PMPCB	QDPR
NADK2	NFASC	PDXK	PNP	QRSL1
NAGA	NFE2L2	PEPD	PNPLA2	RAB11B
NAGLU	NFS1	PET100	PNPLA4	RAB27A
NAGS	NFU1	PET117	PNPLA6	RAB3GAP1
NALCN	NGLY1	PEX1	PNPLA8	RANBP2
NANS	NKX6-2	PEX10	PNPO	RARS
NARS2	NOTCH2	PEX11B	PNPT1	RARS2
NAXD	NPC1	PEX12	POLG	REPS1
NAXE	NPC2	PEX13	POLG2	RFT1
NBAS	NSDHL	PEX14	POLR1A	RMND1
NDST1	NSUN3	PEX16	POLR1C	RNASEH1
NDUFA1	NT5C3A	PEX19	POLR3A	RNF13
NDUFA10	NUBPL	PEX2	POLR3B	RNF213
NDUFA11	NUP214	PEX26	POLRMT	RPIA
NDUFA12	NUS1	PEX3	POMGNT1	RRM2B
NDUFA13	OAT	PEX5	POMT1	RTN4IP1
NDUFA2	OCRL	PEX6	PPA2	RYR1
NDUFA4	OGDH	PEX7	PPCS	SARS
NDUFA6	OPA1	PFKM	PPM1K	SARS2
NDUFA8	OPA3	PGAM2	PPOX	SC5D

SCN8A	SLC37A4	SURF1	TRMT1	VPS53
SCO1	SLC39A14	SVBP	TRMT10C	WVOX
SCO2	SLC39A4	SYT1	TRMT5	XDH
SCP2	SLC39A8	SZT2	TRMU	XPR1
SCYL1	SLC3A1	TACO1	TRNT1	YARS
SDHA	SLC46A1	TAF1	TRPM6	YARS2
SDHAF1	SLC4A10	TALDO1	TSFM	ZBTB42
SDHAF2	SLC52A1	TANGO2	TTC19	ZNHIT3
SDHB	SLC52A2	TAOK1	TTPA	
SDHC	SLC52A3	TARS2	TUBB2A	
SDHD	SLC5A1	TAT	TUBB3	
SELENOI	SLC5A2	TAZ	TUBB4A	
SERAC1	SLC6A17	TBCD	TUBB4B	
SFXN4	SLC6A19	TBCK	TUFM	
SGSH	SLC6A3	TBL1X	TWNK	
SLC13A3	SLC6A8	TCN1	TXN2	
SLC16A1	SLC6A9	TCN2	TYMP	
SLC16A2	SLC7A1	TDO2	UBQLN2	
SLC17A5	SLC7A2	TFAM	UBTF	
SLC19A2	SLC7A7	TH	UFM1	
SLC19A3	SLC7A9	THAP11	UGT1A1	
SLC1A1	SLC9A1	TIMM50	UMPS	
SLC1A4	SLC9A7	TIMM8A	UNC80	
SLC20A2	SMARCD1	TIMMDC1	UPB1	
SLC22A5	SMPD1	TJP2	UQCC2	
SLC25A1	SMPD4	TK2	UQCC3	
SLC25A10	SMS	TKT	UQCRB	
SLC25A12	SNORD118	TMEM106B	UQCRC2	
SLC25A13	SNX14	TMEM126A	UQCRFS1	
SLC25A15	SOD1	TMEM126B	UQCRH	
SLC25A19	SORD	TMEM165	UQCRQ	
SLC25A20	SPG7	TMEM199	UROD	
SLC25A22	SPR	TMEM240	UROS	
SLC25A24	SPTBN4	TMEM260	USP7	
SLC25A26	SQOR	TMEM63A	VAC14	
SLC25A3	SQSTM1	TMEM70	VAMP2	
SLC25A32	SRD5A3	TMLHE	VAR5	
SLC25A4	SSBP1	TMTC3	VAR52	
SLC25A42	SSR4	TNNI3	WARS2	
SLC25A46	ST3GAL5	TOP3A	WDR45	
SLC27A4	STAT2	TPK1	WDR45B	
SLC2A1	STX3	TPP1	WFS1	
SLC2A2	STXBP5L	TRAPPC11	WIPI2	
SLC30A10	SUCLA2	TRAPPC12	VMA21	
SLC30A9	SUCLG1	TRAPPC2L	VPS11	
SLC35A1	SUGCT	TRIAP1	VPS13D	
SLC35A2	SUMF1	TRIM37	VPS33A	
SLC35C1	SUOX	TRIT1	VPS51	

**EP-panel (379 st)**

Gener för epileptisk encefalopati, 115 ingår i listan för medfödda metabola sjukdomar

AARS	CACNA1C	DMXL2	GOSR2	KMT2A
ABAT	CACNA1D	DNAJA1	GOT2	KMT2E
ACTL6B	CACNA1E	DNAJC5	GPAA1	LGI1
ADAM22	CACNA1G	DNM1	GPHN	LIAS
ADAR	CACNA2D1	DNM1L	GPT2	LMNB2
ADCY5	CACNA2D2	DNMT3A	GRIA2	LNPK
ADRA2B	CAD	DOCK7	GRIA3	MAGEL2
ADSL	CAMK2A	DOLK	GRIA4	MAGI2
AFG3L2	CAMK2B	DPM1	GRIN1	MAPK10
AGTR2	CASK	DPM2	GRIN2A	MBD5
AIMP1	CDKL5	DPYD	GRIN2B	MBOAT7
AKT3	CERS1	DYNC1H1	GRIN2D	MDH2
ALDH5A1	CERT1	DYRK1A	GRN	MECP2
ALDH7A1	CHD2	EEF1A2	GUF1	MED13L
ALG1	CHRNA2	EHMT1	HACE1	MED17
ALG11	CHRNA4	EIF2S3	HCN1	MED23
ALG13	CHRNA7	EPM2A	HECW2	MEF2C
ALG14	CHRN2	FBXO28	HIVEP2	MFF
ALG3	CLCN4	FGF12	HNRNPH2	MFSD8
ALG6	CLN3	FGF13	HNRNPU	MID2
ALKBH8	CLN5	FGFR3	HSD17B10	MOCS1
AMT	CLN6	FLNA	IQSEC2	MOCS2
ANKRD11	CLN8	FOLR1	IRF2BPL	MTHFS
AP2M1	CLTC	FOXP1	ITPA	MTOR
AP3B2	CNKSR2	FOXP1	KANSL1	NAA10
AP4S1	CNNM2	FRRS1L	KCNA1	NACC1
APP	CNPY3	GABBR2	KCNA2	NAPB
ARHGEF9	CNTN2	GABRA1	KCNB1	NBEA
ARID1B	CNTNAP2	GABRA2	KCNC1	NECAP1
ARV1	COL4A1	GABRA3	KCND2	NEDD4
ARX	COL4A2	GABRA5	KCNH1	NEUROD2
ASAH1	CPA6	GABRB1	KCNH5	NEXMIF
ASNS	CPLX1	GABRB2	KCNJ10	NF1
ASXL3	CRADD	GABRB3	KCNMA1	NGLY1
ATP13A2	CSNK2B	GABRG2	KCNQ1	NHLRC1
ATP1A2	CSTB	GAL	KCNQ2	NPRL2
ATP1A3	CTSD	GAMT	KCNQ3	NPRL3
ATP6AP2	CTSF	GATM	KCNQ5	NR2F1
ATP6V1A	CUX2	GCSH	KCNT1	NRXN1
ATP7A	CYFIP2	GLDC	KCNT2	NTRK2
ATXN2	DCX	GLRA1	KCTD3	NUP214
BRAT1	DDX3X	GLS	KCTD7	NUS1
BRWD3	DEAF1	GLYCTK	KDM5C	OPHN1
BSCL2	DENND5A	GNAO1	KIF1A	OTUD6B
CACNA1A	DEPDC5	GNB1	KIF5A	PACS2
CACNA1B	DHDDS	GNB5	KIF5C	PAFAH1B1

PAK1	RFT1	ST3GAL5	YWHAG
PAK3	RHOBTB2	STAMBP	ZBTB18
PARS2	RNASEH2A	STRADA	ZEB2
PAX7	RNASEH2B	STT3B	ZNF142
PCDH12	RNASEH2C	STX1B	ZNHIT3
PCDH19	RNF13	STXBP1	
PHACTR1	ROGDI	STYXL1	
PHF21A	RORA	SUMF1	
PIGA	RORB	SV2A	
PIGB	RUSC2	SYN1	
PIGC	SAMD12	SYNGAP1	
PIGG	SAMHD1	SYNJ1	
PIGH	SCARB2	SYP	
PIGL	SCN1A	SZT2	
PIGN	SCN1B	TBC1D24	
PIGO	SCN2A	TBCD	
PIGP	SCN3A	TBL1XR1	
PIGQ	SCN8A	TCF4	
PIGS	SCN9A	TDP2	
PIGT	SEPSECS	THOC2	
PIGU	SERPINI1	TMTC3	
PIGW	SETBP1	TNRC6A	
PLCB1	SETD1B	TPP1	
PLP1	SGCE	TRAK1	
PLPBP	SHANK3	TRAPPC12	
PNKP	SIK1	TRAPPC6B	
PNPO	SLC12A5	TREX1	
POLG	SLC13A5	TRIM8	
PPP1CB	SLC1A2	TRPM3	
PPP2CA	SLC1A4	TRPM6	
PPP2R1A	SLC25A12	TSC1	
PPP3CA	SLC25A22	TSC2	
PPT1	SLC2A1	TUBA1A	
PRDM8	SLC35A2	TUBB2A	
PRICKLE1	SLC35A3	TUBB2B	
PRICKLE2	SLC39A8	UBA5	
PRRT2	SLC45A1	UBE3A	
PRUNE1	SLC6A1	UFC1	
PTPN23	SLC6A8	UNC80	
PURA	SLC9A6	VARS	
QARS	SMARCA2	WASF1	
RAB11A	SMC1A	WDR26	
RAB11B	SNAP25	WDR37	
RAB39B	SORCS3	WDR45	
RAPGEF2	SPATA5	WDR45B	
RARS2	SPTAN1	WDR62	
RBSN	SRPX2	WWOX	
RELN	ST3GAL3	YEATS2	

**MIT-panel (254 st)**

**Gener för mitokondriella sjukdomar, alla ingår i listan för medfödda metabola sjukdomar**

AARS2	COX4I2	MFN2	NDUFB10	PTCD1
ACAD9	COX5A	MGME1	NDUFB11	PTCD3
ADCK2	COX6B1	MICOS13	NDUFB3	PUS1
AFG3L2	COX8A	MICU1	NDUFB6	QRSL1
AGK	CRAT	MICU2	NDUFB8	RARS2
AIFM1	DARS2	MIEF2	NDUFB9	REPS1
ALKBH1	DCAF17	MIPEP	NDUFC2	RMND1
ANTXR2	DGUOK	MPC1	NDUFS1	RNASEH1
APOPT1	DNA2	MPV17	NDUFS2	RRM2B
ARNT2	DNAJC19	MRM2	NDUFS3	RTN4IP1
ATAD3A	DNM1L	MRPL12	NDUFS4	SARS2
ATAD3B	EARS2	MRPL44	NDUFS5	SCO1
ATAD3C	ECHS1	MRPL57	NDUFS6	SCO2
ATP5F1D	ECSIT	MRPS14	NDUFS7	SDHA
ATP5F1E	ELAC2	MRPS16	NDUFS8	SDHAF1
ATP5MD	EPG5	MRPS2	NDUFV1	SDHAF2
ATP6AP1	ERAL1	MRPS22	NDUFV2	SDHB
ATPAF2	ETHE1	MRPS23	NDUFV3	SDHC
BCS1L	FARS2	MRPS25	NFS1	SDHD
BOLA3	FASTKD2	MRPS34	NFU1	SFXN4
C12orf65	FDX2	MRPS7	NSUN3	SLC25A1
C1QBP	FDXR	MSTO1	NUBPL	SLC25A10
CA5A	FOXRED1	MTO1	OPA1	SLC25A19
CARS2	FTL	MTPAP	OPA3	SLC25A24
CEP89	FXN	NADK2	OXA1L	SLC25A3
CHCHD10	GARS	NARS2	PARK7	SLC25A32
CISD2	GATB	NAXD	PARS2	SLC25A4
CLPB	GATC	NAXE	PDE12	SLC25A42
CLPP	GFER	NDUFA1	PDHA1	SLC25A46
CMPK2	GFM1	NDUFA10	PDHB	SPG7
COA5	GTPBP3	NDUFA11	PDHX	SQOR
COA6	HSPD1	NDUFA12	PKD3	SUCLA2
COA7	IARS2	NDUFA13	PDPR	SUCLG1
COASY	IBA57	NDUFA2	PDSS1	SURF1
COQ2	ISCA1	NDUFA4	PDSS2	TACO1
COQ4	ISCA2	NDUFA6	PET100	TAOK1
COQ5	ISCU	NDUFA8	PET117	TARS2
COQ6	KARS	NDUFA9	PITRM1	TAZ
COQ7	KIF5A	NDUFAF1	PMPCB	TFAM
COQ8A	LARS2	NDUFAF2	PNPLA4	TIMM50
COQ9	LONP1	NDUFAF3	PNPLA8	TIMM8A
COX10	LRPPRC	NDUFAF4	PNPT1	TIMMDC1
COX14	LYRM7	NDUFAF5	POLG	TK2
COX15	MCAT	NDUFAF6	POLG2	TMEM126A
COX20	MECR	NDUFAF8	POLRMT	TMEM126B
COX4I1	MFF	NDUFB1	PPA2	TMEM70

TNNI3  
TOP3A  
TRIAP1  
TRIT1  
TRMT10C  
TRMT5  
TRMU  
TSFM  
TTC19  
TUFM  
TWNK  
TYMP  
UQCC2  
UQCC3  
UQCRB  
UQCRC2  
UQCRFS1  
UQCRH  
UQCRQ  
VARS2  
WARS2  
VMA21  
VPS13D  
YARS2



**PEDHEP-panel**

Gener för leversjukdomar hos barn (134st), 53 ingår i listan för medfödda metabola sjukdomar

ABCA1	GALT	PRKCSH
ABCB11	GANAB	PYGL
ABCB4	GATA6	RINT1
ABCC2	GBE1	RPGRIP1L
ABCD3	GDF1	SC5D
ACOX2	GPD1	SEC63
AGL	GUCY2D	SERPINA1
AKR1D1	GYS2	SKIV2L
ALAS2	HAMP	SLC25A13
ALDOB	HFE	SLC25A20
ALG3	HJV	SLC27A5
ALG8	HSD17B4	SLC2A2
ALMS1	HSD3B7	SLC37A4
AMACR	IFT140	SLCO1B1
ANKS6	IFT172	SLCO1B3
ARG1	IFT43	SMPD1
ASAH1	IFT52	TALDO1
ASS1	IFT81	TCTN2
ATP6AP1	INPP5E	TFR2
ATP7B	INTU	TJP2
ATP8B1	JAG1	TMEM107
BAAT	KIF12	TMEM199
BBS1	KRT18	TMEM216
BCS1L	KRT8	TMEM67
CC2D2A	LIPA	TRAF3IP1
CCDC115	LRP5	TRMU
CEP164	LSR	TTC21B
CEP290	MPI	TTC26
CEP41	MPV17	TTC37
CEP83	MYO5B	TWNK
CLDN1	NEK1	UGT1A1
CSPP1	NOTCH2	UNC13D
CYP27A1	NPC1	UNC45
CYP7A1	NPC2	USP53
CYP7B1	NPHP3	WDR19
DCDC2	NR1H4	WDR34
DGUOK	OFD1	WDR35
DNAJB11	PHKA2	WDR60
DYNC2H1	PHKB	WDR83OS
DYNC2LI1	PHKG2	VIPAS39
EIF2AK3	PKD1	VPS33B
FAH	PKD2	XPNPEP3
FBP1	PKHD1	
FECH	POLG	
G6PC	PPM1F	
GALE	PRF1	