

IEM-panel (944 st):
Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild

AARS2	ALDH6A1	ATP1A1	CLCN2	CP
AASS	ALDH7A1	ATP5F1A	CLDN16	CPOX
ABAT	ALDOA	ATP5F1D	CLN3	CPS1
ABCB7	ALDOB	ATP5F1E	CLN5	CPT1A
ABCC8	ALG1	ATP5MD	CLN6	CPT2
ABCD1	ALG11	ATP6AP1	CLN8	CRAT
ABCD4	ALG12	ATP6V0A2	CLPB	CRPPA
ABHD12	ALG13	ATP7A	CLPP	CSF1R
ABHD5	ALG14	ATP7B	CLPX	CTCF
ACAD8	ALG2	ATPAF2	CLTC	CTH
ACAD9	ALG3	AUH	CLTRN	CTNS
ACADM	ALG6	B4GALNT1	CMPK2	CTSA
ACADS	ALG8	B4GALT1	CNCSR2	CTSD
ACADSB	ALG9	B4GALT7	COA3	CTSF
ACADVL	ALKBH1	BBOX1	COA5	CTSK
ACAT1	ALMS1	BCAT2	COA6	CUBN
ACBD5	ALPL	BCKDHA	COA7	CWF19L1
ACER3	ALS2	BCKDHB	COA8	CYC1
ACO2	AMACR	BCKDK	COASY	CYP7B1
ACOX1	AMN	BCS1L	COG1	D2HGDH
ACSF3	AMPD1	BLOC1S3	COG2	DARS1
ACY1	AMPD2	BOLA3	COG4	DARS2
ADA	AMT	BSCL2	COG5	DBH
ADAR	ANO10	BTD	COG6	DBT
ADCK2	ANTXR2	C12orf65	COG7	DCAF17
ADCY5	AP1S1	C19orf12	COG8	DDC
ADK	AP3B1	C1QBP	COLGALT1	DEGS1
ADPRS	AP3D1	CA5A	COQ2	DGAT1
ADSL	AP5Z1	CACNA1A	COQ4	DGUOK
ADSS1	APRT	CACNA1E	COQ5	DHCR24
AFG3L2	ARG1	CACNA1S	COQ6	DHCR7
AGA	ARL3	CAD	COQ7	DHDDS
AGK	ARNT2	CARS1	COQ8A	DHFR
AGL	ARSA	CARS2	COQ9	DHODH
AGPS	ARSB	CAT	COX10	DHPS
AGTPBP1	ASAH1	CBLIF	COX14	DHTKD1
AGXT	ASL	CBS	COX15	DIAPH1
AHCY	ASNS	CCDC115	COX20	DLAT
AIFM1	ASPA	CCDC174	COX411	DLD
AIMP1	ASS1	CCDC88C	COX412	DMGDH
ALAD	ASXL3	CD320	COX5A	DNA2
ALAS2	ATAD3A	CEP89	COX6A1	DNAJC12
ALDH18A1	ATAD3B	CERS1	COX6A2	DNAJC19
ALDH3A2	ATAD3C	CHCHD10	COX6B1	DNAJC3
ALDH4A1	ATIC	CHKB	COX7B	DNAJC5
ALDH5A1	ATP13A2	CISD2	COX8A	DNM1L

DOLK	FCSK	GLB1	HLCS	LARS2
DPAGT1	FDFT1	GLDC	HMBS	LBR
DPM1	FDX2	GLRA1	HMGCL	LDHA
DPM2	FDXR	GLRX5	HMGCS2	LHX4
DPM3	FECH	GLS	HOGA1	LIAS
DPYD	FH	GLUD1	HPD	LIPA
DPYS	FIG4	GLUL	HPRT1	LIPE
DTNBP1	FLAD1	GLYCTK	HPS1	LIPT1
DTYMK	FMO3	GM2A	HPS3	LIPT2
EARS2	FOLR1	GMPPA	HPS4	LMBRD1
EBF3	FOXI1	GMPPB	HPS5	LNPK
EBP	FOXRED1	GNAO1	HPS6	LONP1
ECHS1	FTCD	GNE	HSD17B10	LPIN1
ECM1	FTL	GNMT	HSD17B4	LRP2
ECSIT	FUCA1	GNPAT	HSPD1	LRPPRC
EIF2B1	FUT8	GNPTAB	HSPE1	LYRM4
EIF2B2	FXN	GNPTG	HTRA2	LYRM7
EIF2B3	G6PC	GNS	HYAL1	MAG
EIF2B4	GAA	GOT2	HYOU1	MAGED2
EIF2B5	GABBR2	GPD1	IARS1	MAGEL2
ELAC2	GABRG2	GPHN	IARS2	MAGT1
ELOVL4	GALC	GPR161	IBA57	MAN1B1
ELOVL5	GALE	GRHPR	IDH2	MAN2B1
ENO3	GALK1	GRN	IDH3A	MANBA
EPG5	GALM	GSS	IDI1	MAOA
EPHX1	GALNS	GSX2	IDS	MARS1
EPM2A	GALNT3	GTPBP3	IDUA	MARS2
EPRS1	GALT	GUSB	IER3IP1	MAT1A
ERAL1	GAMT	GYG1	IFIH1	MBD5
ERCC6	GARS1	GYG2	INPP5K	MBOAT7
ETFA	GATB	GYS1	INSR	MCAT
ETFB	GATC	GYS2	IRF2BPL	MCCC1
ETFDH	GATM	HAAO	ISCA1	MCCC2
ETHE1	GBA	HACE1	ISCA2	MCEE
EXOSC3	GBA2	HADH	ISCU	MCOLN1
EXOSC8	GBE1	HADHA	ITPA	MDH1
EXT1	GCDH	HADHB	IVD	MECP2
EXT2	GCH1	HAL	JAG1	MECR
FA2H	GCK	HARS1	JAM2	MFF
FAH	GCLC	HCFC1	KARS1	MFN2
FAM126A	GCSH	HEPACAM	KCNA4	MFSD8
FAM149B1	GFER	HEXA	KCNJ11	MGAT2
FAR1	GFM1	HEXB	KIF5A	MGME1
FARS2	GFM2	HGD	KYNU	MICOS13
FARSB	GGT1	HGSNAT	L2HGDH	MICU1
FASTKD2	GJC2	HIBCH	LAMA1	MICU2
FBP1	GK	HIKESHI	LAMP2	MIEF2
FBXL4	GLA	HK1	LARS1	MIPEP

MLC1	NANS	NKX6-2	PEX1	PNPLA8
MLPH	NARS2	NMNAT1	PEX10	PNPO
MLYCD	NAXD	NPC1	PEX11B	PNPT1
MMAA	NAXE	NPC2	PEX12	POLG
MMAB	NBAS	NSDHL	PEX13	POLG2
MMACHC	NDST1	NSUN3	PEX14	POLR1A
MMADHC	NDUFA1	NT5C3A	PEX16	POLR1C
MMUT	NDUFA10	NUBPL	PEX19	POLR3A
MOCS1	NDUFA11	NUP214	PEX2	POLR3B
MOCS2	NDUFA12	NUS1	PEX26	POLRMT
MOCS3	NDUFA13	OAT	PEX3	POMGNT1
MOGS	NDUFA2	OCRL	PEX5	POMT1
MPC1	NDUFA4	OGDH	PEX6	PPA2
MPDU1	NDUFA6	OPA1	PEX7	PPCS
MPI	NDUFA8	OPA3	PFKM	PPM1K
MPV17	NDUFA9	OPLAH	PGAM2	PPOX
MRM2	NDUFAF1	OTC	PGAP2	PPP1R15B
MRPL12	NDUFAF2	OXA1L	PGK1	PPP1R21
MRPL3	NDUFAF3	OXCT1	PGM1	PPP2R1A
MRPL44	NDUFAF4	OXR1	PGM3	PPP2R5D
MRPL57	NDUFAF5	PAH	PHGDH	PPT1
MRPS14	NDUFAF6	PAK1	PHKA1	PRKAG2
MRPS16	NDUFAF8	PANK2	PHKA2	PRKN
MRPS2	NDUFB1	PARK7	PHKB	PRMT7
MRPS22	NDUFB10	PARS2	PHKG2	PRODH
MRPS23	NDUFB11	PAX7	PHYH	PRODH2
MRPS25	NDUFB3	PAXBP1	PIGA	PRPS1
MRPS34	NDUFB6	PC	PIGM	PRUNE1
MRPS36	NDUFB8	PCBD1	PIGN	PSAP
MRPS7	NDUFB9	PCCA	PIGO	PSAT1
MSL3	NDUFC2	PCCB	PIGQ	PSPH
MSTO1	NDUFS1	PCK1	PIGS	PTCD1
MTFMT	NDUFS2	PCLO	PIGT	PTCD3
MTHFD1	NDUFS3	PCYT1A	PIGV	PTRH2
MTHFR	NDUFS4	PDE10A	PIGW	PTS
MTO1	NDUFS5	PDE12	PIK3R4	PURA
MTPAP	NDUFS6	PDHA1	PITRM1	PUS1
MTR	NDUFS7	PDHB	PKLR	PUS7
MTRR	NDUFS8	PDHX	PLA2G6	PYCR1
MVK	NDUFV1	PDK3	PLAA	PYCR2
MYO5A	NDUFV2	PDP1	PLIN5	PYGL
MYORG	NDUFV3	PDPR	PMM2	PYGM
NAA10	NEU1	PDSS1	PMPCA	PYROXD1
NADK2	NFASC	PDSS2	PMPCB	QARS1
NAGA	NFE2L2	PDXX	PNP	QDPR
NAGLU	NFS1	PEPD	PNPLA2	QRSL1
NAGS	NFU1	PET100	PNPLA4	RAB11B
NALCN	NGLY1	PET117	PNPLA6	RAB27A

RAB3GAP1	SLC22A12	SLC9A1	TH	TYMP
RALGAPA1	SLC22A5	SLC9A7	THAP11	UBQLN2
RANBP2	SLC25A1	SMARCD1	TIMM50	UBTF
RARS1	SLC25A10	SMPD1	TIMM8A	UFM1
RARS2	SLC25A12	SMPD4	TIMMDC1	UMPS
REPS1	SLC25A13	SMS	TK2	UNC80
RFT1	SLC25A15	SNORD118	TKFC	UPB1
RMND1	SLC25A19	SNX14	TKT	UQCC2
RNASEH1	SLC25A20	SOD1	TMEM106B	UQCC3
RNASEH2A	SLC25A21	SOD2	TMEM126A	UQCRB
RNASEH2B	SLC25A22	SORD	TMEM126B	UQCRC2
RNASEH2C	SLC25A24	SPG7	TMEM165	UQCRFS1
RNASET2	SLC25A26	SPR	TMEM199	UQCRH
RNF13	SLC25A3	SPTBN4	TMEM240	UQCRQ
RNF213	SLC25A32	SQOR	TMEM260	UROC1
RPIA	SLC25A4	SQSTM1	TMEM63A	UROD
RRM2B	SLC25A42	SRD5A3	TMEM65	UROS
RTN4IP1	SLC25A46	SSBP1	TMEM70	USP7
RYR1	SLC27A4	SSR4	TMLHE	VAC14
SAMHD1	SLC2A1	ST3GAL5	TMTC3	VAMP2
SARS1	SLC2A2	STAT2	TNNI3	VARS1
SARS2	SLC2A9	STT3A	TOP3A	VARS2
SC5D	SLC30A10	STX3	TPK1	WARS2
SCN8A	SLC30A9	STXBP5L	TPP1	WDR45
SCO1	SLC35A1	SUCLA2	TRAPPC11	WDR45B
SCO2	SLC35A2	SUCLG1	TRAPPC12	WFS1
SCP2	SLC35C1	SUGCT	TRAPPC2L	WIPI2
SCYL1	SLC37A4	SUMF1	TREX1	VMA21
SDHA	SLC39A14	SUOX	TRIAP1	VPS11
SDHAF1	SLC39A4	SURF1	TRIM37	VPS13D
SDHAF2	SLC39A8	SVBP	TRIT1	VPS33A
SDHB	SLC3A1	SYT1	TRMT1	VPS51
SDHC	SLC46A1	SZT2	TRMT10C	VPS53
SDHD	SLC4A10	TACO1	TRMT5	WVOX
SELENOI	SLC52A1	TAF1	TRMU	XDH
SERAC1	SLC52A2	TALDO1	TRNT1	XPR1
SFXN4	SLC52A3	TANGO2	TRPM6	YARS1
SGSH	SLC5A1	TAOK1	TSEN54	YARS2
SIL1	SLC5A2	TARS2	TSFM	YME1L1
SLC13A3	SLC6A17	TAT	TTC19	ZBTB42
SLC16A1	SLC6A19	TAZ	TTPA	ZNHIT3
SLC16A2	SLC6A3	TBCD	TUBB2A	
SLC17A5	SLC6A8	TBCK	TUBB3	
SLC19A2	SLC6A9	TBL1X	TUBB4A	
SLC19A3	SLC7A1	TCN1	TUBB4B	
SLC1A1	SLC7A2	TCN2	TUFM	
SLC1A4	SLC7A7	TDO2	TWNK	
SLC20A2	SLC7A9	TFAM	TXN2	

EP-panel (395 st)

Gener för epileptisk encefalopati, 125 ingår i listan för medfödda metabola sjukdomar

AARS1	BSCL2	DDX3X	GLS	KCTD7
ABAT	CACNA1A	DEAF1	GLYCTK	KDM5C
ABCA2	CACNA1B	DENND5A	GNAO1	KIF1A
ACTL6B	CACNA1C	DEPDC5	GNB1	KIF5A
ADAM22	CACNA1D	DHDDS	GNB5	KIF5C
ADAR	CACNA1E	DMXL2	GOSR2	KMT2A
ADARB1	CACNA1G	DNAJA1	GOT2	KMT2E
ADCY5	CACNA1H	DNAJC5	GPAA1	LGI1
ADRA2B	CACNA2D1	DNM1	GPHN	LIAS
ADSL	CACNA2D2	DNM1L	GPT2	LMNB2
AFG3L2	CAD	DNMT3A	GRIA2	LNPK
AGTR2	CAMK2A	DOCK7	GRIA3	MAGEL2
AIMP1	CAMK2B	DOLK	GRIA4	MAGI2
AKT3	CASK	DPM1	GRIN1	MAPK10
ALDH5A1	CDK19	DPM2	GRIN2A	MARCHF6
ALDH7A1	CDKL5	DPYD	GRIN2B	MBD5
ALG1	CERS1	DYNC1H1	GRIN2D	MBOAT7
ALG11	CERT1	DYRK1A	GRN	MDH2
ALG13	CHD2	EEF1A2	GUF1	MECP2
ALG14	CHRNA2	EHMT1	HACE1	MED13L
ALG3	CHRNA4	EIF2S3	HCN1	MED17
ALG6	CHRNA7	EPM2A	HECW2	MED23
ALKBH8	CHRN2	FBXO28	HIVEP2	MEF2C
AMT	CLCN4	FGF12	HNRNPH2	MFF
ANKRD11	CLN3	FGF13	HNRNPU	MFSD8
AP2M1	CLN5	FGFR3	HSD17B10	MID2
AP3B2	CLN6	FLNA	IQSEC2	MOCS1
AP4S1	CLN8	FOLR1	IRF2BPL	MOCS2
APP	CLTC	FOXP1	ITPA	MTHFS
ARHGGEF9	CNKSR2	FOXP1	KANSL1	MTOR
ARID1B	CNNM2	FRRS1L	KCNA1	NAA10
ARV1	CNPY3	GABBR2	KCNA2	NACC1
ARX	CNTN2	GABRA1	KCNB1	NAPB
ASAH1	CNTNAP2	GABRA2	KCNC1	NBEA
ASH1L	COL4A1	GABRA3	KCND2	NECAP1
ASNS	COL4A2	GABRA5	KCNH1	NEDD4
ASXL3	CPA6	GABRB1	KCNH5	NEUROD2
ATP13A2	CPLX1	GABRB2	KCNJ10	NEXMIF
ATP1A2	CRADD	GABRB3	KCNMA1	NF1
ATP1A3	CSNK2B	GABRG2	KCNQ1	NGLY1
ATP6AP2	CSTB	GAL	KCNQ2	NHLRC1
ATP6V1A	CTSD	GAMT	KCNQ3	NPRL2
ATP7A	CTSF	GATM	KCNQ5	NPRL3
ATXN2	CUX2	GCSH	KCNT1	NR2F1
BRAT1	CYFIP2	GLDC	KCNT2	NRROS
BRWD3	DCX	GLRA1	KCTD3	NRXN1

NTRK2	PURA	SLC45A1	TUBB2B
NUP214	QARS	SLC6A1	UBA5
NUS1	RAB11A	SLC6A8	UBE3A
OPHN1	RAB11B	SLC9A6	UFC1
OTUD6B	RAB39B	SMARCA2	UGDH
OXR1	RALGAPA1	SMC1A	UGP2
PACS2	RAPGEF2	SNAP25	UNC80
PAFAH1B1	RARS2	SORCS3	VAR5
PAK1	RBSN	SPATA5	WASF1
PAK3	RELN	SPTAN1	WDR26
PARS2	RFT1	SRPX2	WDR37
PAX7	RHOBTB2	ST3GAL3	WDR45
PCDH12	RNASEH2A	ST3GAL5	WDR45B
PCDH19	RNASEH2B	STAMBP	WDR62
PHACTR1	RNASEH2C	STARD7	WWOX
PHF21A	RNF13	STRADA	YEATS2
PIGA	ROGDI	STT3B	YWHAG
PIGB	RORA	STX1B	ZBTB18
PIGC	RORB	STXBP1	ZEB2
PIGG	RUSC2	STYXL1	ZNF142
PIGH	SAMD12	SUMF1	ZNHIT3
PIGK	SAMHD1	SV2A	
PIGL	SCARB2	SYN1	
PIGN	SCN1A	SYNGAP1	
PIGO	SCN1B	SYNJ1	
PIGP	SCN2A	SYP	
PIGQ	SCN3A	SZT2	
PIGS	SCN8A	TBC1D24	
PIGT	SCN9A	TBCD	
PIGU	SEMA6B	TBL1XR1	
PIGW	SEPSECS	TCF4	
PLCB1	SERPINI1	TDP2	
PLP1	SETBP1	THOC2	
PLPBP	SETD1A	TMTC3	
PNKP	SETD1B	TNRC6A	
PNPO	SGCE	TPP1	
POLG	SHANK3	TRAK1	
PPP1CB	SIK1	TRAPPC12	
PPP2CA	SLC12A5	TRAPPC4	
PPP2R1A	SLC13A5	TRAPPC6B	
PPP3CA	SLC1A2	TREX1	
PPT1	SLC1A4	TRIM8	
PRDM8	SLC25A12	TRPM3	
PRICKLE1	SLC25A22	TRPM6	
PRICKLE2	SLC2A1	TSC1	
PRRT2	SLC35A2	TSC2	
PRUNE1	SLC35A3	TUBA1A	
PTPN23	SLC39A8	TUBB2A	

MIT-panel (264 st)

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

AARS2	COX20	LARS2	NDUFAF1	PMPCB
ACAD9	COX411	LONP1	NDUFAF2	PNPLA4
ADCK2	COX412	LRPPRC	NDUFAF3	PNPLA8
AFG3L2	COX5A	LYRM7	NDUFAF4	PNPT1
AGK	COX6A1	MCAT	NDUFAF5	POLG
AIFM1	COX6B1	MECR	NDUFAF6	POLG2
ALKBH1	COX7B	MFF	NDUFAF8	POLRMT
ANTXR2	COX8A	MFN2	NDUFB1	PPA2
APOPT1	CRAT	MGME1	NDUFB10	PTCD1
ARNT2	CYC1	MICOS13	NDUFB11	PTCD3
ATAD3A	DARS2	MICU1	NDUFB3	PUS1
ATAD3B	DCAF17	MICU2	NDUFB6	QRSL1
ATAD3C	DGUOK	MIEF2	NDUFB8	RARS2
ATP5F1A	DNA2	MIPEP	NDUFB9	REPS1
ATP5F1D	DNAJC19	MPC1	NDUFC2	RMND1
ATP5F1E	DNM1L	MPV17	NDUFS1	RNASEH1
ATP5MD	EARS2	MRM2	NDUFS2	RRM2B
ATP6AP1	ECHS1	MRPL12	NDUFS3	RTN4IP1
ATPAF2	ECSIT	MRPL3	NDUFS4	SARS2
BCS1L	ELAC2	MRPL44	NDUFS5	SCO1
BOLA3	EPG5	MRPL57	NDUFS6	SCO2
C12orf65	ERAL1	MRPS14	NDUFS7	SDHA
C1QBP	ETHE1	MRPS16	NDUFS8	SDHAF1
CA5A	FARS2	MRPS2	NDUFV1	SDHAF2
CARS2	FASTKD2	MRPS22	NDUFV2	SDHB
CEP89	FDX2	MRPS23	NDUFV3	SDHC
CHCHD10	FDXR	MRPS25	NFS1	SDHD
CISD2	FOXRED1	MRPS34	NFU1	SFXN4
CLPB	FTL	MRPS7	NSUN3	SLC25A1
CLPP	FXN	MSTO1	NUBPL	SLC25A10
CMPK2	GARS	MTO1	OPA1	SLC25A19
COA3	GATB	MTPAP	OPA3	SLC25A21
COA5	GATC	NADK2	OXA1L	SLC25A24
COA6	GFER	NARS2	PARK7	SLC25A3
COA7	GFM1	NAXD	PARS2	SLC25A32
COASY	GFM2	NAXE	PDE12	SLC25A4
COQ2	GTPBP3	NDUFA1	PDHA1	SLC25A42
COQ4	HSPD1	NDUFA10	PDHB	SLC25A46
COQ5	HSPE1	NDUFA11	PDHX	SPG7
COQ6	IARS2	NDUFA12	PDK3	SQOR
COQ7	IBA57	NDUFA13	PDPR	SUCLA2
COQ8A	ISCA1	NDUFA2	PDSS1	SUCLG1
COQ9	ISCA2	NDUFA4	PDSS2	SURF1
COX10	ISCU	NDUFA6	PET100	TACO1
COX14	KARS	NDUFA8	PET117	TAOK1
COX15	KIF5A	NDUFA9	PITRM1	TARS2

TAZ
TFAM
TIMM50
TIMM8A
TIMMDC1
TK2
TMEM126A
TMEM126B
TMEM65
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 (135st), 47 ingår i listan för medfödda metabola sjukdomar

ABCA1	GALE	PRF1
ABCB11	GALT	PRKCSH
ABCB4	GANAB	PYGL
ABCC2	GATA6	RINT1
ABCD3	GBE1	RPGRIP1L
ACOX2	GDF1	SC5D
AGL	GPD1	SEC63
AKR1D1	GUCY2D	SERPINA1
ALAS2	GYS2	SKIV2L
ALDOB	HAMP	SLC25A13
ALG3	HFE	SLC25A20
ALG8	HJV	SLC27A5
ALMS1	HSD17B4	SLC2A2
AMACR	HSD3B7	SLC37A4
ANKS6	IFT140	SLCO1B1
ARG1	IFT172	SLCO1B3
ASAH1	IFT43	SMPD1
ASS1	IFT52	TALDO1
ATP6AP1	IFT81	TCTN2
ATP7B	INPP5E	TFR2
ATP8B1	INTU	TJP2
BAAT	JAG1	TMEM107
BBS1	KIF12	TMEM199
BCS1L	KRT18	TMEM216
CC2D2A	KRT8	TMEM67
CCDC115	LIPA	TRAF3IP1
CEP164	LRP5	TRMU
CEP290	LSR	TTC21B
CEP41	MPI	TTC26
CEP83	MPV17	TTC37
CLDN1	MYO5B	TWNK
CSPP1	NEK1	UGT1A1
CYP27A1	NOTCH2	UNC13D
CYP7A1	NPC1	UNC45A
CYP7B1	NPC2	USP53
DCDC2	NPHP3	WDR19
DGUOK	NR1H4	DYNC2I2
DNAJB11	OFD1	WDR35
DYNC2H1	PHKA2	DYNC2I1
DYNC2LI1	PHKB	WDR83OS
EIF2AK3	PHKG2	VIPAS39
FAH	PKD1	VPS33B
FBP1	PKD2	XPNPEP3
FECH	PKHD1	
G6PC	POLG	
G6PD	PPM1F	

MOVE-panel

Gener för medfödda rörelsesjukdomar hos barn (165st), 97 ingår i listan för medfödda metabola sjukdomar

AARS1	DDC	LIPT2	SPR
AARS2	DLAT	MARS2	SQSTM1
ACER3	DNAJC12	MCOLN1	STAMBP
ACTB	DNAJC19	MDH2	STXBP1
ADAR	DNAJC5	MECP2	SURF1
ADCY5	DNAJC6	MECR	SYNJ1
AFG3L2	EARS2	MLC1	TAF1
AGTPBP1	ECHS1	MRE11	TBC1D24
ALDH18A1	EMC1	MTFMT	TH
ALDH6A1	FA2H	MYORG	THAP1
ANO3	FBXL4	NPC1	TIMM8A
APTX	FBXO7	NPC2	TOR1A
ARSA	FOXG1	NUP62	TPK1
ARV1	FRRS1L	PANK2	TRAPPC11
ARX	FTL	PARK7	TREX1
ATM	FXN	PCDH12	TTPA
ATN1	GABBR2	PDGFB	TUBB4A
ATP13A2	GAMT	PDGFRB	TWNK
ATP1A3	GATM	PDHA1	UBA5
ATP6AP2	GBA	PINK1	UBAP1
ATP7A	GCDH	PLA2G6	VAC14
ATP7B	GCH1	PLEKHG2	WDR45
AUH	GJC2	PLP1	VPS13A
BCAP31	GLRA1	PNKD	VPS13B
BCS1L	GNAL	PNKP	VPS13C
C19orf12	GNAO1	POLG	XPR1
CACNA1A	GNB1	POLR3B	ZNF142
CACNA1B	GRIN1	PRKN	
CACNA1E	GRIN2B	PRKRA	
CACNA1G	HEPACAM	PRRT2	
CAMTA1	HEXA	RELN	
CARS2	HPCA	SCN8A	
CASK	HPRT1	SCP2	
CDKL5	HTRA2	SGCE	
CLN3	HTT	SLC16A2	
COASY	IREB2	SLC18A2	
COL6A3	KCNA1	SLC19A3	
COQ2	KCNA2	SLC20A2	
COX10	KCNC3	SLC25A15	
COX15	KCNMA1	SLC2A1	
COX20	KCNQ2	SLC30A10	
CP	KCTD17	SLC39A14	
CSF1R	KIF1A	SLC6A3	
CWF19L1	KIF1C	SLC6A8	
CYP27A1	KMT2B	SMPD1	
DCAF17	LIPT1	SNCA	

ENDO-panel

Gener för medfödda endokrina sjukdomar (356st), 45 ingår i listan för medfödda metabola sjukdomar

AAAS	CDC6	EIF2B5	GNRH1	LHX4
AARS2	CDC73	EIF2S3	GNRHR	LIG4
ABCC8	CDC48	ENPP1	GPC3	LIPE
ABCD1	CDKN1B	EP300	HADH	LMNA
AGPAT2	CDKN1C	ERCC6	HAMP	LRBA
AIRE	CDT1	ERCC8	HDAC8	MAMLD1
AKT2	CEL	FANCA	HESX1	MAP2K1
ALB	CENPJ	FANCB	HFE	MAP2K2
ALMS1	CEP19	FANCC	HFM1	MAP3K1
ALPL	CHD7	FANCD2	HJV	MC2R
AMH	CISD2	FANCE	HNF1A	MC4R
AMHR2	CLCN5	FANCF	HNF1B	MCM8
AMMECR1	CLCNKA	FANCG	HNF4A	MCM9
ANKRD11	CLCNKB	FANCI	HRAS	MEN1
ANOS1	CLDN16	FANCL	HS6ST1	MKKS
AP2S1	CLPP	FEZF1	HSD11B2	MKRN3
APPL1	CREBBP	FGD1	HSD17B3	MKS1
AQP2	CRIP1	FGF23	HSD17B4	MNX1
AR	CUL4B	FGF8	HSD3B2	MRAP
ARL6	CUL7	FGFR1	IER3IP1	MYRF
ARMC5	CYB5A	FHL1	IGF1	MYT1L
ARX	CYP11A1	FOXA2	IGF1R	NBN
ATRX	CYP11B1	FOXE1	IGFALS	NDN
AVP	CYP11B2	FOXL2	IGSF1	NEUROD1
AVPR2	CYP17A1	FOXP3	IL17RD	NEUROG3
BBS1	CYP19A1	FSHB	IL2RA	NIPBL
BBS10	CYP21A2	FSHR	INS	NKX2-1
BBS12	CYP24A1	GALT	INSR	NKX2-2
BBS2	CYP27B1	GATA3	IRS4	NKX2-5
BBS4	CYP2R1	GATA4	IYD	NNT
BBS5	DCAF17	GATA6	JAG1	NOBOX
BBS7	DCC	GCK	KCNJ1	NR0B1
BBS9	DCHS1	GCM2	KCNJ11	NR3C1
BLK	DHCR7	GH1	KCNJ5	NR3C2
BLM	DHH	GH2	KDM6A	NR5A1
BMP15	DIO1	GHR	KIF14	NRAS
BRAF	DIO2	GHRH	KISS1R	NSMF
BSCL2	DIO3	GHRHR	KLF11	NTN1
BSND	DLK1	GHSR	KMT2D	NTRK2
BTK	DMP1	GLI2	KRAS	OBSL1
CACNA1H	DNAJC3	GLI3	LARS2	ORC1
CASR	DUOX1	GLIS3	LEP	ORC4
CAVIN1	DUOX2	GLUD1	LEPR	ORC6
CBL	DUOXA2	GNA11	LHB	OTX2
CCDC141	DYRK1B	GNAS	LHCGR	PAPPA2
CCDC8	EIF2AK3	GNAS-AS1	LHX3	PAX4

PAX6	SCNN1A	TBX19
PAX8	SCNN1B	TFR2
PCBD1	SCNN1G	TG
PCK1	SDCCAG8	THRA
PCNT	SECISBP2	THRB
PCSK1	SERPINA6	TOE1
PCYT1A	SERPINA7	TPO
PDE11A	SGPL1	TRH
PDE8B	SHOC2	TRHR
PDX1	SHOX	TRIM37
PDZK1IP1	SIM1	TRMT10A
PGM1	SLC12A1	TRPM6
PHEX	SLC12A3	TSHB
PHF6	SLC16A1	TSHR
PIK3R1	SLC16A2	TTC8
PITX2	SLC19A2	TWNK
PLIN1	SLC26A4	UCP2
PMM2	SLC26A7	VDR
PNPLA6	SLC29A3	WDR11
POLD1	SLC2A2	WFS1
POLG	SLC34A1	WNK1
POMC	SLC34A3	WNK4
POR	SLC40A1	VPS13B
POU1F1	SLC5A2	WRN
PPARG	SLC5A5	WT1
PPP1R15B	SMC1A	XRCC4
PRKAR1A	SMC3	ZBTB20
PRMT7	SNRPN	ZFP57
PROK2	SOHLH1	ZFPM2
PROKR2	SOS1	ZMPSTE24
PROP1	SOX10	
PTF1A	SOX2	
PTH	SOX3	
PTH1R	SOX9	
PTPN11	SRA1	
RAD21	SRCAP	
RAF1	SRD5A2	
RBBP8	SRY	
RET	STAG3	
RFX6	STAR	
RIT1	STAT3	
RNU4ATAC	STAT5B	
ROBO2	STX16	
ROR2	TAC3	
RPL10	TACR3	
RPS6KA3	TBCE	
RSPO1	TBL1X	
SAMD9	TBX1	