

臺大醫院 基因醫學部

全外顯子次世代定序帶因篩檢－檢測基因參考列表

ABCA4	ATP13A2	CFAP74	DNAH11	GALT
ABCD1	ATP2A1	CFTR	DNAH5	GBE1
ACADM	ATP5MK	CHRNE	DNAI2	GCDH
ACADS	ATP7B	CLCN1	DNAJC30	GCH1
ACADVL	ATR 不包含 deletion 等拷貝數變異	CLCNKB	DOCK7	GIMAP5
ACAT1	AUH	CLRN1	DPH1	GJB2
ACE	AVIL	CNGA1	DPYS	GLA 不包含 c.639+919G>A (IVS4+919G>A)
ADAMTS2	BBS1	CNGA3	DUOX2 不包含 exon 5-8	GLB1
ADAMTSL4	BBS2	COA8	DUOXA2	GLDC
ADSS1	BCKDHA	COL18A1	DYNC2LI1	GLE1
AFF2	BCKDHB	COPB1	DYSF	GMPPB
AFG2B (SPATA5L1)	BCS1L	COQ4	ELP1	GNPTAB
AGA	BSCL2	CPS1	ERCC3	GNRHR
AGXT	BTD	CPT1A	ETFA	GP1BA
AHSG	C12orf57	CPT2	ETFB	GRM6
AIRE	C2	CRADD	ETFDH	GUCY2D
ALDOB	C6	CRB2	EVC2	GYS2
ALG1	C8B	CTC1	EYS 不包含 exon 12	HADHA
ALG8	CA5A	CYP17A1	F8 不包含轉位序列 (inversion)	HBB
ALMS1	CABP2	CYP1B1	F9	HCFC1
ALPL	CAD	CYP2R1	FAH	HERC2
ANO5	CAPN3	DAAM2	FANCC	HEXA
AP4S1	CBS	DARS2	FGA	HEXB
APTX	CC2D1A	DBT 不包含 deletion 等拷貝數變異	FKRP	HIKESHI
ARG1	CC2D2A	DCAF17	FKTN 不包含反轉錄 轉座子插入序列(retro transposonal insertion)	HKDC1
ARSA	CCDC65	DCPS	FMO3	HLCS
ARSB	CDH23	DDC	G6PC1	HMGCL
ASL	CDK10	DDX11	G6PD	HOGA1
ASPA	CEP250	DGAT1	GAA	HPS1
ASPH	CEP290	DHCR7	GALE	HSD17B10
ASS1	CERKL	DHDDS	GALK1	HSD3B2
ATG7	CERS1	DLD	GALM	HYLS1

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ATM	CFAP298	DMD	GALNS	IDUA
IQCE	MTHFR	PIGO	RTEL1	THG1L
IVD	MTR	PIK3CG	RYR1	TKT
IYD	MTRR	PKHD1	SBDS	TMC1
KCNE1	MTTP	PKLR	SCARF2	TMEM107
KERA	MYO15A	PLEKHG2	SCO2	TMEM216
KIF26A	MYO6	PLOD2	SEMA7A	TMEM260
KIZ	MYO7A	PLP1	SERPINA1	TMEM67
KRT14	MYO9A	PMM2	SERPINB7	TMPRSS3
KRT18	NADSYN1	POLG	SGSH	TOMM7
L1CAM	NAGLU	POLRMT	SHQ1	TPO
LAMA2	NAGS	PPM1K	SLC12A3	TPP1
LAMA5	NHLRC2	PPT1	SLC17A5	TRIM37
LCT	NPHS1	PRDX1	SLC1A4	TRIT1
LIPT1	NR0B1	PREPL	SLC22A5	TRPV6
LMBRD1	NR2E3	PRF1	SLC25A13	TSFM
LOXHD1	NSUN3	PRKRA	SLC25A15	TSHR
MAP3K20	OAT	PROC	SLC26A3	TUBGCP4
MCCC1	OBSCN	PRORP	SLC26A4	TULP1
MCCC2	OCA2	PROS1	SLC2A10	TWINK
MCOLN1	OTC	PTS	SLC34A3	TYR 不包含 exon 4-5
MCPH1	OTOG	QDPR	SLC3A1	TYRP1
MECR	OXCT1	RAB27A	SLC45A1	UBA5
MEFV	P3H1	RAD50	SLC5A5	UNC45B
MID1	PAH	RAG2	SLC6A8	USH1C
MITF	PC	RBM8A	SLC7A7	USH2A
MKS1	PCBD1	RECQL4	SLCO2A1	VPS11
MMAA	PCCA	RMRP	SMARCA1	VPS13B
MMAB	PCCB	RNASEH2B	SMPD1	VPS41
MMACHC	PCDH15	RNASET2	SPINK5	WARS1
MMADHC	PDHB	RNF168	SRD5A2	WNT10A
MMUT	PFKM	RP1L1	TECPR2	XPA
MPL	PGAM2	RPGR	TG	YARS2
MPZL2	PHYH	RS1	TGM1	ZNHIT3