سهامی خاص
شماره ثبت : ۴۱۰۴۵

Musculoskeletal disease
Gene List
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تاریخ : .....
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AARS	CACNA1S	CSTB	FLNC	LITAF	NRAS	PRODH	SLC25A22	VPS13A
ABCB7	CACNB4	CTSD	FTL	LMNA	NSD1	PRPS1	SLC26A2	VPS13B
ABCD1	CAPN3	CYP27A1	FUS	LMX1B	OAT	PRX	SLC35D1	XPA
ACADM	CAV3	CYP7B1	GALC	MAP2K1	OFD1	PTPN11	SMN1	XPC
ACADS	CBS	DBT	GAMT	MAT1A	PAH	PTS	SMPD1	YARS
ACADVL	CDKL5	DDB2	GAN	MATN3	PANK2	QDPR	SOD1	ZEB2
ACAT1	CFL2	DES	GARS	MCC1	PCBD1	RAB23	SOS1	ZFYVE26
ACTA1	CHAT	DMD	GCDH	MCC2	PCCA	RAB7A	SPAST	ZFYVE27
AGA	CHRNA1	DNM2	GCH1	MCEE	PCCB	RAF1	SPG11	
ALDH4A1	CHRNB1	DYSF	GCSH	MECP2	PDHA1	RAPSN	SPG20	
ALDH7A1	CHRND	EBP	GDAP1	MED25	PEX1	RECQL4	SPG21	
ALS2	CHRNE	EGR2	GJB1	MEFV	PEX10	REEP1	SPG7	
AMT	CLCN1	EMD	GLA	MEGF8	PEX12	RNASEH2A	TAF1	
ANG	CLCN7	EP300	GLB1	MFN2	PEX13	RNASEH2B	TARDBP	
ANO5	CLN3	EPM2A	GLDC	MFSD8	PEX14	RNASEH2C	TAT	
APTX	CLN5	ERCC2	GLI3	MMAA	PEX16	RYR1	TCAP	
ARSA	CLN6	ERCC3	GM2A	MMAB	PEX19	SACS	TCfor1	
ARSE	CLN8	ERCC4	GNE	MMADHC	PEX2	SAMHD1	TFAP2A	
ARX	COL11A1	ERCC5	HPD	MPZ	PEX26	SBDS	TGFBR1	
ASPA	COL11A2	ERCC6	HRAS	MTHFR	PEX3	SBF2	TGFBR2	
ASS1	COL1A1	ERCC8	HSPB1	MTM1	PEX5	SCN1A	TNNT1	
ATL1	COL1A2	ETFA	HSPB8	MTMR2	PEX6	SCN4A	TPM2	
ATM	COL6A1	ETFB	HSPD1	MUT	PEX7	SEPN1	TPM3	
ATP1A3	COL6A2	ETFDH	IDS	MYOT	PHYH	SETX	TPP1	
ATP7A	COL6A3	FAH	IVD	NDRG1	PLEC	SGCA	TREX1	
ATP7B	COL9A1	FBN1	KCNA1	NEB	PLP1	SGCB	TRIM32	
BAG3	COL9A2	FGD4	KIAA0196	NEFL	PMP22	SGCD	TRPV4	
BCKDHA	COL9A3	FGFR1	KIF1B	NF1	POLG	SGCE	TSC1	
BCKDHB	COLQ	FGFR2	KIF5A	NF2	POLH	SGCG	TSC2	
BLM	COMP	FGFR3	KRAS	NHLRC1	POMGNT1	SH3TC2	TTN	
BRAF	CPS1	FHL1	L1CAM	NIPA1	POMT1	SLC12A6	TTPA	
BSCL2	CREBBP	FIG4	LAMA2	NOTCH3	POMT2	SLC16A2	VAPB	
BTD	CRTAP	FKRP	LARGE	NPC1	PPT1	SLC1A3	VCP	
CACNA1A	CRYAB	FKTN	LDB3	NPC2	PRNP	SLC25A13	VLDLR	

نشانی: شیراز، خیابان انقلاب، خیابان شکیبا، روبه روی پارک انقلاب، ساختمان رز، طبقه اول، واحد۲ / کد پستی: ۷۱۶۳۷۶۶۵۱۵
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شماره‌ثبت : ۴۱۰۴۵

تاریخ : .....
شماره : .....
پیوست : .....

Importnat Notes:

1- Only known exons of these genes will be examined

2- Repeat expansion disorders will not be covered

3- Genomic regions beside exons of protein-coding genes, genes that are not listed here in this list, repeat expansions and mutations in the upstream and downstream regulatory regions will not be investigated.

4- Additional Comments:

- **Although next generation sequencing (NGS) is a method of choice for high throughput sequencing purposes, NGS has not been approved for clinical and diagnostic use; therefore, Sanger sequencing must be done to confirm the sequencing data, particularly on identified mutations.**
- **Genetic counseling is recommended to explain risks and potential**

5- pitfalls of the experiment.

- **It is of utmost importance for all clinicians involved in the care of families requesting molecular genetic diagnostic tests and the families themselves to be aware of the risk of errors in DNA analysis. Incorrect analysis may result from 1) incorrect data and clinical diagnosis 2) incomplete family studies and history 3) mix-up of DNA samples and mislabeling 4) rare molecular events5) new or spontaneous mutations 6) paternity problems, adaptation, IVF, egg donor, bone marrow transplantation, recent blood product transfusion 7) maternal DNA contamination of CVS or amniotic fluid samples 8) technical errors. The risk of errors from various reasons mentioned above and several others is about 0.5%, while the chance of technical errors of all types is estimated to be around 0.5%. The risk of errors due to DNA recombination in diagnosis is approximately 0.3%. We take no responsibility about patient identity and possible mis-labeling of the DAN samples. Any feedback from our colleagues in the clinical field would be most welcomed. Comments can be given in writing or by calling my number listed below or by e-mail to: Mohammad.ali.faghihi@gmail.com**

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