سهامی خاص
شماره ثبت : ۴۱۰۴۵

Metabolic Disorders Panel
Gene List

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

ABCD1	DBT	MCEE	PPT1
ABCG5	DPYD	MFSD8	PRODH
ABCG8	ENO3	MLYCD	PTS
ACADM	EPM2A	MMAA	PYGM
ACADS	ETFA	MMAB	QDPR
ACADVL	ETFB	MMADHC	SERPINA1
ACAT1	ETFDH	MTHFR	SLC22A5
AGA	FAH	MUT	SLC25A13
AGL	G6PC	NHLRC1	SLC25A15
AGPAT2	G6PD	NPC1	SLC25A20
ALDH4A1	GAA	NPC2	SMPD1
ALDOA	GALC	OAT	TAT
ALDOB	GALT	PAH	TH
ARG1	GBE1	PC	TPP1
ARSA	GCDH	PCBD1	VPS13A
ASL	GCH1	PCCA	
ASS1	GLA	PCCB	
ATP7A	GLB1	PDHA1	
ATP7B	GM2A	PDHX	
BCKDHA	GNE	PDP1	
BCKDHB	GRHPR	PEX1	
BSCL2	GYS1	PEX10	
BTD	GYS2	PEX12	
CBS	HADH	PEX13	
CLN3	HADHA	PEX14	
CLN5	HAL	PEX16	
CLN6	HGD	PEX19	
CLN8	HPD	PEX2	
CPS1	IDS	PEX26	
CPT1A	IVD	PEX3	
CPT2	LDHA	PEX5	
CTNS	LPL	PEX6	
CTSD	MAT1A	PGM1	
CYP21A2	MCCC1	PHKG2	
CYP27A1	MCCC2	PMM2	

نشانی: شیراز، خیابان انقلاب، خیابان شکیبا، روبه روی پارک انقلاب، ساختمان رز، طبقه اول، واحد۲ / کد پستی: ۷۱۶۳۷۶۶۵۱۵
تلفن: ۰۷۱-۳۸۲۶۳۰۶۹ / (۳۲۴۲) ۰۷۱-۲۳۰۵۸۸۶

Work Phone: +98-(071) 2305886 (3242)
www . Parsgen . com
E-mail: info@parsgen.com

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

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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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www . Parsgen . com
E-mail: info@parsgen.com

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