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Exonic Deletion/Duplication analysis by oligonucleotide array-CGH

This oligonucleotide microarray-based assay allows for the detection of exonic deletions or duplications of 105 genes currently tested in our laboratory. This is a custom designed array by Agilent technologies that contains ~140,000 probes present in a 4x180K format with probes more densely spaced in the exons of the genes being tested. The array has been designed to detect copy number changes as small as 300-400 bp. Single genes and custom panels of clinically related genes can be analyzed for deletions and duplications and results may be confirmed by qPCR, MLPA or alternative methodologies.

This assay will allow deletion/duplication analysis for disorders known to be caused by deletions or duplications within a single gene as well as for disorders for which the frequency of gene deletions/duplications is currently not well established. The array-CGH test is particularly indicated for disorders resulting from loss of function or haploinsufficiency. In addition, testing for exonic deletions/duplications is useful in autosomal recessive conditions in which only one mutation is identified by sequence analysis. This assay will detect exonic deletions/duplications of over 300 genes tested for in our laboratory that may not be detected by whole genome array CGH.

The following list contains the genes that are available for deletion/duplication analysis by array-CGH

ABCC8	ABCD1	ACTA1	ACTB	ACTG1	ADSL	AGPS	AGRN	AHI1
AKT2	AKT3	ALDH7A1	ALMS1	ANO5	AP4B1	AP4E1	AP4M1	AP4S1
ARFGEF2	ARHGEF9	ARID1A	ARID1B	ARL13B	ARSE	ARX	ASPM	ATP13A2
ATP1A2	ATP1A3	ATP6AP2	ATP7A*	ATP7B*	ATR	ATRX	BCKDK	BGN
BIN1	BLK	C19orf12	C5orf42	CA8	CACNB4	CAPN3	CASC5	CASK
CAV3	CC2D1A	CC2D2A	CCDC78	CCDC88C	CDC6	CDK5RAP2	CDKL5	CDKN1C
CDT1	CEL	CENPJ	CEP135	CEP152	CEP290	CEP41	CEP63	CETN2
CFL2	CHAT	CHD7*	CHKB	CHMP1A	CHRNA1	CHRNA2	CHRNA4	CHRNB1
CHRNB2	CHRND	CHRNE	CHST14	CISD2	CLN3	CLN5	CLN6	CLN8
CNTN1	CNTNAP2	COL4A1	COL6A2	COL6A3	COLQ	CP	CPA6	CRBN
CREBBP*	CSTB	CTSD	DAB1	DAG1	DCAF17	DCX*	DDHD2	DES
DIAPH1	DKC1	DNAJB6	DNM2	DOK7	DPAGT1	DPM2	DPM3	DYRK1A
DYSF	EBP	ECEL1	EFHC1	EHMT1	EIF2AK3	EMD	EP300*	EPM2A
ERLIN2	ESCO2	EXOSC3	EXT1	EZH2	F8	FA2H	FBN2	FKRP
FKTN	FLNC	FOLR1	FOXG1	FOXP3	FRKP	FTL	G6PD	GAA
GABRA1	GABRA3	GABRE	GABRG2	GABRQ	GAMT	GATA6	GATM	GCK
GFPT1	GJB2 (CX26)*	GLIS3	GLUD1	GNPAT	GPIbβ	GPR56	GPR98	GRIK2
GRIN2A	GTDC2	HADH	HDAC8	HIC1	IDH3G	IER3IP1	INPP5E	INS
INSR	IRAK1	ISPD	ITGA7	ITGA9	KBTBD13	KCNJ11	KCNMA1	KCNQ2
KCNQ3	KCNT1	KCTD7	KDM6A	KIAA1279	KIF7	KLF11	KLHL9	L1CAM
LAMA2	LAMC3	LARGE	LARP7	LGI1	LIS1*	LMNA	LRP2	MAGEA1
MAGI2	MAN1B1	MAPK10	MBD5	MCPH1	MCT8	MECP2*	MED17	MEF2C
MFSD8	MKS1	MLL	MLL2	MPDZ	MTM1	MUSK	MYBPC1	MYF6
MYH3	MYH7	MYH8	MYOT	NDE1	NEB	NEUROD1	NEUROG3	NHLRC1
NIN	NIPBL*	NKX2.5	NPHP1	NRXN1	NSD1*	NSDHL	OCLN	OCR4
OFD1	OPHN1	OPN1LW	OPN1MW	ORC1	ORC4	ORC6	PAFAH1B1	PANK2*
PASD1	PAX4	PAX6	PCDH19	PCNT	PDX1	PEX7	PIGL	PIK3CA
PIK3R2	PLA2G6*	PLCB1	PLEC	PNKP	PNMA3	PNPO	POC1A	POLG
POMGNT1	POMT1	POMT2	PPT1	PRICKLE1	PRICKLE2	PRRT2	PRSS12	PTF1A
NSUN2	RAB18	RAB3GAP1	RAB3GAP2	RAD21	RAPSN	RARS2	RBBP8	RELN
RFX6	ROR2*	RPGRIP1L	RTTN	RYR1	SCN1A	SCN1B	SCN2A	SCN4A
SCN8A	SCN9A	SEPN1	SEPSECS	SETBP1	SGCA	SGCB	SGCD	SGCE
SGCG	SLC12A6	SLC19A2	SLC19A3	SLC25A19	SLC25A19	SLC25A22	SLC29A3	SLC2A1*
SLC2A2	SLC9A6	SMARCA2	SMARCA4	SMARCB1	SMARCE1	SMC1A	SMC3	SNAP29
SPTAN1	SRCAP	SRPX2	ST3GAL3	ST3GAL5	STAMBP	STIL	STXBP1	SYN1
SYNE1	SYNE2	TAZ	TBC1D24	TCAP	TCF4*	TCTN1	TCTN2	TCTN3
TECR	TKTL1	TMEM138	TMEM216	TMEM231	TMEM237	TMEM43	TMEM5	TMEM67
TNNI2	TNNT1	TNNT3	TPM2	TPM3	TPP1	TRAPPC9	TRIM32	TRPS1
TSC1	TSC2	TSEN2	TSEN34	TSEN54	TTC21B	TTN	TUBA1A	TUBA8
TUBB2B	TUBB3	TUSC3	UBE3A	UGT1A1	VLDLR	VPS13B	VRK1	WDR45
WDR62	WFS1	WNT5A	YWHAE	ZC3H14	ZEB2*	ZFP57	ZNF335	ZNF526

* indicates that deletion/duplication analysis for this gene is also available by MLPA

Test methods:
Deletion/duplication analysis involving the coding region of the above listed genes is performed by oligonucleotide array CGH. This exonic array is not appropriate to detect the following abnormalities: a) Low level mosaicism, point or intronic mutations that may be responsible for the clinical phenotype; b) Balanced translocations, inversions, imbalances smaller than the resolution of this array; c)Mutations in genes or intragenic regions that are not represented on the microarray. The sensitivity of this assay may be reduced when DNA is extracted by an outside laboratory.

Deletion/duplication analysis by array-CGH (one gene)

Sample specifications:	3 to10 cc of blood in a purple top (EDTA) tube
Cost:	$1000
CPT codes:	gene specific
Turn-around time:	4 weeks

Deletion/duplication analysis by array-CGH (two or more genes)*

Sample specifications:	3 to10 cc of blood in a purple top (EDTA) tube
Cost:	$1545
CPT codes:	81479
Turn-around time:	4 – 6 weeks

Prenatal testing for a known mutation by deletion/duplication analysis

Sample specifications:	2 T25 flasks of cultured cells from amnio or CVS or 10ml of amniotic fluid
Cost:	$700-950
CPT codes:	Please contact us for CPT codes
Turn-around time:	2 - 3 weeks

Please note: Deletion/duplication analysis by array-CGH is not available for prenatal testing

Please note that for deletion/duplication for two or more genes by array-CGH or by array-CGH and MLPA, analysis will be performed utilizing array-CGH. Cost will be $1545 instead of the sum of the list price of each individual deletion/duplication test.

*Cost of certain deletion/duplication panels vary. Please see our quickguide or specific test information sheets for more details

Results:
Results, along with an interpretive report, are faxed to the referring physician as soon as they are completed. Additional reports are available as requested. All abnormal results are reported by telephone.

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