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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 54 genes currently tested by sequence analysis in our laboratory. This is a custom designed array by Agilent technologies that contains ~50,000 probes present in an 8x60K 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 the 54 genes on the array 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

Disorder	Gene	Disorder	Gene
Allan-Herndon-Dudley syndrome	*MCT8*	Neonatal diabetes	*KCNJ11*
Angelman syndrome	*UBE3A, SLC9A6*	Neurodegeneration with Brain iron accumulation (NBIA)	*PANK2*, PLA2G6**
Autosomal recessive primary microcephaly	*ASPM, CDK5RAP2, CENPJ, MCPH1, STIL,*	Oral Facial Digital syndrome	*OFD1*
Bernard-Soulier syndrome	*GPIbβ*	Pitt-Hopkins syndrome	*TCF4**
Bilateral frontoparietal polymicrogyria	*GPR56*	Pontocerebellar hypoplasia	*TSEN54*
Centronuclear/myotubular myopathy	*BIN1, DNM2, MTM1*	Rett syndrome	*MECP2**
CHARGE syndrome	*CHD7**	Retty syndrome, congenital	*FOXG1*
CHILD sydnrome	*NSDHL*	Roberts syndrome	*ESCO2*
Chondrodysplasia punctata	*ARSE, EBP*	Robinow syndrome	*ROR2**
Cornelia de Lange sydnrome	*NIPBL*, SMC1A*	Rubinstein-Taybi syndrome	*CREBBP**
Early infantile epileptic encephalopathy	*ARX*, CDKL5*, STXBP1*	Sotos syndrome	*NSD1**
Glucose transporter type 1 deficiency	*SLC2A1**	Wilson disease	*ATP7B**
Hearing loss	*GJB2 (CX26)**	X-linked lissencephaly	*DCX**
Lissencephaly	*LIS1**	XLAG lissencephaly	*ARX**
MEF2C testing	*MEF2C*	XLMR and microcephaly with pontine and cerebellar hypoplasia	*CASK*
Menkes disease	*ATP7A**	XLMR with cerebellar hypoplasia	*OPHN1*
Mowat-Wilson disease	*ZEB2**		

* 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. Limitations of this assay include: a) deletions/duplications of less than 2 kb may not be detected by this methodology, b) array-CGH will not detect low level mosaicism, balanced translocations, inversions, or point mutations that may be responsible for the clinical phenotype, c) 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:	83891, 88386 x 2
Turn-around time:	4 – 6 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:	83891,83894, 88386 x 3
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 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.

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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