



ExonArrayDx

Gene list for research purposes

Exon-level deletion/duplication testing is available for disorders due to haploinsufficiency or loss of function for a specific gene in which the frequency of gene deletions/duplications is currently not well established. ExonArrayDx is an oligonucleotide microarray-based (oligo array CGH) test developed by GeneDx to examine one or more genes for full or partial gene deletions or duplications **at the exon level**. The following list contains genes and their associated disorders that are available for deletion/duplication testing by ExonArrayDx. Note that few or no deletions or duplications of these genes have been reported in the literature. ExonArrayDx can be ordered for one specific gene or for a custom panel of genes (up to 10 genes selected by the ordering physician or genetic counselor). Please use the ExonArrayDx submission form to request testing. **This list is searchable—please use the search function in any PDF reader software to find specific entries.**

Genes listed individually by disease name

Disorder	Gene	Disorder	Gene
Achondroplasia	FGFR3	Hypertrophic cardiomyopathy/Wolff-Parkinson-White syndrome	PRKAG2
Alzheimer disease	PSEN1	Hypertrophic Cardiomyopathy, Dilated Cardiomyopathy	ACTC1
Alzheimer disease	PSEN2	Hypertrophic cardiomyopathy, mid-left ventricular chamber type	MYL2
Alzheimer disease, early-onset	APP	Hypertrophic cardiomyopathy, midventricular digenic	MYLK2
Apert syndrome	FGFR2	Hypertrophic cardiomyopathy/LQTS 9	CAV3
Arrhythmogenic right ventricular dysplasia type 5	TMEM43	LDB3-Related Dilated Cardiomyopathy	LDB3
Arrhythmogenic right ventricular dysplasia, familial type 10	DSG2	Noonan syndrome	SOS1
Arrhythmogenic right ventricular dysplasia, familial type 11	DSC2	Noonan syndrome	RAF1
Arrhythmogenic right ventricular dysplasia, familial type 12	JUP	Parkinson disease	SNCA
Arrhythmogenic right ventricular dysplasia, familial, 9	PKP2	Parkinson disease	PARK2
Arrhythmogenic right ventricular dysplasia, type 1	TGFB3	Schizophrenia susceptibility	DISC1
Cardiofaciocutaneous syndrome	BRAF	Serotonin transporter activity (increased/decreased)	SLC6A4
Cardiofaciocutaneous syndrome	KRAS	Split-hand/foot malformation	DLX5
Cardiofaciocutaneous syndrome	MAP2K1		
Cardiofaciocutaneous syndrome	MAP2K2		
Danon disease	LAMP2		
Dilated cardiomyopathy	TNNC1		
Dilated cardiomyopathy	PLN		
Dilated cardiomyopathy 1C	LDB3 (ZASP)		
Dilated cardiomyopathy type 1	DES		
Dilated cardiomyopathy type 1L	SGCD		
Dilated cardiomyopathy type 3A	TAZ		
Dilated cardiomyopathy with wooly hair and keratoderma	DSP		
Emery-Dreifuss muscular dystrophy, Hutchinson-Gilford progeria syndrome, Mandibuloacral dysplasia with type A lipodystrophy	LMNA		
Familial cutaneous melanoma	P16=CDKN2A		
Familial hypertrophic cardiomyopathy	TNNI3		
Familial hypertrophic cardiomyopathy	TNNT2		
Familial hypertrophic cardiomyopathy	TPM1		
Hay-Wells and related disorders	P63 (TP63)		
Hirschsprung disease, Multiple Endocrine Neoplasia Type 2A, Multiple Endocrine Neoplasia Type 2B	RET		
Hypertrophic cardiomyopathy	MYBPC3		
Hypertrophic cardiomyopathy	MYH7		
Hypertrophic cardiomyopathy	MYL3		

