

ExonArrayDx Gene List - Clinical

ExonArrayDx is a microarray-based (array CGH) test developed by GeneDx to examine one or more genes for full or partial gene deletions or duplications **at the exon level** for a list of 433 genetic disorders. The ExonArrayDx test can be ordered for one specific gene or for a custom panel of clinically related genes (up to 10 genes selected by ordering physician or genetic counselor). The following list contains genes (and their associated disorders) tested by ExonArrayDx. Please use the ExonArrayDx submission form to request a test. **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

Disorder	Gene
2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency	HSD17B10
3-methylcrotonyl-CoA carboxylase deficiency	MCCC1
3-methylcrotonyl-CoA carboxylase deficiency	MCCC2
6-pyruvoyltetrahydropterin synthase deficiency	PTS
9q34 subtelomeric deletion syndrome	EHMT1
Achromatopsia	CNGA3
Achromatopsia	CNGB3
Achromatopsia	GNAT2
Adenomatous polyposis colorectal cancer	MUTYH
Adrenoleukodystrophy	ABCD1
Agammaglobulinemia, X-linked	BTX
Alagille syndrome	JAG1
Albright Hereditary Osteodystrophy, PHP1A,PPHP, POH	GNAS
Alpha thalassemia mental retardation syndrome, X-linked	ATRX
Alport syndrome	COL4A5
Amyloid polyneuropathy Andrade or Portuguese type	TTR
Andersen cardiodyrhythmic periodic paralysis	KCNJ2
Androgen insensitivity	AR
Angelman syndrome	UBE3A
Aniridia	PAX6
Aniridia	DCDC1
Aniridia	ELP4
Aniridia (WAGR)	WT1
Anophthalmia/microphthalmia	SOX2
Anophthalmia/microphthalmia	SIX6
Anophthalmia/microphthalmia	BMP4
Anophthalmia/microphthalmia	CHX10 (VSX2)
Anophthalmia/microphthalmia	OTX2
Anophthalmia/microphthalmia	STRA6
Anophthalmia/microphthalmia	RAX
Anterior segment mesenchymal dysgenesis	PITX3
Anterior segment ocular dysgenesis	FOXE3
Anterior segment ocular dysgenesis	MAF
Anterior segment ocular dysgenesis (microphthalmia)	BCOR
Anterior segment ocular dysgenesis /Axenfeld-Rieger syndrome	PITX2
Anterior segment ocular dysgenesis/Axenfeld-Rieger syndrome	FOXC1
Antithrombin III deficiency (thrombophilia)	SERPINC1
Aplastic anemia, dyskeratosis congenita, pulmonary fibrosis	TERT
Ataxia telangiectasia	ATM
Ataxia telangiectasia-like disorder	MRE11A
Atrial septal defect with atrioventricular conduction defects	NKX2-5
Autism spectrum; atypical Rett; epileptic encephalopathy	CDKL5
Autism susceptibility locus	NRXN1
Autism susceptibility locus	AHI1
Autism susceptibility locus	CACNA1C
Autism susceptibility locus	CNTNAP2
Autism susceptibility locus	GABRB3
Autism susceptibility locus	AUTS2
Autism susceptibility locus	CNTN4
Autism susceptibility locus	PCDH9
Autism susceptibility locus	SLC4A10
Autism susceptibility locus, X-linked	NLGN3
Autism susceptibility locus, X-linked	NLGN4
Autism susceptibility locus; 16p13.3 deletion syndrome	A2BP1
Autism susceptibility locus; 22q13.3 deletion syndrome	SHANK3
Autoimmune polyendocrinopathy	AIRE
Bannayan-Riley-Ruvalcaba syndrome	PTEN
Best macular dystrophy	VMD2 (BEST1)
Beta Thalassemia	HBB
Beta-ketothiolase deficiency	ACAT1
Biotinidase deficiency	BTD
Birt-Hogg-Dube syndrome	FLCN
Blackfan-Diamond syndrome	RPS19
Blepharophimosis, ptosis, and epicanthus inversus (BPES)	FOXL2
Bloom syndrome	RECQL3 (BLM)
Branchio-oto-renal syndrome type 1	EYA1
Branchiootic syndrome	SIX1
Campomelic dysplasia	SOX9
Canavan disease	ASPA
Carbamoyl phosphate synthetase I deficiency	CPS1

Disorder

Disorder	Gene
Carnitine palmitoyl transferase I deficiency	CPT1A
Carpenter disease	RAB23
Cerebral cavernous malformation	CCM1 (KRIT1)
Cerebral cavernous malformation	CCM2
Cerebral cavernous malformations 3	PDCD10
Charcot-Marie-Tooth disease type 1A	PMP22
CHARGE syndrome	CHD7
Chondrodysplasia punctata	ARSE
Choroideremia	CHM
Chronic granulomatous disease	CYBA
Chronic granulomatous disease	CYBB
Chronic granulomatous disease	NCF1
Chronic granulomatous disease	NCF2
Citrullinemia type 1	ASS1
Cleidocranial dysplasia	RUNX2
Cobalamin C deficiency	MMACHC
Cockayne syndrome A	ERCC8
Cockayne syndrome B	ERCC6
Coffin-Lowry syndrome	RSK2
Cohen syndrome	COH1
Combined pituitary hormone deficiency	POU1F1
Combined pituitary hormone deficiency	PROP1
Cone-rod dystrophy, autosomal dominant	GUCY2D
Cone-rod dystrophy, autosomal dominant	CRX
Cone-rod dystrophy, autosomal dominant	PRPH2 (RDS)
Cone-rod dystrophy, autosomal recessive	ABCA4
Cornelia-DeLange syndrome	NIPBL
Cortical dysplasia-focal epilepsy syndrome	CNTNAP2
Cowden syndrome	PTEN
Craniofrontonasal syndrome	EFNB1
Craniosynostosis, type 2	MSX2
Currarino syndrome	HLXB9 (MNX1)
D-bifunctional protein deficiency	HSD17B4
Dandy-Walker malformation	ZIC1
Dandy-Walker malformation	ZIC4
Dent disease	CLCN5
Developmental verbal dyspraxia	FOXP2
DiGeorge syndrome type 2	NEBL
DiGeorge-like phenocopy	CRKL
DiGeorge/velocardiofacial syndromes	TBX1
Dihydropteridine reductase deficiency	QDPR
Duane-radial ray syndrome	SALL4
Dyskeratosis congenita	DKC1
Dyskeratosis congenita	TERC
Ehlers-Danlos syndrome type 4	COL3A1
Emery-Dreifuss muscular dystrophy	LMNA
Epilepsy with learning disability and behavior disorders/X-linked	SYN1
Epilepsy, benign neonatal	KCNQ3
Ethylmalonic aciduria	ETHE1
Fabry disease	GLA
Familial exudative vitreoretinopathy	FZD4
Familial exudative vitreoretinopathy	LRP5
Familial gastric cancer	CDH1
Familial hypercholesterolemia	LDLR
Fanconi anemia	FANCA
Fanconi anemia	FANCD2
Fanconi anemia type N	PALB2
Febrile convulsions, migraine	SCN1A
Feingold syndrome	MYCN
Focal dermal hypoplasia (Goltz syndrome)	PORCN
Fragile X syndrome	FMR1
Fumarate hydratase deficiency	FH
Galactokinase deficiency	GALK1
Galactosemia (galactosyltransferase deficiency)	GALT
Glanzmann thrombasthenia	ITGB3
Glutaric aciduria type I	GCDH
Glutaric aciduria type II (Multiple acyl-CoA dehydrogenase def.)	ETFA
Glutaric aciduria type II (Multiple acyl-CoA dehydrogenase def.)	ETFB
Glutaric aciduria type II (Multiple acyl-CoA dehydrogenase def.)	ETFDH
Glutathione synthetase deficiency	GSS
Glycerol kinase deficiency, isolated	GK

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<i>Disorder</i>	<i>Gene</i>
GM1 gangliosidosis	GLB1
Gonadal dysgenesis	SRY
Gorlin syndrome	PTCH
Greig cephalopolysyndactyly syndrome	GLI3
Growth hormone insensitivity syndrome	GHR
Growth hormone insensitivity syndrome	GHRHR
GTP cyclohydrolase 1 deficiency; dopa-responsive dystonia	GCH1
Harlequin ichthyosis	ABCA12
Hemophilia A	F8
Hemophilia B	F9
Hereditary angioedema	C1INH
Hereditary hemorrhagic telangiectasia	ENG
Hereditary leiomyomatosis and renal cell cancer	FH
Hereditary multiple exostoses	EXT1
Hereditary multiple exostoses	EXT2
Hereditary neuropathy with pressure palsies	PMP22
Hereditary nonpolyposis colon cancer	MSH2
Hereditary nonpolyposis colon cancer / Turcot syndrome	MLH1
Hereditary nonpolyposis colon cancer 4	PMS2
Hereditary nonpolyposis colon cancer, ovarian cancer	MSH6
Hereditary paraganglioma-pheochromocytoma syndromes	SDHB/C/D
Hereditary polyposis coli (colon cancer)	APC
Hereditary schwannomatosis/malignant rhabdoid tumor	SMARCB1
Hirschsprung disease	RET
HMG co-A lyase deficiency	HMGCL
Holocarboxylase synthetase deficiency	HLCS
Holoprosencephaly	SHH
Holoprosencephaly	SIX3
Holoprosencephaly	TGIF
Holoprosencephaly	ZIC2
Holt-Oram syndrome	TBX5
Homocystinuria (cystathionine beta-synthase deficiency)	CBS
Hunter syndrome (mucopolysaccharidosis type II)	IDS
Hutchinson-Gilford progeria syndrome	LMNA
Hydrocephalus, X-linked	L1CAM
Hyper-IgM syndrome type 1	CD40LG
Hyperparathyroidism-jaw tumor syndrome	HRPT2
Hypohidrotic ectodermal dysplasia with immune deficiency	IKBKG
Hypohidrotic ectodermal dysplasia, X-linked	EDA1
Hypoparathyroidism, deafness, and renal disease	GATA3
Hypophosphatemic rickets, X-linked	PHEX
Ichthyosis, X-linked	STS
Immunodeficiency with Hyper IgM	AICDA
IRAK4 deficiency	IRAK4
Isovaleric acidemia	IVD
Joubert syndrome	AHI1
Joubert syndrome	CEP290
Junctional epidermolysis bullosa	LAMB3
Juvenile polyposis syndrome	BMPR1A
Juvenile polyposis syndrome; pancreatic cancer	SMAD4
Kallmann syndrome	FGFR1
Kallmann syndrome, X-linked	KAL1
Lamellar ichthyosis	TGM1
Leber congenital amaurosis, autosomal dominant	CRX
Leber congenital amaurosis, autosomal recessive	AIP1L1
Leber congenital amaurosis, autosomal recessive	CEP290
Leber congenital amaurosis, autosomal recessive	GUCY2D
Leber congenital amaurosis, autosomal recessive	RDH12
Leber congenital amaurosis, autosomal recessive	RPGRIP1
Leber congenital amaurosis, autosomal recessive	TULP1
Leber congenital amaurosis, autosomal recessive	CRB1
Leber congenital amaurosis, autosomal recessive	RPE65
Leri-Weill dyschondrosteosis; short stature	SHOX
Lesch-Nyhan syndrome	HPRT1
Leukocyte adhesion deficiency	ITGB2
Li-Fraumeni syndrome	TP53
Li-Fraumeni syndrome type 2	CHEK2
Limb-girdle muscular dystrophy type 2A	CAPN3
Lissencephaly 1	PAFAH1B1
Lissencephaly syndrome, Norman-Roberts type	RELN
Loeys-Dietz syndrome	TGFBR1
Loeys-Dietz syndrome	TGFBR2
Long-chain acyl-CoA dehydrogenase deficiency	HADHA
Long QT syndrome 1	KCNQ1
Long QT syndrome 2	KCNH2
Long QT syndrome 3	SCN5A
Long QT syndrome 4	ANK2

<i>Disorder</i>	<i>Gene</i>
Long QT syndrome 5	KCNE1
Long QT syndrome 6 (acquired susceptibility)	KCNE2
Long QT syndrome 7	KCNJ2
Long QT syndrome 8	CACNA1C
Long QT syndrome 9	CAV3
Long QT syndrome 11	AKAP9
Low eye oculocerebrorenal syndrome	OCRL1
Malonic aciduria	MLYCD
Mandibuloacral dysplasia with type A lipodystrophy	LMNA
Maple syrup urine disease	BCKDHA
Maple syrup urine disease	BCKDHB
Maple syrup urine disease	DBT
Marfan syndrome	FBN1
Meckel syndrome	CEP290
Medium-chain acyl-CoA dehydrogenase deficiency	ACADM
Menkes disease	ATP7A
Mental retardation, autosomal recessive	GRIK2
Metaphyseal chondrodysplasia	COL10A1
Methylmalonic acidemia - cbl A type	MMAA
Methylmalonic acidemia - cbl B type	MMAB
Methylmalonic acidemia - mutase deficiency	MUT
Microphthalmia and linear skin defects	HCCS
Mitochondrial complex I deficiency	NDUFA1
Mitochondrial trifunctional protein deficiency	HADHA
Mitochondrial trifunctional protein deficiency	HADHB
Molybdenum cofactor deficiency	MOCS1
Molybdenum cofactor deficiency	MOCS2
Morquio B syndrome	GLB1
Morquio syndrome A (mucopolysaccharidosis type IVA)	GALNS
Mowat-Wilson syndrome; Hirschsprung disease with MR	ZEB2
MR/microcephaly with pontine and cerebellar hypoplasia	CASK
MR/microcephaly with pontine and cerebellar hypoplasia	EFHC2
Mucopolidosis IV	MCOLN1
Multiple endocrine neoplasia type 1	MEN1
Myoclonic epilepsy of Lafora	EPM2A
Myoclonic epilepsy of Lafora	NHLRC1
Myotubular myopathy, X-linked	MTM1
Nail-patella syndrome	LMX1B
Nance-Horan syndrome	NHS
Nephrogenic diabetes insipidus	AVPR2
Nephronophthisis	NPHP1
Nephropathic cystinosis	CTNS
Neurofibromatosis type 1	NF1
Neurofibromatosis type 2	NF2
Niemann Pick type A and B	SMPD1
Niemann Pick type C	NPC1
Niemann Pick type C	NPC2
Nijmegen breakage syndrome	NBS1 (NBN)
Nocturnal frontal lobe epilepsy type 1	CHRNA4
Nocturnal frontal lobe epilepsy type 3	CHRN2
Norrie disease	NDP
Ocular coloboma	GDF6
Oculocutaneous albinism	OCA2
Opitz syndrome	MID1
Opitz-Kaveggia syndrome	MED12
Optic atrophy 1	OPA1
Optic nerve hypoplasia / septooptic dysplasia	HESX1
Ornithine transcarbamylase	OTC
Oro-facial-digital syndrome (OFD1)	CXORF5
Osler-Rendu-Weber syndrome type 1	ENG
Osler-Rendu-Weber syndrome type 2	ACVRL1
Osteogenesis imperfecta type 1 and 4	COL1A1
Pallister-Hall syndrome	GLI3
Pelizaeus-Merzbacher disease	PLP1
Periventricular nodular heterotopia	FLNA
Peters-plus syndrome	B3GALT1
Peutz-Jeghers syndrome	STK11
Phenylketonuria	PAH
Phosphoglycerate kinase 1 deficiency, Munchen	PGK1
Phosphoribosylpyrophosphate synthetase superactivity	PRPS1
Pituitary dwarfism	LHX3
Pituitary dwarfism type 1, short stature	GH1
Pompe disease (glycogen storage disease type II)	GAA
Potocki-Lupski syndrome	RAI1
Potocki-Shaffer syndrome	ALX4
Prader-Willi syndrome	snoRNA clus-

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Disorder	Gene
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Prader-Willi-like syndrome 2	SIM1
Primary carnitine deficiency (carnitine uptake defect)	SLC22A5
Primary congenital glaucoma	CYP1B1
Primary open angle glaucoma	MYOC
Primary open angle glaucoma	OPTN
Propionic acidemia	PCCA
Propionic acidemia	PCCB
Pseudochondroplasia	COMP
Pseudoxanthoma elasticum	ABCC6
Pyruvate dehydrogenase E1-alpha deficiency	PDHA1
Renal cell carcinoma	FHIT
Renal coloboma syndrome	PAX2
Renal cysts and diabetes syndrome	HNF1B
Retinitis Pigmentosa, autosomal dominant	IMPDH1
Retinitis Pigmentosa, autosomal dominant	RP1
Retinitis Pigmentosa, autosomal dominant	PRPF8
Retinitis Pigmentosa, autosomal dominant	PRPF31
Retinitis pigmentosa, autosomal recessive	TULP1
Retinitis pigmentosa, autosomal recessive	CNGA1
Retinitis pigmentosa, autosomal recessive	EYS (EGFL11)
Retinitis pigmentosa, autosomal recessive	PDE6A
Retinitis pigmentosa, autosomal recessive	PDE6B
Retinitis pigmentosa, autosomal recessive	USH2A
Retinitis pigmentosa, autosomal recessive	CRB1
Retinitis pigmentosa, autosomal recessive	RPE65
Retinitis pigmentosa, autosomal recessive	ABCA4
Retinitis Pigmentosa, X-linked	RP2
Retinitis Pigmentosa, X-linked	RPGR
Retinoblastoma	RB1
Retinoschisis, X-linked	XLRS1
Rett syndrome/male progressive neurodevelopmental syndrome	MECP2
Roberts syndrome	ESCO2
Rothmund-Thompsen syndrome	RECQL4
Rubenstein-Taybi syndrome	CREBBP
Saethre-Chotzen syndrome	Twist1
Senior-Loken syndrome	CEP290
Severe combined immune deficiency	JAK3
Severe combined immune deficiency	RAG1
Severe combined immune deficiency	RAG2
Severe combined immune deficiency	ADA
Severe combined immune deficiency with radiation sensitivity	DCLRE1C
Short stature, pituitary and cerebellar defects, small sella turcica	LHX4
Short-chain acyl-CoA dehydrogenase deficiency	ACADS
Shprintzen-Goldberg craniosynostosis	FBN1
Simpson-Golabi-Behmel syndrome type 1; Wilms' tumor	GPC3
Sjogren-Larsson syndrome	ALDH3A2
Smith-Lemli-Opitz syndrome	DHCR7
Smith-Magenis syndrome	RAI1
Sotos syndrome	NSD1
Split hand-foot malformation type 1	SHFM1
Split hand-foot malformation type 3	FBXW4
Stargardt disease, autosomal recessive	ABCA4
Stargardt like macular dystrophy, autosomal dominant	ELOVL4
Stickler syndr., Kniest dyspl., spondyloepiphyseal dyspl. congenita	COL2A1
Symphodactyly type 1 / syndactyly	HOXD13
Tay-Sachs disease (hexosaminidase A deficiency)	HEXA
Tetralogy of Fallot	ZFPM2
Timothy syndrome	CACNA1C
Townes-Brock syndrome	SALL1
Treacher-Collins syndrome	TCOF1
Trichorhinophalangeal syndrome	TRPS1
Tuberous sclerosis type 1	TSC1
Tuberous sclerosis type 2	TSC2
Tyrosinemia type 2	TAT
Tyrosinemia type I	FAH
Ulnar-mammary syndrome	TBX3
Van der Woude syndrome	IRF6
Ventricular tachycardia, catecholaminergic polymorphic 1	RYR2
Very long-chain acyl-CoA dehydrogenase deficiency	ACADVL
Vohwinkel syndrome	GJB2
von Hippel Lindau syndrome	VHL
Waardenburg syndrome type 2A	MITF
Waardenburg syndrome, type I, 3	PAX3
WAGR; Denys-Drash syndrome; Frasier syndrome	WT1
Weaver syndrome	NSD1
Weill-Marchesani syndrome	FBN1

Disorder	Gene
Werner syndrome	RECQL2 (WRN)
Williams-Beuren syndrome	ELN
Wilson disease	ATP7B
Wiskott-Aldrich syndrome	WAS
X-linked heterotaxy	ZIC3
X-linked lissencephaly type 1	DCX
X-linked lymphoproliferative syndrome	SH2D1A
X-linked mental retardation	FACL4 (ACSL4)
X-linked mental retardation	GDI1
X-linked mental retardation	KIAA2022
X-linked mental retardation	RPL10
X-linked mental retardation	SIZN1 (ZCCHC12)
X-linked mental retardation	SLC38A5
X-linked mental retardation	VCX3A
X-linked mental retardation MRX21	IL1RAPL1
X-linked mental retardation MRX45	ZNF81
X-linked mental retardation MRX58	TM4SF2
X-linked mental retardation MRX89	ZNF41
X-linked mental retardation MRX92	ZNF674
X-linked mental retardation	FMR2
X-linked mental retardation 30	PAK3
X-linked mental retardation 44	FTSJ1
X-linked mental retardation 46	ARHGFE6
X-linked mental retardation 59	AP152
X-linked mental retardation 88	AGTR2
X-linked mental retardation 90	DLG3
X-linked mental retardation 93	BRWD3
X-linked mental retardation 94	GRIA3
X-linked mental retardation (Allan-Herndon-Dudley syndrome)	MCT8 (SLC16A2)
X-linked MR (Borjeson-Forssman-Lehmann syndrome)	PHF6
X-linked mental retardation (Cabezas syndrome)	CUL4B
X-linked mental retardation (Creatine deficiency syndrome)	SLC6A8
X-linked mental retardation (Hyperekplexia and epilepsy)	ARHGFE9
X-linked MR (spasms, epileptic encephalopathy, atypical Rett)	ARX
X-linked mental retardation (Mohr-Tranebjaerg syndrome)	TIMM8A
X-linked mental retardation (Renpenning syndrome)	PQBP1
X-linked MR (Rolandic epilepsy, MR and speech delay)	SRPX2
X-linked mental retardation (Siderius syndrome)	PHF8
X-linked mental retardation (Stocco dos santos syndrome)	KIAA1202
X-linked mental retardation with cerebellar hypoplasia	OPHN1
X-linked mental retardation with epilepsy	ATP6AP2
X-linked mental retardation with growth deficiency	SOX3
X-linked mental retardation with growth deficiency	SOX3
X-linked mental retardation, JARID1C-related	SMCX (JARID1C)
X-linked mental retardation, Snyder-Robinson type	SMS
X-linked mental retardation, syndromic 14	UPF3B
X-linked mental retardation, syndromic	UBE2A
X-linked mental retardation, syndromic Christianson type	SLC9A6
X-linked mental retardation, syndromic, Turner type	HUWE1
X-linked mental retardation, ZDHHC9-related	ZDHHC9
X-linked mental retardation/faciogenital dysplasia	FGD1
X-linked VACTERL association with hydrocephalus	FANCB
Xeroderma pigmentosum	XPA
Xeroderma pigmentosum	XPB (ERCC3)
Xeroderma pigmentosum	XPC
Xeroderma pigmentosum	XPD (ERCC2)
Xeroderma pigmentosum	XPE (DDB2)
Xeroderma pigmentosum	XPF (ERCC4)
Xeroderma pigmentosum	XPG (ERCC5)
XY sex reversal and gonadal dysgenesis	DMRT1