

GenomeDx v.3.0 enables a high-resolution, genome-wide microarray analysis to evaluate for known as well as novel genomic deletion and duplication syndromes. The custom-designed oligonucleotide microarray utilizes 105,000 probes that are placed across the non-repetitive sequence of the genome and offers increased probe density and technical sensitivity. This table provides brief summaries of clinically relevant loci for which genomic loss or gain has been associated with a disease phenotype.

The loci are grouped and color-coded into 4 major categories, based on the clinical utility of these tests:

- Well-established microdeletion/duplication syndromes with a high detection rate.
- Microdeletion/duplication syndromes with a lower detection rate.
- Newly recognized microdeletion/duplication syndromes.
- Single gene disorders only rarely caused by partial or complete gene deletion/duplication. These disorders are predominantly caused by mutations detectable by DNA sequencing.
- Autism susceptibility loci

Autism Susceptibility Loci Covered by the GenomeDx 105K array

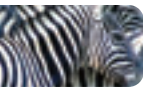
LOCUS	GENE	LOCUS	GENE
● 16p13.2	A2BP1	● Xq13.1	NLGN3
● 6q23.3	AHI1	● Xp22.33	NLGN4X
● 12p13.33	CACNA1C	● 7q22.1	NPTX2
● 7q31.32	CADPS2	● 2p16.	NRXN1
● 3p26.3	CNTN4	● 13q21.32	PCDH9
● 7q35	CNTNAP2	● 10q23.3	PTEN
● 11q13.4	DHCR7	● 7q22.1	RELN
● 1q42.2	DISC1	● 22q13.33	SHANK3
● 7q36.3	EN2	● 2q31.	SLC25A12
● 15q12	GABRB3	● 2q24.2	SLC4A10
● 6q16.3	GRIK2	● 17q11.2	SLC6A4
● 17q21.32	ITGB3	● 9q34.13	TSC1
● Xq28	MECP2	● 16p13.3	TSC2/PKD1
● 7q31.2	MET	● 15q11.2	UBE3A

● All reported intrachromosomal gains or losses linked to autism susceptibility (see microdeletions/duplications on page 1)

Chromosomal and Single Gene Disorders Covered by the GenomeDx 105K array

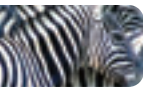
click on OMIM number for more information about that disorder

DISORDER	LOCUS	GENE	OMIM
● 1p36 deletion syndrome	1p36	Multiple	607872
● 1q21 microdeletion (TAR)	1q21.1	Multiple	274000
● 1q41-1q42 deletion	1q41-1q42	Multiple	
● 2p15 microdeletion	2p15	Multiple	
● 2q32.2-2q33 microdeletion	2q32.2-2q33	SATB2	608148
● 2q37 subtelomeric deletion	2q37	Multiple	600430



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DISORDER	LOCUS	GENE	OMIM
● 3q29 microdeletion/duplication	3q29	PAK2	609425
● 6p25 microdeletion	6p25	Multiple	601631
● 7q11 Williams-Beuren region duplication	7q11.32	ELN	609757
● 8p23 microdeletion/duplication	8p23	Multiple	
● 9q34 subtelomeric deletion syndrome	9q34	EHMT1	
● 10q22-10q23 microdeletion	10q22.3-10q23.31	Multiple	
● 12q24.21 microduplication	12q24	Multiple	
● 13q32 deletion	13q32	ZIC2	609637
● 14q11.2 deletion	14q11.2	Multiple	
● 14q22-14q23 deletion	14q22.1-q23.1	Multiple	
● 15q11.2-15q13.1 microduplication	15q11.2-q13.1	Multiple	608636
● 15q13.3 microdeletion	15q13.1-15q13.3	Multiple	
● 15q24 microdeletion	15q24	Multiple	
● 16p11.2 microdeletion/duplication (AUTS14)	16p11.2	Multiple	611913
● 16p11.2-16p12.2 microdeletion	16p11.2-p12.2	Multiple	
● 16p13.1 microdeletion/duplication	16p13.1	Multiple	610543
● 17q21 microdeletion/duplication	17q21	MAPT	610443
● 22q11.2 microduplication	22q11.2	Multiple	608363
● 22q11.2 distal microdeletion	22q11.2	BCR	
● 22q13.3 microdeletion	22q13.3	SHANK3	606232
● Xp11.3 microdeletion	Xp11.3	ZNF674	300578
● Alagille syndrome	20p12.2	JAG1	118450
● Alport syndrome	Xp22.3	ACSL4	301050
● Alpha thalassemia/mental retardation	16p13.3	HBA1 / HBA2	141750
● Androgen insensitivity syndrome	Xq12	AR	300068
● Angelman syndrome	15q11.2-15q13.1	UBE3A	105830
● Aniridia type 2	11p13	PAX6	106210
● Axenfeld-Rieger syndrome	6p25	FOXC1	602482
● Axenfeld-Rieger syndrome type 1	4q25	PITX2	180500
● Bannayan-Riley-Ruvalcaba syndrome	10q23.31	PTEN	153480
● Basal cell nevus syndrome, Gorlin Syndrome	9q22.32	PTCH1	109400
● Beckwith-Wiedeman syndrome	11p15	IGF2	130650
● Blepharophimosis, ptosis epicanthus inversus (BPE)	3q22.3	FOXL2	110100
● Branchio-oto-renal Syndrome	8q13.3	EYA1	113650
● Bruton's agammaglobulinemia	Xp22.1	BTK	300300
● Campomelic dysplasia	17q24.3	SOX9	114290
● Cat-eye syndrome	22q11.21	CRKL	115470
● Charcot-Marie tooth disease	17p11.2	PMP22	118220
● CHARGE syndrome	8q12.2	CHD7	214800
● Chronic granulomatous disease, X-linked	Xp11.4	CYBB	306400
● Cleidocranial dysplasia	6p21.1	RUNX2	119600
● Congenital adrenal hypoplasia	Xp21.2	NROB1	300200
● Congenital adrenal hyperplasia	6p21.32	CYP21A2	119600
● Congenital diaphragmatic hernia	15q26.2	NR2F2	142340
● Cornelia DeLange syndrome	5p13.1	NIPBL	122470
● Cowden syndrome	10q23.31	PTEN	158350
● Cri-du-chat syndrome	5p15.2	TERT	123450
● Cystinosis	17p13.3	CTNS	219800
● Dandy-Walker malformation	3q24	ZIC1/ZIC4	220200
● Diabetes insipidus, X-linked	Xq28	AVPR2	304800



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DISORDER	LOCUS	GENE	OMIM
● DiGeorge/Velocardiofacial syndrome	22q11.2	TBX1	188400
● DiGeorge locus 2	10p13-10p14	NEBL	601362
● Dosage-sensitive sex reversal	Xp21.2	NROB1	300018
● Down syndrome critical region duplication	21q22.3	DSCR3	605298
● Duane radial ray syndrome	20q13.2	SALL4	607323
● Duchenne/Becker muscular dystrophy (large del/dup)	Xp21.2	DMD	310200
● Familial adenomatous polyposis/Gardner syndrome	5q22.2	APC	175100
● Feingold syndrome	2p24.3	MYCN	164280
● Glycerol kinase deficiency	Xp21.3	GK1	300474
● Gonadal dysgenesis	Yp11.3	SRY	480000
● Gorlin Syndrome	9q22.32	PTCH1	610828
● Greig cephalopolysyndactyly	7p14.1	GLI3	175700
● Hereditary neuropathy with pressure palsies/ Charcot-Marie-Tooth disease, Type 1A (CMT1A)	17p11.2	PMP22	162500
● Hypoparathyroidism, deafness and renal dysplasia (HDR) syndrome	10p14	GATA3	146255
● Hemophilia A	Xq28	F8	306700
● Hemophilia B	Xq27.1	F9	306900
● Holoprosencephaly type 1	21q22.3	TMEM1	236100
● Holoprosencephaly type 2	2p21	SIX3	157170
● Holoprosencephaly type 3	7q36	SHH	142945
● Holoprosencephaly type 4	18p11.3	TGIF1	142946
● Holoprosencephaly type 5	13q32	ZIC2	609637
● Holt-Oram syndrome-related microdeletion	12q24.21	TBX5 / TBX3	142900
● Infantile spasm syndrome, X-linked	Xp22.13	CDKL5	308350
● Infantile spasm syndrome, X-linked	Xp21.3	ARX	308350
● Jacobsen syndrome	11q23-11q25	Multiple	147791
● Juvenile retinoschisis, X-linked	Xp22.13	RS1	312700
● Kallmann syndrome	Xp22.31	KAL1	308700
● Langer-Giedion syndrome; Hereditary multiple exostoses (HME)	8q24.11	EXT1 / TRPS1	150230
● Leri-Weill dyschondrosteosis	Xp22.33/Yp11.32	SHOX	127300
● Lesch-Nyhan syndrome	Xq26.2	HPRT1	300322
● Lowe syndrome	Xq25	OCRL	309000
● Marfan syndrome	15q21.1	FBN1	154700
● MECP2 duplication mental retardation syndrome	Xq28	MECP2	300260
● Microphthalmia with linear skin defects	Xp22.2	HCCS	309801
● Microphthalmia with cataract type 2	14q23.1	SIX6	212550
● Microphthalmia type 3, syndromic	3q26.33	SOX2	206900
● Miller-Dieker syndrome	17p13.3	PAFAH1B1	247200
● Mowat-Wilson syndrome	2q22.3	ZEB2	235730
● Nablus mask-like facial syndrome	8q21.3-8q22.1	Multiple	608156
● Nail-patella syndrome	9q33.3	LMX1B	161200
● Nephronophthisis	2q13	NPHP1	256100
● Neurofibromatosis type 1	17q11.2	NF1	162200
● Neurofibromatosis type 2	22q12.2	NF2	101000
● Noonan syndrome	12q24	PTPN11	176876
● Oculocutaneous albinism 2	15q13.1	OCA2	203200
● Opitz syndrome	Xp22	MID1	300000
● Ornithine transcarbamylase deficiency	Xp11.4	OTC	311250
● Oral-facial-digital syndrome type 1	Xp22.2	OFD1	311200
● Pallister-Killian syndrome	12p arm	Multiple	601803



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DISORDER	LOCUS	GENE	OMIM
● Pelizaeus-Merzbacher syndrome	Xq22.2	PLP1	312080
● Pseudovaginal perineoscrotal hypospadias	2p23.1	SRD5A2	607306
● Polycystic kidney disease	16p13.3	PKD1	601313
● Potocki-Shaffer syndrome	11p11.2	EXT2 / ALX4	601224
● Potocki-Lupski syndrome	17p11.2	RAI1	610883
● Prader-Willi syndrome	15q11.2-15q13.1	SNRPN	176270
● Prader-Willi-like syndrome	6q16.3	SIM1	176270
● Renal cysts and diabetes syndrome	17q12	HNF1B	137920
● Retinoblastoma	13q14.2	RB1	180200
● Rett syndrome	Xq28	MECP2	312750
● Rubinstein-Taybi syndrome	16p13.3	CREBBP	180849
● Saethre-Chotzen syndrome	7p21.1	TWIST1	101400
● Sex reversal, autosomal	9p24.3	Multiple	154230
● Smith-Magenis syndrome	17p11.2	RAI	182290
● Sotos syndrome	5q35.3	NSD1	117550
● Speech language disorder	7q3.1	FOXP2	602081
● Split-hand/foot malformation type 1	7q21.3	SHFM1	183600
● Split-hand/foot malformation type 3	10q24.32	FBXW4	600095
● Steroid sulfatase deficiency	Xp22.31	STS	308100
● Stickler syndrome type 1	12q13.11	COL2A1	108300
● Stickler syndrome type 2	1p21.1	COL11A1	604841
● Synpolydactyly	2q31.1	HOX cluster	186000
● Townes-Brocks syndrome	16q12.1	SALL1	107480
● Trichorhinophalangeal syndrome type 1	8q24.12	TRPS1	190350
● Tuberous sclerosis type 2	16p13.3	TSC2	191100
● Ulnar-mammary syndrome	12q24.21	TBX3	181450
● Van der Woude syndrome	1q32.2	IRF6	119300
● Waardenburg syndrome type 1	2q36.1	PAX3	193500
● Waardenburg syndrome type 2/Tietz syndrome	3p14.1	MITF	193510
● WAGR syndrome	11p13	PAX6 / WT1	194072
● Williams-Beuren syndrome	7q11.23	ELN	194050
● Wilms tumor	11p13	WT1	194070
● Wolf-Hirschhorn syndrome	4p16.3	Multiple	194190
● X-linked heterotaxy	Xq26.3	ZIC3	306955
● X-linked lissencephaly	Xq23	DCX	300067
● X-linked lymphoproliferative syndrome	Xq25	SH2D1A	308240
● X-linked mental retardation MRX21	Xp21.3	IL1RAPL1	300143
● X-linked MR with growth hormone deficiency	Xq27.1	SOX3	300123
● XIST deletion on ring X chromosomes	Xq13.2	XIST	314670

● Whole chromosome and segmental aneuploidies	All chromosomes
● Subtelomeric rearrangements	41 regions
● Pericentromeric rearrangements	43 regions

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