



Conditions included in the Genzyme Genetics microarray	Probe Location	Critical Gene(s)
1p31.1-p22 microdeletion	1p31.1-p22	Multiple
1p36 microdeletion	1p36	Multiple
1q21.1 microdeletion	1q21.1	Multiple
1q41-q42 microdeletion	1q41-q42	Multiple
2p21 microdeletion	2p21	SLC3A1, PREPL, PPM1B, C2orf34
2q32.2 microdeletion (cleft palate)	2q32.2-q33	SATB2
3q29 microdeletion syndrome	3q29	Multiple
8p23.1 microdeletion (Diaphragmatic hernia 2, DIH2)	8p23.1	Multiple
8q21.3-q22.1 microdeletion	8q21.3-q22.1	Multiple
9q22.3 microdeletion	9q22.3	Multiple
9q34.3 deletion syndrome	9q34.3	Multiple, EHMT1 candidate
14q11.2 microdeletion	14q11.2	Multiple
14q22-q23 microdeletion	14q22-q23	Multiple
15q13.3 microdeletion	15q13.3	Multiple
15q21 microdeletion	15q21	Multiple
15q24 microdeletion	15q24	Multiple
15q26.1-overgrowth & developmental disorders	15q26.1	Multiple
16p11.2 microdeletion/microduplication	16p11.2	Multiple
16p11.2-p12.1 microdeletion	16p11-p12.1	Multiple
16q11.2-q12.2 microdeletion	16q11.2-q12.2	Multiple
17p11.2 microduplication (Potocki-Lupski syndrome)	17p11.2	Multiple
17q11.2-q13.2 deletion	17q11.2-q13.2	Multiple
17q12 microdeletion	17q12	Multiple
17q21.31 microdeletion/duplication	17q21.31	Multiple
22q11.2 microduplication syndrome	22q11.2	Multiple
22q13.3 deletion syndrome (Phelan-McDermid)	22q13.3	Multiple (SHANK3 candidate)
Aarskog-Scott syndrome (Faciogenital dysplasia)	Xp11.22-p11.21	FGD1
Alagille syndrome 1 (ALGS1)	20p12.2	JAG1
Alagille syndrome 2 (ALGS2)	1p12	NOTCH2
Albright hereditary osteodystrophy-like syndrome/Brachydactyly-mental retardation syndrome	2q37	Multiple
Androgen insensitivity syndrome	Xq12	AR
Angelman syndrome	15q11.2	UBE3A
Aniridia II	11p13	PAX6
Bannayan-Riley-Ruvalcaba syndrome (BRRS)	10q23.31	PTEN
Basal cell nevus syndrome (Gorlin-Goltz syndrome)	9q22.32	PTCH1
Becker-Duchenne Muscular Dystrophy	Xp21.1	DMD
Beckwith-Wiedemann syndrome	11p15.5	IGF2
Blepharophimosis, ptosis, epicanthus inversus (BPES)	3q22.3	FOXL2
Branhiootorenal (BOR) syndrome 1 (Melnick-Frasier syndrome)	8q13.3	EYA1
Campomelic dysplasia (CMPD)	17q24.3	SOX9
Cat eye syndrome	22q11.1	Multiple
Charcot-Marie-Tooth disease type 1A (CMT1A)/Heredity neuropathy with liability to pressure palsies	17p12	PMP22
CHARGE	8q12.2	CHD7
Cleidocranial dysplasia	6p21.1	RUNX2
Congenital adrenal hypoplasia	Xp21.3	NROB1
Congenital diaphragmatic hernia (CDH)	15q26.1-q26.2	NR2F2, CHD2
Congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD syndrome)	Xq28	NSDHL
Cornelia de Lange syndrome 1	5p13.2	NIPBL
Cowden syndrome	10q23.31	PTEN
Cri-du-chat syndrome	5p15.2	TERT, multiple
Dandy-Walker malformation	3q24	ZIC1/ZIC4
DiGeorge/Velocardiofacial syndrome	22q11.21	TBX1, HIRA
DiGeorge/velocardiofacial syndrome spectrum of malformation 2 (DGS2)	10p14-p13	Multiple
Dosage-sensitive sex reversal	Xp21.3	NROB1
Down Syndrome Critical Region gene 3	Xp11.23	GATA1
Down syndrome critical region gene 4	21q22.13	DSCR4
Doyle honeycomb retinal dystrophy	2p16.1	EFEMP1
Familial adenomatous polyposis (FAP)/Gardner/MR	5q21-q22	APC
Feingold syndrome	2p24.3	MYCN
Fragile X syndrome - rare microdeletion	Xq27.3	FMR1
Glycerol kinase deficiency (hyperglycerolemia)	Xp21.2	GK
Greig cephalopolysyndactyly syndrome	7p14.1	GLI3
Holoprosencephaly 1 (Epilepsy)	21q22.3	TMEM1
Holoprosencephaly 2	2p21	SIX3
Holoprosencephaly 3	7q36.3	SHH
Holoprosencephaly 4	18p11.31	TGIF
Holoprosencephaly 5	13q32.3	ZIC2
Holoprosencephaly 6	2q37.1-q37.3	Multiple
Holoprosencephaly 7	9q22.32	PTCH1
Holoprosencephaly 8	14q13.1-q13.2	Multiple
Holt-Oram syndrome	12q24.21	TBX5
Hypomyelinating leukodystrophy (HLD4)	2q33.1	HSPD1
Hypoparathyroidism, sensorineural deafness, and renal disease (HDR syndrome)	10p14	GATA3
Infantile spasm syndrome, X-linked	Xp21.3	ARX
Jacobsen syndrome	11q23-11qter	Multiple
Joubert syndrome 4	2q13	NPHP1
Kallmann syndrome 1	Xp22.31	KAL1
Leigh syndrome, French-Canadian type	2p21	LRPPRC
Leri-Weil dyschondrosteosis	Xp22.33; Yp11.32	SHOX
Limb-girdle muscular dystrophy type 2J (LGMD2J)	2q31.2	TTN
Loeys-Dietz syndrome, type 2B	3p24.1	TGFBR2
Lowe oculocerebrorenal syndrome	Xq25	OCRL1
Marfan syndrome	15q21.1	FBN1
Menkes disease	Xq12-q13	ATP7A
Metachromatic leukodystrophy	22q13.33	ARSA
Microphthalmia 7 with linear skin defects	Xp22.2	HCCS
Miller-Dieker lissencephaly syndrome	17p13.3	PAFAH1B1
Mitochondrial complex deficiency 1/leukodystrophy	11q13.2	NDUFV1

Conditions included in the Genzyme Genetics microarray	Probe Location	Critical Gene(s)
Mowat-Wilson syndrome	2q22.3	ZEB2
Muscle-eye-brain disease	1p34.1	POMGNT1
Nail-patella syndrome	9q33.3	LMX1B
Nephronophthisis 1	2q13	NPHP1
Nephropathic cystinosis	17p13.3	CTNS
Neurofibromatosis type 1	17q11.2	NF1
Neurofibromatosis type 2	22q12.2	NF2
Noonan syndrome 1	12q24.13	PTPN11
Noonan syndrome 4	2p22.1	SOS1
Oculocutaneous albinism 2	15q13.1	OCA2
Opitz GBBB syndrome, X-linked	Xp22.2	MID1
Ornithine transcarbamylase deficiency	Xp11.4	OTC
Oro-facial-digital	Xp22.2	OFD1
Otofaciocervical (OFC) syndrome	8q13.3	EYA1
Pallister-Killian	12p	Multiple
Pelizaeus-Merzbacher disease	Xq22.2	PLP1
Pitt-Hopkins syndrome	18q21.2	TCF4
Pituitary Hormone Deficiency, combined, 4	1q25.2	LHX4
Polycystic kidney disease (PKD1)	16p13.3	PKD1
Potocki-Shaffer syndrome	11p11.2	EXT2, ALX4
Prader-Willi syndrome	15q11.2	SNRPN
Prader-Willi-like phenotype	6q16.3	SIM1
Renal cysts and diabetes syndrome	17q12	HNF1B
Retinoblastoma/MR	13q14.2	RB1
Reff syndrome	Xq28	MECP2
Rieger syndrome type 1	4q25	PITX2
Rubinstein-Taybi syndrome	16p13.3	CREBBP
Saethre-Chatzen syndrome	7p21.1	TWIST1
Short stature, pituitary and cerebellar defects, small sella turcica	1q25	LHX4
Smith-Magenis syndrome	17p11.2	RAI1
Sotos syndrome	5q35.3	NSD1
Split-hand/foot malformation 1	7q21.3	SHFM1
Split-hand/foot malformation 3	10q24.32	FBXW4
Split-hand/foot malformation 4	3q28	TP63
Split-hand/foot malformation 5	2q31.1	DLX1, DLX2
Steroid sulfatase deficiency	Xp22.32	STS
Stickler syndrome 1	12q13.11	COL2A1
Synpolydactyly & Syndactyly II	2q31.1	HOXD gene cluster
Trichorhinophalangeal syndrome, type 1	8q23.3	TRPS1
Tuberous sclerosis 2	16p13.3	TSC2
Ulnar-mammary syndrome	12q24.21	TBX3
Van der Woude syndrome	1q32.2	IRF6
Waardenburg syndrome IIA	3p14.1	MITF
Walker-Warburg syndrome	9q34.13	POMT1
Williams-Beuren syndrome	7q11.23	ELN
Wilms Tumor 1	11p13	WT1
Wilms tumor, aniridia, genitourinary anomalies, mental retardation syndrome; WAGR	11p13	WT1, PAX6
Wolf-Hirschhorn syndrome	4p16.3	Multiple
X-inactivation center (XIST)	Xq13.2	XIST
X-linked agammaglobulinemia (Bruton-type agammaglobulinemia)	Xq22.1	BTX
X-linked congenital adrenal hypoplasia	Xp21.2	NROB1
X-linked early infantile epileptic encephalopathy 2, X-linked infantile spasm syndrome 2	Xp22.13	CDKL5
X-linked lissencephaly-1	Xq22.3-q23	DCX
X-linked lymphoproliferative syndrome	Xq25	SH2D1A
X-linked mental retardation 21	Xp21.3	IL1RAPL1
X-linked MR with isolated growth hormone deficiency	Xq27.1	SOX3
X-linked visceral heterotaxy	Xq26.3	ZIC3
Xp11.3 microdeletion	Xp11.3	RP2, ZNF674
XX male	Yp11.31	SRY
XY gonadal dysgenesis (XY female)	Yp11.31	SRY
All 41 unique subtelomeric regions	41 sites	
All 43 unique pericentromeric regions	43 sites	
Chromosomal aneuploidy for 24 chromosomes	24 chromosomes	

Regions included in the Genzyme Genetics microarray associated with Autism Spectrum Disorders

Chromosome band where probes are located	Critical Genes(s)
1q21.1	
1q23.3-q24.2	
1q24	
2p16.3	NRXN1
2p15-p16.1	
2q37.2-q37.3	
2q37.3	AGAP1
3p14.2	FHIT
3q29	PAK2, DLG1
5p13.2	NIPBL
5p15	
5q35.3	NSD1
6q16.3	GRIK2
6q25.1-q25.3	
7q11.23	ELN, GTF2I
7q21.13	
7q31.1	FOXP2
7q31.32	
7q35-q36	CNTNAP2
8q24.11	EXT1
9q34.1	TSC1
9q34.3	
10p14	GATA3
10q11.23-q21.2	
10q22.3-q23.31	
10q23.31	PTEN
12p13.33	CACNA1C

Chromosome band where probes are located	Critical Genes(s)
13q12.2	
13q21.32	PCDH9
15q11.2	CYFIP1
15q11.2	SNRPN
15q11.2-q12	GABRA5, GABRG3
15q11.2-q13	UBE3A, GABRB3
16p11.2	
16p13.3	CACNA1H, TSC2
17p11.2	RAI1
17q11.2	NF1
18q12	
18q21.2-qter	
20p11.22-p11.23	
20p13	
trisomy 21	
22q11.2	
22q11.21	HIRA, TBX1
22q13.31	
22q13.33	SHANK3
Xp11.23	MAOA
Xp21.2	
Xp21.3	ARX, IL1RAPL1
Xp22.31-p22.32	NLGN4
Xq13.1	NLGN3
Xq27.3	FMR1
Xq28	MECP2