

Maximise information in known disease regions

CytoChip constitutional arrays

BlueGnome's constitutional CytoChips are disease focused microarrays that include extra tiling on regions that are associated with recognised genetic disorders. Disease regions are selected through the OMIM database, consultation with the cytogenetic community and from recent publications. CytoChip and CytoChip Oligo arrays include 101 registered OMIM diseases and a further 9 newly recognised disease regions; on the CytoChip Focus Constitutional array there are 97 OMIM diseases. A total of 143 disease associated genes are covered at high resolution. Each gene is tiled by 3 BAC probes on the CytoChip and CytoChip Focus Constitutional arrays and by 20 oligos on the Cytochip Oligo array.

Disease	OMIM	Locus	Cyto band	Disease	OMIM	Locus	Cyto band
1p36 DELETION SYNDROME	607872	P21127-10	1p36.33	DIGEORGE SYNDROME/VELOCARDIOFACIAL SYNDROME SPECTRUM OF MALFORMATION 2	601362	D10S293	10p14
1q21.1 DELETION SYNDROME, 1.35-MB	612474	N/A	1q21.1	NEBL			10p12.31
1q21.1 DELETION SYNDROME, 200-KB ²	274000	N/A	1q21.1	DIGEORGE SYNDROME; DGS	188400	HIRA, TBX1	22q11.21
1q41-q42 MICRODELETION ²	N/A	N/A	1q41	DOSAGE-SENSITIVE SEX REVERSAL; DSS	300018	NROB1	Xp21.2
2p15-p16.1 MICRODELETION ²	N/A	N/A	2p15	DOWN SYNDROME	190685	DSCR2	21q22.2
3q29 MICRODELETION SYNDROME	609425	DLG1, PAK2	3q29	GATA1			Xp11.23
8q21.3-q22.1 MICRODELETION ²	N/A	N/A	8q21.3-q22.1	FEINGOLD SYNDROME	164280	MYCN	2p24.3
9q22.3 MICRODELETION ²	N/A	N/A	9q22.31-q22.33	FRAGILE X MENTAL RETARDATION SYNDROME	300624	FMR1	Xq27.3
15q13.3 MICRODELETION SYNDROME	612001	N/A	15q13.2-q13.3	GREIG CEPHALOPOLYSYNDACTYLY SYNDROME; GGPS	175700	GLI3	7p14.1
15q24 MICRODELETION ²	N/A	N/A	15q24.1-q24.2	HETEROTAXY, VISCERAL, 1, X-LINKED; HTX1	306955	ZIC3	Xq26.3
16p11-p12.1 MICRODELETION ²	N/A	N/A	16p11.2-p12.2	HIRSCHSPRUNG DISEASE, SUSCEPTIBILITY TO, 1; HSCR1	142623	RET ²	10q11.21
17q11.2-q12 MICRODELETION ²	N/A	N/A	17q11.2-q12	HOLOPROSENCYPHALY	236100	TMEM1	21q22.3
17q12 MICRODELETION ²	N/A	N/A	17q12	HOLOPROSENCYPHALY 2; HPE2	157170	SIX3	2p21
17q21.31 MICRODELETION SYNDROME	610443	CRHR1, MAPT	17q21.31	HOLOPROSENCYPHALY 3; HPE3	142945	SHH	7q36.3
20q13.13-q13.2 MICRODELETION ²	N/A	N/A	20q13.2	HOLOPROSENCYPHALY 4; HPE4	142946	TGIF1	18p11.31
22q11.2 DELETION SYNDROME, DISTAL	611867	N/A	22q11.21-q11.23	HOLOPROSENCYPHALY 5; HPE5	609637	ZIC2	13q32.3
22q13.3 DELETION SYNDROME	606232	SHANK3	22q13.33	HOLT-ORAM SYNDROME; HOS	142900	TBX5 ² , TBX3 ²	12q24.21
ADENOMATOUS POLYPOSIS OF THE COLON; APC	175100	APC	5q22.2	HYPERGLYCEROLEMIA	307030	GK3P	Xp21.2
ADRENAL HYPOPLASIA, CONGENITAL; AHC	300200	NROB1	Xp21.2	HYPOPARATHYROIDISM, SENSORINEURAL DEAFNESS, AND RENAL DISEASE	146255	GATA3	10p14
ALAGILLE SYNDROME 1; ALGS1	118450	JAG1	20p12.2	ICHTHYOSIS, X-LINKED; XLI	308100	STS	Xp22.31
ANGELMAN SYNDROME; AS	105830	UBE3A	15q11.2	JACOBSEN SYNDROME; JBS	147791	N/A	11q23.1-q24.1
		ATP10A	15q12	JOHANSON-BLIZZARD SYNDROME; JBS	243800	UBR1	15q15.2
		MECP2	Xq28	JOUBERT SYNDROME 4; JBTS4	609583	NPHP1	2q13
ANIRIDIA; AN	106210	PAX6	11p13	KABUKI SYNDROME	147920	N/A	8p22
AUTISM	209850	N/A	16p11.2	KALLMANN SYNDROME 1; KAL1	308700	KAL1	Xp22.31
		RPL10	Xq28	LERI-WEILL DYSCHONDROSTEOSIS; LWD	127300	SHOX ¹	Xp22.33
AUTISM, X-LINKED, SUSCEPTIBILITY TO, 2	300495	NLGN4X	Xp22.31-p22.32	LISSENCEPHALY, X-LINKED, 1; LISX1	300067	DCX	Xq22.3-q23
AUTISM, X-LINKED, SUSCEPTIBILITY TO, 1	300425	NLGN3	Xq13.1	MENTAL RETARDATION, X-LINKED, WITH PANHYPOPITUITARISM	300123	SOX3	Xq27.1
AUTISM, X-LINKED, SUSCEPTIBILITY TO, 3	300496	MECP2	Xq28	METACHROMATIC LEUKODYSTROPHY	250100	ARSA	22q13.33
BASAL CELL NEVUS SYNDROME; BCNS	109400	PTCH1	9q22.32	MICROPTHALMIA, SYNDROMIC 7; MCOPS7	309801	HCCS, ARHGAP6	Xp22.2
BECKWITH-WIEDEMANN SYNDROME; BWS	130650	NSD1	5q35.2-q35.3	MILLER-DIEKER LISSENCEPHALY SYNDROME; MDLS	247200	PAFAH1B1, YWHAE, HIC1	17p13.3
		H19, IGF2	11p15.5	MITOCHONDRIAL COMPLEX I DEFICIENCY	252010	NDUFS2	1q23.3
		KCNQ1	11p15.4-p15.5	NDUFS1			2q33.3
		CDKN1C	11p15.4	NDUFS6			5p15.33
BRACHYDACTYLY-MENTAL RETARDATION SYNDROME; BDMR	600430	Z51342	2q37.3	NDUFS4			5q11.2
BRANCHIOOTORENAL SYNDROME 1; BOR1	113650	EYA1	8q13.3	NDUFA12L			5q12.1
BRUTON AGAMMAGLOBULINEMIA TYROSINE KINASE; BTK	300300	BTK	Xq22.1	PTPMT1			11p11.2
BUSCHKE-OLLENDORFF SYNDROME	166700	N/A	12q14.2-q15	NDUFS8, NDUFV1			11q13.2
CAMPOMELIC DYSPLASIA	114290	SOX9	17q24.3	NDUFV2			18p11.22
CAT EYE SYNDROME; CES	115470	CECR5, CECR1, CECR6	22q11.1	NDUFS7			19p13.3
CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A; CMT1A	118220	PMP22	17p12	MUSCULAR DYSTROPHY, BECKER TYPE; BMD	300376	DMD	Xp21.1-p21.2
CHARCOT-MARIE-TOOTH DISEASE, X-LINKED, 1; CMTX1	302800	GJB1	Xq13.1	DXS7			Xp11.3
CHARGE SYNDROME	214800	CHD7	8q12.2	MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD	310200	DMD	Xp21.1-p21.2
CLEIDOCRANIAL DYSPLASIA; CCD	119600	RUNX2	6p12.3	NAIL-PATELLA SYNDROME; NPS	161200	LMX1B	9q33.3
CORNELIA DE LANGE SYNDROME 1; CDLS1	122470	NIPBL	5p13.2	NEPHRONOPHTHISIS 1; NPHP1	256100	NPHP1	2q13
CRI-DU-CHAT SYNDROME	123450	TERT	5p15.33	NEUROFIBROMATOSIS, TYPE I; NF1	162200	NF1	17q11.2
		Z23908	5p15.2	NEUROFIBROMATOSIS, TYPE II; NF2	101000	NF2	22q12.2
DANDY-WALKER SYNDROME; DWS	220200	ZIC1, ZIC4	3q24	NEUROPATHY, HEREDITARY, WITH LIABILITY TO PRESSURE PALSIES; HNPP	162500	PMP22	17p12
DIAPHRAGMATIC HERNIA, CONGENITAL	142340	CHD2	15q26.1	NOONAN SYNDROME 1; NS1	163950	PTPN11	12q24.13
		NR2F2	15q26.2				

Disease	OMIM	Locus	Cyto band
PELIZAEUS-MERZBACHER DISEASE; PMD	312080	PLP1	Xq22.2
POLYCYSTIC KIDNEY DISEASE, INFANTILE SEVERE, WITH TUBEROUS SCLEROSIS; PKDTS	600273	PKD1	16p13.3
POTOCKI-LUPSKI SYNDROME; PTLIS	610883	RAI1, MFAP4, FLII	17p11.2
POTOCKI-SHAFFER SYNDROME	601224	ALX4, EXT2	11p11.2
PRADER-WILLI SYNDROME; PWS	176270	SIM1	6q16.3
PRADER-WILLI SYNDROME; PWS	176270	SNRPN, NDN	15q11.2
RETINOBLASTOMA; RB1	180200	RB1	13q14.2
RETT SYNDROME; RTT	312750	CDKL5 MECP2	Xp22.13 Xq28
RIEGER SYNDROME, TYPE 1; RIEG1	180500	PITX2	4q25
RUBINSTEIN-TAYBI SYNDROME; RSTS	180849	CREBBP	16p13.3
SAETHRE-CHOTZEN SYNDROME; SCS	101400	TWIST1	7p21.1
SEX-DETERMINING REGION Y; SRY	480000	SRY	Yp11.31
SMITH-MAGENIS SYNDROME; SMS	182290	RAI1, MFAP4, FLII	17p11.2
SOTOS SYNDROME	117550	NSD1	5q35.2-q35.3
SPERMATOGENIC FAILURE, NONOBSTRUCTIVE, Y-LINKED	415000	USP9Y, UTY CDY2B ¹ JARID1D NR_001537, DAZ3 ¹ , DAZ1 ¹ , DAZ2 ¹	Yq11.21 Yq11.221 Yq11.222 Yq11.223
SPLIT-HAND/FOOT MALFORMATION 1; SHFM1	183600	SHFM1	7q21.3
SPLIT-HAND/FOOT MALFORMATION 3; SHFM3	600095	FBXW4	10q24.32
SPLIT-HAND/FOOT MALFORMATION 4; SHFM4	605289	TP63	3q28
SPLIT-HAND/FOOT MALFORMATION 5; SHFM5	606708	DLX1, EVX2	2q31.1

Disease	OMIM	Locus	Cyto band
SYNPOLYDACTYLY 1; SPD1	186000	HOXD13	2q31.1
TOWNES-BROCKS SYNDROME; TBS	107480	SALL1	16q12.1
TRICHORHINOPHALANGEAL SYNDROME, TYPE I; TRPS1	190350	TRPS1	8q23.3
TRICHORHINOPHALANGEAL SYNDROME, TYPE II; TRPS2	150230	TRPS1 EXT1	8q23.3 8q24.11
TUBEROUS SCLEROSIS; TS	191100	TSC1 TSC2	9q34.13 16p13.3
VELOCARDIOFACIAL SYNDROME	192430	ARVCF, TBX1	22q11.21
WILLIAMS-BEUREN REGION DUPLICATION SYNDROME	609757	N/A	7q11.23
WILLIAMS-BEUREN SYNDROME; WBS	194050	GTF2IRD1, MLXIPL, BAZ1B, ELN, RFC2, WBSOR22, FKBP6, GTF2I, LAT2, BCL7B, TBL2, CLIP2, EIF4H, LIMK1, WBSOR27, WBSOR16, FZD9, WBSOR23	7q11.23
WILMS TUMOR 1; WT1	194070	WT1	11p13
WILMS TUMOR, ANIRIDIA, GENITOURINARY ANOMALIES, AND MENTAL RETARDATION	194072	PAX6	11p13
WOLF-HIRSCHHORN SYNDROME; WHS	194190	WHSC1 MSX1	4p16.3 4p16.2
X INACTIVATION-SPECIFIC TRANSCRIPT; XIST	314670	XIST ²	Xq13.2

¹not targeted by CytoChip Oligo, ²not targeted by CytoChip Focus Constitutional

CytoChip Focus Haematology array

The CytoChip Focus Haematology array includes extra tiling on regions that are associated with haematological malignancies.

Included in the list below are genes or loci found in regions that are reported to be deleted or amplified in haematological samples.

Cyto band	Locus	Mb start	Mb end
1p34	FUCA1	24.17	24.19
1p13	TRIM33	114.94	115.05
1q12		128.90	142.60
1q21		142.60	155.00
1q22		155.00	156.50
1q43-44		236.60	249.25
2p24	NAG	15.31	15.70
3p24.2	TOP2B	25.64	25.71
4q12		52.70	59.50
5q13		66.70	76.90
5q31		130.60	144.50
6q23.3	C-MYB	135.50	135.54
7q22		98.00	107.40
7q32		127.10	132.60
7q34		138.20	143.10
8q24	C-MYC	128.75	128.75
9p21	CDKN2_2B	22.00	22.01
9q34		130.30	141.21
11q13	CCND1 (BCL1)	69.46	69.47
11q22.3	ATM	108.09	108.24
11q23.3	D11S1341 (MPZL2)	118.13	118.14
12q15	MDM2	69.20	69.24
13q12.2	FLT3	28.58	28.67
13q14.3	RB1	48.88	49.06
13q14.3	D13S319-D13S272	50.71	50.71
17p13.1	p53	7.57	7.59
17q21.1	TOP2A	38.54	38.57
20q12-q13.12	D20S107-D20S17	38.88	44.99
20q12	TOP1	39.66	39.75
21q22	AML1 (RUNX1)	36.19	36.26
22q11.2	Distal to DiGeorge	17.90	22.20

Genomic locations taken from Feb.2009 build hg19

Tiling is included over genes that are associated with balanced rearrangements traditionally detected using FISH or RT-PCR. Important gene fusions at balanced rearrangements covered on the array are listed in the table below. Truly balanced translocations cannot be detected by arrayCGH, however, there is evidence that some cytogenetically balanced translocations result in detectable copy number imbalances at the higher resolution provided by arrays.

Sample	Translocation	Fusion
Acute Lymphoblastoid Leukaemia	t(9;22)(q34;q11)	BCR-ABL
	t(12;21)(p12;q22)	TEL-AML1
Acute Myeloid Leukaemia	t(8;21)(q22;q22.3)	AML1-ETO
	t(15;17)(q22;q21)	PML-RARA
Non-Hodgkin Lymphoma	t(2;5)(p23;q35)	ALK-NPM1
	t(11;14)(q13;q32)	IGH-BCL1
	t(14;18)(q32;q21)	BCL2-IGH
MLL rearranged leukaemia	t(8;14)(q24;q32)	MYC-IGH
	t(4;11)(q21;q23)	MLL-AF4
	t(6;11)(q27;q23)	MLL-AF6
	t(9;11)(p21;q23)	MLL-AF9
	t(10;11)(p12;q23)	MLL-AF10
t(11;19)(q23;p13.1)	MLL-ELL	
t(X;11)(q13;q23)	MLL-AFX1	

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