Regiões alvo de doenças detectadas por CGH:

Doenças Relacionadas	Gene	Chr/Banda	OMIM #
1P31deleção	DIRAS3	1p31	605193
1p36microdeleção	SKI	1p36	607872
1p36microdeleção	TP73	1p36	607872
2p15-16.1microdeleção	múltiplo	2p15-16.1	612513
2p37.3deleção	HDAC4	2q37	600430
3q29microdeleção	PAK2	3q29	609425
6p24microdeleção	FKHL7	6p24	612852
8p23deleção	CTSB	8p23	116810
9pdeleção	DMRT1	9p24	158170
9pdeleção	DMRT2	9p24	158170
9q34.3microdeleção	EHMT1	9q34.3	610253
10q22-23deleção	GRD1	10q22	610659
12q14.1-q15deleção	GRP1	12q14	604597
13qdeleção	GPC5	13q31	602446
13qdeleção	GPC6	13q31	604404
13qdeleção	PCDH9	13q21	603581
14q11-q22deleção	CHD8	14q11	613457
14q11-q22deleção	SUPT16H	14q11	613457
14q11-q22microdeleção	SIX1	14q22	608389
(Síndrome Branchicotic 3)			
14q11-q22microdeleção	SIX4	14q22	608389
14q11-q22microdeleção	SIX6	14q22	212550
14q11-q22microdeleção	OTX2	14q22	610125
14q22microd/orofacial 11	BMP4	14q22	600625
15q13.3microdeleção	CHRNA7	15q13	612001
17q21.31microdeleção	CRHR1	17q21.31	610443
17q21.31microduplicação	MAPT	17q21.31	613533
18qmicrodeleção	TCE838	18q21	609522
18qdeleção	ZNF407	18p22	301808
18qdeleção	GALR1	18q23	600377
22q11.2	BCR	22q11	611867
Aarskog-Scott	FGD1	Xp11	305400
1q44microdeleção	AKT3		
Adrenal hypoplasia congenital	NR081	Xp21	300200
Adult-onset autosomal domina	LMNB1	5q23	169500
Agammaglobulnemia ligX	BTK	Xq22.1	300755
Alagille	JAG1	20p12	118450
Alexander disease	NDUFV1	11q13	203450
Alfa-talassemia-RM	HBA1&HBA2	16p13	141750
Alfa-talassemia-RM	SOX8	16p13.3	141750
Alfa-talassemi-RM-LigX	ATRX	Xq21	300032
Alport lig-X	COL4A5	Xq22.3	301050
Androgen insensitivity	AR	Xq12	300068
Angelman/Prader-Willi	MAGEL2	15q11	105830
Angelman/Prader-Willi	MKRN3	15q11	105830

Regiões alvo de doenças detectadas por CGH:

Angelman/Prader-Willi	NDN	15q11	105830
Angelman/Prader-Willi	NIPA1	15q11	105830
Angelman/Prader-Willi	NIPA2	15q11	105830
Angelman/Prader-Willi	PWS IC	15q11	105830
Angelman/Prader-Willi	SNORD10/7/64/1	15q11	105830
Angelman/Prader-Willi	SNORD10 9B	15q11	105830
Angelman/Prader-Willi	SnoRNA	15q11	105830
Angelman/Prader-Willi	SNRPN	15q11	105830
Angelman/Prader-Willi	SNURF	15q11	105830
Angelman/Prader-Willi	UBE3A	15q11	105830
Aniridia	PAX6	11p13-p14	106210
Anxiety-related personality tra	SLC6A4	17p11	607834
Atrial fibrilation	GJA5	1q21	121013
Atrial septal defect	NKX2-5	5q35	108900
Autism	A2BP1	16p13	209850
Autism	ATP10A	15q12	209850
Autism	CADPS2	7q31	209850
Autism	CNTN4	3p25	209850
Autism	CNTNAP2	7q35-36	209850
Autism	DLGAP2	8p23	209850
Autism	EGR2	10q21	209850
Autism	EN2	7q36	209850
Autism	GABRB3	15q12	209850
Autism	MET	7q31	209850
Autism	NLGN3	Xq13	209850
Autism	NLGN4X	Xp22	209850
Autism	SHANK3	22q13	209850
Autism	SLC4A10	2q24	209850
Autism	DISC1	1q42	209850
Autism	NRXN1	2p16	209850
Autism	NPTX2	7q22	600750
Bartler 1	SLC12A1	15q21	601678
Bartler 2	KCNJ1	11q24	241200
Bartler 3	CLCNKB	1p36	607364
Bartler 4A	BSND	1p32	602522
Bartler 4B	CLCNKA	1p36	613090
Bartler AD hipocalcemia	CASR	3q13.33-	601199
Beckwith - Wiedemann	H19 IC	11p15	130650
Beckwith - Wiedemann	KCNQ1 IC	11p15	130650
Beckwith - Wiedemann	CDKN1C	11p15	130650
Beckwith - Wiedemann	H19	11p15	130650
Beckwith - Wiedemann	IGF2	11p15	130650
Beckwith - Wiedemann	INS	11p15	130650
Beckwith - Wiedemann	PHLDA2	11p15	130650
Beckwith - Wiedemann	SLC22A18	11p15	130650
Bilateral frontparietal polymic	GPR56	16q13	606854

Regiões alvo de doenças detectadas por CGH:

Birk-Barel	KCNK9	8q24	605874
Blepharophimosis	FOXL2	3q22	110100
Borjeson-Forssman-Lehmann	PHF6	Xq26	301900
Brachydactyly type C	GDF5	20q11	113100
Branchiootorenal	EYA1	8q13	113650
Breast cancer	BRCA2	13q13	114480
Brunner	MAOA	Xp11	300615
Campomelic dysplasia	SOX9	17q24	114290
Carnitine biosynthesis pathwa	PAR2	Xq28	300777
Cataract juvenile with microc	SLC16A12	10q23	612018
Cat Eye Syndrome-typeI	ATP6V1E1	22q11	115470
Cerebral amyloid angiopathy	APP	21q21	605714
Charcot-marie-Tooth disease 1	PRPS1	Xq22	311070
Charcot-marie-Tooth disease t	PMP22	17p11	118220
CHARGE	CHD7	8q12	214800
Chondrodysplasia punctatarec	CDPX1	Xp22	302950
Choroideremia	CHM	Xp21	303100
Chronic granulomatous diseas	CYBB	Xp11	306400
Chronic pancreatitis	SPINK1	5p32	167800
Cleft lip	MSX1	4p16	608874
Cleft palate	SATB2	2p32	119540
Cleidocranial dysplasia	RUNX2	6p21	119600
Coffin-Lowry	RPS6KA3	Xp22	303600
Congenital adrenal hyperplasia	CYP21A2	6p21.32	201910
Congenital diaphagmatic herni	NR2F2	15q26	142340
Cornelia de Lange 1	NIPBL	5p13	122470
Cornelia de Lange 2	SMC1L1	Xp11	300590
Cowden	BMPR1A	10q23	158350
Craniosynostosis	MSX2	5q35	604757
Craniosynostosis	SOX6	11p15.1-p15.2	218350
Creatine deficiency/ X-lig.mer	SLC6A8	16p11	300352
Cri-du-chat	TERT	5p15	123450
Cryptorchidism idiopathic	LGR8	13q13	219050
Currarino	HLXB9	7q36	176450
Cystinosis	CTNS	17p13	219800
Cystinuria with mitochondrial	PREPL	2p21	606407
Cystinuria with mitochondrial	SLC3A1	2p21	606407
Dandy-Walker	ZIC1	3q24	220200
Dandy-Walker	ZIC4	3q24	220200
Danon disease	LAMP2	Xq24	300257
Deafness autos.ressev. 1A	GJB2	13q12	220290
Deafness sanscrineural and ma	CATSPER2	15q15	611102
Deafness dystonia optic neuro	TIMM8A	Xq22	304700
Delayed cranial ossification	CBFB	16q22	121360
Diabetes insipidus nephrogeni	AVPR2	Xq28	304800
Diabetes melitus transient neo	ZAC	6q24.2	601410

Regiões alvo de doenças detectadas por CGH:

Diabetes melitus transient neo	HYMAI	6q24	601410
Diamond Blackfan anemia	RPS19	19q13	105650
Diaphragmatic hernia 3	ZFPM2	8q23	610187
DiGeorge1	CRKL	22q11	188400
DiGeorge1	GATA4	8p23	188400
DiGeorge1	HIRA	22q11	188400
DiGeorge1	MFHAS1	8p23	188400
DiGeorge1	TBX1	22q11	188400
DiGeorge2	GATA3	10p14	601362
DiGeorge2	NEBL	10p12	601362
Down syndrome critical regior	DSCR1	21q22	190685
Down syndrome critical regior	DSCR3	21q22	190685
Down syndrome critical regior	DYRK1A	21q22	190685
Down syndrome critical regior	RUNX1	21q22	190685
Duane radial ray	SALL4	20q13	607323
Duchenne muscular dystrophy	DMD	Xp21.1-p21.2	310200
Dyggve melchiorClausen dise	DYM	18q21	223800
Dystonia 11	PEG10	7p21	159900
Dystonia 11	PEG10IC	7p21	159900
Dystonia 11	SGCE	7p21	159900
Ectodemal dysplasia	EDA	Xq13	305100
Ehlers-Danlos	COL1A2	7q21	225320
Exudative vireoretinopathy	FZD4	11q14	133780
Fabry disease	GLA	Xq22	301500
Fanconi anemia	FANCA	16q24	227650
Fanconi anemia	FANCB	Xp22	300515
Feingold	MYCN	2q24	164280
FG syndrome 4	CASK	Xp11	300172
Forebrain defects	TDGF1	3p21	187395
Focal dermal hypoplasia	PORCN	Xp11	300651
FMR1 sindrome microdeleção	FMR1	Xq27	300624
FMR1 sindrome microdeleção	FMR2	Xq28	300624
Fryns 1q41	DISP1	1q41	229850
Gardner	APC	5q22	175100
Gitelman	SLC12A3	16q13	263800
Glycerol kinase deficiency	GK	Xp21	307030
Gonadal dysgenesis	SRY	Yp11.31	480000
Greig cephalopolysyndactyly	GLI3	7p14	175700
Hemophilia A	F8	Xq28	306700
Hemophilia B	F9	Xq27	306900
Hepatocellular carcinoma	IGF2R	6q25	114550
Hereditary hemorrhagic leslan	MADH4	18q21	175050
Hereditary pancreatitis	PRSS1	7q34	276000
Heterotopia periventricular do	FLNA	Xq28	300019
Hirschsprung disease	EDNRB	13q22	600155
Hirschsprung disease plus	RET	10q11	142623

Regiões alvo de doenças detectadas por CGH:

Holoprosencephaly 1	LSS	21q22	600909
Holoprosencephaly 1	TMEM1	21q22	602108
Holopronsencephaly 2	SIX3	2p21	157170
Holopronsencephaly 3	SHH	7q36	142945
Holopronsencephaly 4	TGIF1	18p31	142946
Holopronsencephaly 5	ZIC2	13q32	609637
Holoproncensephaly 7	PTCH1	9q22	610828
Holoproncensephaly 9	GLI2	2q14	610829
Holopronsencephaly and preax	FBXW11	5q35	264480
Holt-Oram	TBX5	12q14	142900
HSAS,MASA,CRASH	L1CAM	Xq28	303350
Hunter, mucopolysaccharidosi	IDS	Xq28	309900
Hyperekplexia and epilepsy	ARHGEF9	Xq11	300607
Hypomyelination global cereb	SLC25A12	2q31	612949
Hypophosphatemic rickets	PHEX	Xp22	307800
Ichthyosis lig. X	STS	Xp22.31	308100
Ichthyosis lig. X	VCX3A	Xp22	308100
IGF-1 resistance	IGF1R	15q25	147370
Incontinentia pigmenti	IKBKG	Xq28	308300
Infertility and deafness	KIAACB77	15q15	611102
Joubert 3	AHI1	6q23	608894
Joubert 5	CEP290	12q21	610188
Kallmann 1	KAL1	Xp22.31	308700
Kallmann 2	FGFR1	8p12	147950
Kenny-Caffey	TBCE	1q42	24460
Langer-Giedion	EXT1	8q24.11	150230
Leri-Weill Dyschondrosteosis	PAR1	Xp22	127300
Lesch-Nyhan	HPRT1	Xq26.2	300322
Li-Fraumeni 1	TP53	17p13.1	151623
Lissencephaly with cerebellar	RELN	7q22.1	257320
Lissencephaly lig.X	DCX	Xq22.3-q23	300067
Loeys-Dietz tipo 1A	TGFBR1	9q22	609192
Loeys-Dietz tipo 2B	TGFBR2	3p24	610380
Long QT	KCNQ1	11p15	192500
Lowe	OCRL	Xq25	309000
Macrocefalia/autismo	PTEN	10q23	605309
Major effective disorder	IMPA1	8q21	125480
Marfan 1	FBN1	15q21.1	154700
Meningioma	NF2	22q12	607174
Doença de Menkes	ATP7A	Xq21.1	309400
Retardo mental	DMXL2	15q21	612186
Retardo mental Aut.Dom. 5	SYNGAP1	6p21.3	612621
Retardo mental Aut.Rec. 6	GRIK	6q16	611092
Metachromatic leukodystroph	ARSA	22q13	250100
Microphthalmia syndromic 2	BCOR	Xp11	300166
Microphthalmia syndromic 7	HCCS	Xp22	300056

Regiões alvo de doenças detectadas por CGH:

Microphthalmia	SOX2	3q26.3-q27	206900
Migraine familial hemiplegic 1	SCN1A	2q24.3	609634
Miller-Dieker	ABR	17p13.3	247200
Miller-Dieker	PAFAH1B1	17p13.3	247200
Miller-Dieker	PITPNA	17p13.3	247200
Miller-Dieker	YWHAE	17p13.3	247200
Mitochondrial complex deficie	NDUFA1	Xq24	252010
Mowat-Wilson	ZEB2	2q22	235730
Nail-patella	LMX1B	9q33.3	161200
Nephonophthisis	NPHP1	2q13	256100
Neurofibromatose tipo 1	JJAZ1	17q11.2	162200
Neurofibromatose tipo 1	NF1	17q11.2	162200
NFIA haploinsuficiency	NFIA	1p31.3	600727
Noonan 1	PTPN11	12q24	163950
Noonan4	SOS1	2p22.1	610733
Noonan5	RAF1	3p25	611553
Doença de Norrie	NDP	Xp11	310600
Obesidade severa	SM1	6q16	603128
Oculocutaneous albinism tipo	OCA2	15q13	203200
Oligodontia	PAX9	14q13	604625
Opitz G	MID2	Xq22	300204
Opitz G	MID1	Xq22	300000
Opitz-Kaveggia	MED12	Xq13.1	305450
Optic atrophy	TP73L	3q28	165500
Deficiência ornitina transcarba	OTC	Xp11.4	311250
Orofaciodigital 1	OFD1	Xp22.2	311200
Osteogenesis imperfecta	COL1A1	17q21	120150
Osteokilosis	LEMD3	12q14.3	166705
Doença Pelizaeus-Merzbacher	PLP1	Xq22.2	312080
Phosphoglycerate kinase defic	PGK1	Xp21	300653
Pitt-Hopkins	TCF4	18q21.2	610954
Doença do Rim Policístico	PKD1	16p13	601313
Doença do Rim Policístico 2	PKD2	4q22.1	613095
Polidactilia preaxial 2	LMER1	7q36	174500
Potocki-Shaffer	ALX4	11p11.2	601224
Potocki-Shaffer	EXT2	11p11.2	601224
Pseudohypoparathyroidism Ia	GNAS	20q13	108580
Pseudohypoparathyroidism Ia	GNASIC	20q13	108580
Pseudovaginal perineoscrotal l	SRD5A2	2p23	264600
Pulmonary vencocclusive dise	BMPR2	2q32	265450
Pyruvate dehydrogenase defici	PDHA1	Xp22	312170
Renal cysts and diabetes	TCF2	17q12	137920
Retinoblastoma	RB1	13q14.2	180200
Rett syndrome congenital vari	FOXG 1B	14q12	613454
Rett	MECP2	Xq28	312750
Rieger	PITX2	4q25	180500

Regiões alvo de doenças detectadas por CGH:

Rolandic epilepsy, mental reta	SRPX2	Xq22	300643
Rubinstein-Taybi	CREBBP	16p13	610543
Rubinstein-Taybi	DNASE1	16p13	610543
Russel silver	CPA4	7q32	607635
Russel silver	GRB10	7p12	180860
Schizophrenia 11	NRG3	10p22	608078
Sclerosteosis	SOST	17q21.31	269500
Scoliosis	CHD2	15q25	602119
Short stature	LHX4	1q25.2	262700
SHOX	SHOX	Xq22.33	312865
Simpson-Golabi-Behmel tipo1	GPC3	Xq26	312870
Smith-Lemli-Opitz	DHCR7	11q13.4	270400
Small size	PEG3	19q13	601483
Small size	PEG3IC	19q13	
Small size	ZIM2	19q13	601483
Smith-Magenis	RAI1	17p11.2	182290
Sotos	NSD1	5p35.3	117550
Split-hand/footmalformation1	DLX5	7q21.3	220600
Split-hand/footmalformation1	SHFM1	7q21.3	220600
Split-hand/footmalformation3	BTRC	10q24.32	600095
Split-hand/footmalformation3	FBXW4	10q24.32	600095
Split-hand/footmalformation3	LBX1	10q24.32	600095
Split-hand/footmalformation3	POLL	10q24.32	600095
Spli-hand/footmalformation	DLX1	2q31	606708
Spli-hand/footmalformation	DLX2	2q31	606708
Stickler 1	COL2A1	12q13.11	108300
Stickler 2	COL11A1	1p21	604840
Synpolydactyly	EVX2	2q31	186000
Synpolydactyly	HOXD13	2q31.1	186000
Synpolydactyly	HOXD9	2q31	186000
Thrombastheria of Glanzmann	ITGB3	17q21	273800
Thrombocytopenia absent radi	GJA8	1q21.1	274000
Timdhy	CACNA1C	12p13	601005
Treacher collins mandibulofac	TCOF1	5q33	154500
Townes-Brocks	SALL1	16q12.1	107480
Trichorhinophalangeal	TRPS1	8q23.3	150230
Tuberous sclerosis	TSC1	9q34.13	191100
Tuberous sclerosis 2	TSC2	16p13.3	613254
Ulnar-mammary	TBX3	12q24.21	181450
Usher	ABCC8	11p15	276904
Usher	USH1C	11p15	276904
van der Woude	IRF6	1q32.2	119300
Vascular endothelial growth fa	VEGFA	6p21	192240
Verbal dyspraxia	FOXP2	7q31.1	602081
Visceral heterdaxy	CFC1	2q21	605376
von Hippel -Lindau	VHL	3p25.3	193300

Regiões alvo de doenças detectadas por CGH:

WaardenburgI	PAX3	2p36.1	193500
Waardenburg IIA	MITF	3p14.1	193510
WAGR	WT1	11p13	194072
Williams-Beuren	ELN	7q11.23	194050
Williams-Beuren	GTF2I	7q11.23	194050
Williams-Beuren	GTF2IRD1	7q11.23	104050
Williams-Beuren	LIMK1	7q11.23	194050
Williams-Beuren	NCF1	7q11.23	194050
Wolf-Hirschhom	LETM1	4p16.3	194190
Wolf-Hirschhom	WHSC1	4p16.3	194190
Wolf-Hirschhom	WHSC2	4p16.3	194190
Inativação do X- Familial skev	XIST	Xq13	314670
Inativação do X - dyskeratosis	DKC1	Xq28	305000
Heterotaxia lig X	ZIC3	Xq26.3	306955
Juvenil retinoschisis lig.X	RS1	Xp22	312700
Lymphoproliferativo tipo 1 lig	SH2D1A	Xq25	308240
Lymphoproliferativo tipo 2 lig	BIRC4	Xq25	308240
Retardo mental ligado ao X	ARX	Xp21	300419
Retardo mental ligado ao X	IL1RAPL1	Xp21	300143
Retardo mental ligado ao X	PAK3	Xp22	300558
Retardo mental ligado ao X	SOX3	Xq27	300123
Espasmos ligado ao X	CDKL5	Xp22.13	300672
Retardo mental ligado ao X	ACSL4	Xp22	300387
Retardo mental ligado ao X	AGTR2	Xq23	300084
Retardo mental ligado ao X	AP1S2	Xq22	300630
Retardo mental ligado ao X	ARHGEF6	Xq26	300436
Retardo mental ligado ao X	ATP6AP2	Xp11	300423
Retardo mental ligado ao X	BRWD3	Xp21	300659
Retardo mental ligado ao X	CUL4B	Xp24	300354
Retardo mental ligado ao X	DLG3	Xq13	300189
Retardo mental ligado ao X	FTSJ1	Xp11	309549
Retardo mental ligado ao X	GDI1	Xq28	309541
Retardo mental ligado ao X	GRIA3	Xq24	300699
Retardo mental ligado ao X	HADH2	Xp11	300220
Retardo mental ligado ao X	JARID1C	Xp11	300534
Retardo mental ligado ao X	KIAA2022	Xq13	300524
Retardo mental ligado ao X	OPHN1	Xq12	300486
Retardo mental ligado ao X	PHF8	Xp11	300263
Retardo mental ligado ao X	SLC38A5	Xp11	309549
Retardo mental ligado ao X	SMS	Xp22	309583
Retardo mental ligado ao X	SYN1	Xp11	300491
Retardo mental ligado ao X	TM4SF2	Xp11	300210
Retardo mental ligado ao X	UBE2A	Xq23	312180
Retardo mental ligado ao X	UPF3B	Xq24	300676
Retardo mental ligado ao X	VCX3A	Xp22	300533
Retardo mental ligado ao X	ZDHHC15	Xq13	300577

Regiões alvo de doenças detectadas por CGH:

Retardo mental ligado ao X	ZDHHC9	Xq25	300799
Retardo mental ligado ao X	ZNF261	Xq13	300061
Retardo mental ligado ao X	ZNF41	Xp11	314995
Retardo mental ligado ao X	ZNF81	Xp11	300498
Retardo mental ligado ao X	GRIA3	Xq24	300699
Retardo mental lig.X- Allan H	SLC16A2	Xq13	300523
Retardo mental lig. X - Christi	SLC9A6	Xq26	300243
Retardo mental lig.X-Stocco d	SHROOM4	Xp11	300434
Retardo mental lig. X- Turner	HUWE1	Xp11	300706
Retinoblastoma	RB1	13q14.2	180200
Xq28 microdeleção	ABCD1	Xq28	300475
Xq28 microduplicação	RPL10	Xq28	300815
Deleção Xp11.3	RP2	Xp11	300578
Deleção Xp11.3	ZNF674	Xp11	300578
XY Sex reversal	NR5A1	9q33.3	184757
Unique subtelomeric regions	múltiplo	43 sítios	
Unique pericentromeric regior	múltiplo	43 sítios	
Aneuploidia	múltiplo	todos	

O a-CGH HD realiza o rastreamento de todo o genoma para investigação de anomalias cromossômicas. Esta lista sofre alterações constantemente, visto que muitas alterações genômicas são descobertas, com a inclusão de novos genes,síndromes e regiões cromossômicas de interesse.