qChip Post®

qgenomics
Genomics for Human Health

Dado que ***qChip Post®*** es un microarray de cobertura global, existe la posibilidad de identificar alteraciones cromosómicas no asociadas a patologías descritas en esta tabla. El array puede determinar la presencia de alteraciones de número de copia asociadas a fenotipos de aparición tardía no relacionados con las actuales manifestaciones clínicas del paciente y que pueden tener implicaciones para otros miembros de la familia. En caso que exista una historia familiar de enfermedad con posible origen genético no relacionada con la consulta, es conveniente comunicarlo al laboratorio.

	CONDITION	CANDIDATE	OMIM	BAND
●	* 1p36 deletion syndrome	Multiple	607872	1p36
●	* 1q21.1 Distal microdeletion (with susceptibility for mental retardation, autism or congenital anomalies)	Multiple: ACP6, GJA5 , GJA8	612474	1q21.1
●	1q21.1 Microdeletion with susceptibility for thrombocytopenia-absent radius (TAR)	Multiple	274000	1q21.1
●	1q41-q42 microdeletion/Fryns syndrome	Multiple	612530	1q41-q42
●	1q43-q44 deletion syndrome	Multiple: AKT3	612337	1q44
●	* 22q11.2 distal microdeletion syndrome	Multiple	611867	22q11.21
●	22q11.21 duplication syndrome	Multiple: TBX1	608363	22q11.21
●	* 22q13.3 microdeletion syndrome (Phelan-McDermid syndrome)	Multiple: SHANK3	606232	22q13.3
●	2p15-p16.1 Microdeletion syndrome	Multiple	612513	2p15-p16.1
○	* 2p16.3 Microdeletion/Pitt-Hopkins-like 2	NRXN1	614325	2p16.3
●	2p21 homozygous microdeletion syndrome	Multiple	606407	2p21
●	* 2q11.2 deletion	Multiple: LMAN2L, ARID5A		2q11.2
●	2q11.2-q13 deletion	Multiple: NCK2, FHL2		2q11.2-q13
●	2q23.1 Microdeletion	MBD5, EPC2	156200	2q23.1
●	2q32.2-q33 microdeletion syndrome	Multiple: SATB2	612313	2q32.2-q33
●	2q37 microdeletion	Multiple	600430	2q37
●	3q29 deletion syndrome	Multiple: FBX045, DLG1, PAK2	609425	3q29
●	3q29 microduplication syndrome	Multiple	611936	3q29
●	5p13 microduplication syndrome	NIPBL	613174	5p13.2
○	5q14.3 Microdeletion	MEF2C	613443	5q14.3
●	5q22 microdeletion / Familial adenomatous polyposis (FAP)	Multiple: APC	175100	5q22.2
●	5q35.2-q35.3 microduplication syndrome	NSD1		q35.2-q35.3
●	6pter-p24 microdeletion syndrome	Multiple	612582	6pter-p24
●	6q24-q25 microdeletion syndrome	Multiple	612863	6q24-q25
●	6q24.3 microdeletion	Multiple	612863	6q24.3
●	* 7q11.23 distal deletion syndrome	Multiple	613729	7q11.23
●	7q11.23 microduplication syndrome	Multiple	609757	7q11.23
●	8p23.1 microdeletion syndrome / Congenital diaphragmatic hernia 2 (CDH2)	Multiple: GATA4	22400	8p23.1
●	8p23.1 microduplication syndrome	Multiple: GATA4		8p23.1
●	8q12 microduplication syndrome	Multiple:CHD7		8q12
●	8q21.11 microdeletion syndrome	8q21.11	614230	Multiple: ZFHX4
●	9q22.32-q22.33 microdeletion syndrome	Multiple: TGFBR1, GABBR2		9q22.32-q22.33
○	* 9q34 microdeletion / Kleefstra syndrome	EHMT1	610253	9q34
●	* 10q11.21-q11.23 deletion syndrome	Multiple: CHAT, SLC18A3		10q11.21-q11.23
●	10q22-q23 deletion syndrome	Multiple	612242	10q22.3-q23.31
●	11p15-p14 homozygous deletion syndrome	USHC1, ABCC8	606528	11p15.1
●	* 12q14.1-q15 microdeletion syndrome	Multiple: GRIP1, LEMD3, HMGA2		12q14.3
●	12q14.3-related primordial dwarfism/ Russell-Silver-like	HMGA2		12q14.3
●	12q24.21-q24.23 Microduplication	Multiple		12q24.21-q24.23
●	14q11.2 microdeletion syndrome	Multiple	613457	14q11.2
●	14q12 microduplication syndrome	Multiple: FOXG1		14q12

	CONDITION	CANDIDATE	OMIM	BAND
●	14q22-q23 microdeletion syndrome	Multiple		14q22-q23
●	14q32.2 microdeletion causing upd(14)mat phenotype	Multiple: DLK1, RTL1		14q32.2
●	14q32.2 microdeletion causing upd(14)pat phenotype	Multiple:MEG3, MEG8, RTL1	608149	14q32.2
●	15q11-q13 microduplication	Multiple	608636	15q11-q13
●	15q13.3 microdeletion syndrome	Multiple: CHRNA7	612001	15q13.3
●	15q24 microdeletion syndrome	Multiple	613406	15q24.1-q24.3
●	15q24 microduplication syndrome	Multiple	613406	15q24.1-q24.3
●	15q25.2 microdeletion	Multiple	614294	15q25.2
●	* 15q26 overgrowth syndrome	Multiple		15q26
●	15q26-qter deletion syndrome	Multiple	612626	15q26
●	16p11.2 distal microdeletion	Multiple: SH2B1	613444	16p11.2
●	* 16p11.2 microdeletion syndrome (ASD susceptibility)	Multiple	611913	16p11.2
●	* 16p11.2-p12.2 microdeletion syndrome	Multiple	613604	16p11.2-p12.2
●	16p12.1 microdeletion syndrome	Multiple: CDR2, EEF2K, UQCRC2	136570	16p12.1
●	* 16p13.11 microdeletion	Multiple: MYH11		16p13.11
●	16p13.3 deletion syndrome/Severe Rubinstein-Taybi syndrome	Multiple: CREBBP	610543	16p13.3
●	16p13.3 microduplication	CREBBP	613458	16p13.3
●	16q11.2-q12.2 microdeletion syndrome	Multiple: SALL1, ZNF423		16q11.2-q12.2
●	16q21-q22 microdeletion syndrome	CBFB	614541	16q21-q22
●	17p13.1 microdeletion syndrome	Multiple: TP53	613776	17p13.1
●	17p13.3 distal microdeletion	YWHAE, CRK		17p13.3
●	17p13.3 distal microduplication including lissencephaly region (PAFAH1B1)	PAFAH1B1, YWHAE, CRK	613215	17p13.3
●	17p13.3 distal microduplication not including lissencephaly region (PAFAH1B1)	YWHAE, CRK	613215	17p13.3
●	17p13.3 microduplication syndrome	Multiple: PAFAH1B1, YWHAE, CRK	613215	17p13.3
●	17q12 microdeletion	Multiple: HNF1B, LHX1	614527	17q12
●	17q12 microduplication syndrome	Multiple: HNF1B, LHX1	614526	17q12
●	17q21.31 microdeletion syndrome	Multiple: MAPT	610443	17q21.3
●	17q21.31 microduplication syndrome	Multiple	613533	17q21.31
●	17q23.1-q23.2 microdeletion syndrome	Multiple: TBX2, TBX4	613355	17q23.1-q23.2
●	19p13.12 microdeletion syndrome	Multiple	613638	19p13.2
●	19q13.11 microdeletion syndrome	Multiple	613026	19q13.11
●	Adrenal hypoplasia congenital (AHC)	NR0B1 (DAX1)	300200	Xp21.2
○	Adult-onset autosomal dominant leukodystrophy (ADLD)	LMNB1	169500	5q23.2
○	Agammaglobulinemia, X-linked	BTK	300755	Xq22.1
●	Alagille syndrome, type 1	JAG1	118450	20p12.2
●	Alpha Thalassemia / Mental Retardation syndrome	Multiple	141750	16p13.3
●	Alport syndrome, X-linked	COL4A5	301050	Xq22.3
⊙	Alzheimer disease, early onset with cerebral amyloid angiopathy	APP	104300	21q21.3
⊙	Androgen insensitivity syndrome	AR	300068	Xq12
●	Angelman syndrome	Multiple: UBE3A	105830	15q11-q13
⊙	Aniridia, type II	PAX6	106210	11p13
●	ANKRD11 haploinsufficiency / 16q24.3 Microdeletion	ANKRD11		16q24.3

* Microduplicación asociada a fenotipo reconocible

⊙ Alteración de número de copia es un evento raro (<5%)

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	CONDITION	CANDIDATE	OMIM	BAND
⊙	Atrial septal defect (ASD) with atrioventricular conduction defects	NKX2-5	108900	5q35.2
⊙	Autism with intellectual disability, SHANK2-related	SHANK2	613436	11q13.3
●	Bannayan-Riley-Ruvalcaba syndrome	PTEN	153480	10q23.31
●	Barakat syndrome / Hypoparathyroidism, sensorineural deafness, and renal disease (HDR)	GATA3	146255	10p14
○	Bardet-Biedl syndrome 14 (BBS)	CEP290	209900	12q21.32
○	Bartter syndrome, type 1, antenatal	SLC12A1	146200	15q21.1
○	Bartter syndrome, type 2, antenatal	KCNJ1	241200	11q24.3
○	Bartter syndrome, type 3, classic	CLCNKB	607364	1p36.13
○	Bartter syndrome, type 4 (infantile with sensorineural deafness)	Multiple: CLCNKA, CLCNKB, BSND	602522	1p32.3
⊙	Basal cell nevus/Gorlin-Goltz syndrome	PTCH1	109400	9q22.32
○	Beckwith-Wiedemann syndrome (BWS)	KCNQ1OT1, IGF2	130650	11p15.5
⊙	Beckwith-Wiedemann syndrome (BWS), IGF2-related	IGF2	130650	11p15.5
⊙	Beta thalassemia	HBB	613985	11p15.4
○	Bilateral frontoparietal polymicrogyria (BFPP)	GPR56	606854	16q13
○	Blepharophimosis, ptosis, epicanthus inversus (BPE)	FOXL2	110100	3q22.3
●	Brachydactyly-mental retardation syndrome / Albright hereditary osteodistrophy-like	HDAC4	600430	2q37.3
●	Branchio-oto-renal syndrome (BOR)/ Melnick-Fraser syndrome	EYA1	113650	8q13.3
○	Buschke-Ollendorff syndrome	LEMD3	166700	12q14.3
⊙	Campomelic dysplasia (CMPD)	SOX9	114290	17q24.3
⊙	CardioFacioCutaneous syndrome (CFC)	Multiple	115150	7q34, 12p12.1, 19p13.3
○	Cat-Eye Syndrome	Multiple	115470	22q11.1
⊙	Cerebellar ataxia and MR, recessive (Hutterite form)	VLDLR	224050	9p24.2
○	Cerebral cavernous malformations, type 1 (CCM1)	KRIT1	116860	7q21.2
●	Cerebral cavernous malformations, type 2 (CCM2)	CCM2	603284	7p13
⊙	Cerebral cavernous malformations, type 3 (CCM3)	PDCD10	603285	3q26.1
●	Charcot-Marie-Tooth Disease, type 1A (CMT1A) / Microduplication 17p12	PMP22	118220	17p12
●	Charge syndrome	CHD7	214800	8q12.2
●	Chondrodysplasia punctata, X-linked recessive (CDPX1)	ARSE	302950	Xp22.33
⊙	Choroideremia, X-linked	CHM	303100	Xq21.2
●	Chronic granulomatous disease, X-linked	CYBB	306400	Xp11.4
⊙	Cleft Palate, Isolated (CPI)	SATB2	119540	2q33.1
⊙	Cleidocranial dysplasia (CCD)	RUNX2	119600	6p12.3
○	Cohen, autosomal recessive	VPS13B	216550	8q22.2
○	Congenital adrenal hyperplasia (CAH)	CYP21A2	201910	6p21.32
⊙	Congenital diaphragmatic hernia (CDH)	Multiple: CHD2, NR2F2	142340	15q26.1-q26.3
⊙	Congenital hemidysplasia with ichthyosiform erythroderma & limb defects (CHILD)	NSDHL	308050	Xq28
⊙	Cornelia de Lange syndrome	NIPBL	122470	5p13.2
○	Cornelia de Lange syndrome 2	SMC1A	300590	Xp11.22
⊙	Cornelia de Lange syndrome, X-linked	SMC1L1	300590	Xp11.22
○	Costello syndrome	HRAS	218040	11p15.5
⊙	Cowden syndrome / PTEN hamartoma tumor	PTEN	158350	10q23.31
○	Craniofrontonasal syndrome (CFNS)	EFNB1	304110	Xq13.1
○	Craniosynostosis, type 2 / Boston-type craniosynostosis	MSX2	604757	5q35.2
●	Cri-du-Chat syndrome	Multiple: TERT, CTNND2	123450	5p15.2
⊙	Currarino syndrome	MNX1	176450	7q36.3
⊙	Dandy-Walker syndrome (DWS)	Multiple (ZIC1,ZIC4)	220200	3q24
○	Deafness 22, autosomal recessive	OTOA	607039	16p12.2
⊙	Diamond-Blackfan anemia 1	RPS19	105650	19q13.2
●	DiGeorge syndrome / VeloCardioFacial syndrome / 22q11.2 deletion syndrome	Multiple: HIRA, TBX1	188400	22q11.21
⊙	DiGeorge syndrome, type 2	Multiple	601362	10p14
●	Dopa-responsive dystonia (DRD)/Segawa	GCH1	128230	14q22.2-q22.3

	CONDITION	CANDIDATE	OMIM	BAND
○	Dosage-sensitive sex reversal X-linked	NR0B1 (DAX1)	300018	Xp21.2
○	* Early-onset Alzheimer disease with cerebral amyloid angiopathy	APP	605714	21q21.3
⊙	Early-onset ataxia with oculomotor apraxia & hypoalbuminemia, autosomal recessive	APTX	208920	9p13.3
⊙	Emery-Dreifuss muscular dystrophy, X-linked (EDMD)	EMD	181350	Xq28
○	EPHA7 haploinsufficiency / 6q16.1 Microdeletion	EPHA7		6q16.1
⊙	Epilepsy and intellectual disability restricted to females, X-linked/Juberg-Hellman, X-linked/Dravet-like, X-linked	PCDH19	300088	Xq22.1
⊙	Epilepsy, benign neonatal	KCNQ2	121200	20q13.33
⊙	Faciogenital dysplasia, X-linked/Aarskog-Scott, X-linked	FGD1	305400	Xp11.22
●	Familial adenomatous polyposis due to 5q22 microdeletion / Gardner syndrome	APC	175100	5q22
○	Familial hypocalciuric hypercalcemia, type 1 (HHC1)	CASR	145980	3q21.1
⊙	Feingold syndrome	MYCN	164280	2p24.3
⊙	* FG syndrome, type 5 (FGS5)	MID2	300581	Xq22.3
⊙	FMR1 Microdeletion syndrome	FMR1	300624	Xq27.3
●	Focal dermal hypoplasia, X-linked/ Gorlin-Goltz syndrome	PORCN	305600	Xp11.23
○	Generalized epilepsy with febrile seizures plus 2	SCN1A	604403	2q24.3
○	Gitelman syndrome	SLC12A3	263800	16q13
⊙	GLUT1 deficiency	SLC2A1	606777	1p34.2
⊙	Glycerol kinase deficiency (GKD)	GK	300679	Xp21.2
○	Gonadal dysgenesis / Sex reversal, autosomal dominant (SRA2)	Multiple (DMRT1)	154230	9p24.3
●	Greig cephalopolysyndactyly	GLI3	175700	7p14.1
⊙	Growth hormone insensitivity, autosomal recessive / Laron, autosomal recessive	GHR	262500	5p12
⊙	Hemophilia A, X-linked	F8	306700	Xq28
⊙	Hemophilia B, X-linked	F9	306900	Xq27.1
⊙	Hereditary hemorragic telangiectasia, type 2 (HHT2)	ACVRL1	600376	12q13.13
●	Hereditary Neuropathy with liability to Pressure Palsies (HNPP)/ Microdeletion 17p12	PMP22	162500	17p12
⊙	Hereditary paraganglioma-pheochromocytoma, SDHB-related	SDHB	115310	1p36.13
⊙	Hereditary paraganglioma-pheochromocytoma, SDHD-related	SDHD	168000	11q23.1
⊙	Heterotaxy, X-linked	ZIC3	306955	Xq26.3
○	Hirschsprung, RET-related	RET	142623	10q11.21
⊙	Holoprosencephaly, type 1 (HPE1)	Multiple	236100	21q22.3
⊙	Holoprosencephaly, type 2 (HPE2)	SIX3	157170	2p21
⊙	Holoprosencephaly, type 3 (HPE3)	SHH	142945	7q36.3
⊙	Holoprosencephaly, type 4 (HPE4)	TGIF	142946	18p11.31
⊙	Holoprosencephaly, type 5 (HPE5)	ZIC2	609637	13q32.3
○	Holoprosencephaly, type 6	Multiple	605934	2q37.1-q37.3
○	Holoprosencephaly, type 7 (HPE7)	PTCH1	610828	9q22.32
○	Holoprosencephaly, type 8 (HPE8)	Multiple	609408	14q13.1-q13.2
⊙	Holt-Oram syndrome (HOS)	TBX5	142900	12q24.21
○	Hydrocephalus with nephrogenic diabetes insipidus, X-linked	L1CAM, AVPR2	304800	Xq28
●	Hyper-IgE recurrent infection, autosomal recessive	DOCK8	243700	9p24.3
○	Hypertrichosis terminalis, generalized, with or without gingival hyperplasia	Multiple	135400	17q24
⊙	Hypohidrotic ectodermal dysplasia, X-linked (XHED)	EDA	305100	Xq13.1
○	Hypospadias 2 isolated, X-linked	MAMLD1	300758	Xq28
●	Hypotonia-cystinuria	SLC3A1, PREPL	606407	2p21
●	Idiopathic short stature, X/Y-linked (ISSX)	SHOX	300582	Xp22.33/Yp11.32 (PAR1)
○	Infantile spasm syndrome-1 (ISSX1), X-linked / Early infantile epileptic encephalopathy-1	ARX	308350	Xp21.3
○	Infantile spasms, MAGI2-related	MAGI2		7q21.11
⊙	Infantile spasms,CDKL5-related, X-linked / Atypical Rett	CDKL5	300672	Xp22.13
⊙	Intellectual disability and microcephaly with pontine and cerebellar hypoplasia, X-linked	CASK	300749	Xp11.4

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○	Intellectual disability with cerebellar hypoplasia & distinctive facial appearance, X-linked	OPHN1	300486	Xq12
○	Intellectual disability with language impairment and autistic features	FOXP1	613670	3p14.1
⊙	Isolated Ectopia Lentis	FBN1	129600	15q21.1
○	Jacobsen syndrome / 11q terminal deletion	Multiple	147791	11q23-qter
○	Joubert syndrome, type 3	AHI1	608629	6q23.3
○	Joubert syndrome, type 4 (autosomal recesive) / Joubert syndrome, type 4 / 2q13 deletion syndrome	NPHP1	609583	2q13
○	Joubert syndrome, type 5 (JBTS5)	CEP290	610188	12q21.32
●	Juvenile polyposis syndrome (JPS), BMPR1A-related	BMPR1A	174900	10q23.2
●	Juvenile polyposis syndrome (JPS), SMAD4-related	SMAD4	174900	18q21.2
⊙	Juvenile retinoschisis, X-linked	RS1	312700	Xp22.13
●	Kallmann syndrome, type 1	KAL1	308700	Xp22.31
⊙	Kniest dysplasia / Stickler syndrome, type I	COL2A1	156550	12q13.11
○	Langer mesomelic dysplasia (LMD)	SHOX	249700	Xp22.3/Yp11.32
●	Langer-Giedion syndrome / trichorhinophalangeal syndrome, type II	EXT1, TRPS1	150230	8q24.11q23.3
⊙	Leber congenital amaurosis, type X (LCA10)	CEP290	611755	12q21.32
○	Leiomyomatosis, diffuse, with Alport syndrome (DL-ATS)	COL4A5, COL4A6	308940	Xq22.3
●	Leri-Weill dyschondrosteosis (LWD)	SHOX	127300	Xp22.3/Yp11.32
○	Lesch-Nyhan, X-linked (LNS)	HPRT1	300322	Xq26.2
⊙	Li-Fraumeni syndrome, type 1 (LFS)	TP53	151623	17p13.1
○	Lissencephaly with ambiguous genitalia, X-linked 2 (LISX2)	ARX	300215	Xp21.3
○	Lissencephaly with cerebellar hypoplasia	RELN	257320	7q22.1
●	Lissencephaly, type 1	PAFAH1B1 (LIS1)	607432	17p13.3
○	Lissencephaly, X-linked	DCX	300067	Xq22.3
⊙	Loeys-Dietz syndrome type 1A (LDS), TGFBR1-related	TGFBR1	609192	9q22.33
⊙	Loeys-Dietz syndrome type 1B (LDS), TGFBR1-related	TGFBR2	610168	3p24.1
⊙	Lowe syndrome	OCRL	309000	Xq25
●	Lubs syndrome / MECP2 duplication syndrome	MECP2	300260	Xq28
●	Lymphoproliferative syndrome, X-linked (XLP)	SH2D1A	308240	Xq25
⊙	Macrocephaly-autism syndrome	PTEN	605309	10q23.31
⊙	Marfan syndrome, type 1 (MFS1)	FBN1	154700	15q21.1
⊙	Marfan syndrome, type 2 (MFS2)	TGFBR2	610380	3p24.1
⊙	MASS phenotype	FBN1	604308	15q21.1
●	Mc Leod neuroacanthocytosis syndrome	XK	314850	Xp21.1
○	Meckel syndrome type 4	CEP290	611134	12q21.32
⊙	Menkes syndrome	ATP7A	309400	Xq21.1
○	Mental retardation 1, autosomal dominant (MRD1)	MBD5	156200	2q23.1
○	Mental retardation 21, X-linked	IL1RAPL1	300143	Xp21.3
⊙	Mental retardation 30, X-linked	PAK3	300558	Xq22.3
○	Mental retardation 5, autosomal dominant	SYNGAP1	612621	6p21.32
⊙	Mental retardation 54. X-linked	ARX	300419	Xp21.3
○	Mental retardation 6, autosomal recessive	GRIK2	611092	6q16.3
○	Mental retardation 7, autosomal recessive	TUSC3	611093	8p22
○	Mental retardation 9, X-linked/Mental retardation 44, X-linked	FTSJ1	309549	Xp11.23
○	Mental retardation 94, X-linked	GRIA3	300699	Xq25
○	Mental retardation with isolated growth hormone deficiency, X-linked	SOX3	300123	Xq27.1
○	Mental retardation, X-linked 17/31, microduplication	Multiple: HSD17B10, HUWE1	300705	Xp11.22
○	Mesomelic dysplasia Kantaputra type	HOXD gene cluster	156232	2q31.1
⊙	Metachromatic leukodystrophy, autosomal recessive (MLD)/Arylsulfatase A deficiency	ARSA	250100	22q13.3

	CONDITION	CANDIDATE	OMIM	BAND
○	Microphthalmia 7 with linear skin defects (MCOPS7)	HCCS	309081	Xp22.2
⊙	Microphthalmia syndromic, type 3 (MCOPS3)	SOX2	206900	3q26.33
●	Miller-Dieker syndrome	PAFAH1B1 (LIS1)	247200	17p13.3
⊙	Mohr-Tranebjaerg syndrome	TIMM8A	304700	Xq22.1
●	Mowat-Wilson syndrome	ZEB2	235730	2q22.3
●	Myoclonus dystonia	SGCE	159900	7q21.3
⊙	Myotubular myopathy 1, X-linked	MTM1	310400	Xq28
○	Nablus mask-like facial syndrome	Multiple	608156	8q22.1
⊙	Nail-patella syndrome (NPS)	LMX1B	161200	9q33.3
○	Neonatal severe primary hypoparathyroidism (NSHPT)	CASR	239200	3q21.1
●	Nephronophthisis, type 1	NPHP1	256100	2q13
●	Nephropatic cystinosis, autosomal recessive	CTNS	219800	17p13.3
●	Neurofibromatosis, type 1 (NF1) / Von Recklinghausen Disease	Multiple - NF1	162200	17q11.2
●	Neurofibromatosis, type 2 (NF2)	NF2	101000	22q11.2
○	Neurosensory deafness, autosomal recessive	GJB6	220290	13q12.11
○	NFIA Haploinsufficiency / 1p32p31 microdeletion syndrome	NFIA	613735	1p31.3
○	Noonan syndrome, type 1 (NS1)	PTPN11	163950	12q24.13
○	Noonan syndrome, type 4 (NS4)	SOS1	610733	2p22.1
●	Norrie disease, X-linked	NDP	310600	Xp11.3
⊙	Oculocutaneous albinism, type II (OCA2)	OCA2	203200	15q13.1
○	Oculofaciocardiodental, X-linked/ Microphthalmia 2	BCOR	300166	Xp11.4
●	Okihiro syndrome / Duane-Radial Ray syndrome (DRRS)	SALL4	607323	20q13.2
⊙	Opitz GBBB syndrome, X-linked	MID1	300000	Xp22.2
⊙	Oral-facial-digital syndrome type 1	OFD1	311200	Xp22.2
●	Ornithine transcarbamylase deficiency	OTC	311250	Xp11.4
●	Osteopathia striata with cranial sclerosis, X-linked	FAM123B	300373	Xq11.1
○	Oto-dental syndrome	FGF3	166750	11q13.3
○	Oto-facio-cervical syndrome (OFC)	EYA1	166780	8q13.3
○	Pallister-Killian syndrome / Tetrasomy 12p	Multiple	601803	12p
○	Parietal foramina, type 1	MSX2	168500	5q35.2
●	Pelizaeus-Merzbacher (PMD), X-linked	PLP1	312080	Xq22.2
●	Peutz-Jeghers syndrome	STK11	175200	19p13.3
●	Pitt-Hopkins syndrome	TCF4	610954	18q21.2
○	Pitt-Hopkins-like 1, autosomal recessive / Cortical dysplasia-focal epilepsy, autosomal recessive (CDFE syndrome)	CNTNAP2	610042	7q35-q36.1
⊙	Polycystic kidney disease 1 (PKD1)	PKD1	173900	16p13.3
●	Potocki-Lupski syndrome (PTLS)	Multiple: RAI1	610883	17p11.2
●	Potocki-Shaffer syndrome	EXT2, ALX4	601224	11p11.2
●	Prader-Willi syndrome	Multiple	176270	15q11-q13
⊙	Prader-Willi-like phenotype	Multiple: SIM1	176270	6q16.1-q16.3
○	Proteus/Proteus-like syndrome	PTEN	176920	10q23.31
○	PTEN Hamartoma Tumor syndrome	PTEN		10q23.31
●	Renal cysts and diabetes (RCAD)	HNF1B	137920	17q12
●	Retinoblastoma with intellectual disability	RB1	180200	13q14.2
○	Rett syndrome	MECP2	312750	Xq28
●	Rett, congenital variant	FOXG1	613454	14q12
○	Rieger 1 (RIEG1)/Axenfeld-Rieger	PITX2	180500	4q25
●	Rubinstein-Taybi syndrome (RTS)	CREBBP	180849	16p13.3
○	Rubinstein-Taybi syndrome 2	EP300	613684	22q13.2
●	Saethre-Chotzen syndrome	TWIST1	101400	7p21.1
○	Schizencephaly	EMX2	269160	10q26.11

* Microduplicación asociada a fenotipo reconocible

⊙ Alteración de número de copia es un evento raro (<5%)

● Alteración de número de copia es una causa común (>5%) y reconocida del fenotipo

○ Alteración de número de copia es un evento muy raro o sólo predicho

	CONDITION	CANDIDATE	OMIM	BAND
○	Senior-Loken syndrome, type 6 (SLSN6)	CEP290	610189	12q21.32
⊙	Severe myoclonic epilepsy of infancy (SMEI) / Dravet syndrome	SCN1A	607208	2q24.3
○	Short stature, pituitary and cerebellar defects, small sella turcica/ Pituitary hormone deficiency, combined, 4	LHX4	262700	1q25.2
○	Shprintzen-Goldberg craniosynostosis	FBN1	182212	15q21.1
○	Siderius type intellectual disability, X-linked	PHF8	300263	Xp11.22
○	Simpson-Golabi-Behmel syndrome, type 1	GPC3	312870	Xq26.2
⊙	Smith-Lemli-Opitz syndrome (SLOS)	DHCR7	270400	11q13.4
●	Smith-Magenis syndrome	Multiple: RAI1	182290	17p11.2
●	Sotos syndrome	NSD1	117550	5q35.3
○	Speech-language disorder, type 1	FOXP2	602081	7q31.1
○	Split-hand/foot malformation, type 1 (SHFM1) / Ectrodactyly	Multiple: SHFM1	183600	7q21.3
○	Split-hand/foot malformation, type 3 (SHFM3) / Ectrodactyly	Multiple: FBXW4	246560	10q24.32
⊙	Split-hand/foot malformation, type 4 (SHFM4)	TP63	605289	3q28
○	Split-hand/foot malformation, type 5 (SHFM5)	EVX2, HOXD gene cluster	606708	2q31.1
○	SRY dosage abnormalities / XX male	SRY	278850	Yp11.31
⊙	SRY dosage abnormalities / XY gonadal dysgenesis	SRY	400044	Yp11.31
●	Steroid sulfatase deficiency / X-linked ichtyosis	STS	308100	Xp22.31
○	Stickler syndrome, type I	COL2A1	108300	12q13.11
⊙	Synpolydactyly / Syndactyly II	HOXD cluster	186000	2q31.1
●	Tetrasomy 15q26 (Levy-Shanske Syndrome)	Multiple	614846	15q26
●	Toe syndactyly, telecantus, anogenital and renal malformations, X-linked (STAR)	FAM58A	300707	Xq28
⊙	Townes-Brocks Syndrome (TBS)	SALL1	107480	16q12.1
●	Trichorhinophalangeal syndrome, type I	TRPS1	190350	8q23.3
⊙	Tuberous sclerosis, type 1 (TSC1)	TSC1	191100	9q34.13
●	Tuberous sclerosis, type 2 (TSC2)	TSC2	613254	16p13.3
⊙	Ulnar-Mammary syndrome (UMS)	TBX3	181450	12q24.21
⊙	Usher IIC, autosomal recessive	GPR98	605472	5q14.3
⊙	Van der Woude syndrome	IRF6	119300	1q32.2
○	Vascular endothelial growth factor (VEGFA)-related disorders	VEGFA	192240	6p21.1
⊙	von Hippel-Lindau syndrome	VHL	193300	3p25.3
⊙	Waardenburg syndrome, type I	PAX3	193500	2q36.1
○	Waardenburg syndrome, type IIA	MITF	193510	3p14.1
●	WAGR syndrome (Wilms tumor+Aniridia+Genitourinary Anomalies+MR)	PAX6, WT1	194072	11p13
⊙	Walker-Warburg syndrome (WWS)	LARGE	236670	22q12.3
○	Weill-Marchesani syndrome 2	FBN1	608328	15q21.1
●	* Williams-Beuren syndrome	Multiple: ELN	194050	7q11.23
⊙	Wilms tumor, type 1	WT1	194070	11p13
●	Wolf-Hirschhorn syndrome (WHS)	Multiple	194190	4p16.3
●	Xp11.23-p11.22 duplication syndrome	Multiple	300801	Xp11.23-p11.22
●	Xp11.3 microdeletion	Multiple: ZNF674, RP2	300578	Xp11.3
●	Xp11.4-p21.2 deletion	Multiple: IL1RAPL1, OTC		Xp11.4-p21.2
●	Xq28 microduplication syndrome	Multiple:GD1, IKBKG	300815	Xq28
○	XY sex-reversal 3; (SRXY3), +/- adrenal failure	NR5A1	612965	9q33.3

●	All Subtelomeric regions		
●	All euchromatic pericentromeric regions		
●	Complete aneuploidy for any chromosome	Multiple	24 chromosomes

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Genomics for Human Health

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