qChip Pre®

qgenomics
Genomics for Human Health

Although ***qChip Pre®*** [redacted]

CONDITION	CANDIDATE	OMIM	BAND
* 1p36 deletion syndrome	Multiple	607872	1p36
1q43q44 deletion syndrome	Multiple		1q43-q44
2p15-p16.1 microdeletion syndrome	Multiple		2p15-p16.1
* 3q29 microdeletion	Multiple	609425	3q29
4ptel deletion	Multiple		4p16.3-p16.1
5q22 microdeletion/Famililal Adenomatous Polyposis/Gardner syndrome	Multiple	175100	5q22.2
6p25.3 microdeletion syndrome	Multiple		6p25.3
* 7q11.23 microduplication syndrome (WBS reciprocal)	Multiple	609757	7q11.23
10q22-q23 deletion syndrome	Multiple	612242	10q22.3
* 12q14.1-q15 microdeletion syndrome	Multiple, GRIP1		12q14.3
* 12q24.21-q24.23 microduplication syndrome	Multiple		12q23.2-q24.2
* 15q11-q13 microduplication	Multiple	608636	15q11-q13
15q13.3 microdeletion syndrome	Multiple, KLF13	612201	15q13.3
15q24 microdeletion syndrome	Multiple		15q24.1-q24.3
* 16p11.2-p12.2 microdeletion syndrome	Multiple	611913	16p11.2-p12.2
16p11.2 autism susceptiblity locus	Multiple		16p11.2
16p13.3 deletion syndrome/Severe Rubinstein Taybi	DNASE1, CREBBP	180849, 610543	16p13.3
16q11.2q12.2 microdeletion syndrome	Multiple, SALL1, ZNF423	121360	16q11.2-q12.2
* 17q21.3 microdeletion syndrome	Multiple, MAPT	610443	17q21.3
*22q11.21 duplication syndrome	Multiple, TBX1	608363	22q11.21
*22q13.3 microdeletion syndrome (Phelan-McDermid Syndrome)	Multiple, ARSA, SHANK3		22q13.3
22q11.2 microdeletion syndrome, distal	Multiple	611867	22q11.21
Alagille syndrome, type 1	JAG1	118450	20p12.2
Angelman syndrome	Multiple, UBE3A	105830	15q11-13
Barakat syndrome / Hypoparathyroidism, sensorineural deafness, and renal disease (HDR)	GATA3	146255	10p14
Beckwith-Wiedemann syndrome (BWS)	KCNQ1OT1	130650	11p15.5
Brachydactyly-mental retardation syndrome / Albright hereditary osteodistrophy-like	Multiple	600430	2q37.3
Campomelic dysplasia (CMPD)	SOX9	114290	17q24.3
CardioFacioCutaneous syndrome (CFC)	Multiple	115150	7q34, 12p12.1, 19p13.3
* Cat-Eye syndrome	Multiple	115470	22q11.1
Charcot-Marie-Tooth Disease, type 1A (CMT1A)	PMP22	118200	17p12
Charge syndrome	CHD7	214800	8q12.2
Cleidocranial dysplasia (CCD)	RUNX2	119600	6p12.3
Cornelia de Lange syndrome	NIPBL	122470	5p13.2
Cornelia de Lange syndrome, X-linked	SMC1L1	300590	Xp11.22
Cri-du-Chat syndrome	Multiple	123450	5p15.2
Dandy-Walker syndrome (DWS)	ZIC4, ZIC1	220200	3q24
* DiGeorge syndrome / VeloCardioFacial syndrome	Multiple, HIRA, TBX1	188400	22q11.21
DiGeorge syndrome, type 2	Multiple	601362	10p14
Dosage sensitive sex reversal/Adrenal hypoplasia congenital	NR0B1		Xp21.2
Feingold syndrome	MYCN	164280	2p24.3
Glycerol kinase deficiency (GKD)	GK	300474	Xp21.2
Greig cephalopolysyndactyly	GLI3	175700	7p14.1
* Hereditary Neuropathy with liability to Pressure Palsies (HNPP)	PMP22	162500	17p12
Holoprosencephaly, type 1 (HPE1)	TMEM1	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 7 (HPE7) / Gorlin-Goltz syndrome	PTCH1	610828	9q22.32
Jacobsen syndrome / 11q terminal deletion	Multiple	147791	11q23-qter
Kallmann syndrome, type 1	KAL1		Xp22.31
Langer mesomelic dysplasia (LMD)	SHOX	249700	Xpter-p22.3 & Ypter-p11.32
Langer-Giedion syndrome / trichorhinophalangeal syndrome, type II	EXT1, TRPS1	150230	8q24.11
Leri-Weill dyschondrosteosis (LWD)	SHOX	127300	Xpter-p22.3 & Ypter-p11.32
Mieller-Diecker, lyssencephaly syndrome	PAFAH1B1 (LIS1)	247200; 607432	17p13.3
Mowat-Wilson syndrome	ZFHX1B	235730	2q22.3
Neurofibromatosis, type 1 (NF1) / Von Recklinghausen Disease	Multiple	162200	17q11.2
Noonan syndrome, type 1 (NS1)	PTPN11	163950	12q24.13
Norrie disease	NDP		Xp11.3
Pallister-Killian syndrome	Multiple	601803	12p
Pelizaeus-Merzbacher (PMD)	PLP1	312080	Xq22.2
Polycystic kidney disease 1 (PKD1)	PKD1	601313	16p13.3
Potocki-Lupski syndrome (PTLS)	Multiple	610883	17p11.2
Potocki-Shaffer syndrome	EXT2, ALX4	601224	11p11.2
Prader-Willi syndrome, type II	SNRPN (Multiple)	176270	15q11-q13
Retinoblastoma, type I / Mental Retardation	RB1	180200	13q14.2
Rieger syndrome, type I	PITX2	180500	4q25
Rubinstein-Taybi syndrome (RTS)	CREBBP	180849	16p13.3
Saethre-Chotzen syndrome	TWIST1	101400	7p21.1
Smith-Magenis syndrome	Multiple	182290	17p11.2
Sotos syndrome	NSD1	117550	5q35.3
Split-hand/foot malformation, type 3 (SHFM3)	Multiple	600095	10q24.32
Steroid sulfatase deficiency	STS	308100	Xp22.31
Syndactyly, type II	HOXD cluster	186000	2q31.1
Tuberous sclerosis, type 1 (TSC1)	TSC1	191100	9q34.13
Tuberous sclerosis, type 2 (TSC2)	TSC2	191100	16p13.3
Waardenburg syndrome, type IIA	MITF	193510	3p14.1
WAGR syndrome (Wilms tumor+Aniridia+Genitourinary Anomalies+MR)	PAX6, WT1	194072	11p13
Williams-Beuren syndrome	Multiple, ELN	194050	7q11.23
Wolf-Hirschhorn syndrome (WHS)	Multiple	194190	4p16.3
X-linked Alport syndrome	COL4A5		Xq22.3
X-linked heterotaxy	ZIC3	306955	Xq26.3
X-linked ichtyosis / Steroid sulfatase deficiency (STS)	STS	308100	Xp22.31
X-linked idiopathic short stature (ISSX)	SHOX	300582	PAR1
X-linked lissencephaly	DCX	300067	Xq22.3
X-linked mental retardation with isolated growth hormone deficiency	SOX3	300123	Xq27.1
All Subtelomeric regions			
All euchromatic pericentromeric regions			
Aneuploidy for any chromosome			

Contact us. Telf.: (+34) 932 301 270
info@qgenomics.com · http://www.qgenomics.com

* Microduplication associated to a clinically recognizable phenotype