PATOLOGIA	OMIM	LOCALIZACION
1p36 DELETION SYNDROME	607872	1p36.33
1q21.1 DELETION SYNDROME	612474	1q21.1
15q13.3 MICRODELETION SYNDROME	612001	15q13.2-q13.3
22q11.2 DELETION SYNDROME, DISTAL	611867	22q11.21-q11.23
22q13.3 DELETION SYNDROME	606232	22q13.33
17q21.31 MICRODELETION SYNDROME	610443	17q21.31
22q13.3 DELETION SYNDROME	606232	22q13.3
3q29 MICRODELETION SYNDROME	609425	3q29
ADENOMATOUS POLYPOSIS OF THE COLON	175100	5q22.1, 5q22.2
ADRENAL HYPOPLASIA, CONGENITAL	300200	Xp21.3-p21.2
ALAGILLE SYNDROME 1	118450	20p12.2
ANGELMAN SYNDROME	105830	15q11-q13, Xq28
ANIRIDIA, TYPE II	106210	11p13
AUTISM	209850	7q22, 6p21.3-p21.2, 2q32
AUTISM, X-LINKED, SUSEPTIBILITY TO, 1	300425	Xq13
AUTISM, X-LINKED, SUSCEPTIBILITY TO, 2	300495	Xp22.33
AUTISM, X-LINKED, SUSEPTIBILITY TO, 3,	300496	Xq28
BASAL CELL NEVUS SYNDROME	109400	9q22.3
BECKWITH-WIEDEMANN SYNDROME	130650	11p15.5, 5q35
BRACHYDACTYLY-MENTAL RETARDATION SYNDROME	600430	2q37
BRANCHIOOTORENAL SYNDROME 1	113650	8q13.3
BRUTON AGAMMAGLOBULINEMIA TYROSINE KINASE	300300	Xq21.3-q22
CAMPOMELIC DYSPLASIA	114290	17q24.3-q25.1
CAT EYE SYNDROME	115470	22q11
CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A	118220	17p11.2
CHARCOT-MARIE-TOOTH DISEASE, X-LINKED, 1	302800	Xq13.1
CHARGE SYNDROME	214800	8q12.1, 7q21.1
CLEIDOCRANIAL DYSPLASIA	119600	6p21
CORNELIA DE LANGE SYNDROME 1	122470	5p13.1
CRI-DU-CHAT SYNDROME	123450	5p15.2
DANDY-WALKER SYNDROME	220200	3q24
DIAPHRAGMATIC HERNIA, CONGENITAL	142340	15q26.1
DIGEORGE SYNDROME	188400	22q11.2
DIGEORGE SYNDROME/VELOCARDIOFACIAL SYNDROME SPECTRUM OF MALFORMATION 2	601362	10p14-p13
DOSAGE-SENSITIVE SEX REVERSAL	300018	Xp21.3-p21.2
DOWN SYNDROME	190685	Xp11.23, 21q22.3, 1q43
FEINGOLD SYNDROME	164280	2p24.1
FRAGILE X MENTAL RETARDATION SYNDROME	300624	Xq27.3
GREIG CEPHALOPOLYSYNDACTYLY SYNDROME	175700	7p13
HETEROTAXY, VISCERAL, X-LINKED	306955	Xq26.2
HOLOPROSENCEPHALY 1	236100	2q37.1-q37.3, 21q22.3
HOLOPROSENCEPHALY 2	157170	2p21
HOLOPROSENCEPHALY 3	142945	7q36
HOLOPROSENCEPHALY 4	142946	18p11.3
HOLOPROSENCEPHALY 5	609637	13q32
HYPERGLYCEROLEMIA	307030	Xp21.3-p21.2
HYPOPARATHYROIDISM, SENSORINEURAL DEAFNESS, AND RENAL DISEASE	146255	10p15
ICHTHYOSIS, X-LINKED	308100	Xp22.32
JACOBSEN SYNDROME	147791	11q23
JOHANSON-BLIZZARD SYNDROME	243800	15q15-q21.1

JOUBERT SYNDROME 4	609583	2q13
KABUKI SYNDROME	147920	8p22
KALLMANN SYNDROME 1	308700	Xp22.3
LERI-WEILL DYSCHONDROSTEOSIS	127300	Ypter-p11.2, Xpter-p22.32
LISSENCEPHALY, X-LINKED	300067	Xq22.3-q23
MENTAL RETARDATION, X-LINKED, WITH PANHYPOPITUITARISM	300123	Xq26.3
METACHROMATIC LEUKODYSTROPHY	250100	22q13.31-qter
MICROPHTHALMIA, SYNDROMIC 7	309801	Xp22
MILLER-DIEKER LISSENCEPHALY SYNDROME	247200	17p13.3
MITOCHONDRIAL COMPLEX I DEFICIENCY	252010	1q23, 11q13, 6q16.1, 5q12.1, 5q11.1, 5pter-p15.22, 2q33-q34, Xq24
MONOSOMY 1p36 SYNDROME	607872	1p36
MUSCULAR DYSTROPHY, BECKER TYPE	300376	Xp21.1
MUSCULAR DYSTROPHY, DUCHENNE TYPE	310200	12q21, Xp21.2
NAIL-PATELLA SYNDROME	161200	9q34.1
NEPHRONOPHTHISIS 1	256100	2q13
NEUROFIBROMATOSIS, TYPE I	162200	17q11.2
NEUROFIBROMATOSIS, TYPE II	101000	22q12.2
NEUROPATHY, HEREDITARY, WITH LIABILITY TO PRESSURE PALSIES	162500	17p11.2
NOONAN SYNDROME 1	163950	12q24.1
PELIZAEUS-MERZBACHER DISEASE	312080	Xq22
POLYCYSTIC KIDNEY DISEASE, INFANTILE SEVERE, WITH TUBEROUS SCLEROSIS	600273	16p13.3
POTOCKI-LUPSKI SYNDROME	610883	17p11.2
POTOCKI-SHAFFER SYNDROME	601224	11p11.2
PRADER-WILLI SYNDROME	176270	15q12, 15q11-q13
PRADER-WILLI-LIKE SYNDROME ASSOCIATED WITH CHROMOSOME 6	176270	6q16.3
RETINOBLASTOMA	180200	13q14.1-q14.2
RETT SYNDROME	312750	Xq28, Xp22
RIEGER SYNDROME, TYPE 1	180500	4q25-q26
RUBINSTEIN-TAYBI SYNDROME	180849	16p13.3, 22q13
SAETHRE-CHOTZEN SYNDROME	101400	10q26, 7p21.1
SEX-DETERMINING REGION Y	480000	Yp11.3
SMITH-MAGENIS SYNDROME	182290	17p11.2
SOTOS SYNDROME	117550	5q35
SPERMATOGENIC FAILURE, NONOBSTRUCTIVE, Y-LINKED	415000	Yq11.2
SPLIT-HAND/FOOT MALFORMATION 1	183600	7q21.2-q21.3, 2q31
SPLIT-HAND/FOOT MALFORMATION 3	600095	10q24
SPLIT-HAND/FOOT MALFORMATION 4	605289	3q27
SPLIT-HAND/FOOT MALFORMATION 5	606708	2q31

SYNPOLYDACTYLY 1,	186000	2q31-q32
TOWNES-BROCKS-BRANCHIOOTORENAL-LIKE-SYNDROME	107480	16q12.1
TRICHORHINOPHALANGEAL SYNDROME, TYPE I	190350	8q24.12
TRICHORHINOPHALANGEAL SYNDROME, TYPE II	150230	8q24.11, q24.13
TUBEROUS SCLEROSIS	191100	16p13.3, 12q14, 9q34
VELOCARDIOFACIAL SYNDROME	192430	22q11.2
WAGR SYNDROME	194072	11p13
WILLIAMS-BEUREN SYNDROME	194050	7q11.23
WILMS TUMOR 1,	194070	13q12.3, 11p13, Xq26
WOLF-HIRSCHHORN SYNDROME	194190	4p16.2, 4p16.3