

Seznam mikrolečních syndromů detekovatelných pomocí Cytochip Focus Constitutional

17q21.31 MICRODELETION SYNDROME, (610443), 17q21.31	LERHWEILL DYSCHONDROSTEOSIS, (127300), Ypter-p11.2, Xpter-p22.32
22q13.3 DELETION SYNDROME, (606232), 22q13.3	LISSENCEPHALY, X-LINKED, (300067), Xq22.3-q23
3q29 MICRODELETION SYNDROME, (609425), 3q29	MENTAL RETARDATION, X-LINKED, WITH PANHYPOPHYTARISM, (300123), Xq26.3
ADENOMATOUS POLYPOSIS OF THE COLON, (175100), 5q22.1, 5q22.2	METACHROMATIC LEUKODYSTROPHY, (250100), 22q13.31-qter
ADRENAL HYPOPLASIA, CONGENITAL, (300200), Xp21.3-p21.2	MICROPHTHALMIA, SYNDROMIC 7, (309801), Xp22
ALAGILLE SYNDROME 1, (118450), 20p12.2	MILLER-DIEKER LISSENCEPHALY SYNDROME, (247200), 17p13.3
ANGELMAN SYNDROME, (105830), 15q11-q13, Xq28	MITOCHONDRIAL COMPLEX I DEFICIENCY, (252010), 1q23, 11q13, 6q16.1, 5q12.1, 5q11.1, 5pter-p15.22, 2q33-q34, Xq24
ANIRIDIA, TYPE II, (106210), 11p13	MONOSOMY 1p36 SYNDROME, (607872), 1p36
AUTISM, (209850), 7q22, 6p21.3-p21.2, 2q32	MUSCULAR DYSTROPHY, BECKER TYPE, (300376), Xp21.1
AUTISM, X-LINKED, SUSCEPTIBILITY TO, 1, (300425), Xq13	MUSCULAR DYSTROPHY, DUCHENNE TYPE, (310200), 12q21, Xp21.2
AUTISM, X-LINKED, SUSCEPTIBILITY TO, 2, (300495), Xp22.33	NAIL-PATELLA SYNDROME, (161200), 9q34.1
AUTISM, X-LINKED, SUSCEPTIBILITY TO, 3, (300496), Xq28	NEPHRONOPHTHISIS 1, (256100), 2q13
BASAL CELL NEVUS SYNDROME, (109400), 9q22.3	NEUROFIBROMATOSIS, TYPE I, (162200), 17q11.2
BECKWITH-WIEDEMANN SYNDROME, (130650), 11p15.5, 5q35	NEUROFIBROMATOSIS, TYPE II, (101000), 22q12.2
BRACHYDACTYLY-MENTAL RETARDATION SYNDROME, (600430), 2q37	NEUROPATHY, HEREDITARY, WITH LIABILITY TO PRESSURE PALSIES, (162500), 17p11.2
BRANCHIOOTORENAL SYNDROME 1, (113650), 8q13.3	NOONAN SYNDROME 1, (163950), 12q24.1
BRUTON AGAMMAGLOBULINEMIA TYROSINE KINASE, (300300), Xq21.3-q22	PELIZAEUS-MERZBACHER DISEASE, (312080), Xq22
CAMPOMELIC DYSPLASIA, (114290), 17q24.3-q25.1	POLYCYSTIC KIDNEY DISEASE, INFANTILE SEVERE, WITH TUBEROUS SCLEROSIS, (600273), 16p13.3
CAT EYE SYNDROME, (115470), 22q11	POTOCKI-LUPSKI SYNDROME, (610883), 17p11.2
CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A, (118220), 17p11.2	POTOCKI-SHAFFER SYNDROME, (601224), 11p11.2
CHARCOT-MARIE-TOOTH DISEASE, X-LINKED, 1, (302800), Xq13.1	PRADER-WILLI SYNDROME, (176270), 15q12, 15q11-q13
CHARGE SYNDROME, (214800), 8q12.1, 7q21.1	PRADER-WILLI-LIKE SYNDROME ASSOCIATED WITH CHROMOSOME 6, (176270), 6q16.3
CLEIDOCRANIAL DYSPLASIA, (119600), 6p21	RETINOBLASTOMA, (180200), 13q14.1-q14.2
CORNELIA DE LANGE SYNDROME 1, (122470), 5p13.1	RETT SYNDROME, (312750), Xq28, Xp22
CR1-DU-CHAT SYNDROME, (123450), 5p15.2	RIEGER SYNDROME, TYPE 1, (180500), 4q25-q26
DANDY-WALKER SYNDROME, (220200), 3q24	RUBINSTEIN-TAYBI SYNDROME, (180849), 16p13.3, 22q13
DIAPHRAGMATIC HERNIA, CONGENITAL, (142340), 15q26.1	SAETHRE-CHOTZEN SYNDROME, (101400), 10q26, 7p21.1
DIGEORGE SYNDROME, (188400), 22q11.2	SEX-DETERMINING REGION Y, (480000), Yp11.3
DIGEORGE SYNDROME/VELOCARDIOFACIAL SYNDROME SPECTRUM OF MALFORMATION 2, (601362), 10p14-p13	SMITH-MAGENIS SYNDROME, (182290), 17p11.2
DOSAGE-SENSITIVE SEX REVERSAL, (300018), Xp21.3-p21.2	SOTOS SYNDROME, (117550), 5q35
DOWN SYNDROME, (190685), Xp11.23, 21q22.3, 1q43	SPERMATOGENIC FAILURE, NONOBSTRUCTIVE, Y-LINKED, (415000), Yq11.2
FEINGOLD SYNDROME, (164280), 2p24.1	SPLIT-HAND/FOOT MALFORMATION 1, (183600), 7q21.2-q21.3, 2q31
FRAGILE X MENTAL RETARDATION SYNDROME, (300624), Xq27.3	SPLIT-HAND/FOOT MALFORMATION 3, (600095), 10q24
GREG CEPHALOPOLYSYNDACTYLY SYNDROME, (175700), 7p13	SPLIT-HAND/FOOT MALFORMATION 4, (605289), 3q27
HETEROTAXY, VISCERAL, X-LINKED, (306955), Xq26.2	SPLIT-HAND/FOOT MALFORMATION 5, (606708), 2q31
HOLOPROSENCEPHALY 1, (236100), 2q37.1-q37.3, 21q22.3	SYNPOLYDACTYLY 1, (186000), 2q31-q32
HOLOPROSENCEPHALY 2, (157170), 2p21	TOWNES-BROCKS-BRANCHIOOTORENAL-LIKE-SYNDROME, (107480), 16q12.1
HOLOPROSENCEPHALY 3, (142945), 7q36	TRICHO RHINOPHALANGEAL SYNDROME, TYPE I, (190350), 8q24.12
HOLOPROSENCEPHALY 4, (142946), 18p11.3	TRICHO RHINOPHALANGEAL SYNDROME, TYPE II, (150230), 8q24.11-q24.13
HOLOPROSENCEPHALY 5, (609637), 13q32	TUBEROUS SCLEROSIS, (191100), 16p13.3, 12q14, 9q34
HYPERGLYCEROLEMIA, (307030), Xp21.3-p21.2	VELOCARDIOFACIAL SYNDROME, (192430), 22q11.2
HYPOPARATHYROIDISM, SENSORINEURAL DEAFNESS, AND RENAL DISEASE, (146255), 10p15	WAGR SYNDROME, (194072), 11p13
ICHTHYOSIS, X-LINKED, (308100), Xp22.32	WILLIAMS-BELUREN SYNDROME, (194050), 7q11.23
JACOBSEN SYNDROME, (147791), 11q23	WILMS TUMOR 1, (194070), 13q12.3, 11p13, Xq26
JOHANSON-BLIZZARD SYNDROME, (243800), 15q15-q21.1	WOLF-HIRSCHHORN SYNDROME, (194190), 4p16.2, 4p16.3
JOUBERT SYNDROME 4, (609583), 2q13	
KABUKI SYNDROME, (147920), 8p22	
KALLMANN SYNDROME 1, (308700), Xp22.3	