CytoSure™ Syndrome Plus

Genetic Disorder	Band(s)	OMIM
1p36 Microdeletion	1p36	607872
2q32.2 Micordeletion	2q33.1-q33.2	119540
2q37 .3 Monosomy	2q37.3	
3q29 Microdeletion	3q29	609425
6p25.3 Microdeletion	6p25.3	220210
8p23.1 Microdeletion	8p23.1	600576
8p23.1 Microdeuplication	8p23.1	
9q22.32-q22.33 / Loeys-Dietz Microdeletion	9q22.33	609192
9q34.3 Microdeletion	9q34.3	610253
9q34.3 Microdeletion (severe)	9q34.3	610253
10q22-23 Microdeletion	10q23.1-q23.2	
11q14 Microdeletion	11q14.2	
11q23 Jacobsen Deletion Syndrome	11q23.3-q24.3	147791
13q14 Deletion	13q14.2-q21.1	
14q22 Microdeletion	14q22.2-q23.1	607932
15q13.3 Microdeletion	15q13.3	
15q15.3 Infertility and Deafness	15q15.3	
15q24 Microdeletion	15q24.1	
16p11.2 Deletion	16p11.2	
16p11.2-p12.2 Microdeletion	16p11.2-p12.2	
16p13.1 Microdeletion predisposition to Autism	16p13.11	
17q21.31 Microdeletion	17q21.31	
17q21.31 Microduplication	17q21.31	
22q11.2 Microduplication	22q11.2	608363
22q13 Microdeletion (Phelan McDermid syndrome)	22q13.33	606232
Abnormal growth	15q26.3	610254
Adrenal hypoplasia congenita	Xp21.2	300200
Adult-onset autosomal dominant leukodystrophy	5q23.2	169500
Alagille Syndrome	20p12.2	118450
Albright hereditary osteodystrophy	20q13.32	103580
Alpha thalassemia mental retardation	16p13.3	141750
Alport syndrome (X)	Xq22.3	301050
Angelman	15q11.2	176270
Aniridia	11p13-p14.1	106210
Atrial septal defect (ASD) with atrioventricular conduction defects	5q35.1-q35.2	108900
Autism genes		
AZFa	Yq11.21	415000
AZFb (azoospermia factor b)	Yq11.221-q11.222	415000
AZFb + AZFc	Yq11.222	415000
Bartter 1	15q21.1	601678
Bartter 2	11q24.3	241200
Bartter 3	1p36.13	607364
Bartter 4	1p36.13	602522
Beckwith-Wiedemann	11p15.4-p15.5	130650
Bilateral frontoparietal polymicrogyria	16q13	606854
Blepharophimosis	3q22.3	110100
Branchio-Oto-Renal Syndrome	8q13.3	113650
CADASIL	19p13.12	125310
Campomelic dysplasia	17q24.3	114290
Canavan	17p13.3	271900
Cat-eye	22q11.1	115470

Genetic Disorder	Band(s)	OMIM
Cerebral Carvernous Malformations (CCM)	3q26.1	603285
Cerebral Carvernous Malformations (CCM)	7p13	603285
Cerebral Carvernous Malformations (CCM)	7q21.2	603285
Charcot-Marie Tooth Disease	17p11.2-p12	118220
CHARGE syndrome	8q12.2	214800
Cleidocranial dysplasia	6p21.3	119600
Congenital adrenal hyperplasia (CAH)	6p21.32	201910
Congenital diaphragmatic hernia	15q26.1-q26.2	142340
Congental heart disease genes		
Cornelia de Lange	5p13.2	122470
Cowden	10q23.2-q23.31	158350
Craniosynostosis SOX6	11p15.1-p15.2	607257
Craniosynostosis type2	5q35.2	604757
Cri-du-Chat	5p15.2-p15.33	123450
Currarino	7q36.3	176450
Cystic Fibrosis	7q31.2	219700
Cystinosis	17p13.3	219800
Cystinuria with mitochondrial disease	2p21	606407
Cystinuria	19q13.11	604144
Dandy-Walker Syndrome	3q24	220200
DCX-Associated Lissencephaly/Subcortical Band Heterotopia	Xq22.3-q23	300067
Deafness-Dystonis-Optic Neuronopathy	Xq22.1	304700
DiGeorge / VCF	22q11.21	188400
DiGeorge Duplication		
DiGeorge 2	10p14	601362
DMD	Xp21.1-p21.2	310200
Dopa resistant dystonia / Parkinsons	14q22.2-q22.3	
Down Syndrome Critical Region	21q22.13	190685
Emery-Dreifuss muscular dystrophy	Xq28	310300
Eye disorder genes		
Fanconi Anemia	3p25.3	227646
Feingold	2q24.3	164280
FLNA-Related Disorders	Xq28	300017
FMR1 Microdeletion	Xq27.3	309550
FMR2 Microdeletion	Xq28	309548
Fryns 1q41	1q41	229850
Gardner	5q22.2	175100
Glucose Transport defect	1p34.2	606777
Glycerol kinase	Xp21.2	300474
Greig cephalopolysyndactyly	7p14.1	175700
Hereditary hemorrhagic teangiectasia	9q34.11	187300
Hereditary non-polyposis colon cancer	2p16.3-21	120435
Hirschsprung Disease Plus	2q22.3	142623
HNPP	17p11.2-p12	162500
Holoprosencephaly 1	21q22.3	236100
Holoprosencephaly 2	2p21	157170
Holoprosencephaly 3	7q36.3	142945
Holoprosencephaly 4	18p11.31	142946
Holoprosencephaly 5	13q32.3	609637
Holoprosencephaly 7	9q22.32	610828
Holoprosencephaly and preaxial polydactly	5q35.1	605651
Holt-Oram	12q14.21	142900
Hypocalcemia with Bartter	3q13.33-q21.1	601199
Infantile hyperinulinism, enteropathy and deafness (Ushers disease)	11p15.1	276904
Joubert 5	12q21.32	610188
Kallmann 1	Xp22.31	308700

Genetic Disorder	Band(s)	OMIM
Langer-Giedion Syndrome	8q24.11	150230
Lesch-Nyhan Syndrome	Xq26.2	300322
Li-Fraumeni 1	17p13.1	151623
Lissencephaly with cerebellar hypoplasia	7q22.1	257320
LMNA	1q22	150330
Lowe	Xq25	309000
Marfan 1	15q21.1	154700
Marfan 2	3p24.1	190182
MECP2 / Retts	Xq28	312750
MENKES	Xq21.1	309400
Mental retardation genes incl XLMR		
Mental retardation X linked Xq27.1 dup and del	Xq27.1	300123
Microphthalmia with linear skin defects	Xp22.2	309801
Microphthalmia	3q26.3-q27	206900
Miller-Dieker	17p13.3	247200
Muenke / Achondroplasia	4p16.3	134934
Muscle-Eye-Brain Disease	1p34.1	253280
Nail-Patella	9q33.3	161200
Nebulette	10p12.31	605491
Nephrolithiasis 1, X-linked, Dent disease	Xp11.22-p11.23	310468
Nephronophthisis	2q13	256100
Neurofibromatosis 1 (NF1)	17q11.2	162200
NF1 Microduplication		
Neurofibromatosis 2 (NF2)	22q12.2	607379
NFIA Haploinsufficiency	1p31.3	600727
Noonan 1	12q24.13	163950
Noonan 4	2p22.1	610733
Norrie disease	Xp11.3	310600
Obesity, severe	6q16.3	603128
Okihiro	20q13.2	607323
Oligodontia	14q13.3	604625
Opitz	Xq22.3	300000
Opitz - Kaveggia syndrome	Xq13.1	305450
Optic Atrophy	3q28-q29	165500
Orofaciodigital 1	Xp22.2	311200
Osteopoikilosis	12q14.3	166700
OTC	Xp11.4	311250
Pallistar-Killian	12p12.1	601803
Pelizaeus-Merzbacher disease	Xq22.2	312080
Peters Plus Syndrome	13p13-q12.3	261540
Peutz-Jeghers Syndrome	19p13.3	175200
Pitt Hopkins	18q21.2	610954
Polycystic kidney disease	4q22.1	173910
Potocki-Lupski	17q11.2	610883
Potocki-Shaffer	11p11.2	601224
Prader-Willi	15q11.2	176270
Renal cysts and diabetes	17q12	137920
Retinoblastoma	13q14.2	180200
Rieger	4q25	180500
Rubinstein-Taybi Syndrome	16p13.3	610543
Saethre-Chotzen	7p21.1	101400
Schizencephaly	10q26.11	269160
Sclerostin	17q21.31	605740
SCN1A-related seizures	2q24.3	609634
Short stature	1q25.2	606606
SHOX	Xp22.33	312865

Genetic Disorder	Band(s)	OMIM
Smith-Lemli-Opitz	11q13.4	270400
Smith-Magenis	17p11.2	182290
Smith Magenis Microduplication		
SOTOS	5q35.3	117550
Split/hand foot	7q21.3	220600
Split/hand foot malformation -3	10q24.32	600095
Split/hand foot malformation -5	2q31.1	606708
SRY deletion	Yp11.31	480000
Steroid sulfatase	Xp22.31	308100
Stickler 1	12q13.11	108300
STS Microduplication		
Synpolydactyly	2q31.1	186000
Thrombocytopenia absent radius syndrome	1q21.1	274000
Townes Brocks	16q12.1	107480
Transient Neonatal diabetes	6q24.2	601410
Trischororhinophlangeal, Langer-Giedion Syndrome probes	8q23.3	150230
Tuberous sclerosis-1	9q34.13	191100
Tuberous sclerosis-2	16p13.3	191100
Ulnar-Mammary	12q24.21	181450
Van der Woude Syndrome	1q32.2	119300
VEGFA disorders	6p21.1	192240
Verbal dyspraxia	7q31.1	605317
Von Hippel Lindau	3p25.3	193300
Waardenburg syndrome I	2q36.1	193500
Waardenburg syndrome 4	22q13.1	609136
Waardenburg syndrome IIA	3p14.1	193510
WAGR Syndrome probes	11p13	194072
Walker/Warburg Syndrome	9q34.13	236670
Williams-Beuren	7q11.23	194050
Williams Microduplication	7q11.23	609757
Wilsons disease	13q14.3	277900
Wolf-Hirschhorn	4p16.3	194190
X linked agammaglobulinemia	Xq22.1	300300
X linked heterotaxy	Xq26.3	306955
X linked juvenille retinoschisis	Xp22.13	312700
X linked lymphoproliderative syndrome	Xq25	308240
X linked spasms/ atypical Rett	Xp22.13	300672
Xp11.3 deletion	Xp11.3	300578
XY sex reversal	9q33.3	184757

All Aneupliodies
All Subtelomeres

200 + Syndromes*, Probe every 3Kb within gene, 33 probes per gene
Subtelomeres probe every 40Kb
Recombination hotspots, Probe every 40Kb
Autism/Heart Disease/MR/Eye*. 410 genes, Probe every 3Kb, 33 probes per gene
Genome backbone, Probe every 40Kb

* Average figures for gene regions including 5Kb of flanking region