

Gene	Disease	Reference	#Diseases	Highly Sign.	Significant	Specific	Highest rank	aBandApart concept	Band	voc	bands	links	docs	p-value
chromosome 1			5											
UROD	porphyria cutanea tarda			1	1	0	1	porphyria cutanea tarda	1p34.1	omim	1p34	1/402	2	7,00E-05
GBA	Gaucher disease			1	1	1	1	Gaucher disease	1q21	omim	1q21	13/858	14	2,40E-22
GLC1A	Glaucoma			1	1	1	0	Glaucoma	1q24.3	omim	1q24	24/383	123	2,21E-26
HPC1	Prostate Cancer			1	1	1	0	Prostate Cancer	1q25.3	omim	8p22	52/474	486	0,00E+00
PS2	Alzheimer disease	D		0	1	1	0	Alzheimer disease	1q42.13	omim	1q42.1	2/276	46	2,38E-03
			5	4	5	4	2							
chromosome 2			4											
ETM2	Essential Tremor	A		1	1	1	1	tremor	2p24.1	omim	2p24.1	6/169	54	8,29E-10
MSH2	Colon cancer	B		1	1	1	0	Colon cancer	2p21	omim	5q21	21/368	151	2,26E-20
MSH6	Colon cancer	B		1	1	1	0	Colon cancer	2p16.3	see B	see B	see B	see B	see B
PAX3	Waardenberg syndrome			1	1	1	1	waardenburg syndrome	2q35	omim	2q35	7/317	22	8,05E-11
			4	4	4	4	2							
chromosome 3			4											
VHL	Von Hippel-Lindau			1	1	1	1	von hippel lindau disease	3p26-p25	omim	3p26.2	36/284	77	0,00E+00
MLH1	Colon Cancer	B		1	1	1	0	Colon cancer	3p21.3	omim	see B	see B	see B	see B
SCLC1	Lung Cancer			1	1	1	0	Lung Cancer	3p23-p21	omim	3p14	49/391	570	0,00E+00
ETM1	Essential Tremor	A		1	1	1	1	tremor	3q13	omim	see A	see A	see A	see A
			4	4	4	4	2							
chromosome 4			6											
EVC	Ellis-van-Crevelde			1	1	1	1	ellis van creveld syndrome	4p16	omim	4p16	7/229	11	1,67E-14
HD	Huntington Disease			1	1	1	1	huntington disease	4p16.3	omim	4p16.3	42/311	63	0,00E+00
FGFR3	Achondroplasia			1	1	1	1	Achondroplasia	4p16.3	omim	4p16.3	10/311	17	2,11E-18
NRCLP	Narcolepsy			1	1	1	1	Narcolepsy	4p13-q21	omim	4cen	1/57	6	2,07E-05
SNCA	Parkinson Disease			1	1	1	0	Parkinson Disease	4q21	omim	1ptel	4/338	17	9,72E-08
FOP	Fibrodysplasia ossificans progressiva			1	1	1	0	Fibrodysplasia ossificans progressiva	4q27-q31	omim	17q22	2/400	5	5,68E-06

		6	6	6	6	4						
chromosome 5		5										
SRD5A1	Steroid 5-alpha reductase	0	0	0	0	0	Steroid 5-alpha reductase	5p15 / 2p23				
CKN1	Cockayne syndrome	0	0	0	0	0	cockayne syndrome	5q12	omim	10q21.1	1/104	19 7,77E-04
SMN1	Spinal muscular atrophy	1	1	1	1	1	spinal muscular atrophy	5q12.2-q13.3	omim	5q11.2	45/176	185 0,00E+00
Asthma	Asthma	1	1	1	1	1	asthma	2q22 5q31 5q31.1	omim	5q31.3	54/384	252 0,00E+00
DTD	Diastrophic dysplasia	1	1	1	0	0	Diastrophic dysplasia	5q32-q33.1	omim	5q31	6/629	10 9,95E-10
		5	3	3	3	2						
chromosome 6		5										
SCA1	Spinocerebellar ataxia	1	1	1	0	0	Spinocerebellar ataxia	6p23	omim	12q24.1	16/258	129 1,89E-17
HFE	Hereditary hemochromatosis	1	1	1	1	1	hemochromatosis	6p21.3	omim	6p21.3	22/535	55 6,58E-29
CYP21A	Adrenal hyperplasia, congenital	1	1	1	1	1	adrenal hyperplasia	6p21.3	omim	6p21.3	7/535	29 7,12E-10
IDDM1	Diabetes, type 1	1	1	0	0	0	diabetes	6q24.31	omim	1q21.2	18/473	292 1,32E-10
EPM2A	Epilepsy	1	1	1	0	0	Epilepsy	6q24	omim	15q12	33/668	346 6,35E-18
		5	5	5	4	2						
chromosome 7		5										
GCK	Diabetes, type 1	C	0	0	1	0	diabetes mellitus	7p13	omim			444
ELN	Williams syndrome		1	1	1	1	Williams syndrome	7q11.2	omim	7q11.23	88/301	106 0,00E+00
CFTR	Cystic fibrosis		1	1	0	1	Cystic fibrosis	7q31.2	omim	7q31	29/459	86 0,00E+00
Pendrin	Pendred syndrome		1	1	1	1	Pendred syndrome	7q31	omim	7q31	16/459	23 8,72E-28
OB	Obesity		1	1	1	0	obesity	7q31.3		15q12	37/668	289 2,17E-24
		5	4	4	4	3						
chromosome 8		2										
WRN	Werner syndrome		1	1	1	1	Werner syndrome	8p12-p11.2	omim	8p12	17/281	25 1,00E-32
MYC	Burkitt lymphoma		1	1	0	1	Burkitt lymphoma	8q24.12-q24.13		8q24	21/653	68 4,85E-23
		2	2	2	1	2						
chromosome 9		5										
CDKN2	Malignant melanoma		1	1	1	1	Malignant melanoma	9p21	omim	9p21	40/1178	149 5,23E-30
FRDA	Friedreich's ataxia		1	1	1	1	friedreich ataxia	9q13	omim	9q13	23/189	27 0,00E+00
ABC1	Tangier disease		1	1	1	1	Tangier disease	9q22-q31	omim	9q31	6/255	9 1,76E-12
TSC1	Tuberous sclerosis		1	1	1	1	Tuberous sclerosis	9q34	omim	16p13.3	62/369	145 0,00E+00
ABL	Leukemia, chronic myeloid F		1	1	0	1	myeloid leukemia	9q34.1 / 22	omim	9q34	42/676	235 0,00E+00

			5	5	5	4	5							
chromosome 10			2											
PAHX	Refsum disease		0	0	0	0	0	Refsum disease	10p13 -- 10pter-p11.2	omim	xq21.1	1/206	5,49E-05	
OAT	Gyrate atrophy of the choroid and retina		0	0	0	0	0	chorioretinal atrophy	10q26	omim	6q13	3/142	11 2,38E-08	
			2	0	0	0	0							
chromosome 11			6											
HRAS	Harvey Ras oncogene		0	1	1	0	0	oncogenesis	11p15.5		GO (biolo	13q14	14/773	187 4,45E-07
IDDM2	Diabetes, type 1	C	1	1	1	0	0	diabetes mellitus	11p15.5	see C	see C	see C	see C see C	
LQT	Long QT syndrome		1	1	1	1	1	long qt syndrome	11p15.5	omim	11p15.5	32/630	44 0,00E+00	
VMD2	Best disease		1	1	1	0	0	vitelliform macular dystrophy	11q13	omim	11q12.3	5/180	9 8,74E-11	
MEN1	Multiple endocrine neoplasia		0	0	0	0	0	multiple endocrine neoplasia iia	11q13	omim	20p12.2	1/55	1 1,15E-03	
ATM	Ataxia telangiectasia		1	1	1	1	1	Ataxia telangiectasia	11q22.3	omim	11q23.3	74/559	0,00E+00	
			6	4	5	5	2							
chromosome 12			2											
PXR1	Zellweger syndrome		0	0	0	0	0	zellweger syndrome	12p13.3	omim	5q2.3	1/2	14 7,90E-08	
PAH	Phenylketonuria		1	1	1	1	1	Phenylketonuria	12q24.1	omim	12q24.1	6/258	10 4,70E-12	
			2	1	1	1	1							
chromosome 13			4											
BRCA2	Breast and ovarian cancer	E	1	1	1	1	1	breast cancer	13q12.3	omim	13q12	61/326	1098 0,00E+00	
CX26	Deafness		1	1	1	0	0	Deafness	13q11-q12	omim	xq21	16/242	293 2,87E-12	
RB1	Retinoblastoma		1	1	0	1	1	Retinoblastoma	13q14.1-q14.2	omim	13q14	233/773	461 0,00E+00	
ATP7B	Wilson's disease		1	1	1	1	1	Wilson disease	13q14.3-q21.1	omim	13q14.3	19/283	32 0,00E+00	
			4	4	4	3	3							
chromosome 14			2											
PS1 (AD3)	Alzheimer disease	D	1	1	1	1	1	Alzheimer disease	14q24.3	omim	14q24.3	7/265	46 1,60E-10	
SERPINA1	Alpha-1-antitrypsin deficiency		0	0	0	0	0		14q32.1					
			2	1	1	1	1							
chromosome 15			4											
SNRPN	Prader-Willi syndrome		1	1	1	1	1	prader willi syndrome	15q12, 15q11-q13, 15q11	omim	15q11	249/638	310 0,00E+00	
UBE3A	Angelman syndrome		1	1	1	1	1	Angelman syndrome	15q11-q13, Xq28	omim	15q11	214/638	249 0,00E+00	
FBN1	Marfan syndrome		1	1	1	1	1	marfan syndrome	15q21.1	omim	15q21.1	22/143	47 0,00E+00	

SGLT1	Glucose galactose malabsorption	1	1	1	1	glucose galactose malabsorption	22q13.1	omim	22q13.1	2/265	2	3,04E-05
DGS	DiGeorge syndrome	1	1	1	1	DiGeorge syndrome	22q11.2	omim	22q11	156/1092	242	0,00E+00
NF2	Neurofibromatosis	1	1	1	0	Neurofibromatosis	22q12.2	omim	22q12	42/405	212	0,00E+00
BCR	Leukemia, chronic myeloid F	1	1	1	1	myeloid leukemia	22q11.21 / 9q34.1	see F	see F	see F	see F	see F
		4	4	4	4	3						

chromosome X

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PIG-A	Paroxysmal nocturnal hemoglobinuria	1	1	1	1	Paroxysmal nocturnal hemoglobinuria	Xp22.1	omim	xp22.1	2/174	5	4,66E-07
DMD	Duchenne muscular dystrophy	1	1	1	0	duchenne muscular dystrophy	Xp21.2	omim	xp21	121/360	181	0,00E+00
ATP7A	Menkes syndrome	1	1	1	1	menkes disease	Xq12-q13	omim	xq13.3	14/243	21	5,63E-28
COL4A5	Alport syndrome	1	1	1	0	alport syndrome	Xq22.3	omim	xq22	27/292	50	0,00E+00
IL2RG	Severe combined immunod G	1	1	1	0	scid	Xq13	see G	see G	see G	see G	see G
TNFSF5	Immunodeficiency with hyper-IgM	1	1	1	1	hyper igm	Xq26	omim	xq26	9/336	13	2,54E-17
HPRT1	Lesch-Nyhan syndrome	1	1	1	1	lesch nyhan syndrome	Xq26-q27.2	omim	xq26.2	1/228	2	2,25E-05
FMR1	Fragile X syndrome	1	1	1	1	fragile x syndrome	Xq27.3	omim	xq27	49/416	170	0,00E+00
ALD	Adrenoleukodystrophy	1	1	1	1	Adrenoleukodystrophy	Xq28	omim	xq28	31/908	39	0,00E+00
MECP2	Rett syndrome	1	1	1	1	rett syndrome	Xq28, Xp22	omim	xq28	41/908	76	0,00E+00
HEMA	Hemophilia A	1	1	1	1	Hemophilia	Xq28	omim	xq28	2/908	3	6,75E-06
		11	11	11	11	8						

chromosome Y

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SRY	Sex determination / Testes-determini	1	1	1	0	sex determination	Yp11.3	GO biol	xp21	10/360	39	2,61E-13
		1	1	1	1	0						

number of diseases 93 80 84 79 57