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Characterizing Genetic Syndromes Involved in Cancer and Radiogenic Cancer Risk

Caractérisation des syndromes génétiques liés au cancer et au risque de cancer radiogénique

P. Unrau, K. Doerffer

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ABSTRACT

The COG project 2806A (1995), reviewed the On-line Mendelian Inheritance in Man (OMIM) database of genetic syndromes to identify those syndromes, genes, and DNA sequences implicated in some way in the cancer process, and especially in radiogenic cancer risk. The current report describes a recent update of the survey in light of two years of further progress in the Human Genome project, and is intended to supply a comprehensive list of those genetic syndromes, genes, DNA sequences and map locations that define genes likely to be involved in cancer risk. Of the 8203 syndromes in OMIM in 1997 June, 814 are associated, even if marginally, with cancer. Of the 814 syndromes so linked, 672 have been mapped to a chromosome, and 476 have been mapped to a chromosome and had a DNA sequence associated with their messenger RNA (or cDNA) sequences. In addition, 35 syndromes have sequences not associated with map locations, and the remaining 107 have neither been mapped nor sequenced. We supply the list of the various genetic syndromes sorted by chromosome location and by OMIM descriptor, together with all the associated but unmapped and unsequenced syndromes.

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RÉSUMÉ

Le projet 2806A (1995) du GPC a permis d'examiner la base de données OMIM (*On-line Mendelian Inheritance in Man*) des syndromes génétiques afin de distinguer les syndromes, les gènes et les séquences de l'ADN mis en cause d'une manière quelconque dans le processus du cancer, et en particulier dans le risque de cancer radiogénique. Le rapport actuel signale une mise à jour récente de l'étude à la lumière des résultats de deux années de travaux supplémentaires dans le cadre du projet Human Genome et vise a fournir une liste exhaustive de ces syndromes génétiques, des gènes, des séquences de l'ADN et des lieux sur les cartes factorielles qui définissent les gènes susceptibles d'intervenir dans le risque de cancer. Parmi les 8 203 syndromes relevés dans la base de données OMIM en juin 1997, 814 sont associés, même si ce n'est que de façon marginale, au cancer. Des 814 syndromes ainsi liés, 672 ont été repérés sur la carte factorielle d'un chromosome et 476 ont été repérés sur la carte factorielle d'un chromosome et avaient une séquence de l'ADN associée à leurs séquences d'ARN messager (ou ADNc). En outre, 35 syndromes ont des séquences non associées à des emplacements sur les cartes et, pour les 107 autres, on n'a pas pu établir leur séquence ni leur carte factorielle. Nous fournissons la liste des divers syndromes génétiques triés selon l'emplacement chromosomique et selon le descripteur OMIM, ainsi que tous les syndromes associés mais non repérés sur les cartes factorielles et non séquencés.

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1. SUMMARY

The COG project 2806A (1995), report reviewed the On-line Mendelian Inheritance in Man (OMIM) database of genetic syndromes to identify those syndromes, genes, and DNA sequences implicated in some way in the cancer process, and especially in radiogenic cancer risk. The following report describes a recent update of the survey in light of two years of further progress in the Human Genome project, and is intended to supply a comprehensive list of those genetic syndromes, genes, DNA sequences and map locations that define genes likely to be involved in cancer risk.

In the past two years, the OMIM database has grown by 20% and the number of syndromes in some way associated with cancer risk has increased to a similar extent. This means that 10% of all genetic syndromes yet defined have some links to cancer; this may reflect the fact that 10% of all genes and genetic systems in humans have to do with the control of cell division, cell to cell communications, and the other types of functions implicated in the cancer process. If that is true, then one would expect 6500 to 8000 genes (of the total of about 65 000 in the human genome) to be involved directly or peripherally in the cancer process. In that case, variants of genes in some way affecting cancer risk would be expected to be so common that predictive assays would be unlikely to be developed.

Alternatively, because only about 10-15% of the genes that specify a human have already been described, it may be that the relative proportions of genes associated with cancer may decrease; the reasons for any possible enrichment at this early stage of analysis is that genetic factors in cancer development are of great interest to research oncologists. This is because it is evident that somatic genetic changes occur in cells that give rise to cancers such that the series of changes involved in the cancer process are becoming defined.

Of the 8203 syndromes in OMIM in 1997 June, 814 are associated, even if marginally, with cancer. Of the 814 syndromes so linked, 672 have been mapped to a chromosome, and 476 have been mapped to a chromosome and had a DNA sequence associated with their messenger RNA (or cDNA) sequences. In addition, 35 syndromes have sequences not associated with map locations, and the remaining 107 have neither been mapped nor sequenced. We supply the list of the various genetic syndromes sorted by chromosome location and by OMIM descriptor, together with all the associated but unmapped and unsequenced syndromes.

Identification of the 511 sequence associated syndromes provides a molecular epidemiological database of those genes to be tested for their involvement in cancer and radiogenic cancer risk. Two approaches are being developed. We are working on the isolation and sequencing of cDNA from the FA (C) gene with a view to developing molecular cDNA separation techniques to target those 511 cDNAs we presently associate with cancer risk. The idea is that if we can separate out these 511 cDNAs from the up to

65 000 others expected in a cell, we will have performed an initial 100-fold purification of the genes we are interested in. Analysis of these cDNAs by DNA chip or array technologies would then be relatively straightforward. We are initiating experiments with knockout mice to examine the effects on radiogenic cancer risk of heterozygosity for a limited subset of our top 511 cancer risk genes.

2. INTRODUCTION

Cancer risk arises from interactions between an individual's genotype and factors in the environment. It has long been apparent that somatic changes in the cellular genetic apparatus are connected with carcinogenesis; Knudson's multi-step model of carcinogenesis is consistent with such observations. Because cellular systems are genetically determined, people who carry genetic variants of genes that affect, control, or impinge on cellular division may be at additional cancer risk because the initiation steps of cancer have already occurred. That is, carrying a prior, inherited mutation in a gene that determines cell division systems is itself a genetic cancer risk factor.

In recognizing that cancer is a genetic disease of somatic cells, we recognize that genetic factors may be decisive determinants of cancer risk. Because there is a high natural rate of cancer death, estimated at about 25% of present deaths, it has been impossible to obtain data about cancer induction by ionizing radiation delivered at low doses and low dose rates in human populations. The situation is one in which the biological "noise" of natural cancer incidence has masked any increases that may be due to, for example, low doses at low dose rates of ionizing radiation to genetically at-risk members of the Atomic Radiation Workers population. There are good reasons to believe that no increase in effort in classical epidemiological studies will ever be adequate to establish reasonable estimates of low-dose ionizing radiation risks.

Two solutions exist for this dilemma: to continue to apply present radiation protection standards as they now exist, or to attempt to distinguish between naturally and radiogenically caused cancers. To attempt the second discrimination, two things are needed: a complete list of genes associated with or involved in the cancer process, and molecular signatures of radiogenic somatic genetic changes occurring in cancers.

Improvements in isolating, mapping, sequencing, and determining the functions of genes have come from the availability and public access to genomic databases. The major present example is the complete 12 million base pair genome of the yeast *Saccharomyces cerevisiae*. With the sequencing of the genetic blueprints of the 5800 genes of a whole, eucaryotic (nucleated) organism, the Human Genome community has access to information about genes that are also found in the human blueprint, and by analogy, can assign functions based on those assigned in yeast. Thus, between our last survey of the human genetic syndromes associated with cancer risk and the present survey, functions have been assigned to many human genes. The next phase of the Yeast Genome project is to knockout and determine the functions of all 5800 genes found in that organism by

combinations of genetics, physiology and molecular biology, so that information will be applied to human genes and genetic syndromes.

In our cancer gene lists, we already see this process at work in the assignment of human genes to the RAD51 and RAD52 complexes (defined by functional and sequence homology to the yeast DNA repair and metabolism gene complexes of the same name), and involved in ionizing radiation damage repair in both humans and yeast. The fundamental evolutionary relationships implied by these cross-species functional and DNA sequence relationships means that information from one organism may be applied at least to thinking about others, and that principles worked out in one organism may apply broadly to other or all organisms. So, as was rightly intuited many years ago, studies of DNA damage and repair in simple nucleated cells can be used to establish the fundamental principles that apply to humans, including ARWs.

We expect then a pyramid of functional relationships, based on the hereditary molecule DNA itself and its maintenance, replication and repair, which should be common in all nucleated organisms. Then we expect additional functions associated with the growth and development of more complicated clonal organisms, such as ourselves, and the identification of the genes that make these systems function (and that, when mutated, may lead to malfunction). These will include genes that govern the life and death of an injured cell or nucleus, and those that govern internal cellular communications, as well as external cellular and tissue communications broadly defined. Genes at all these functional levels are in our list, and are associated with the cancer process.

Note that when we say the cancer process, we are not implying that there is one pathway to cancer, or one type of cancer; we are saying that the process by which cells become cancerous is marked by somatic genetic changes that may well be unique for each type of cancer, and may even depend upon the exact mutational changes present in a particular gene for the type and location of the ultimate cancer. That cancers arise in association with increasing instability of the cellular genetic apparatus is becoming overwhelmingly clear.

The list of genes associated with DNA sequences and with cancer risk has several functions:

1. In collaboration with biotechnology firms, we expect the DNA sequence data to be useful in designing methods to separate and analyze large numbers of cDNAs from large numbers of genes involved in cancer risk simultaneously. This is because we expect to be able to use the base pairing properties of the cDNAs from normal cell lines to isolate the cDNAs from at-risk cells, or from actual tumors, for mutational analysis.

2. This catalogue of genes, functions and sequences can serve as a first index to the characterization of the series of genetic changes, and the genes involved, in different types of cancers. We would expect the patterns of gene activation and inactivation in

different tumors to be different, depending upon the initial genetic risk factors present, and their evolution during the cancer process.

3. Furthermore, we expect there to be radiogenic signatures of the types of somatic genetic changes seen in tumors, which may be used to distinguish induced tumors from spontaneous ones. It may, in fact, be possible not only to reduce the uncertainties associated with the radiogenic signature, but to apply tests of radiogenic signature to tumors in individual ARWs. The ability to classify early events in the tumorigenic process as radiogenic or not may prove to be a very useful capability, both for workers and management.

4. Finally, because the various genes listed can be classified as to cellular function, we expect to be able to define those cellular functions that affect cell division and control functions to prioritize the genes to be tested in animal knockout studies. That is, we expect to be able to rank the defined genes in order of cancer risk, radiogenic cancer risk, and function, to define a subset of genes covering the most interesting and relevant genetic cancer risk factors, and use that list to define the order of testing of mouse knockouts for radiogenic cancer risk.

3. METHODS

Our first OMIM review prepared for COG project 2806A (1995), generated a list of 641 syndromes associated with cancer risk, of which 256 syndromes were linked to DNA sequences in GenBank. The Internet supplied a further list of syndromes associated with breast cancer, containing OMIM numbers but not associated GenBank numbers. Finally, we searched OMIM in its latest version for "cancer genes" and extracted a further 328 syndromes for classification. By combining the lists and removing redundancies, we generated a list of syndromes with OMIM numbers and expanded it by including genes known to be involved in DNA metabolism in yeast, which therefore are good candidates for similar functions in humans. Our updated list contains 814 entries. They are listed in Table 1, sorted primarily by chromosome location and secondarily by OMIM number. Remember that this is a list of genetically based syndromes in some way associated with cancers and/or with DNA metabolism.

Each of these entries was then searched for associated GenBank sequence acquisition numbers, map location, and textual evidence of association with cancer risk or occurrence. Of the 814 syndromes, 476 were assigned a chromosomal location, a GenBank number, and indication of association with cancer and/or radiogenic cancer. A further 35 were not assigned a chromosomal location, but were assigned a GenBank number, and are thus accessible for molecular analysis. The 511 OMIM# syndromes assigned either a GenBank accession number and a chromosomal location, or only the GenBank number, comprise our present set of genes, which are in some way associated with cancer risk, and for which molecular assays are being developed.

4. FUNCTIONAL GROUPINGS

We are using an iterative Venn-diagram-based approach to classify the genes associated with cancer risk in functional groupings. This means that we initially center our classification on DNA and DNA metabolic functions, including replication, repair and recombination. An even earlier stage, which we have not yet considered, but which we know impinges upon the details of replication and repair, is the synthesis of precursors for nucleic acid replication and repair. But the replicative and repair functions must be integrated with the nuclear signals that coordinate the replication cycle and the cell cycle, as indicated in Figure 1.

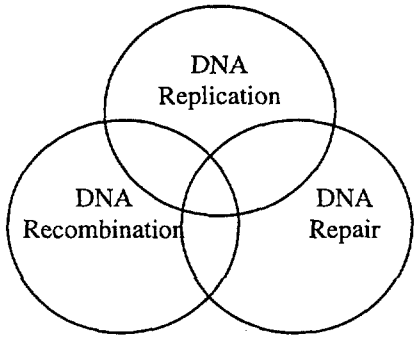

Figure 1: DNA metabolism.

There is a requirement for the functions of the cell cycle to be coordinated with those of other cells in a tissue, and this integration requires cell to cell communications. Some of these intracellular signaling systems have been implicated in carcinogenesis, and the subsetting of Figure 2 provides one way to integrate this data.

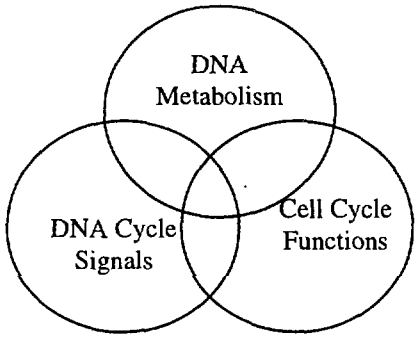

Figure 2: DNA - cell cycle integration - cell cycle functions.

A tissue must then become a functional organ that uses intercellular cell signaling systems of hormones, and other cellular effectors, receptors, and so forth, as indicated in Figure 3.

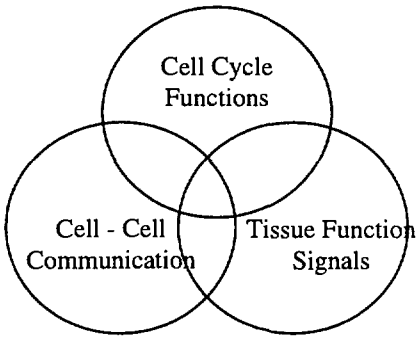

Figure 3: Cell cycle - tissue integration.

Finally, because cell, tissue, and organ functions are integrated systems within the whole organism, there are additional levels of control associated with the functioning of the immune system in self-surveillance. It should be remembered that the recombinational mechanisms of the immune system are also cross-functional in DNA damage repair (as judged by the radiation sensitivity of cells from severe combined immune deficiency mice, or Swiss Type Aggamaglobulinemia patients). This implies interconnectivity in the systems required for a whole organism to function, as shown in Figure 4. This means that each level at which control is exerted is a potential candidate for loss of function control leading to, causing, or abetting the cancer process.

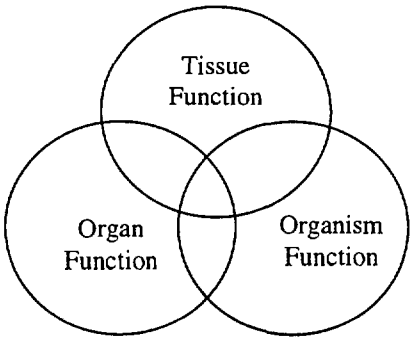

Figure 4: Tissue - organ - organism integration.

If we recognize then that there are many interconnections that are not obvious on first analysis, we can generate a series of nested sets of functions, nested in Venn diagram terms, in which at each advancing level of organismic organization we can see new interactions. Thus, Figure 1 indicates the types of genes involved in the core DNA metabolic functions of replication, repair, and recombination (which is a key somatic genetic event in the cancer progression process).

Because we have chosen a Venn analysis of genes and gene functions in cell cycle control, and hence cancer initiation and progression, we always have sets that include prior sets. Thus, for instance, even though we picked the DNA cycle set as our primary

set, we could have extended the analysis down to the level of the synthesis of DNA precursors, which are known to affect the distribution of DNA damage in the various DNA repair pathways, which are implicated in the repair of spontaneous and radiogenic DNA lesions. Similarly, we could have focused exclusively on the sorts of DNA damage induced by the environment and by ionizing radiation. However, such subsetting in detail is premature to the general organization of the arguments about genes involved in the cancer process.

Similarly, as each tissue represents the product of developmental pathways as yet not understood, it might be expected, by analogy with somatic differentiation pathways deduced from Drosophila Aristopedelia mutations, that the loss of control of differentiation leading to cancers in different tissues would themselves follow unique patterns. That is, the occurrence of a breast cancer arising from the risk source being a variant of the Ataxia Telangiectasia gene may follow a very different developmental path from one occasioned by the risk associated with carriers of variants of the BRCA1 or BRCA2 genes.

These distinctions will become clear as the methods for determining pathways of cancer development as a function of the genetic risk factors carried by an individual are worked out. Of course, in crosses between inbred strains of mice, we would expect to see consistent cancer development pathways if the genes involved do not generate genetic variants in other genes. Thus, for example, if the absence of p53 function in tumors developing in p53+/- knockout mice leads to higher mutation rates in other genes during the cell cycle, then a wide spectrum of secondary mutations might be observed, leading to a large number of potential pathways to cancer progression. But if the absence of function leads to, for example, the successive loss of function of a set of genes that are heterozygous between the two parental strains, then we would expect the spectrum of genetic changes within tumors to be broadly consistent in this example, in inbred mice.

We know, for example, that in yeast loss of the RAD51 and RAD52 functions is associated with hypermutability during the normal DNA replication cycle, implicating recombinational functions in copy-editing after DNA replication. In the case of RAD51 or RAD52 knockouts, we would then expect to see higher post-replication DNA mutation rates. Certainly, in yeast, the spontaneous rates are enhanced at least 10-fold in such strains. We might, therefore, expect tumors in RAD51 or RAD52 knockouts to reflect this higher post-replication mutation rate by mutating other genes in the spectrum of those that occur in tumors, so the pathways to cancer development may be different in RAD51 and p53 knockout strains. This of course remains to be tested.

We have therefore established an algorithm for the functional classification of genes into the systems they are involved in, and the interrelationships of these functional systems in living things. Because of the underlying functional and evolutionary relationships between different classes and types of living organisms, it has become apparent that such an integrated functional systems approach will be valuable in setting research priorities in cancer and radiogenic cancer risk gene studies. For the long-term goals of being able to

figure out radiogenic cancer risks in individual ARWs, a combined gene risk profile and dose approach will need to be used.

Table 1: List of genes involved in cancer process sorted by chromosome and OMIM#.

GENE NAME	GENE LOCATION	OMIM#	SEQUENCE			
PHEO	1p	171300				
NHLH2	1p12	162361	M96740		M97508	M97507
CSF1	1p13	120420	HUMCSF1M	M37435	X05825	
GALBP	1p13	137033	M64303			
NGFB	1p13.1	162030	VO1511			
NRAS	1p13.2	164790	X02751			
GST1	1p13.3	138350	M96233	J03817	X08020	
ARH9	1p21	165380				
T-UURIA	1p22	274270	U09178	HSU20938		
MTS1	1p22.1	154280				
MIS1	1p32	137290	J04152	X77753		
BLYM	1p32	164830	K01884			
JUN	1p32	165160	J04111			
PTPRF	1p32	179590	Y00815			
LMYC	1p32	180610	X58480			
SIL	1p32	181590	X57515			
T-cell leuk/lymph - 5	1p32	187040	M81078			
FABP3	1p33	134651	U57623	X56549		
DDIT1	1p34	126335	M60974			
EDN2	1p34	131241	M65199	X55177		
MPLV	1p34	159530	HSTPRMPL	M90103		
Paraneoplas Enceph	1p34	168360	U12431	M62843		
LCK	1p34.3	153390	S72855			
MYCL1	1p34.3	164850	M19720			
EBVIS	1p35	132850				
CSF3R	1p35	138971	M59819	M59820	M59818	
PLA2G2A	1p35	172411	M22430	HUMRASFA		
Inf. Gene. Agranuloc	1p35	202700	S78382			
ERPL1	1p36	131190	M11348			
CD30	1p36	153243	M83554			
Malig. Mel.	1p36	155600				
	1p36	157975				
E2F1	1p36	189971	L48996	M96577		
Breast Canc Ductal	1p36	211420				
LAP18	1p36.1	151442	J04991			
FGR	1p36.2	164940	HUMSRC	HUMFGR0		

continued....

GENE NAME	GENE LOCATION	OMIM#	SEQUENCE		
PAX7	1p36.2	167410	L09745	HUMPAX20	X15044
HKR3	1p36.3	165270	HSHFR3SO		
Neuroblastoma	1p36.3	256700	D28124		
S100A8	1q12	123885	X06234	X06233	Y00278
HPTHYR	1q21	145001			
MUC1	1q21	158340	J05582	J05581	HUMEPISL
MCL1	1q21	159552	L08246		
PE1	1q21	164873	L16464		
Pap.Renal Cell Carc	1q21	179755	X99720	X97124	
	1q21	191315	X79910		
FCGR1A	1q21.2	146760			
Charcot-Marie-Tooth	1q22	118200	D14583	D14720	D14584
NHLH1	1q22	162360	M96739		
SKI	1q22	164780	X15218	X15217	X15219
Tropomyosin	1q22	191030	X79910		
Antithromb III Def	1q23	107300	HUMAT3X	X00238	X68793
TRK	1q23	164970	X79910	X79909	X03541
PBX1	1q23	176310	M31523		
ABL2	1q24	164690	M14904	M14903	
PROSC	1q24-25	601518			
RNS4	1q25	180435	X74987	X76388	
Transl. Prom reg.	1q25	189940	M15326		
CTSE	1q31	116890	HUMCTSE0		
	1q41	190220	M19154		
HRES1	1q42	143025			
ADPRT	1q42	173870	M32721	HUMADPRT	
Chediak Higashi	1q42.1	214500	U67615	L77889	
5SRNA	1q42.11	180420	V00589	J01867	
Z F P 124	1q44	194631	S54641		
VSNL1	2p	600817	U14747		
InV	2p12	147200	HUMIGKC		
ARH6	2p12	165370			
PAP	2p12	167805	L15533	S51768	
CD8B	2p12	186730	M17514		
Pulm Alv. Proteinosis	2p12	265120			
REL	2p13	164910			
TGFA	2p13	190170			
GTBP	2p16	600678	U28946		
Fam. Nonpoly Colon	2p22	120435	HSHMSH	L47580	L47581
HTLF	2p22	143089	U57029		
ALK	2p23	105590	U04946		
XANTHU	2p23	278300			
FAP	2p23	600403			
DDX1	2p24	601257	HSCL1042		

continued.....

GENE NAME	GENE LOCATION	OMIM#	SEQUENCE			
MYCN	2p24.1	164840	XO2363			
Xeroderma Pig 111	2p25	278720	D21090			
MG50	2p25.3	600134				
BRCA1A	2q	601593				
NF2	2q12.2	101000	X72664	X72663	Z22664	L11353
GLI2	2q14	165230	HUMGLI2	HUMHKR4		
LCO	2q14	165320				
	2q14	601248				
ERCC3	2q21	133510	M31899			
ADCP2	2q23	102720	HSDPPIV	X60708		
CHRNA1	2q24	100690	X74624	Y00762		
PMS1	2q31	600258				
CREB1	2q32.3	123810	M27691			
IGFBP5	2q33	146734	L27560	HUMIGFBPO		
INHA	2q33	147380	M13437	M13436	M13144	
TCL-4	2q34	186860				
Waardenberg Synd	2q35	193500	X15251			
HEK	3p11.2	179611	M83941			
Hyperneph	3p14.2	144700	U46922			
PTPRG	3p14.2	176886	M34668			
FHIT	3p14.2	601153				
APH	3p21	102645	J03068			
CDC25A	3p21	116947				
CCK	3p21	118440	HUMCCK0			
WNT5A	3p21	164975	L20861			
ARH12	3p21	165390				
UBE2	3p21	191325	L13852			
TCTA	3p21	600690	L41143			
ACY1	3p21.1	104620	L07548			
SEMA-V	3p21.2	601281				
CTNNB1	3p21.3	116806	X87838	X89448		
Col7A1	3p21.3	120120	L02870	M65158		
NPFCC	3p21.3	120436	U07418	U07343		
Muir - Torre	3p21.3	158320				
Epidermolysis Bullo	3p21.3	226600	M22613	K01886		
Turcot Syndrome	3p21.3	276300				
SEMA3F	3p21.3	601124	U17279	U17278		
TGFBR2	3p22	190182	M85079			
VIPR	3p22	192321	HSV1RG	U13989		
ZNF35	3p22	194533	HUMZNF35P			
Small Cell Lung Can	3p23	182280				
RARB	3p24	180220	X07282	X51650		
THRB	3p24.3	190160	X04707			
RAF1	3p25	164760	X03484			

continued.....

GENE NAME	GENE LOCATION	OMIM#	SEQUENCE			
TR4	3p25	601426				
von Hippel Lindau	3p26	193300	L15409			
CD80	3q21	112203	HUMB7AN0			
CDCL-1	3q21	116945	X67334			
CALLA	3q21	120520	HUMCALLA			
	3q21	191155	M90657			
EVI1	3q26	165215	S82592			
RPL22	3q26	180474	L21756			
B-cell lymphoma-6	3q27	109565	U00115			
FIM3	3q27	136770				
MAP97	3q29	155750	M12530	HUMP97		
CD38	4p15	107270				
ALB	4q11	103600	HUMALBAF			
AFP	4q11	104150	M16110			
GRO2	4q12	139110	M36821	M36820	J03561	X53799
GRO1	4q12	155730	J03561	X12510		
KIT	4q12	164920	L04143	X06182		
PDGFRA	4q12	173490	HUMCSF1R0			
INP10	4q21	147310				
JCH	4q21	147790				
Myeloid/lymph/leuk-2	4q21	159557	L22179	L04731		
FGF5	4q21	165190	HUMFGF5			
RAP1GDS1	4q21	179502				
GNRHR	4q21.2	138850	L03380			
EGF	4q25	131530	K01166			
IL2	4q26	147680	J00264	K02056	V00564	
CCNA	4q27	123835	X68303	X51688		
Schler. Kerat.Derm	4q28	181600				
MN	4q28.2	111300	U00179	U00178	U00177	M12857
Hepatocell Carc-2	4q32.1	142380				
IRF2	4q35.1	147576	D14082			
C9	5p13	120940	K02766	X02176		
LIFR	5p13	151443				
	5p13	176761	M31661			
OV CA.	5p13	601236	U53446	U53445		
MMLV12	5p14	157960				
Renal cell carc.3	5q	179770				
MSH3	5q11	600887	JO4810			
RASA1	5q13.3	139150	M23612	M23379		
HERDES	5q21	135290				
MCC	5q21	159350	M62397			
APC	5q21	175100	M73548	M73547	M74088	
CTNNA1	5q31	116805	Z37994	D14705		
PDGFRB	5q31	173410	HUMCSF1R0	M21574		

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GENE NAME	GENE LOCATION	OMIM#	SEQUENCE			
EGR1	5q31.1	128990				
CSF2	5q31.1	138960	L07488	M11734	M10663	
IL9	5q31.1	146931	M86593			
IRF1	5q31.1	147575				
IL3	5q31.1	147740	M14743	L07488		
CD14	5q31.1	158120	X13334			
ADRA1B	5q33	104220	L31773	U03865		
CSF1R	5q33.2	164770	HUMCSF1R0			
NPM	5q35	164040	U04946	M23613		
FGFR4	5q35.1	134935	M58051	X57205		
KRAS	6p12	190070	HUMRASKO			
VEGF	6p12	192240	HUMVEGF			
TFAP2B	6p12	601601				
DSP	6p21	125647				
SKIV1L	6p21	600478	U09877			
CDKN1A	6p21.2	116899	L25610	U03106		
PIM1	6p21.2	164960	M54915			
HLADR	6p21.3	142860	K02405	J00193	J00198	
HLAG	6p21.3	142871	L27836	L27837	L41363	HSHLAGS
Paget Disease of Bo	6p21.3	167250				
TAP1	6p21.3	170260	L21208	X57522		
PBX2	6p21.3	176311	D28769	X80700		
TNF	6p21.3	191160	M10988			
Adrenal Hyper 111	6p21.3	201910	M26856	M26857		
Hemochromatosis	6p21.3	235200	U22963	J04755		
DEK	6p23	125264				
FIM1	6p23	136750				
PI6	6p25	173321	HSSCCA2S	HSSCCA1S	Z22658	
MPSH	6q	601228				
Collagen	6q12	120165	M63597			
	6q14	190920				
EDDR1	6q16	600452	Z29093			
FYN	6q21	137025				
AMD1	6q21	180980	HUMAMDO	U02035		
CGA	6q21.1	118850	HUMGLYCA	V00518		
LAMA2	6q22	156225	Z26653			
ROS1	6q22	165020	M13880			
	6q22	189990	MI5024			
MACS	6q22.2	177061	D10522			
MAS1	6q24	165180				
CYTOVIL	6q25	123900	J05021	X51521		
ESR	6q25.1	133430	X73067	M12674	X03635	
IGF2R	6q26	147280	J03528	Y00285		
	6q26	192320	HUMVIPHM			
MLLT4	6q27	159559	U02478			
THBS2	6q27	188061	L12350			
ARAF2	7p11.4	164710	L24037			

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GENE NAME	GENE LOCATION	OMIM#	SEQUENCE			
INM7	7p12	147830				
EGFR	7p12.3	131550	X00588			
GLI3	7p13	165240				
AMPH	7p14	600418	U07616			
MYCLK1	7p15	164865	M64786			
	7p15	186970				
IL6	7p21	147620	X04430	X04602		
PMS2	7p22	600259	U24169	U38980		
TPTN12	7q11.23	600079	D13380	M93425		
EPO	7q21	133170	M11319	X02157	X02158	
HGF	7q21.1	142409	M73240	M73239	HUMHGFG	M55379
PGY1	7q21.1	171050	M14758			
PGY3	7q21.1	171060	X06181			
SRI1	7q21.1	182520				
CTR1	7q21.3	114131	L00587			
Acetylcholin.	7q22	100740	HUMACHE0	L42812		
CDP	7q22	116896	M74099			
DRA	7q22	126650	L02785			
MUC3	7q22	158371	M55406	M55405		
CLD	7q22	214700				
CYP3A4	7q22.1	124010	D00003	M18907	M13785	X12387
MET	7q31	164860	J02958	M37519	X54559	
Thyroid Hormonogen	7q31	274600				
PCTT	7q32	167800	L36190			
PIP	7q32	176720	X51503			
EPH	7q32	179610	BTTGR			
BRAF	7q34	164757	X65187			
TCRB	7q35	186930				
Presacral Teratoma	7q36	176450				
Werner Synd.	8p11	277700	L76937			
FLG	8p11.2	136350	U10888	X61692		
PolB1	8p11.2	174760	HSDPOLB			
TPA	8p12	173370	HUMTPA	X02901	M26666	
HGL	8p22	142445	M94168	M94167	M94166	
PPCTS	8p22	601385	HUMN33S	U42349		
DEF1	8p23	125220	M23281	M26602	M21130	
ISONAS	8p23.1	243400	D90042	D90040		
LYN	8q	165120	M16038			
	8q11	190060	J00119			
AGAMA S T	8q11	202500	L27425			
PRKDC	8q11	600899	U47077	U34994		
OPRK1	8q11.2	165195	U17298			
TELBF1	8q13	600951	U40705			
Microceph/normal	8q21	251260				
BBS	8q21	600885				
CBFA2T1	8q22	133435	D43969	D43968	D43638	

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GENE NAME	GENE LOCATION	OMIM#	SEQUENCE		
MYBL1	8q22	159405	X13293	X13294	
GPT1	8q24	138200			
PVT1	8q24	165140	X04620		
	8q24	167959			
NOV	8q24.1	164958			
Mult Exostoses	8q24.11	133700	U70539		
MYC	8q24.12	190080	J00120		
GLI4	8q24.3	165280			
McKusick Chond.	9p13	250250			
IFNW1	9p21	147553			
Cutan Malig Mel	9p21	155601	L11248		
MTAP	9p21	156540	U22233		
CDKN2A	9p21	600160	L27211	HSPCDK	
CDKN2B	9p21	600431	L36844		
ACO1	9p22	100880			
IFNA1	9p22	147660	J00215	J00216	V00532
HAM	9p22	159558	L13744	L13773	L13743
Acute lympho leuk	9p22	247640			
TRYP1	9p23	115501	X51420		
OVC	9p24	164759	S69805		
CTSL	9q21	116880	M20496	X12451	
GAS1	9q21.3	139185	L13698		
Basal Nevus Cell Ca	9q22.3	109400	U59464		
Fanc Pancyt. C	9q22.3	227645	X66894	X66893	
PTCH	9q22.3	601309	U59464		
TAL-2	9q31	186855	M81078		
PBX3	9q33	176312	X59842		
ABO	9q34	110300	J05175	U22302	
SURF-1	9q34	185620	X61923		
CAN	9q34.1	114350			
C5DEF	9q34.1	120900	M65134		
ENG	9q34.1	131195	HSENDOGL		
NPS1	9q34.1	161200			
ABL1	9q34.1	189980	U07000		
Xeroderma Pig 1	9q34.1	278700	D14533		
DAPK1	9q34.1	600831	X76105	X76104	
Notch 1	9q34.3	190198	X79439	X80115	
GLIOMA	10p12	137800			
BMI1	10p13	164831	L13689		
ERCC6	10q11	133540	U28413	L04791	
Thyroid Carc Pap	10q11	188550	M31213		
Med. Thyroid Carc	10q11.2	155240			
MSMB	10q11.2	157145	HUMMSPO	MI5885	
Neuromata/Endo	10q11.2	162300	U05016	U05017	
RET	10q11.2	164761	M57464		
Pheo. Islet cell tum	10q11.2	171420			
Mult Hamartoma Sn	10q22	158350			

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GENE NAME	GENE LOCATION	OMIM#	SEQUENCE		
Term. Deoxy. transf.	10q23	187410	HUMDONT		
ZNF32	10q23	194539			
DEL10	10q23.3	601728	U93051		
NFKB2	10q24	164012			
T-cell leuk -3	10q24	186770	S38742	M75952	
URK	10q24	191840	DOO244		
APT1	10q24.1	134637	M67454	X63717	
CYP2E	10q24.3	124040	J02843	M17398	
Prostate Cancer	10q25	176807	U20770		
MXI1	10q25	600020	S78470		
MGMT	10q26	156569	M29971	M31767	
BEK,FGFR2	10q26	176943	M55614		
SPI1	11p11.2	165170	X52056		
KAI1	11p11.2	600623			
Xeroderma Pig V	11p12	278740			
CD44	11p13	107269	HUMSCG	M69040	M69039
MIC4	11p13	143040			
AL-A1	11p13	151250			
Acute T-cell Leuk	11p13	151390	M15853	M14906	M14905
RAG1	11p13	179615	M29474		
LMO2	11p13	180385	X61118		
Wilms	11p13	194070	HUMWT1		
HEPAT	11p14	114550	M23190		
ST2	11p14.3	185440			
HTS1	11p15	140750			
CALCA	11p15.2	114130	HUMCALCR	X02330	
CALCB	11p15.2	114160	X02404		
TSG101	11p15.2	601387	U52945		
Prarthyrin	11p15.3	168450	HUMPTH		
EMG Synd.BWS	11p15.4	130650			
Ad. Skel. Mus.	11p15.5	103280			
CTSD	11p15.5	116840	M11233		
HRC1	11p15.5	143023	M91083		
IGF2	11p15.5	147470	X00910	J03242	M17863
LSP1	11p15.5	153432	S58726		
MUC2	11p15.5	158370	M74027	M22404	M22405
MER2	11p15.5	179620			
RRM1	11p15.5	180410	L10342		
HRAS	11p15.5	190020	J00277	VOO574	
Wilms Tumor T2	11p15.5	194071			
Adenocort. Carc.	11p15.5	202300			
Rhabdomyosarc	11p15.5	268210			
HRAS	11p15.5	310990			
CDKN1C	11p15.5	600856			
M4F2	11q	158070	J02939	J03569	
Here. Angioneurotic	11q11	106100	M13656	M13203	
CD20	11q13	112210	M27394		
Mult Endocr. Adeno	*11q13*	*131100*			

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GENE NAME	GENE LOCATION	OMIM#	SEQUENCE		
GSTP1	11q13	138370	X65032		
LEU1	11q13	153340	X04391		
NDUFV1	11q13	161015	S67973		
EMS1	11q13	164765			
FGF3	11q13	164950			
FGF4	11q13	164980	J02986		
SEA	11q13	165110			
CCND1	11q13	168461	Z23022	M64349	HUMCCNDBO
PPP1A	11q13	176875	J04759		
ST3	11q13	191181			
FOLR1	11q13.3	136430	U20391	M28099	X62753
Chronic Lymphatic L	11q13.3	151400	M350537	M30535	M18806
MRE11	11q21	600814	U37359		
Anal Canal Carc.	11q22	105580			
PgR	11q22	264080	M15716		
CASP1	11q22.2	147678	M87507		
Ataxia Telangiec	11q22.3	208900	U33841		
Ataxia T grp D	11q22.3	208905	L24203		
Myeloid/lymph/leuk	11q23	159555	L22179	L01986	L04285
PGL1	11q23	168000			
SRPR	11q23	182180	X06272		
CD3G	11q23	186740	X04145		
BRCA3	11q23	600048			
PLZF	11q23.1	176797	Z19002		
ETS1	11q23.3	164720	J04102	JO4101	
RNA Helicase	11q23.3	164905	D17532	Z11685	
CBL	11q23.3	165360			
Ewing Sarc Bk Pt	11q24	193067	M93255	X67001	
BB1	12p	185595			
RBBP2	12p11	180202			
RecQl	12p12	600537	L36140		
PTHLH	12p12.1	168470	M17183	J03580	
IAPP	12p12.3	147940	J04422		
MPE	12p13	131440			
FGF6	12p13	134921	X63454		
MIC3	12p13	143030	M38690		
PTPN6	12p13	176883	M74903	X62055	
CD27	12p13	186711			
RAD52	12p13	600392	U12134		
TNF1	12p13.2	191190	M60275	M58286	
TCF13L1	12p13.2-2	601714			
CD63	12q12	155740	M58485	X07982	
WNT1	12q12	164820	X03072	X07876	
ATF1	12q13	123803	X55543	X55544	X5545
PVIS2	12q13	167960			
poly-A bind. prot.	12q13	173865			
SAS	12q13	181035	S78432		
ERBB3	12q13	190151	M29366		

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GENE NAME	GENE LOCATION	OMIM#	SEQUENCE			
APR	12q13.1	107770	X13916			
DDIT	12q13.1	126337	X71428	S40706		
GLI1	12q13.2	165220	X07384			
MDM2	12q14.3	164785	Z12020			
Mult Lipomatosis	12q15	151900	X71428	X71427	U29107	
ASCL1	12q22	100790	L08424			
BTG1	12q22	109580	X61123	X52870		
Testicular tumors	12q22	273300				
PKU1	12q24.1	261600	K03020	D28124		
RSN	12q24.31	179838	X64838			
HIECTDYS	13q	129500				
FLT3	13q12	136351	L36162	U02687		
FLT	13q12	165070				
TDPX1	13q12	600538	U06099			
BRCA2	13q12.3	600185				
Adamantinoma	13q14	102660				
B-Cell Mal.	13q14	109543				
Bladder Cancer	13q14.1	109800				
RETINO	13q14.1	180200	L11910	L41870		
Osteogenic Sarc	13q14.1	259500				
Rhabdomyosarc-2	13q14.1	268220				
ESD	13q14.11	133280	M13450			
Psudochol E1	13q26.1	177400	M16541			
Renal cell carc.2	14q	179760				
Angiogenin	14q11	105850				
MMP14	14q11	600754	D26512			
CTSG	14q11.2	116830	M16117	J04990		
TCRD	14q11.2	186810	Z24462			
TCRA	14q11.2	186880				
FKHL1	14q13	164874	X74143			
Nuc. Phos.	14q13.1	164050	HUMPNU	K02574		
NSP	14q21	600865	U22398	D64137		
Max prot	14q23	154950	U19775	M64240		
FOS	14q24.3	164810				
OV CA.	14q31	601404				
CHGA	14q32	118910	J03483			
CKBB	14q32	123280	L47647	M60806	M16451	
US1	14q32	276900				
	14q32	600009	X67325			
P1	14q32.1	107400	J02619	HUMA1ATP	K022212	M11465
T-cell lymph/leuk	14q32.1	186960	X82241			
AKT1	14q32.3	164730	K02777			
ELK2	14q32.3	165350				
IGHG1	14q32.33	147100	Z49801			
MIC7	15q11	108990				
Prader Willi Synd	15q11	176270	HSPPROT	M99564		
Albinism II	15q11.2	203200	M99564			
B2MR	15q13	109710				

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GENE NAME	GENE LOCATION	OMIM#	SEQUENCE			
LTK	15q15.1	151520	D16105			
RecA	15q15.1	179617	D13804			
B2M	15q21	109700				
ANX2	15q21	151740	M62899	M62898	M62896	
PML	15q22	102578	X64800	S50916	S50913	
CYP1A1	15q22	108330	M12078	K03191	X02612	Z00036
CYP1A2	15q22	124060				
CSK	15q23	124095	X60114			
Tyrosinemia t-1	15q23	276700	M55150			
Thyroid hor. b.p.	15q24	188555				
PACE	15q25	136950	X04329			
IGF1R	15q25	147370	X04434			
AGC1	15q26.1	155760	X96753			
	15q26.1	190030	X06292			
Bloom Synd.	15q26.1	210900	U39817	HUMLIG10		
CD19	16p11.2	107265				
FUS	16p11.2	137070	X71427	X71428	S62140	
STX1B	16p11.2	601485	L27586			
REG ENT	16p12	266600				
BCM	16p13.1	109545	Z14955	Z14320	Z14954	
MRP1	16p13.1	158343	HUMMRPX			
SMHC	16p13.13	160745	D10667	L20298		
XPF	16p13.2	278760				
MPG	16p13.3	156565	M99626	L10752		
TSC2	16p13.3	191092	X75621			
TTD	16p13.3	601675				
CAR	16q	116935	X65784	D14075		
WT Limb/Blood Synd	16q	194350				
RBL2	16q12.2	180203	X74594			
CFBA	16q22	121360	L20298			
FRA16A	16q22	136580	S70397			
MAF	16q22	177075				
ZNF23	16q22	194527				
HPR	16q22.1	140210	HUMHPARS			
CDH1	16q22.1	192090	HUMCDHO			
MOV34	16q23	157970				
CDH13	16q24	601364				
MC1R	16q24.3	155555	X67594	L08603	X65634	X65633
Renal Dipeptidase	16q24.3	179780	J05257			
Fanc. Pancyt. 1	16q24.3	227650	X99226	HUMFA		
MIC6	17p11	143060				
Cancer of colon	17p13.1	114500				
Medulloblastoma	17p13.1	155255				
PEDF	17p13.1	172860	M76979			
RCV1	17p13.1	179618	S43855			
TP53	17p13.1	191170	L25610			
BCPR	17p13.3	113721				
CRC17	17p13.3	120460				

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GENE NAME	GENE LOCATION	OMIM#	SEQUENCE			
CRK	17p13.3	164762	S65701			
HTLVR	17q	143090				
CYB561	17q11	600019	HUMCYTO			
SCYA7	17q11.2	158106	X72309	X72304	X72308	
EVI2	17q11.2	158380	HUMEV12			
EVI2B	17q11.2	158381	M60830	M60829		
van Reck Neuofibro	17q11.2	162200	HSNF1GEN	M89914	D12625	HUMNFX
THRA1	17q11.2	190120	X04707			
17BHyd Dehydrog.	17q12	109684	U05659			
HYPKAR	17q12	144200	X75015			
IGFBP4	17q12	146733				
ERBB2	17q12	164870	X03363	M11730		
PNP	17q12	167780	M15788	M15789	X00491	
RARA	17q12	180240	X06614	X06538		
DLG2	17q12	600723	X82895			
BRCA1	17q21	113705	L78833			
TOP2A	17q21	126430	J04088			
MAPT	17q21	157140	J03778			
INT4	17q21	165330				
JUP	17q21	173325	Z68228	M23410		
PHB	17q21	176705	S85655	L14273		
Glycogen storage	17q21	232200				
DUSP3	17q21	600183	L05147			
Ovarian Tumor	17q21.1	167000	X76952			
MDC prot	17q21.3	155120	D17390			
NM23	17q21.3	156490	X75598	L16785		
NM23B	17q21.3	156491	X58965			
Chonic Lymph/leukB	17q22	151441	M31826	S67782		
IGAB29 prot	17q23	147245	L27587			
Kerat Palm Plant	17q23	148500				
	17q23.1	254600	J02694			
ZFP147	17q23.1	600453	D21205			
SSTR2	17q24	182452	L05521			
Russell-Silver Synd	17q25	180860				
TIMP2	17q25	188825				
	17q25	264470	HSAOX01			
LGALS3BP	17q25	600626	U20770			
EVPL	17q25	601590	U53786			
CD-7	17q25.2	186820	X06180			
CF	17q31.2	219700	M55106	M55131		
ERV1	18p11.21	131150	HUMERVM			
YES1	18p11.3	164880	D00333			
Thymidylate synth.	18p11.32	188350	D00517	X02308		
TGFR	18q	275355				
Cancer	18q11	114400				
Hepatit B Int Site	18q11.1	142333				
DSG2	18q12.1	125671	M77830			
GRP	18q21	137260	K02054	M12550		

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GENE NAME	GENE LOCATION	OMIM#	SEQUENCE			
GLV	18q21	182090	J03061			
MADH4	18q21	600993	U44378			
MADH2	18q21	601366	U76622	U65019		
DCC	18q21.3	120470	X76132			
FVT1	18q21.3	136440	X63657			
Chr. L. Leuk, T2	18q21.3	151430	M15878	M13995	M14745	M15699
MASPIN	18q21.3	154790	HSSCCA2S	HSSCCA1S	U04313	
SCCA1	18q21.3	600517	HSSCCA2S			
JUND	19p13.1	165162				
RAB3A	19p13.1	179490				
LYL1	19p13.2	151440				
MEL	19p13.2	165040				
JUNB	19p13.2	165161				
RAD23A	19p13.2	600061	D21090	D21235		
RENT1	19p13.2	601430	U65533			
FUT3	19p13.3	111100	D89325	D89324		
CDC34	19p13.3	116948	L22005			
DNMT	19p13.3	126375	X63692	U70051		
TCF3	19p13.3	147141	HUMIGCD	HUMIGCC	Z49801	
MMLT1	19p13.3	159556	U16282	L04284		
VAV1	19p13.3	164875	X16316			
Int Hamart. Polypos	19p13.3	175200				
BCL3	19q13	109560				
Pros spec anti	19q13	176820	M24543	X05332		
AXL	19q13.1	109135	S65125			
Lu	19q13.1	111200	X83425	X80026		
CCNE	19q13.1	123837	HSCCNC	M74093	M73812	M74092
AKT2	19q13.1	164731	M95936			
PSG1	19q13.1	176390	HUMPSBGB			
MKN28	19q13.1	600765	Z48615			
CD66	19q13.2	109770	J03858			
CEA	19q13.2	114890	M15042			
LIG1	19q13.2	126391	HUMLIG10	M36067		
NCRA	19q13.2	163980	M29541	M29540		
LPSA	19q13.2	164953				
ZNF42	19q13.2	194550	M58297			
Xeroderma Pig IV	19q13.2	278730				
CD33	19q13.3	159590				
FOSB	19q13.3	164772	L49169			
BAX	19q13.3	600040				
PolD1	19q33.2	174761	M81735			
PYGB	20p11.1	138550	J03544			
Cholestasis w Sten	20p11.2	118450	X83384			
PCNA	20p12	176740	J04718			
OXT	20p13	167050				
AVP	20p13	192340	M11166			
HCK	20q11	142370				
GHRF	20q11.2	139190	HUMGRFP			

continued.....

GENE NAME	GENE LOCATION	OMIM#	SEQUENCE			
CD40	20q12	109535	X60592			
TOP1	20q12	126420	J03250	HUMTOP		
SRC	20q12	190090	HUMSRC			
APOP	20q13	601342				
PTP1B	20q13.1	176885	M31724			
BMYB	20q13.1	310305				
BRCA2	20q13.2	600125				
TFAP2C	20q13.2	601602	X95693	X95694		
Myeloprolif. Synd.	21q11.2	159595				
CBR	21q22.12	114830	J04056			
TFF1	21q22.3	113710				
Acute Myeloid Leuk	21q22.3	151385	D43969	D43968	D43967	L34598
Melan Tu Anti	21q22.3	155770	M15395			
ETS2	21q22.3	164740	M11921	M11922		
ERG	21q22.3	165080	M17254	HUMERG1		
TFF2	21q22.3	182590	X51698			
Trisomy 21	21q22.3	190685				
TFF3	21q22.3	600633	LO8044			
DSBB	22q11	152690	J04611			
IGLC1	22q11.12	147220	HUMIGLCB	M61769	J00255	
IGLV	22q11.2	147240				
MMP11	22q11.2	185261				
C.M.Leukemia	22q11.21	151410	M17543	U07000	HSABLGR	
Ewing Sarc	22q12	133450	U17163	X66899	X67001	
LIF	22q12.1	159540	X13967			
OSM	22q12.1	165095				
MGM	22q12.2	156100	X85026	X85024	X82209	M55987
	22q12.3	190040	M12783			
CYP2D	22q13.1	124030	M20403	M33387	M33388	
COL1AR	Chr. 15	120340				
Keratin KD1	Chr. 17	148020	M33101	J03607	Y00503	
LIG3	Chr. 17	600940	U40671			
ITGB6	Chr. 2	147558	M35198			
LAG5	Chr. 4	151450				
FEA	Chr. 6	137010				
Mito ATPase	Chr.10	164360				
Ductal Breast Canc.	Chr.13	211410	M33647	L11910		
HKR1	Chr.19	165250				
HKR2	Chr.19	165260				
PKCSH	Chr.19	177060	J03075			
PST1	Chr.5	167790	HUMPSTIA			
Cockayne synd	Chr.5	216400	U28413			
PolD2	Chr.7	600815	U21096			
HTK	Cr7	600011				
Rothmund Thomson	Cr8	268400				
ELK!	Xp11.2	311040	M25269			
RCCXL	Xp11.2	312390	X86175			
Synovial Sarc.	Xp11.2	312820	X86175	X86174	X79200	

continued.....

GENE NAME	GENE LOCATION	OMIM#	SEQUENCE	
Aldrisch Synd	Xp11.23	301000	D84454	U19927
ARAF1	Xp11.4	311010	L24037	
UBIQUIT	Xp21.2	300050	U44839	
X-Link Agammaglob	Xp22	300310		
Phosphotidylinositol	Xp22.1	311770	D11466	
GRPR	Xp22.3	305670		
	Xp22.3	306970		
N Syndrome	Xp22.3	310465		
MIC2	Xp22.3	313470	X16996	
AML-M2	Xp22.32	306250	M59941	M73832
XLICTHY	Xp22.32	308100	J04964	
AIS	Xq11	300068		
SMAX1	Xq11	313200		
AR	Xq11	313700	L29496	
LAMBR	Xq13	150370		
Agammagloulinemia	Xq21.3	300300	U78027	
Basex Synd	Xq24	301845		
X-link Prog. Immunod	Xq25	308240		
	Xq26	312870	L47125	
MCF.2	Xq27	311030	J03639	
CRD1; CDR34	Xq27.1	302650	M16965	
Congen. Dyskeratos	Xq28	305000		
G6PD	Xq28	305900	X55448	
L1CAM	Xq28	308840	X67912	
	Xq28	312173	M64241	1683004
	Yp11	425000		
MIC2Y	Yp11.2	450000		
	Yp11.3	480000	L08063	
Acanthosis Nigricans		100600		
STAT3		102582	L29277	
Alzheimer Disease		104300	L44577	X06989
Amyloidosis(RET)		105250		
Aniridia		106200		
AHH		108340		
Barrett Esophagus		109350		
Basal Cell Carc		109390		
Blu Rub Bleb Nevus		112200		
Bone Dysp. Med Fibr		112250		
DF3Antig.		113720	J03651	
Burkitt Lymphoma		113970	M20583	
CDW52		114280		
Fam Cancw Rad Res		114450		
Fam Breast Can		114480		
Intestinal Carcinoid		114900		
Cerebral Gigantism		117550		

continued.....

GENE NAME	GENE LOCATION	OMIM#	SEQUENCE		
Cerebral Sarc		117600			
Var in Cerumen		117800			
Chelitis Glanularis		118330			
Chemodectoma		118350			
CHK		118491	D10704		
EPX		131399	M26515	HUMEPP	
Fam. Erythroleuk		133180			
Esophageal Canc		133239			
pS2		133420	X00474		
Fibrocystic Pulm Dys		135000			
Gastric Cancer		137215			
Gastric Lymph		137245			
ENPEP		138297	L14721	L12468	
GH		139250	J03071	J00148	V00519
HANDED		139900			
HLF		142385	M95585	M95586	
Hyperkeratosis Lent		144150			
Pri. Fam HyperPara.		145000			
IL4		147684	L15344		
Kaposi Sarc		148000	U75698	U18552	
Fam. Act. Keratoses		148390			
LDHK		150160			
Acute Mono. Leuk		151380			
Lichen Schlerosus		151590			
Li-Frau		151623			
Macroglobulinemia		153600			
Mal Intraocular Mela		155700			
Uveal Melanoma		155720			
MUC18		155735	M29277	M28882	
Mel Astrocyt Synd		155755			
Mal. Mesothelioma		156240			
Chrom. Mosaicism		158250			
Mal Neur Musc Atro		158650			
Mylocerebellar Disor		159550			
		159580			
Nasopharyn Can.		161550			
Duo. Neuro. Pheochr		162240			
Seb Nevus Jadass'n		163200			
HS2		164756			
YUASA		164891	X53292	X53291	
INT3		164951			
RRAS		165090	HUMRASR		
BMYC		165210			
RMYC		165290			
SNO		165340	X15219	X15217	X15218
Oslam Synd.		165660			
Osteochondromatosis		166000			
M17S2		166945			

continued.....

GENE NAME	GENE LOCATION	OMIM#	SEQUENCE		
Paget Disease		167300			
PG		169700	HUMPEPCZ	HUMPEPCY	HUMPEP
Med. Thyroid Carc.		171400			
		171885	L12052		
Poly ADP Rib Synth.		173871	J03473		
PLD		174050			
Mccune Albright Syn		174800			
Juv Polyposis Coli		174900			
PGJ		175050			
Prokeratosis of Mibe		175800			
PRL		176760	V00566	J00299	
TKF		176944	X59932		
RBBP1		180201			
Emb. Rhabdomyosar		180295			
RPS15		180535	M32405		
Rombo Synd		180730			
Leu-2		186910	M12828		
TALLA		186920			
Tetranectin		187520	X70912		
		187680			
Essent. Thrombocyt		187950			
Fam Throglossal Duc		188455			
		188470			
TL Antigen		188850	HUMCD1A		
DRTF1		189902			
ELF1		189973			
		191309	M34309		
Ureter Cancer		191600			
WAGR Synd.		194072			
Wilms Tu/PseudoH		194080			
Wilms Tumor t-3		194090			
Xeroderma Pig		194400			
Cong Hypoplast. An		205900			
Ataxia T w skin pig		208910			
Aust. antigen		209800			
Lung Cancer		211980			
Chondrosarcoma		215300			
		215510			
DS		223200			
Leuk and Dohle Bod		223350			
Dubrowitz Synd.		223370			
Epidermodysplasia		226400			
Fanc. pancyt.type2		227660	HUMFA		
Galatosyl Def		230430			
Mal. Hemangio cyt		234820			

Table 1 (concluded)

GENE NAME	GENE LOCATION	OMIM#	SEQUENCE
Hemihypertrophy		235000	
Hodgkin		236000	
AML + Polyposis		246470	
Neurocutaneous Mel		249400	
		250650	BTATP2
Mycosis Fungoides		254400	
Mult Myeloma		254500	
Pancreatic carc		260350	
Panc. Bone marrow		260400	
Papilloma of Choroid		260500	
Renal Hamartoma		267000	
Ret. Cell Carc.		267730	
Schinzel Gideon		269150	
Thymic neoplasia		274260	
Wiskott Aldrich		277970	
Xero Pig Norm Rep		278750	
Epider. Verruci. X-lin		305350	
Acute X-linked Leuk		308960	
Reticuloendotheliosis		312500	
Sacral Def.Ant Sac		312800	
SAX		313450	
GBY		424500	
		600186	
GSTT1		600436	X79389
BFL1		601056	U27467
ALG2		601057	U49112
dUTPASE		601266	M89913
ARC		601320	X77548
GRB14		601524	L76687
ING1		601566	
TB15		601587	
ABC3		601615	X97187

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