Details of genes contained in the overlapping regions (identified using Decipher patients)

DUPLICATIONS

We decided to check those genes which are present in the overlapping regions, according to these conditions:

Firstly, we considered as more interesting regions those overlapping regions with 7, 6, 5, 4 or 3 overlpas which contains 1-5 genes.

AND

Secondly, we considered also those regions characterized by just 2 overlaps which contains 1-2 genes.

Source: ***GeneCards – The human gene compendium***

C22orf29

Chromosome 22 Open Reading Frame 29

Overlapping region: **chr22:19771891-19928090** (DUP)
Num. of overlaps for that region: 8
How many other genes present: 3 genes

Function(s): This gene encodes a protein which could induce apoptosis in a BH3 domain-dependent manner. The direct interaction network of Bcl-2 family members may play a key role in modulation BOP intrinsic apoptotic signaling activity.

Associated disease(s): nothing

Associated syndrome(s): nothing

TBX1

T-Box 1

Overlapping region: **chr22:19747566-19749385** (DUP)
Num. of overlaps for that region: 8
How many other genes present: 1 gene

Function(s): T-box genes encode transcription factors involved in the regulation of developmental processes. This gene product shares 98% amino acid sequence identity with the mouse ortholog. DiGeorge syndrome (DGS)/velocardiofacial syndrome (VCFS), a common congenital disorder characterized by neural-crest-related developmental defects, has been associated with deletions of chromosome 22q11.2.

Associated disease(s): Defects in this gene are associated with velocardiofacial syndrome (DiGeorge Syndrome) and Gerstmann syndrome.

Associated syndrome(s): DiGiorge Syndrome and Gerstmann syndrome.

GNB1L

Guanine Nucleotide Binding Protein (G Protein), Beta Polypeptide 1-Like

Overlapping region: **chr22:19771891-19928090** (DUP)
Num. of overlaps for that region: 8
How many other genes present: 3 genes

Function(s): This gene encodes a G-protein beta-subunit-like polypeptide which is a member of the WD repeat protein family. WD repeats are minimally conserved regions of approximately 40 amino acids typically bracketed by gly-his and trp-asp (GH-WD), which may facilitate formation of heterotrimeric or multiprotein complexes. This protein contains 6 WD repeats and is highly expressed in the heart.

Associated disease(s): The gene maps to the region on chromosome 22q11, which is deleted in DiGeorge syndrome, trisomic in derivative 22 syndrome and tetrasomic in cat-eye syndrome. Defect in this gene are related to hypopharynx cancer and cat eye syndrome.

Associated syndrome(s): DiGeorge syndrome, trisomic in derivative 22 syndrome and cat-eye syndrome.

TXNRD2

Thioredoxin Reductase 2

Overlapping region: **chr22:19771891-19928090** (DUP)
Num. of overlaps for that region: 8
How many other genes present: 3 genes

Function(s): This gene encodes a member of the class I pyridine nucleotide-disulfide oxidoreductase family. The encoded protein is a selenocysteine-containing flavoenzyme that maintains thioredoxins in a reduced state, thereby playing a key role in regulating the cellular redox environment. This gene encodes a mitochondrial form important for scavenging of reactive oxygen species in mitochondria.

Associated disease(s): Defects in this gene are associated with generalized resistance to thyroid hormone and thyrotoxicosis.

Associated syndrome(s): nothing

DGCR6

DiGeorge Syndrome Critical Region Gene 6

Overlapping region: **chr22:18895226-18919942** (DUP)
Num. of overlaps for that region: 5
How many other genes present: 2 genes

Function(s): DiGeorge syndrome, and more widely, the CATCH 22 syndrome, are associated with microdeletions in chromosomal region 22q11.2. This gene is a candidate for involvement in DiGeorge syndrome pathology and in schizophrenia.

Associated disease(s): Defects in this gene are associated with DiGeorge syndrome and pharyngitis.

Associated syndrome(s): DiGeorge syndrome.

PRODH

Proline Dehydrogenase (Oxidase) 1

Overlapping region: **chr22:18895226-18919942** (DUP)
Num. of overlaps for that region: 5
How many other genes present: 2 genes

Function(s): This gene encodes a mitochondrial protein that catalyzes the first step in proline degradation (proline dehydrogenase activity and FAD binding).

Associated disease(s): Mutations in this gene are associated with hyperprolinemia type 1, monkeypox and susceptibility to schizophrenia 4 (SCZD4). This gene is located on chromosome 22q11.21, a region which has also been associated with the contiguous gene deletion syndromes, DiGeorge syndrome and CATCH22.

Associated syndrome(s): contiguous gene deletion syndromes, DiGeorge syndrome and CATCH22.

ADCYAP1

Adenylate Cyclase Activating Polypeptide 1 (Pituitary)

Overlapping region: **chr18:776907-1802498** (DUP)
Num. of overlaps for that region: 4
How many other genes present: 3 genes

Function(s): This gene encodes a secreted proprotein that is further processed into multiple mature peptides. These peptides stimulate adenylate cyclase and increase cyclic adenosine monophosphate (cAMP) levels, resulting in the transcriptional activation of target genes. The products of this gene are key mediators of neuroendocrine stress responses.

Associated disease(s): Defects in this gene are associated with post-traumatic stress disorder and adenohypophysitis.

Associated syndrome(s): nothing

LINC00470

Long Intergenic Non-Protein Coding RNA 470

Overlapping region: **chr18:776907-1802498** (DUP)
Num. of overlaps for that region: 4
How many other genes present: 3 genes

Function(s): It is an RNA gene, and is affiliated with the lncRNA class.

Associated disease(s): nothing

Associated syndrome(s): nothing

YES1

V-Yes-1 Yamaguchi Sarcoma Viral Oncogene Homolog 1

Overlapping region: **chr18:776907-1802498** (DUP)
Num. of overlaps for that region: 4
How many other genes present: 3 genes

Function(s): This gene is the cellular homolog of the Yamaguchi sarcoma virus oncogene. The encoded protein has tyrosine kinase activity (ion channel binding and enzyme binding) and belongs to the src family of proteins. This gene lies in close proximity to thymidylate synthase gene on chromosome 18 and a corresponding pseudogene has been found on chromosome 22.

Associated disease(s): Defects in this gene are associated with megaesophagus and patellofemoral pain syndrome.

Associated syndrome(s): patellofemoral pain syndrome.

BCRP2

Breakpoint Cluster Region Pseudogene 2

Overlapping region: **chr22:21440514-21461607** (DUP)
Num. of overlaps for that region: 4
How many other genes present: 1 gene

Function(s): This is a pseudogene

Associated disease(s): nothing

Associated syndrome(s): nothing

CYFIP1

Cytoplasmic FMR1 Interacting Protein 1

Overlapping region: **chr15:22821537-23085400** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): Its encoded protein is a component of the CYFIP1-EIF4E-FMR1 complex which binds to the mRNA cap and mediates translational repression. In the CYFIP1-EIF4E-FMR1 complex this subunit is an adapter between EIF4E and FMR1. It promotes the translation repression activity of FMR1 in brain probably by mediating its association with EIF4E and mRNA (by similarity) and it regulates the formation of membrane ruffles and lamellipodia. Furthermore it plays a role in axon outgrowth and it binds to F-actin (but not to RNA). This is the part of the WAVE complex that regulates actin filament reorganization via its interaction with the Arp2/3 complex. Actin remodeling activity is regulated by RAC1. It is also a regulator of epithelial morphogenesis and it may act as an invasion suppressor in cancers.

Associated disease(s): Defects in this gene are associated with Angelman syndrome and fragile x syndrome.

Associated syndrome(s): Angelman syndrome and fragile x syndrome.

NIPA1

Non Imprinted In Prader-Willi/Angelman Syndrome 1

Overlapping region: **chr15:22821537-23085400** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): This gene encodes a magnesium transporter that associates with early endosomes and the cell surface in a variety of neuronal and epithelial cells. This protein may play a role in nervous system development and maintenance.

Associated disease(s): Defects in this gene are associated with spastic paraplegia 6, spastic paraplegia 6 (autosomal dominant), Prader-Willi syndrome and Angelman syndrome.

Associated syndrome(s): Prader-Willi syndrome and Angelman syndrome.

NIPA2

Non Imprinted In Prader-Willi/Angelman Syndrome 2

Overlapping region: **chr15:22821537-23085400** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): This gene encodes a possible magnesium transporter (magnesium ion transmembrane transporter activity).

Associated disease(s): Defects in this gene are associated with microdeletion 15q11.2 syndrome and Prader-Willi syndrome.

Associated syndrome(s): microdeletion 15q11.2 syndrome and Prader-Willi syndrome.

TUBGCP5

Tubulin, Gamma Complex Associated Protein 5

Overlapping region: **chr15:22821537-23085400** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): The encoded protein is a gamma-tubulin complex is necessary for microtubule nucleation at the centrosome.

Associated disease(s): Defects in this gene are associated with Prader-Willi syndrome and Angelman syndrome.

Associated syndrome(s): Prader-Willi syndrome and Angelman syndrome.

SYNM

Synemin, Intermediate Filament Protein

Overlapping region: **chr15:99618190-99759831** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 2 genes

Function(s): The protein encoded by this gene is an intermediate filament (IF) family member. This protein has been found to form a linkage between desmin, which is a subunit of the IF network, and the extracellular matrix and provides an important structural support in muscle.

Associated disease(s): Defects in this gene are associated with olfactory neuroblastoma and pulmonary sclerosing hemangioma.

Associated syndrome(s): nothing

TTC23

Tetratricopeptide Repeat Domain 23

Overlapping region: **chr15:99618190-99759831** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 2 genes

Function(s): Its functions are still unknown.

Associated disease(s): Diseases associated with this gene include cervical cancer and cervicitis.

Associated syndrome(s): nothing

MPV17L

MPV17 Mitochondrial Membrane Protein-Like

Overlapping region: **chr16:15278668-15504454** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 2 genes

Function(s): One of its isoforms (isoform 1) participates in reactive oxygen species metablism by up- or down-regulation of the genes of antioxidant enzymes (receptor binding).

Associated disease(s): Defects in this gene are related to ureteral obstruction and kidney disease.

Associated syndrome(s): nothing

NPIPA5

Nuclear Pore Complex Interacting Protein Family, member A5

Overlapping region: **chr16:15278668-15504454** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 2 genes

Function(s): Its encoded protein seems to be involved in nuclear pore complex pathway. It is an important paralog of this gene is LOC440353.

Associated disease(s): nothing

Associated syndrome(s): nothing

ABCC1

ATP-Binding Cassette, Sub-Family C (CFTR/MRP), Member 1

Overlapping region: **chr16:16127848-16284248** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 2 genes

Function(s): The protein encoded by this gene is a member of the superfamily of ATP-binding cassette (ABC) transporters. This full transporter is a member of the MRP subfamily which is involved in multi-drug resistance. This protein functions as a multispecific organic anion transporter, with oxidized glutatione, cysteinyl leukotrienes and activated aflatoxin B1 as substrates. This protein also transports glucuronides and sulfate conjugates of steroid hormones and bile salts.

Associated disease(s): Defects in this gene are associated with intraocular retinoblastoma and Dubin-Johnson syndrome.

Associated syndrome(s): Dubin-Johnson syndrome.

ABCC6

ATP-Binding Cassette, Sub-Family C (CFTR/MRP), Member 6

Overlapping region: **chr16:16127848-16284248** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 2 genes

Function(s): The protein encoded by this gene is a member of the superfamily of ATP-binding cassette (ABC) transporters. This full transporter is a member of the MRP subfamily which is involved in multi-drug resistance. The encoded protein, a member of the MRP subfamily, is involved in multi-drug resistance (ATPase activity, coupled to transmembrane movement of substances and transporter activity). It may participate directly in the active transport of drugs into subcellular organelles or influence drug distribution indirectly. It transports glutathione conjugates as leukotriene-c4 (LTC4) and N-ethylmaleimide S-glutathione (NEM-GS).

Associated disease(s): Defects in this gene are associated with acquired pseudoxanthoma elasticum and pseudoxanthoma elasticum (forme fruste).

Associated syndrome(s): nothing

DEFB132

Defensin, Beta 132

Overlapping region: **chr20:232000-356270** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 5 genes

Function(s): Its encoded protein has antibacterial activity (potential).

Associated disease(s): nothing

Associated syndrome(s): nothing

RBFOX1

RNA Binding Protein, Fox-1 Homolog (C. Elegans) 1

Overlapping region: **chr16:5865956-6098252** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 1 gene

Function(s): The Fox-1 family of RNA-binding proteins is evolutionarily conserved, and regulates tissue-specific alternative splicing in metazoa. Fox-1 recognizes a (U)GCAUG stretch in regulated exons or in flanking introns. The protein binds to the C-terminus of ataxin-2 and may contribute to the restricted pathology of spinocerebellar ataxia type 2 (SCA2). Ataxin-2 is the product of the SCA2 gene which causes familial neurodegenerative diseases. Fox-1 and ataxin-2 are both localized in the trans-Golgi network.

Associated disease(s): Defects in this gene are associated with intestinal atresia and mental retardation epilepsy.

Associated syndrome(s): nothing

LOC101927488

Antisense RNA (uncharacterized) [RP5-1103G7.4]

Overlapping region: **chr20:232000-356270** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 5 genes

Function(s): RP5-1103G7.4 is an RNA gene, and is affiliated with the antisense RNA class.

Associated disease(s): nothing

Associated syndrome(s): nothing

NRSN2
Neurensin 2

Overlapping region: **chr20:232000-356270** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 5 genes

Function(s): It may play a role in maintenance and/or transport of vesicles.

Associated disease(s): Defects in this gene are associated with hepatocellular carcinoma and neuronitis.

Associated syndrome(s): nothing

SOX12
SRY (Sex Determining Region Y)-Box 12

Overlapping region: **chr20:232000-356270** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 5 genes

Function(s): Members of the SOX family of transcription factors are characterized by the presence of a DNA-binding high mobility group (HMG) domain, homologous to the HMG box of sex-determining region Y (SRY). The protein encoded by this gene was identified as a SOX family member based on conserved domains, and its expression in various tissues suggests a role in both differentiation and maintenance of several cell types.

Associated disease(s): nothing

Associated syndrome(s): nothing

ZCCHC3
Zinc Finger, CCHC Domain Containing 3

Overlapping region: **chr20:232000-356270** (DUP)
Num. of overlaps for that region: 3
How many other genes present: 5 genes

Function(s): Its encoded protein is related to this gene include nucleic acid binding.

Associated disease(s): nothing

Associated syndrome(s): nothing

DGCR6
DiGeorge Syndrome Critical Region Gene 6

Overlapping region: **chr22:18895226-18919942** (DUP)
Num. of overlaps for that region: 5
How many other genes present: 2 genes

Function(s): DiGeorge syndrome, and more widely, the CATCH 22 syndrome, are associated with microdeletions in chromosomal region 22q11.2. This gene is a candidate for involvement in DiGeorge syndrome pathology and in schizophrenia.

Associated disease(s): Defects in this gene are associated with DiGeorge syndrome and pharyngitis.

Associated syndrome(s): DiGeorge syndrome.

BCRP2

Breakpoint Cluster Region Pseudogene 2

Overlapping region: **chr22:21440514-21461607** (DUP)
Num. of overlaps for that region: 4
How many other genes present: 1 gene

Function(s): This is a pseudogene

Associated disease(s): nothing

Associated syndrome(s): nothing

C15orf41

Chromosome 15 Open Reading Frame 41 (uncharacterized)

Overlapping region: **chr15:36606006-37231466** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes (one is MEIS2, see slide 3)

Function(s): Its functions are not known.

Associated disease(s): Defects in this genes are associated with C15orf41 include congenital dyserythropoietic anemia type I(b) and paraplegia.

Associated syndrome(s): nothing

Conclusion: ???

RGS7

Regulator Of G-Protein Signaling 7

Overlapping region: **chr1:240891005-241063575** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): The protein encoded by this gene inhibits signal transduction by increasing the GTPase activity of G protein alpha subunits thereby driving them into their inactive GDP-bound form. Activity on G(o)-alpha is specifically enhanced by the RGS6/GNG5 dimer. It may play a role in synaptic vesicle exocytosis and an important role in the rapid regulation of neuronal excitability and the cellular responses to short-lived stimulations (by similarity).

Associated disease(s): Defects in this genes are related to agoraphobia and leiomyomatosis.

Associated syndrome(s): nothing

PLD5

Phospholipase D Family, Member 5

Overlapping region: **chr1:242554821-242561749** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): The protein encoded by this gene is an enzymatic component of the tricarboxylic acid (TCA) cycle, or Krebs cycle, and catalyzes the formation of L-malate from fumarate. It exists in both a cytosolic form and an N-terminal extended form, differing only in the translation start site used. The N-terminal extended form is targeted to the mitochondrion, where the removal of the extension generates the same form as in the cytoplasm.

Associated disease(s): Defects in this genes are associated with multiple sclerosis and obesity.

Associated syndrome(s): nothing

MRGPRX1

MAS-Related GPR, Member X1

Overlapping region: **chr11:18949929-18959354** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): It is an RNA gene and is affiliated with the lncRNA class.

Associated disease(s): Defects in this gene are associated with nephrogenic diabetes insipidus and diabetes insipidus.

Associated syndrome(s): nothing

PSMD13

Proteasome (Prosome, Macropain) 26S Subunit, Non-ATPase, 13

Overlapping region: **chr11:215420-237805** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): The 26S proteasome is a multicatalytic proteinase complex with a highly ordered structure composed of 2 complexes, a 20S core and a 19S regulator. This gene encodes a non-ATPase subunit of the 19S regulator. An essential function of a modified proteasome, the immunoproteasome, is the processing of class I MHC peptides.

Associated disease(s): Defects in this genes are associated with anisakiasis and spondylolisthesis.

Associated syndrome(s): nothing

SIRT1

Sirtuin 3

Overlapping region: **chr11:215420-237805** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): This gene encodes a member of the sirtuin family of proteins. The functions of human sirtuins have not yet been determined; however, yeast sirtuin proteins are known to regulate epigenetic gene silencing and suppress recombination of rDNA. Studies suggest that the human sirtuins may function as intracellular regulatory proteins with mono-ADP-ribosyltransferase activity. The protein encoded by this gene is included in class I of the sirtuin family (NAD+ ADP-ribosyltransferase activity and NAD+ binding).

Associated disease(s): Defects in this gene are associated with uterine fibroid and diffuse large B-cell lymphoma.

Associated syndrome(s): nothing

BBOX1

Butyrobetaine (Gamma), 2-Oxoglutarate Dioxygenase (Gamma-Butyrobetaine Hydroxylase) 1

Overlapping region: **chr11:27006061-27182736** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): This gene encodes gamma butyrobetaine hydroxylase which catalyzes the formation of L-carnitine from gamma-butyrobetaine, the last step in the L-carnitine biosynthetic pathway. Carnitine is essential for the transport of activated fatty acids across the mitochondrial membrane during mitochondrial beta-oxidation (gamma-butyrobetaine dioxygenase activityand iron ion binding).

Associated disease(s): Defects in this gene are associated with mulibrey nanism and angina pectoris.

Associated syndrome(s): nothing

FIBIN

Fin Bud Initiation Factor Homolog (Zebrafish)

Overlapping region: **chr11:27006061-27182736** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): Its functions are still unknown.

Associated disease(s): Defects in this gene are associated with endotheliitis.

Associated syndrome(s): nothing

OR52N5

Olfactory Receptor, Family 52, Subfamily N, Member 5

Overlapping region: **chr11:5785900-5805665** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): Its protein encoded is a n olfactory receptor. Olfactory receptors interact with odorant molecules in the nose, to initiate a neuronal response that triggers the perception of a smell. The olfactory receptor proteins are members of a large family of G-protein-coupled receptors (GPCR) arising from single coding-exon genes.

Associated disease(s): Defects in this gene are associated with neuronitis.

Associated syndrome(s): nothing

PWRN1

Prader-Willi Region Non-Protein Coding RNA 1

Overlapping region: **chr15:24487787-24823816** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): This gene is located in the Prader-Willi syndrome (PWS) region of chromosome 15, which is known to undergo imprinting. The transcript is believed to be non-coding. It is bi-allelically expressed in testis and kidney, but mono-allelically expressed from the paternal allele in brain. This gene is poly-adenylated and is known to undergo alternative splicing. Transcript variants may represent part of a complex imprinting center-SNURF-SNRPN transcription unit. The contribution of this gene to the PWS phenotype is unknown, but it has been suggested that it may play a role in establishing paternal imprinting in the PWS region, perhaps by maintaining the paternal allele in an open chromatin configuration.

Associated disease(s): Diseases associated with Prader-Willi syndrome and Angelman syndrome.

Associated syndrome(s): Prader-Willi syndrome and Angelman syndrome

OCA2

Oculocutaneous Albinism II

Overlapping region: **chr15:28155036-28280541** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): This gene encodes the human homologue of the mouse p (pink-eyed dilution) gene. The encoded protein is believed to be an integral membrane protein involved in small molecule transport, specifically tyrosine - a precursor of melanin.

Associated disease(s): Defect of this gene are associated with acute conjunctivitis and oculocutaneous albinism type 2.

Associated syndrome(s): nothing

IGF1R

Insulin-Like Growth Factor 1 Receptor

Overlapping region: **chr15:99408987-99618190** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): This receptor binds insulin-like growth factor with a high affinity. It has tyrosine kinase activity. The insulin-like growth factor I receptor plays a critical role in transformation events. Cleavage of the precursor generates alpha and beta subunits. It is highly overexpressed in most malignant tissues where it functions as an anti-apoptotic agent by enhancing cell survival.

Associated disease(s): Diseases associated with insulin-like growth factor i deficiency and insulin-like growth factor 1 resistance to.

Associated syndrome(s): nothing

PGPEP1L

Pyroglutamyl-Peptidase I-Like

Overlapping region: **chr15:99408987-99618190** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): Its main function includes cysteine-type peptidase activity.

Associated disease(s): nothing

Associated syndrome(s): nothing

ABCC6

ATP-Binding Cassette, Sub-Family C (CFTR/MRP), Member 6

Overlapping region: **chr16:16284248-16305736** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): The protein encoded by this gene is a member of the superfamily of ATP-binding cassette (ABC) transporters. This full transporter is a member of the MRP subfamily which is involved in multi-drug resistance. The encoded protein, a member of the MRP subfamily, is involved in multi-drug resistance (ATPase activity, coupled to transmembrane movement of substances and transporter activity). It may participate directly in the active transport of drugs into subcellular organelles or influence drug distribution indirectly. It transports glutathione conjugates as leukotriene-c4 (LTC4) and N-ethylmaleimide S-glutathione (NEM-GS).

Associated disease(s): Defects in this gene are associated with acquired pseudoxanthoma elasticum and pseudoxanthoma elasticum (forme fruste).

Associated syndrome(s): nothing

SNP

Sialophorin

Overlapping region: **chr16:29673954-29683609** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): The protein encoded by this gene is a major sialoglycoprotein found on the surface of thymocytes, T lymphocytes, monocytes, granulocytes, and some B lymphocytes. It may be part of a physiologic ligand-receptor complex involved in T-cell activation. During T-cell activation, this protein is actively removed from the T-cell-APC (antigen-presenting cell) contact site, suggesting a negative regulatory role in adaptive immune response.

Associated disease(s): Defects in this gene are associated with myeloid sarcoma and Richter's syndrome.

Associated syndrome(s): nothing

LOC101926950

ncRNA (uncharacterized) [RP11-124K4.1]

Overlapping region: **chr16:5286857-5865956** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 gene

Function(s): RP11-124K4.1 is an RNA gene and is affiliated with the antisense RNA class.

Associated disease(s): nothing

Associated syndrome(s): nothing

MIR8065

microRNA 8065 [Homo sapiens (human)]

Overlapping region: **chr16:5286857-5865956** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): It is an RNA gene and is affiliated with the miRNA class. Its functions are not known.

Associated disease(s): nothing

Associated syndrome(s): nothing

DEFB129

Defensin, Beta 129

Overlapping region: **chr20:208113-232000** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): Defensins are cysteine-rich cationic polypeptides that are important in the immunologic response to invading microorganisms. The protein encoded by this gene is secreted and is a member of the beta defensin protein family. Beta defensin genes are found in several clusters throughout the genome, with this gene mapping to a cluster at 20p13.

Associated disease(s): nothing

Associated syndrome(s): nothing

DEFB115

Defensin, Beta 115

Overlapping region: **chr20:208113-232000** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): Its main function is related with antibacterial activity (by similarity).

Associated disease(s): nothing

Associated syndrome(s): nothing

HSFY1P1

Heat Shock Transcription Factor, Y-Linked 1 Pseudogene 1

Overlapping region: **chr22:17279961-17309807** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): It is a pseudogene, and is affiliated with the lncRNA class.

Associated disease(s): Diseases associated with this gene include cat eye syndrome.

Associated syndrome(s): cat eye syndrome,.

XKR3

XK, Kell Blood Group Complex Subunit-Related Family, Member 3

Overlapping region: **chr22:17279961-17309807** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): XKRX (MIM 300684) and XKR3 are homologs of the Kell blood group precursor XK (MIM 314850), which is a putative membrane transporter and a component of the XK/Kell complex of the Kell blood group system.

Associated disease(s): nothing

Associated syndrome(s): nothing

GGT3P

Gamma-Glutamyltransferase 3 Pseudogene

Overlapping region: **chr22:18640301-18889039** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): It is a is a pseudogene whose function is related to glutathione hydrolase activity and gamma-glutamyltransferase activity.

Associated disease(s): Defects in this gene are related to benign recurrent intrahepatic cholestasis and Stickler syndrome.

Associated syndrome(s): Stickler syndrome.

USP18

Ubiquitin Specific Peptidase 18

Overlapping region: **chr22:18640301-18889039** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): The protein encoded by this gene belongs to the ubiquitin-specific proteases (UBP) family of enzymes that cleave ubiquitin from ubiquitinated protein substrates. It is highly expressed in liver and thymus, and is localized to the nucleus. This protein efficiently cleaves only ISG15 (a ubiquitin-like protein) fusions.

Associated disease(s): Defects in this gene are associated with lymphocytic choriomeningitis and DiGeorge syndrome.

Associated syndrome(s): DiGeorge syndrome.

TULP4

Tubby Like Protein 4

Overlapping region: **chr6:158872999-158943409** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): Its encoded protein may be a substrate-recognition component of a SCF-like ECS (Elongin-Cullin-SOCS-box protein) E3 ubiquitin ligase complex which mediates the ubiquitination and subsequent proteasomal degradation of target proteins (by similarity).

Associated disease(s): Defects in this gene are associated with Sensenbrenner syndrome and craniosynostosis.

Associated syndrome(s): Sensenbrenner syndrome.

THSD7A

Thrombospondin, Type I, Domain Containing 7A

Overlapping region: **chr7:11485526-12216730** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): The protein encoded by this gene is found almost exclusively in endothelial cells from placenta and umbilical cord. The encoded protein appears to interact with alpha(V)beta(3) integrin and paxillin to inhibit endothelial cell migration and tube formation. This protein may be involved in cytoskeletal organization.

Associated disease(s): Defects in this gene are associated with osteoporosis, and endotheliitis.

Associated syndrome(s): nothing

AGMO

Alkylglycerol Monooxygenase

Overlapping region: **chr7:14849569-15548854** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): The protein encoded by this gene is a tetrahydrobiopterin- and iron-dependent enzyme that cleaves the ether bond of alkylglycerols. Sequence comparisons distinguish this protein as forming a third, distinct class of tetrahydrobiopterin-dependent enzymes.

Associated disease(s): Defects in this gene are associated with intracranial aneurysm and tuberculosis.

Associated syndrome(s): nothing

DGKB

Diacylglycerol Kinase, Beta 90kDa

Overlapping region: **chr7:14849569-15548854** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): The protein encoded by this gene is a diacylglycerol kinase, beta isotype. Two alternatively spliced transcript variants have been found for this gene. Diacylglycerol kinases (DGKs) are regulators of the intracellular concentration of the second messenger diacylglycerol (DAG) and thus play a key role in cellular processes.

Associated disease(s): Defects in this gene are associated with aggressive periodontitis and retinal degeneration.

Associated syndrome(s): nothing

BLK
B Lymphoid Tyrosine Kinase

Overlapping region: **chr8:11391786-11419675** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): This gene encodes a nonreceptor tyrosine-kinase of the src family of proto-oncogenes that are typically involved in cell proliferation and differentiation. The protein has a role in B-cell receptor signaling and B-cell development. The protein also stimulates insulin synthesis and secretion in response to glucose and enhances the expression of several pancreatic beta-cell transcription factors.

Associated disease(s): Defects in this gene are associated with maturity-onset diabetes of the young (type11).

Associated syndrome(s): nothing

CTSB
Cathepsin B

Overlapping region: **chr8:11683633-11805930** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): The protein encoded by this gene is a lysosomal cysteine proteinase composed of a dimer of disulfide-linked heavy and light chains, both produced from a single protein precursor. It is also known as amyloid precursor protein secretase and is involved in the proteolytic processing of amyloid precursor protein (APP).

Associated disease(s): Incomplete proteolytic processing of APP has been suggested to be a causative factor in Alzheimer disease, the most common cause of dementia. Overexpression of the encoded protein, which is a member of the peptidase C1 family, has been associated with esophageal adenocarcinoma and other tumors. Defects in this gene are associated with ileum cancer and occlusion of gallbladder..

Associated syndrome(s): nothing

FDFT1
Farnesyl-Diphosphate Farnesyltransferase 1

Overlapping region: **chr8:11683633-11805930** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): This gene encodes a membrane-associated enzyme located at a branch point in the mevalonate pathway. The encoded protein is the first specific enzyme in cholesterol biosynthesis, catalyzing the dimerization of two molecules of farnesyl diphosphate in a two-step reaction to form squalene.

Associated disease(s): Defects in this gene are associated with keratolytic winter erythema and Smith-Lemli-Opitz syndrome.

Associated syndrome(s): Smith-Lemli-Opitz syndrome.

ADAM3A
ADAM Metallopeptidase Domain 3A (Pseudogene)

Overlapping region: **chr8:39237438-39386158** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): It is a pseudogene, and is affiliated with the lncRNA class.

Associated disease(s): Defects in this gene are associated with composite lymphoma and Addison's disease.

Associated syndrome(s): nothing

ADAM5

ADAM Metallopeptidase Domain 5 (Pseudogene)

Overlapping region: **chr8:39237438-39386158** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): It is a pseudogene, and is affiliated with the lncRNA class. It is a non catalytic metalloprotease-like protein (by similarity).

Associated disease(s): nothing

Associated syndrome(s): nothing

XG

Xg Blood Group

Overlapping region: **chrX:2700078-2703633** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): This gene encodes the XG blood group antigen, and is located at the pseudoautosomal boundary on the short (p) arm of chromosome X. The three 5' exons reside in the pseudoautosomal region and the remaining exons within the X-specific end.

Associated disease(s): Defects in this gene are related to glucosephosphate dehydrogenase deficiency and x-linked ichthyosis.

Associated syndrome(s): nothing

IL1RAPL1

Interleukin 1 Receptor Accessory Protein-Like 1

Overlapping region: **chrX:28801776-29206531** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): The protein encoded by this gene is a member of the interleukin 1 receptor family and is similar to the interleukin 1 accessory proteins. It is most closely related to interleukin 1 receptor accessory protein-like 2 (IL1RAPL2).

Associated disease(s): Defects in this gene are associated with x-linked mental retardation 21 and complex glycerol kinase deficiency.

Associated syndrome(s): nothing

BRWD3

Bromodomain And WD Repeat Domain Containing 3

Overlapping region: **chrX:79993658-80012744** (DUP)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): The protein encoded by this gene contains a bromodomain and several WD repeats. It is thought to have a chromatin-modifying function, and may thus play a role in transcription.

Associated disease(s): Defects in this gene are associated with chronic lymphocytic leukemia and mental retardation X-linked.

Associated syndrome(s): nothing