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

DELETIONS

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

SATB2

Special AT-Rich Sequence-Binding Protein

Overlapping region: **chr2:199818779-200303015** (DEL)
Num. of overlaps for that region: 7
How many other genes present: 1 gene

Function(s): DNA binding protein that specifically binds nuclear matrix attachment regions. The encoding protein is involved in transcription regulation and chromatin remodeling.

Associated disease(s): Defects in this gene are associated with isolated cleft palate and mental retardation.

Associated syndrome(s): *2q33.1 microdeletion syndrome* and *Toriello Carey syndrome*.

NOTE: This gene is already known to be related with isolated CP.

MEIS2

Myeloid Ecotropic Viral Integration Site 1 Homolog 2

Overlapping region: **chr15:37310981-37515525** (DEL)
Num. of overlaps for that region: 5
How many other genes present: 1 gene

Function(s): This gene encodes a homeobox protein belonging to the TALE ('three amino acid loop extension') family of homeodomain-containing proteins. TALE homeobox proteins are highly conserved transcription regulators, and several members have been shown to be essential contributors to developmental programs.

Associated disease(s): Defects in this gene are associated with *attention deficit hyperactivity disorder* and *acute myeloid leukemia*.

Associated syndrome(s): nothing

NOTE: This gene is already known to be related with isolated CP.

TSHZ1

Teashirt Zinc Finger Homeobox 1

Overlapping region: **chr18:72380146-73152159** (DEL)
Num. of overlaps for that region: 5
How many other genes present: 4 genes

Function(s): This gene encodes a colon cancer antigen that was defined by serological analysis. It is involved in transcriptional regulation of developmental processes.

Associated disease(s): Defects in this gene are associated with *congenital aural atresia* and *microtia*.

Associated syndrome(s): congenital aural atresia syndrome.

ZADH2

Zinc Binding Alcohol Dehydrogenase Domain Containing 2

Overlapping region: **chr18:72380146-73152159** (DEL)
Num. of overlaps for that region: 5
How many other genes present: 4 genes

Function(s): This gene encodes for a protein which is involved in oxidoreductase activity.

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

Associated syndrome(s): nothing

ZNF407

Zinc Finger Protein 407

Overlapping region: **chr18:72380146-73152159** (DEL)
Num. of overlaps for that region: 5
How many other genes present: 4 genes

Function(s): This gene encodes a zinc finger protein whose exact function is not known. It may be involved in transcriptional regulation (DNA binding and zinc ion binding).

Associated disease(s): Defects in this gene are associated with *ectodermal dysplasia.*

Associated syndrome(s): nothing

Conclusion:???

PARD6G

Par-6 Family Cell Polarity Regulator Gamma

Overlapping region: **chr18:77982123-78010032** (DEL)
Num. of overlaps for that region: 5
How many other genes present: 1 gene

Function(s): Among its related super-pathways are cell-cell junction organization and cytoskeleton remodeling CDC42 in cellular processes. It is an adapter protein involved in asymmetrical cell division and cell polarization processes, furthermore it could play a role in the formation of epithelial tight junctions.

Associated disease(s): nothing

Associated syndrome(s): nothing

Conclusion: ???

MIR8063

microRNA 8063 [Homo sapiens (human)]

Overlapping region: **chr18:77982123-78010032** (DEL)
Num. of overlaps for that region: 5
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

MEIS2

Myeloid Ecotropic Viral Integration Site 1 Homolog 2

Overlapping region: **chr15:37310981-37515525** (DEL)
Num. of overlaps for that region: 5
How many other genes present: 1 gene

Function(s): This gene encodes a homeobox protein belonging to the TALE ('three amino acid loop extension') family of homeodomain-containing proteins. TALE homeobox proteins are highly conserved transcription regulators, and several members have been shown to be essential contributors to developmental programs.

Associated disease(s): Defects in this gene are associated with *attention deficit hyperactivity disorder* and *acute myeloid leukemia*.

Associated syndrome(s): nothing

NOTE: This gene is already known to be related with isolated CP.

FAM98B

Family With Sequence Similarity 98, Member B

Overlapping region: **chr15:37515525-38781772** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 5 genes

Function(s): Its functions are not known.

Associated disease(s): nothing

Associated syndrome(s): nothing

LOC101928227

ncRNA (uncharacterized)

Overlapping region: **chr15:37515525-38781772** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 5 genes

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

Associated disease(s): nothing

Associated syndrome(s): nothing

RASGRP1

RAS Guanyl Releasing Protein 1 (Calcium And DAG-Regulated)

Overlapping region: **chr15:37515525-38781772** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 5 genes

Function(s): It functions as a diacylglycerol (DAG)-regulated nucleotide exchange factor specifically activating Ras through the exchange of bound GDP for GTP. It activates the Erk/MAP kinase cascade (calcium ion binding) and regulates T-cells and B-cells development, homeostasis and differentiation.

Associated disease(s): Defects in this gene are associated with leukocyte adhesion deficiency and systemic lupus erythematosus.

Associated syndrome(s): nothing

SPRED1

Sprouty-Related, EVH1 Domain-Containing Protein 1

Overlapping region: **chr15:37515525-38781772** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 5 genes

Function(s): It is phosphorylated by tyrosine kinase in response to several growth factors. The encoded protein can act as a homodimer or as a heterodimer with SPRED2 to regulate activation of the MAP kinase cascade. Its functions include protein kinase binding and stem cell factor receptor binding.

Associated disease(s): Defects in this gene are associated with neurofibromatosis type 1-like syndrome (NFLS) and optic disk drusen.

Associated syndrome(s): *legius syndrome*.

Conclusion: this gene could be interesting.

TMCO5A

Transmembrane and Coiled-Coil Domains 5A

Overlapping region: **chr15:37515525-38781772** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 5 genes

Function(s): Its main function is a structural function (not more details about it).

Associated disease(s): Defects in this gene are associated with paraplegia and bipolar disorder.

Associated syndrome(s): nothing

Conclusion: ???

CCDC102B

Coiled-Coil Domain Containing 102B

Overlapping region: **chr18:66525097-66590638** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 1 gene

Function(s): no data.

Associated disease(s): Defects in this gene are associated with congenital diaphragmatic hernia and microphthalmia.

Associated syndrome(s): nothing

BBS12

Bardet-Biedl Syndrome 12

Overlapping region: **chr4:123652619-123668544** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 2 genes

Function(s): Its protein is part of a complex that is involved in membrane trafficking (ciliogenesis regulating transports vesicles to the cilia). The encoded protein is a molecular chaperone that aids in protein folding upon ATP hydrolysis. This protein also plays a role in adipocyte differentiation.

Associated disease(s): Defects in this gene are associated with *bbs12-related Bardet-Biedl syndrome* and *Bardet-Biedl syndrome 12*.

Associated syndrome(s): *Bardet-Biedl syndrome* and *Bardet-Biedl syndrome 12*.

CETN4P

Centrin EF-Hand Protein 4, Pseudogene1

Overlapping region: **chr4:123652619-123668544** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 2 genes

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

Associated disease(s): nothing

Associated syndrome(s): nothing

DCTD

DCMP Deaminase

Overlapping region: **chr4:183698508-184167427** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 5 genes

Function(s): The protein encoded by this gene catalyzes the deamination of dCMP to dUMP, the nucleotide substrate for thymidylate synthase. The encoded protein is allosterically activated by dCTP and inhibited by dTTP, and is found as a homohexamer. This protein uses zinc as a cofactor for its activity.

Associated disease(s): Defects in this gene are associated with lymphosarcoma and neutropenia.

Associated syndrome(s): nothing

FAM92A1P2

Family With Sequence Similarity 92, Member A1 Pseudogene 2

Overlapping region: **chr4:183698508-184167427** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 5 genes

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

Associated disease(s): nothing

Associated syndrome(s): nothing

TENM3

Teneurin Transmembrane Protein 3

Overlapping region: **chr4:183698508-184167427** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 5 genes

Function(s): Involved in neural development, regulating the establishment of proper connectivity within the nervous system. Promotes axon guidance and homophilic cell adhesion. Plays a role in the development of the visual pathway. It seems to be involved in the differentiation of the fibroblast-like cells in the superficial layer of mandibular condylar cartilage into chondrocytes.

Associated disease(s): Defects in this gene are associated with Matthew-Wood syndrome and coloboma.

Associated syndrome(s): Matthew-Wood syndrome.

WWC2
WW And C2 Domain Containing Protein 2

Overlapping region: **chr4:183698508-184167427** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 5 genes

Function(s): Its functions are not known.

Associated disease(s): Diseases associated with this gene include acute lymphocytic leukemia and leukemia.

Associated syndrome(s): nothing

WWC2-AS2
WWC2 antisense RNA 2

Overlapping region: **chr4:183698508-184167427** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 5 genes

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

Associated disease(s): nothing

Associated syndrome(s): nothing

GRAMD3
GRAM Domain Containing Protein 3

Overlapping region: **chr5:125133096-125794639** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 3 genes

Function(s): Its functions are not known.

Associated disease(s): Diseases associated with GRAMD3 include hepatitis C and hepatitis C virus.

Associated syndrome(s): nothing

LOC101927488
ncRNA (uncharacterized)

Overlapping region: **chr5:125133096-125794639** (DEL)
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

LOC102546228

ncRNA (uncharacterized)

Overlapping region: **chr5:125133096-125794639** (DEL)
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

FBN2

Fibrillin 2

Overlapping region: **chr5:127873679-128220098** (DEL)
Num. of overlaps for that region: 4
How many other genes present: 1 gene

Function(s): The protein encoded by this gene is a component of connective tissue microfibrils and it seems to be involved in elastic fiber assembly. It is related to extracellular matrix structural constituents and calcium ion binding.

Associated disease(s): Defects in this genes are related to congenital contractural arachnodactyly, and congenital contractures.

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

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

CSNK1A1P1

Casein Kinase 1, Alpha 1 Pseudogene 1

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

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

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

Associated syndrome(s): nothing

LOC145845

ncRNA (uncharacterized)

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

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

Associated disease(s): nothing

Associated syndrome(s): nothing

MEIS2

Myeloid Ecotropic Viral Integration Site 1 Homolog 2

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

Function(s): This gene encodes a homeobox protein belonging to the TALE ('three amino acid loop extension') family of homeodomain-containing proteins. TALE homeobox proteins are highly conserved transcription regulators, and several members have been shown to be essential contributors to developmental programs.

Associated disease(s): Defects in this gene are associated with *attention deficit hyperactivity disorder* and *acute myeloid leukemia*.

Associated syndrome(s): nothing

NOTE: This gene is already known to be related with isolated CP.

C15orf53

Chromosome 15 Open Reading Frame 53 (uncharacterized)

Overlapping region: **chr15:38781772-39743689** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 3 genes

Function(s): Its functions are not characterized.

Associated disease(s): Defects in this genes are associated with major depressive disorder and paraplegia.

Associated syndrome(s): nothing

C15orf54

Chromosome 15 Open Reading Frame 53 (uncharacterized)

Overlapping region: **chr15:38781772-39743689** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 3 genes

Function(s): Its functions are not characterized.

Associated disease(s): nothing

Associated syndrome(s): nothing

RASGRP1

RAS Guanyl Releasing Protein 1 (Calcium And DAG-Regulated)

Overlapping region: **chr15:38781772-39743689** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 3 genes

Function(s): It functions as a diacylglycerol (DAG)-regulated nucleotide exchange factor specifically activating Ras through the exchange of bound GDP for GTP. It activates the Erk/MAP kinase cascade (calcium ion binding) and regulates T-cells and B-cells development, homeostasis and differentiation.

Associated disease(s): Defects in this gene are associated with leukocyte adhesion deficiency and systemic lupus erythematosus.

Associated syndrome(s): nothing

THOC1

THO Complex 1

Overlapping region: **chr18:140336-220071** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 2 genes

Function(s): Required for efficient export of polyadenylated RNA. Acts as component of the THO subcomplex of the TREX complex which is thought to couple mRNA transcription, processing and nuclear export, and which specifically associates with spliced mRNA and not with unspliced pre-mRNA. Its functions are not known. Regulates transcriptional elongation of a subset of genes. Involved in genome stability by preventing co-transcriptional R-loop formation.

Associated disease(s): Defects in this gene are associated with major depressive disorder and Kaposi's sarcoma.

Associated syndrome(s): nothing

USP14

Ubiquitin Specific Peptidase 14 (TRNA-Guanine Transglycosylase)

Overlapping region: **chr18:140336-220071** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 2 genes

Function(s): The encoded protein is located in the cytoplasm and cleaves the ubiquitin moiety from ubiquitin-fused precursors and ubiquitinylated proteins. It is involved in ubiquitin-specific protease activity and ubiquitin thiolesterase activity. Serves also as a physiological inhibitor of endoplasmic reticulum-associated degradation (ERAD) under the non-stressed condition by inhibiting the degradation of unfolded endoplasmic reticulum proteins via interaction with ERN1. Indispensable for synaptic development and function at neuromuscular junctions (NMJs).

Associated disease(s): Defects in this gene are associated with tremor and serous cystadenocarcinoma.

Associated syndrome(s): nothing

CD28

T-Cell-Specific Surface Glycoprotein CD28

Overlapping region: **chr2:204430764-205639051** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): Its protein encoded by this gene is essential for T-cell proliferation and survival, cytokine production, and T-helper type-2 development.

Associated disease(s): Defects in this gene are associated with tremor and serous cystadenocarcinoma.

Associated syndrome(s): nothing

CTLA4

Cytotoxic T-Lymphocyte-Associated Protein 4

Overlapping region: **chr2:204430764-205639051** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): This gene is a member of the immunoglobulin superfamily and encodes a protein which transmits an inhibitory signal to T cells. It is an inhibitory receptor acting as a major negative regulator of T-cell responses. The affinity of CTLA4 for its natural B7 family ligands, CD80 and CD86, is considerably stronger than the affinity of their cognate stimulatory co-receptor CD28.

Associated disease(s): Diseases associated with CTLA4 include Graves' disease (type 4).

Associated syndrome(s): nothing

ICOS

Inducible T-Cell Co-Stimulator

Overlapping region: **chr2:204430764-205639051** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): The protein encoded by this gene belongs to the CD28 and CTLA-4 cell-surface receptor family. It forms homodimers and plays an important role in cell-cell signaling, immune responses, and regulation of cell proliferation.

Associated disease(s): Defect of this gene are associated with icos-related common variable immune deficiency, and autoimmune myocarditis.

Associated syndrome(s): nothing

PARD3B

Par-3 Family Cell Polarity Regulator Beta

Overlapping region: **chr2:204430764-205639051** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): Putative adapter protein involved in asymmetrical cell division and cell polarization processes. It may play a role in the formation of epithelial tight junctions.

Associated disease(s): Diseases associated with amyotrophic lateral sclerosis 2 (juvenile) and lateral sclerosis.

Associated syndrome(s): nothing

CRYBB2P1

Crystallin, Beta B2 Pseudogene 1

Overlapping region: **chr22:25719287-25911071** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 3 genes

Function(s): It is a pseudogene.

Associated disease(s): Diseases associated with cerulean cataract and neurofibromatosis.

Associated syndrome(s): nothing

LRP5L

Low Density Lipoprotein Receptor-Related Protein 5-Like

Overlapping region: **chr22:25719287-25911071** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 3 genes

Function(s): The functions of its encoded protein include WNT-activated receptor activity and WNT-protein binding.

Associated disease(s): nothing

Associated syndrome(s): nothing

MIR6817

microRNA 6817 [Homo sapiens (human)]

Overlapping region: **chr22:25719287-25911071** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 3 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

MIR5739

microRNA 5739 [Homo sapiens (human)]

Overlapping region: **chr22:28675022-28885620** (DEL)
Num. of overlaps for that region: 3
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

TTC28

Tetratricopeptide Repeat Domain 28

Overlapping region: **chr22:28675022-28885620** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 2 genes

Function(s): During mitosis, it may be involved in the condensation of spindle midzone microtubules, leading to the formation of midbody.

Associated disease(s): Diseases associated with this gene include obesity and malaria.

Associated syndrome(s): nothing

FGF2

Fibroblast Growth Factor 2 (Basic)

Overlapping region: **chr4:123668544-124976381** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 5 genes

Function(s): This protein has been implicated in diverse biological processes, such as limb and nervous system development, wound healing, and tumor growth. FGF family members bind heparin and cytokines and possess broad mitogenic and angiogenic activities.

Associated disease(s): Diseases associated with this gene include intravascular papillary endothelial hyperplasia and chronic tympanitis.

Associated syndrome(s): nothing

LINC01091

Long Intergenic Non-Protein Coding RNA 1091

Overlapping region: **chr4:123668544-124976381** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 5 genes

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

Associated disease(s): nothing

Associated syndrome(s): nothing

NUDT6

Nudix (Nucleoside Diphosphate Linked Moiety X)-Type Motif 6

Overlapping region: **chr4:123668544-124976381** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 5 genes

Function(s): This gene overlaps and lies on the opposite strand from FGF2 gene, and is thought to be the FGF2 antisense gene. The two genes are independently transcribed, and their expression shows an inverse relationship, suggesting that this antisense transcript may regulate FGF2 expression. This gene has also been shown to have hormone-regulatory and antiproliferative actions in the pituitary that are independent of FGF2 expression. It is involved hydrolase activity and growth factor activity. It may contribute to the regulation of cell proliferation.

Associated disease(s): Diseases associated with NUDT6 include gingival overgrowth and bone fracture.

Associated syndrome(s): nothing

SPATA5

Spermatogenesis Associated 5

Overlapping region: **chr4:123668544-124976381** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 5 genes

Function(s): It may be involved in morphological and functional mitochondrial transformations during spermatogenesis (by similarity). It seems to be involved in nucleoside-triphosphatase activity.

Associated disease(s): Diseases associated with SPATA5 include transient cerebral ischemia and alopecia areata.

Associated syndrome(s): nothing

SPRY1

Sprouty Homolog 1, Antagonist Of FGF Signaling (Drosophila)

Overlapping region: **chr4:123668544-124976381** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 5 genes

Function(s): It may be involved in morphological and functional mitochondrial transformations during spermatogenesis (by similarity). It seems to be involved in nucleoside-triphosphatase activity.

Associated disease(s): Diseases associated with legius syndrome and osteoporosis.

Associated syndrome(s): legius syndrome.

ANKRD50

Ankyrin Repeat Domain 50

Overlapping region: **chr4:125106907-126864511** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): Its functions are not characterized.

Associated disease(s): nothing

Associated syndrome(s): nothing

FAT4

FAT Atypical Cadherin 4

Overlapping region: **chr4:125106907-126864511** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): The encoded protein is a protocadherin and it may play a role in regulating planar cell polarity (PCP).

Associated disease(s): Defects in this gene are associated with Van Maldergem Syndrome type 1 and type 2.

Associated syndrome(s): Van Maldergem Syndrome type 1 and type 2.

LOC101927087

ncRNA (uncharacterized)

Overlapping region: **chr4:125106907-126864511** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

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

Associated disease(s): nothing

Associated syndrome(s): nothing

MIR2054
microRNA 2054 [Homo sapiens (human)]

Overlapping region: **chr4:125106907-126864511** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 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

AGA
Aspartylglucosaminidase

Overlapping region: **chr4:178195181-180933534** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): Aspartylglucosaminidase is involved in the catabolism of N-linked oligosaccharides of glycoproteins. It cleaves asparagine from N-acetylglucosamines as one of the final steps in the lysosomal breakdown of glycoproteins.

Associated disease(s): Defects in this gene are associated with aspartylglucosaminuria and lysosomal storage disease.

Associated syndrome(s): nothing

LINC01098
Long Intergenic Non-Protein Coding RNA 1098

Overlapping region: **chr4:178195181-180933534** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

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

Associated disease(s): nothing

Associated syndrome(s): nothing

LINC01099
Long Intergenic Non-Protein Coding RNA 1099

Overlapping region: **chr4:178195181-180933534** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

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

Associated disease(s): nothing

Associated syndrome(s): nothing

NEIL3

Nei Endonuclease VIII-Like 3 (E. Coli)

Overlapping region: **chr4:178195181-180933534** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): It belongs to a class of DNA glycosylases homologous to the bacterial Fpg/Nei family. These glycosylases initiate the first step in base excision repair by cleaving bases damaged by reactive oxygen species and introducing a DNA strand break via the associated lyase reaction (damaged DNA binding and double-stranded DNA binding.).

Associated disease(s): Diseases associated with this gene include meningioma and adenoma.

Associated syndrome(s): nothing

LINC00290

Long Intergenic Non-Protein Coding RNA 290

Overlapping region: **chr4:181005159-183698508** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

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

Associated disease(s): nothing

Associated syndrome(s): nothing

MGC45800

Uncharacterized LOC90768

Overlapping region: **chr4:181005159-183698508** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

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

Associated disease(s): Diseases associated with this gene include pre-eclampsia and eclampsia.

Associated syndrome(s): nothing

MIR1305

microRNA 1305 [Homo sapiens (human)]

Overlapping region: **chr4:181005159-183698508** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 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

TENM3

Teneurin Transmembrane Protein 3

Overlapping region: **chr4:181005159-183698508** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): Involved in neural development, regulating the establishment of proper connectivity within the nervous system. Promotes axon guidance and homophilic cell adhesion. Plays a role in the development of the visual pathway. It seems to be involved in the differentiation of the fibroblast-like cells in the superficial layer of mandibular condylar cartilage into chondrocytes.

Associated disease(s): Defects in this gene are associated with Matthew-Wood syndrome and coloboma.

Associated syndrome(s): Matthew-Wood syndrome.

LETM1

Leucine Zipper-EF-Hand Containing Transmembrane Protein 1

Overlapping region: **chr4:1837887-1985244** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): The encoded protein functions to maintain the mitochondrial tubular shapes and is required for normal mitochondrial morphology and cellular viability. It is crucial for the maintenance of mitochondrial tubular networks and for the assembly of the supercomplexes of the respiratory chain.

Associated disease(s): Diseases associated with this gene include Wolf-Hirschhorn syndrome and malaria.

Associated syndrome(s): Wolf-Hirschhorn syndrome

SCARNA22

Small Cajal Body-Specific RNA 22

Overlapping region: **chr4:1837887-1985244** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

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

Associated disease(s): Diseases associated with this gene include multiple myeloma and myeloma.

Associated syndrome(s): nothing

WHSC1

Wolf-Hirschhorn Syndrome Candidate 1

Overlapping region: **chr4:1837887-1985244** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): It is expressed ubiquitously in early development. The encoded protein functions include chromatin binding and histone-lysine N-methyltransferase activity.

Associated disease(s): Diseases associated with this gene include myeloma, and Wolf-Hirschhorn syndrome.

Associated syndrome(s): Wolf-Hirschhorn syndrome.

NELFA

Negative Elongation Factor Complex Member A

Overlapping region: **chr4:1837887-1985244** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 4 genes

Function(s): This gene is expressed ubiquitously with higher levels in fetal than in adult tissues. The encoded protein is found to be capable of reacting with HLA-A2-restricted and tumor-specific cytotoxic T lymphocytes, suggesting a target for use in specific immunotherapy for a large number of cancer patients. This protein has also been shown to be a member of the NELF (negative elongation factor) protein complex that participates in the regulation of RNA polymerase II transcription elongation.

Associated disease(s): Diseases associated with this gene include Wolf-Hirschhorn syndrome and hiv-1.

Associated syndrome(s): Wolf-Hirschhorn syndrome.

NELFA

Negative Elongation Factor Complex Member A

Overlapping region: **chr4:1988629-2009787** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 1 gene

Function(s): This gene is expressed ubiquitously with higher levels in fetal than in adult tissues. The encoded protein is found to be capable of reacting with HLA-A2-restricted and tumor-specific cytotoxic T lymphocytes, suggesting a target for use in specific immunotherapy for a large number of cancer patients. This protein has also been shown to be a member of the NELF (negative elongation factor) protein complex that participates in the regulation of RNA polymerase II transcription elongation.

Associated disease(s): Diseases associated with this gene include Wolf-Hirschhorn syndrome and hiv-1.

Associated syndrome(s): Wolf-Hirschhorn syndrome.

COL26A1

Collagen, Type XXVI, Alpha 1

Overlapping region: **chr7:101019060-101363597** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 3 genes

Function(s): This gene encodes a protein containing an emilin domain and two collagen stretches. This gene may be associated with aspirin-intolerant asthma.

Associated disease(s): Diseases associated with this gene include asthma and ataxia.

Associated syndrome(s): nothing

LINC01007

Long Intergenic Non-Protein Coding RNA 1007

Overlapping region: **chr7:101019060-101363597** (DEL)
Num. of overlaps for that region: 3
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

MYL10
Myosin, Light Chain 10, Regulatory

Overlapping region: **chr7:101019060-101363597** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 3 genes

Function(s): The encoded protein is involved in calcium ion binding and in regulatory activity.

Associated disease(s): Diseases associated with this gene include hiv-1 and endotheliitis.

Associated syndrome(s): nothing

CYTH3
Myosin, Light Chain 10, Regulatory

Overlapping region: **chr7:6146033-6296948** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 2 genes

Function(s): The members of PSDC family appear to mediate the regulation of protein sorting and membrane trafficking. This encoded protein is involved in the control of Golgi structure and function, and it may have a physiological role in regulating ADP-ribosylation factor protein 6 (ARF) functions, in addition to acting on ARF1.

Associated disease(s): Diseases associated with this gene include thyroiditis and hepatitis.

Associated syndrome(s): nothing

USP42
Ubiquitin Specific Peptidase 42

Overlapping region: **chr7:6146033-6296948** (DEL)
Num. of overlaps for that region: 3
How many other genes present: 2 genes

Function(s): Its encoded protein seems to deubiquitinating enzyme which may play an important role during spermatogenesis (by similarity).

Associated disease(s): Diseases associated with this gene include myelodysplastic syndromes.

Associated syndrome(s): myelodysplastic syndromes.

NPHP4
Nephronophthisis 4

Overlapping region: **chr1:5967499-6023558** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): This gene encodes a protein involved in renal tubular development and function (structural molecule activity). This protein interacts with nephrocystin, and belongs to a multifunctional complex that is localized to actin- and microtubule-based structures.

Associated disease(s): Diseases associated with this gene include nephronophthisis 4 and Senior-Loken syndrome 4.

Associated syndrome(s): Senior-Loken syndrome

ZMYND11

Zinc Finger, MYND-Type Containing 11

Overlapping region: **chr10:158945-313504** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): The nuclear protein encoded by this gene was first identified by its ability to bind the adenovirus E1A protein. It functions as a transcriptional repressor and expression of E1A inhibits this repression. It may be involved in chromatin remodeling through association with several remodeling factors.

Associated disease(s): Diseases associated with this gene include fibrodysplasia ossificans progressiva, and ovarian cancer.

Associated syndrome(s): nothing

LINC00639

Long Intergenic Non-Protein Coding RNA 639

Overlapping region: **chr14:39117362-39310670** (DEL)
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): nothing

Associated syndrome(s): nothing

FSIP1

Fibrous Sheath Interacting Protein 1

Overlapping region: **chr15:39743689-40012522** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): Its protein functions are not characterized.

Associated disease(s): Diseases associated with this gene include intellectual disability and asthma.

Associated syndrome(s): nothing

THBS1

Thrombospondin 1

Overlapping region: **chr15:39743689-40012522** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): The encoded protein is an adhesive glycoprotein that mediates cell-to-cell and cell-to-matrix interactions. This protein can bind to fibrinogen, fibronectin, laminin, type V collagen and integrins alpha-V/beta-1. This protein has been shown to play roles in platelet aggregation, angiogenesis, and tumorigenesis.

Associated disease(s): Diseases associated with this gene include hereditary leiomyomatosis, renal cell cancer and thrombotic thrombocytopenic purpura.

Associated syndrome(s): nothing

CRHR1

Corticotropin Releasing Hormone Receptor 1

Overlapping region: **chr17:43717703-43739782** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): This gene encodes a G-protein coupled receptor that binds neuropeptides of the corticotropin releasing hormone family that are major regulators of the hypothalamic-pituitary-adrenal pathway. The encoded protein is essential for the activation of signal transduction pathways that regulate diverse physiological processes including stress, reproduction, immune response and obesity.

Associated disease(s): Diseases associated with this gene include 17q21.31 microdeletion syndrome and placenta accreta.

Associated syndrome(s): 17q21.31 microdeletion syndrome

CRHR1-IT1

CRHR1 Intronic Transcript 1 (non-protein coding)

Overlapping region: **chr17:43717703-43739782** (DEL)
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 non-coding RNA class.

Associated disease(s): nothing

Associated syndrome(s): nothing

KANSL1

KAT8 Regulatory NSL Complex Subunit 1

Overlapping region: **chr17:44159862-44208312** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): This gene encodes a nuclear protein that is a subunit of two protein complexes involved with histone acetylation, the MLL1 complex and the NSL1 complex. Its functions are involved in GO annotations related to this gene include histone acetyltransferase activity (H4-K16 specific)and histone acetyltransferase activity (H4-K5 specific).

Associated disease(s): Diseases associated with this gene include Koolen-de Vries syndrome and KANSL1-related intellectual disability syndrome.

Associated syndrome(s): Koolen-de Vries syndrome and KANSL1-related intellectual disability syndrome.

CDH7

Cadherin 7, Type 2

Overlapping region: **chr18:62395473-63635289** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): The encoded membrane protein is a calcium dependent cell-cell adhesion glycoprotein. Cadherins mediate cell-cell binding in a homophilic manner, contributing to the sorting of heterogeneous cell types and the maintenance of orderly structures.

Associated disease(s): Diseases associated with this gene include Charge syndrome and mammary paget's disease.

Associated syndrome(s): Charge syndrome.

PARD3B

Par-3 Family Cell Polarity Regulator Beta

Overlapping region: **chr2:205639051-205659589** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): Putative adapter protein involved in asymmetrical cell division and cell polarization processes. It may play a role in the formation of epithelial tight junctions.

Associated disease(s): Diseases associated with amyotrophic lateral sclerosis 2 (juvenile) and lateral sclerosis.

Associated syndrome(s): nothing

IGLL3P

Immunoglobulin Lambda-Like Polypeptide 3, Pseudogene

Overlapping region: **chr22:25689977-25719287** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

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

Associated disease(s): nothing

Associated syndrome(s): nothing

TTC28

Tetratricopeptide Repeat Domain 28

Overlapping region: **chr22:28885620-28959281** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): During mitosis, it may be involved in the condensation of spindle midzone microtubules, leading to the formation of midbody.

Associated disease(s): Diseases associated with this gene include obesity and malaria.

Associated syndrome(s): nothing

SYNPO2

Synaptopodin 2

Overlapping region: **chr4:119910786-119919459** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 1 gene

Function(s): its encoded protein fuctions involve muscle alpha-actinin binding and actin binding.

Associated disease(s): Diseases associated with this gene include myofibrillar myopathy and Duchenne muscular dystrophy.

Associated syndrome(s): nothing

LINC01262

Long Intergenic Non-Protein Coding RNA 639

Overlapping region: **chr4:190431430-190721966** (DEL)
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): nothing

Associated syndrome(s): nothing

MIR943

microRNA 943 [Homo sapiens (human)]

Overlapping region: **chr4:1985244-1988629** (DEL)
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): Diseases associated with this gene include cervical cancer and cervicitis.

Associated syndrome(s): nothing

KPNA7

Karyopherin Alpha 7 (Importin Alpha 8)

Overlapping region: **chr7:98717972-98867726** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): The encoded protein is involved in protein transporter activity (nuclear protein import similarity).

Associated disease(s): Diseases associated with this gene include ulcerative colitis and influenza.

Associated syndrome(s): nothing

SMURF1

SMAD Specific E3 Ubiquitin Protein Ligase 1

Overlapping region: **chr7:98717972-98867726** (DEL)
Num. of overlaps for that region: 2
How many other genes present: 2 genes

Function(s): This gene encodes a ubiquitin ligase that is specific for receptor-regulated SMAD proteins in the bone morphogenetic protein (BMP) pathway. This protein plays a key roll in the regulation of cell motility, cell signalling, and cell polarity.

Associated disease(s): Diseases associated with this gene include cerebral cavernous malformations-2 and Wolfram syndrome.

Associated syndrome(s): Wolfram syndrome.