EXCLI Journal 2014;13:252-264 – ISSN 1611-2156
Received: January 29, 2014, accepted: February 19, 2014, published: March 13, 2014

Supplementary material to:

IMPRINTING GENES ASSOCIATED WITH ENDOMETRIOSIS

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Paternally expressed and maternally imprinted genes: These genes are imprinted, with preferential expression from the paternal allele.

NDUFA4P1, NADH dehydrogenase (ubiquinone) 1 alpha subcomplex, 4, 9kDa, pseudogene 1
NDUFA4 is a component of the cytochrome c oxidase (CcO), which is the terminal oxidase of the mitochondrial electron transport chain (http://www.ncbi.nlm.nih.gov/gene/360165) (Balsa et al., 2012; Srinivasan and Avadhani, 2012). CcO dysfunction or a reduced expression leads to mitochondrial reactive oxygen species production and cellular oxidative stress, and is affected in a wide variety of diseases including preeclampsia, neurodegenerative diseases, myocardial ischemia/reperfusion, bone and skeletal diseases, diabetes and cancer (He et al., 2004).

GFI1, growth factor independent 1 transcription repressor
GFI1 is a transcriptional repressor that plays a critical role in hematopoiesis and lymphomagenesis (http://www.ncbi.nlm.nih.gov/gene/2672) (Du et al., 2013). It is repressed by p53 and inhibits DNA damage-induced apoptosis (Du et al., 2013).

DIRAS3, DIRAS family, GTP-binding RAS-like 3
DIRAS3 is a member of the Ras superfamily, and appears to be a putative tumor suppressor gene (http://www.ncbi.nlm.nih.gov/gene/9077). Up-regulation of DIRAS3 expression is associated with infertility and endometriosis (Li et al., 2011).

FUCA1, fucosidase, alpha-L- 1, tissue
This protein is a lysosomal enzyme involved in the degradation of fucose-containing glycoproteins and glycolipids (http://www.ncbi.nlm.nih.gov/gene/2517). The alpha-fucosidase activity was increased in patients with infertility (Brandelli and Passos, 1998).

BMP8B, bone morphogenetic protein 8b
BMP is a part of the transforming growth factor-beta (TGFB) superfamily that induces ectopic bone growth (http://www.ncbi.nlm.nih.gov/gene/656). Progesterone-dependent Ihh, Wnt, and BMP signaling pathways within the endometrium play a role in decidualization, implantation and embryo attachment (Wetendorf and DeMayo, 2012).

RPL22, ribosomal protein L22
This protein is a component of the 60S subunit and acts as a cytoplasmic ribosomal protein (http://www.ncbi.nlm.nih.gov/gene/6146).

PRDM16, PR domain containing 16
PRDM16, PPARγ and C/EBPα are master regulators of adipocyte development and regulate the process of adipogenesis that include adiponectin secretion and insulin-dependent glucose uptake (http://www.ncbi.nlm.nih.gov/gene/63976) (Kajimura et al., 2010; Farmer, 2008).

OBSCN, obscurin, cytoskeletal calmodulin and titin-interacting RhoGEF
OBSCN has a role in the organization of myofibrils and is essential for normal muscle function (http://www.ncbi.nlm.nih.gov/gene/84033).

OR11L1, olfactory receptor, family 11, subfamily L, member 1
The olfactory receptor proteins are members of a large family of G-protein-coupled receptors (GPCR) (http://www.ncbi.nlm.nih.gov/gene/391189).

LRRTM1, leucine rich repeat transmembrane neuronal 1
This protein is predominantly expressed in thalamus, hippocampus and limbic cortex and functions as a synaptic cell adhesion molecule that influences synaptic excitation (http://www.ncbi.nlm.nih.gov/gene/347730). Lack of LRRTM1 presumably causes the abnormality in behavior (Soler-Llavina et al., 2013).

CCDC85A, coiled-coil domain containing 85A
CCDC85A expression is association with age at menarche, a landmark of the late stage of pubertal development, in the Japanese population. Multiple factors and the interactions between genetic and environmental factors influence the age at which menarche occurs (http://www.ncbi.nlm.nih.gov/gene/114800).

CYP1B1, cytochrome P450, family 1, subfamily B, polypeptide 1
The cytochrome P450 metabolizes procarcinogens and synthesizes cholesterol, steroids and other lipids (http://www.ncbi.nlm.nih.gov/gene/1545). This enzyme is involved in eye development. The gene polymorphisms of CYP1B1 in exon 2 codon 119 are also an associated risk factor for endometriosis (Li and Wang 2009).

GPR1, G protein-coupled receptor 1
GPR1 is regulated in the process of the development of a chronic inflammatory disease such as atherosclerosis (http://www.ncbi.nlm.nih.gov/gene/2825) (Karagiannis et al., 2013).

TIGD1, tigger transposable element derived 1
This protein belongs to the superfamily of DNA-mediated transposons in humans and is similar to the major mammalian centromere protein B (CENP-B) (http://www.ncbi.nlm.nih.gov/gene/200765). CENP-B is a centromeric satellite DNA-binding protein and functions in the assembly of centromeric nucleosomes (Tachiwana et al., 2013).

MYEOV2, myeloma overexpressed 2
MYEOV2 expression is associated with the development of multiple myeloma (http://www.ncbi.nlm.nih.gov/gene/150678) (Ebina et al., 2013).

HES1, hairy and enhancer of split 1
This protein belongs to the transcriptional repressor gene family that requires a basic helix-loop-helix protein for their transcription (http://www.ncbi.nlm.nih.gov/gene/3280). The HES1 gene regulatory networks play a central role in the timing of somitogenesis and can become deregulated in human cancer (Sturrock et al., 2013).

SPON2, spondin 2, extracellular matrix protein
SPON2 is known as mindin. SPON2 is an extracellular matrix protein and can modify the production of proinflammatory cytokines. Aberrant DNA methylation of SPON2 gene was observed in prostate cancer (Kim et al., 2012).

NAP1L5, nucleosome assembly protein 1-like 5
Nucleosome assembly protein 1 (NAP-1) is an integral component associated with the establishment, maintenance, and dynamics of eukaryotic chromatin and its modification states (Park and Luger, 2006). NAP1L5 plays a role for transcriptional interference (Park and Luger, 2006).

GAB1, GRB2-associated binding protein 1
Members of the GAB family are expressed in T cells, B cells, macrophages and mast cells. GAB acts as a downstream effector of receptor-tyrosine kinase-triggered signal transduction and plays a central role in growth, transformation and apoptosis (http://www.ncbi.nlm.nih.gov/gene/2549). GAB1 is also a mediator of branching renal tubulogenesis.

CDH18, cadherin 18, type 2
CDH18 is expressed in the central nervous system and is involved in calcium-dependent synaptic adhesion, axon outgrowth, guidance, neural circuitry and mature synaptic function (http://www.ncbi.nlm.nih.gov/gene/1016). CDH18 is of special interest in the pathogenesis of neuropsychiatric disease (Redies et al., 2012).

FAM50B, family with sequence similarity 50, member B
FAM50B plays a role in spermatogenesis (Zhang et al., 2011).

MRAP2, melanocortin 2 receptor accessory protein 2
MRAP2 modulates energy balance, glucose homeostasis and the development of obesity (Liu et al., 2013a).

LIN28B, lin-28 homolog B
This gene is highly expressed in fetal liver, testis and placenta (http://www.ncbi.nlm.nih.gov/gene/389421). Pubertal timing is under strong genetic control (Leinonen et al., 2012). LIN28B is the puberty-associated gene and influences adult adiposity-related traits, including obesity, type 2 diabetes and cardiovascular disease (Leinonen et al., 2012).

PLAGL1, pleiomorphic adenoma gene-like 1
This protein is a zinc finger protein with DNA-binding activities (http://www.ncbi.nlm.nih.gov/gene/5325). It functions as a tumor suppressor. Overexpression of this gene during fetal development is linked to developmental disorders such as transient neonatal diabetes mellitus, which is a rare disease characterized by intrauterine growth retar-

dation, dehydration, and failure to thrive due to a lack of normal insulin secretion (Abdollahi, 2007). Transient neonatal diabetes mellitus type 1 frequently recurs in later life

HYMAI, hydatidiform mole associated and imprinted
This gene encodes a non-protein coding transcript (http://www.ncbi.nlm.nih.gov/gene/57061). HYMAI is one of the causative genes for transient neonatal diabetes mellitus (Mackay and Temple, 2010). Diabetes frequently recurs in later life. It is caused by overexpression of the imprinted genes PLAGL1 and HYMAI on human chromosome 6q24 (Mackay and Temple, 2010).

PEG10, paternally expressed 10
Four imprinted genes, two paternally expressed (IGF2 and PEG10) and two maternally expressed (PHLDA2 and CDKN1C), play a role in fetal growth and placental development (Dória et al., 2010). Gene expression pattern of IGF2, PHLDA2, PEG10 and CDKN1C imprinted genes is associated with pregnancy loss such as spontaneous miscarriages or fetal deaths (Dória et al., 2010). Overexpression of the PEG10 gene is also associated with hepatocellular carcinomas (http://www.ncbi.nlm.nih.gov/gene/23089).

SGCE, sarcoglycan, epsilon
Sarcoglycans are components of the dystrophin-glycoprotein complex, which link the actin cytoskeleton to the extracellular matrix (http://www.ncbi.nlm.nih.gov/gene/8910). Myoclonus-dystonia is a common clinical sign in many neurogenetic disorders. Mutations in the SGCE gene and its variant (c.1295G > A, p.Ser432His) may have contributed to the development of dystonia (Kurtis et al., 2010).

MEST, mesoderm specific transcript
The encoded protein plays a role in development (http://www.ncbi.nlm.nih.gov/gene/4232). Decreased MEST methylation was observed in gestational diabetes mellitus groups or in adults with morbid obesity compared with controls (El Hajj et al., 2013). The loss of imprinting of MEST gene has been linked to certain types of cancer.

COPG2IT1, COPG2 imprinted transcript 1
COPG2IT1 is a transcript located within intron of COPG2 that is located telomeric to MEST at human chromosome 7q32 in fetal tissues. Embryonic stem cells express COPG2IT1 gene.

MESTIT1, MEST intronic transcript 1, antisense RNA
This gene encodes a non-protein coding antisense RNA and is located in an intron of MEST gene (http://www.ncbi.nlm.nih.gov/gene/317751).

DLGAP2, discs, large (Drosophila) homolog-associated protein 2
The product of this gene is a membrane-associated guanylate kinase and plays a role in synapse organization and signalling in neuronal cells (http://www.ncbi.nlm.nih.gov/gene/9228). DLGAP2 was also associated with eating behavior and obesity (Do et al., 2013).

ZFAT, zinc finger and AT hook domain containing
ZFAT-AS1, ZFAT antisense RNA 1
The encoded protein, ZFAT, is a DNA binding protein and functions as a transcriptional regulator involved in cell survival and apoptosis (http://www.ncbi.nlm.nih.gov/gene/57623). SNPs of the ZFAT gene are associated with an increased risk of autoimmune thyroid disease. ZFAT

expressed in syncytiotrophoblasts is downregulated in placentas from preeclampsia (Barbaux et al., 2012). The ZFAT-AS1 gene encodes a small antisense RNA that regulates the sense strand locus, ZFAT (http://www.ncbi.nlm.nih.gov/gene/594840).

GLIS3, GLIS family zinc finger 3
This gene encodes a transcriptional nuclear protein with zinc finger domains (http://www.ncbi.nlm.nih.gov/gene/169792). GLIS3 regulates the development of pancreatic beta cells, thyroid, eye, liver and kidney. Mutations in this gene are well known to cause neonatal diabetes and congenital hypothyroidism. GLIS3 is a candidate gene for type 1 diabetes (Santin and Eizirik, 2013).

INPP5F, inositol polyphosphate-5-phosphatase F
The encoded protein is an inositol 1,4,5-trisphosphate (InsP3) 5-phosphatase (http://www.ncbi.nlm.nih.gov/gene/22876). INPP5F modulates cardiac myocyte size and cardiac hypertrophy (Zhu et al., 2009).

KCNQ1OT1, KCNQ1 opposite strand/antisense transcript 1
The KCNQ1OT1 transcript is the antisense to the KCNQ1 gene and is a long non-coding RNA (http://www.ncbi.nlm.nih.gov/gene/10984). It interacts with chromatin and regulates transcription of multiple target genes through epigenetic modifications. KCNQ1OT1 plays an important role in patients with Beckwith-Wiedemann syndrome and in colorectal carcinogenesis. Beckwith-Wiedemann syndrome is a loss-of-imprinting syndrome characterized by macrosomia, macroglossia, and abdominal wall defects, which is referred to as genetic overgrowth disorder or large offspring syndrome.

INS, insulin
Insulin (INS) stimulates glucose uptake through the insulin receptor. The INS and insulin-like growth factor-2 (IGF-2) region on chromosome 11p15.5 has been associated with the metabolic syndrome, type 2 diabetes and coronary heart disease (Osada, 2009).

IGF2AS, IGF2 antisense RNA
The IGF2AS gene encodes an antisense transcript of the IGF2 gene (http://www.ncbi.nlm.nih.gov/gene/51214). The transcript is overexpressed in Wilms tumor. Both IGF2 and IGF2AS appear to be part of the same chromatin domain. IGF2 (G13790C and ApaI) and IGF2AS (A1364C and G11711T) are associated with muscle damage (Devaney et al., 2007).

IGF2, insulin-like growth factor 2 (somatomedin A)
The IGF2 gene is involved in cell proliferation, growth, migration, differentiation and survival. Epigenetic changes at this locus are associated with Wilms tumour, Beckwith-Wiedemann syndrome, rhabdomyosarcoma, Silver-Russell syndrome and cardiovascular disease (Bergman et al., 2013) (http://www.ncbi.nlm.nih.gov/gene/3481).

ZC3H12C, zinc finger CCCH-type containing 12C
ZC3H12C suppresses the inflammatory response in endothelial cells through inhibition of NF-kappaB activation and subsequent pro-inflammatory gene expression (Liu et al., 2013b).

DLK1, delta-like 1 homolog
The encoded protein is involved in the suppression of brown adipocyte differentiation (Hudak and Sul, 2013) (http://www.ncbi.nlm.nih.gov/gene/8788).

MAGEL2, MAGE-like 2
This gene lies in the Prader-Willi syndrome chromosomal region 15q11-q12 (http://www.ncbi.nlm.nih.gov/gene/54551). Disruption of this gene at this locus contributes to Prader-Willi syndrome. Prader-Willi Syndrome exhibits neonatal hypotonia, developmental delay, and childhood-onset obesity and is caused by the loss of function of several genes, including MAGEL2. Mice lacking Magel2 display obesity and hypothalamic deficiency (Mercer et al., 2013).

MKRN3, makorin ring finger protein 3
MKRN3 is localized to close proximity, 15q11-q13, to the Prader-Willi syndrome imprinting center on the paternal chromosome (Rodriguez-Jato et al., 2013) (http://www.ncbi.nlm.nih.gov/gene/7681).

SNORD64, small nucleolar RNA, C/D box 64
SNORD107, small nucleolar RNA, C/D box 107
SNORD108, small nucleolar RNA, C/D box 108
SNORD109A, small nucleolar RNA, C/D box 109A
SNORD109B, small nucleolar RNA, C/D box 109B
SNORD115-48, small nucleolar RNA, C/D box 115-48
SNORD115@, small nucleolar RNA, C/D box 115 cluster
SNORD116@, small nucleolar RNA, C/D box 116 cluster
SNORD116-1, also known as PWCR1, Prader-Willi syndrome chromosome region 1
Box C/D class small nucleolar RNAs (snoRNAs) are involved in site-specific methylation of preribosomal RNA precursors and RNA processing (http://www.ncbi.nlm.nih.gov/gene/?term=SNORD107). SNORDs are expressed predominantly in the brain (http://www.ncbi.nlm.nih.gov/gene/347686). The loss of SNORD115 or SNORD116 expression at the locus 15q11.2 has been implicated as a cause for the Prader-Willi syndrome (Zhang et al., 2012, Kishore et al., 2010). Therefore, loss of expression of the snoRNAs confers much or all of the phenotype of Prader-Willi syndrome (Gallagher et al., 2002).

SNRPN, small nuclear ribonucleoprotein polypeptide N
SNURF, SNRPN upstream reading frame
Alternative splicing or deletion in the region 15q11.2 is responsible for Angelman syndrome or Prader-Willi syndrome (Plagge, 2012) (http://www.ncbi.nlm.nih.gov/gene/6638, http://www.ncbi.nlm.nih.gov/gene/8926).

NDN, necdin, melanoma antigen (MAGE) family member
Loss of expression of this intronless gene, NDN, may result in the phenotype of Prader-Willi syndrome (http://www.ncbi.nlm.nih.gov/gene/4692).

ZIM2, zinc finger, imprinted 2
Human chromosome 19q13.4 contains several imprinted gene clusters including PEG3 (paternally expressed gene 3) and ZIM2 (http://www.ncbi.nlm.nih.gov/gene/23619). This transcript is expressed in the brain and testis.

MIMT1, MER1 repeat containing imprinted transcript 1
MIMT1 is a non-protein coding gene that forms part of the imprinted PEG3 (paternally expressed gene 3) domain. Loss of paternal MIMT1 expression results in the phenotype of late term abortion and stillbirth in cattle (Flisikowski et al., 2010).

PEG3, paternally expressed 3
Tuman PEG3 protein belongs to the zinc finger protein family (http://www.ncbi.nlm.nih.gov/gene/5178). The imprinted gene PEG3 plays a role in p53-mediated apoptosis and tumor suppressor activity. PEG3 has shown tumorigenesis in glioma and ovarian cells. PEG3 confers sexual behaviors, alters growth and development, and regulates apoptosis (Jiang et al., 2010).

MIR371A, microRNA 371a
MicroRNAs (miRNAs) recognize target mRNAs and result in translational inhibition of the target mRNA. There is no research linking MIR371A to human phenotype.

PSIMCT-1, malignant T cell amplified sequence 1 pseudogene
PSIMCT-1 is a pseudogene for MCT-1. The MCT-1 oncogene was identified as an amplified gene on chromosome Xq22-24 in a T-cell lymphoma and in a subset of diffuse large B-cell lymphoma (DLBCL), a common form of Non-Hodgkin's Lymphoma (NHL).

NNAT, neuronatin
This protein regulates ion channels during normal brain development (http://www.ncbi.nlm.nih.gov/gene/4826). NNAT plays a role in forming and maintaining the structure of the nervous system. Similar to IGF2, aberrant expression of NNAT gene results in the phenotype of a variety of embryonic neoplasms, including Wilms tumor (Hubertus et al., 2013).

MCTS2, malignant T cell amplified sequence 2
The retrogene MCTS2 is a functional gene resulting from retrotransposition of a mRNA molecule into the genome. There is no research linking MCTS2 to human phenotype.

L3MBTL, l(3)mbt-like 1
The encoded protein functions to regulate gene activity, via chromatin modification. L3MBTL is necessary for mitosis (http://www.ncbi.nlm.nih.gov/gene/26013). Deletion of L3MBTL is a characteristic of myeloid malignancies (Li et al., 2004).

GNASAS, GNAS antisense RNA 1, also known as SANG
Prenatal growth restriction is associated with different epigenetic changes that may lead to later health outcomes, including obesity and metabolic syndrome (Tobi et al., 2011). DNA methylation of GNASAS gene might be associated with small for gestational age and myocardial infarction among women (Tobi et al., 2011). Loss of imprinting of the GNASAS gene is also a cause of pseudohypoparathyroidism (http://www.ncbi.nlm.nih.gov/gene/149775).

MIR296, microRNA 296
MIR298, microRNAs 298
Several miRNAs including MIR296 are found to be dysregulated in placenta of preeclampsia patients (Choi et al., 2013). MIR296 is important for the pathogenesis of preeclampsia (Choi et al., 2013). MIR296 and MIR298 lie within the GNASAS transcription units (Robson et al., 2012).

Maternally expressed and paternally imprinted genes: These genes are imprinted, with preferential expression from the maternal allele.

DVL1, dishevelled segment polarity protein 1
Wnt stimulation induces recruitment of DVL to the G-protein coupled frizzled (FZD) receptors (Kawano et al., 2011). DVL plays a key role in relaying cellular information for several developmental pathways such as cell proliferation, migration, polarity, terminal differentiation, and the self-renewal of stem cells (Dillman et al., 2013). DVL1 encodes a cytoplasmic phosphoprotein and is a substrate of NR1I2 (nuclear receptor subfamily 1, group I, member 2), which is a family of serine/threonine kinases that have been associated with differentiation of epithelial and neuronal cells (http://www.ncbi.nlm.nih.gov/gene/1855) (Elbert et al., 2006). Charcot-Marie-Tooth disease has been mapped to the same region as DVL1. This disease is the hereditary neuropathy characterized by muscular atrophy and weakness in the distal parts of the legs (Ostern et al., 2013). Failures in Wnt signalling are a cause of infertility and endometriosis (Sonderegger et al., 2010).

TP73, tumor protein p73
TP53, TP63, and TP73 genes encode a member of the p53 family of transcription factors (http://www.ncbi.nlm.nih.gov/gene/7161). TP73 is involved in cellular responses to stress and development. Aberrant methylation of gene promoter regions including TP73 and cyclin-dependent kinase inhibitor 2B (CDKN2B) is important for inactivation of tumor suppressor genes in ovarian cancer (Ozdemir et al., 2012).

WRAP73, WD repeat containing, antisense to TP73
The WRAP73 gene encodes a member of the Trp-Asp (WD) motif repeat protein family (http://www.ncbi.nlm.nih.gov/gene/49856). WRAP73, a member of the WD family, is involved in a variety of cellular processes, including cell cycle progression, signal transduction, apoptosis, gene regulation and ossification.

PEX10, peroxisomal biogenesis factor 10
Peroxisomes have an important role in lipid metabolism and the homeostasis of reactive oxygen species (http://www.ncbi.nlm.nih.gov/gene/5192). PEX10 is involved in import of peroxisomal matrix proteins. Mutations in the PEX10 gene are closely associated with the Zellweger spectrum, ranging from neonatal adrenoleukodystrophy to a lethal neurometabolic disorder, Zellweger syndrome (Steinberg et al., 2009).

HSPA6, heat shock 70kDa protein 6
The heat shock proteins are chaperones with central roles in processes of polypeptide remodeling events. HSPA6 induction is associated with cellular stress (http://www.ncbi.nlm.nih.gov/gene/3310) (Wisniewska et al., 2010).

PTPN14, protein tyrosine phosphatase, non-receptor type 14
The protein encoded by the PTPN14 gene is a member of the protein tyrosine phosphatase (PTP) family (http://www.ncbi.nlm.nih.gov/gene/5784). PTPs are known to be signaling molecules that regulate cell growth, differentiation, mitotic cycle, and oncogenic transformation.

HIST3H2BB, histone cluster 3, H2bb
This gene encodes a member of the histone H2B family (http://www.ncbi.nlm.nih.gov/gene/128312). Hypoacetylation of the histone family within promoter regions of candidate genes (e.g., HOXA10, ESR1, CDH1, and p21 WAF1/Cip1) is downregulated in endometriosis (Monteiro et al., 2013).

VAX2, ventral anterior homeobox 2
This gene encodes a homeobox protein and is expressed in the retina during development (http://www.ncbi.nlm.nih.gov/gene/25806). The VAX2 gene was required for the patterning of the eye and the development of optic fissure and retina (Liu et al., 2008).

OTX1, orthodenticle homeobox 1
The OTX1 and OTX2 homeobox genes are crucial for the brain development (http://www.ncbi.nlm.nih.gov/gene/5013) (Larsen et al., 2010). Both genes are important in neuronal cell development and differentiation: OTX1 in the neocortex, and OTX2 in the archicortex, diencephalon, rostral brain stem, and cerebellum (Larsen et al., 2010).

ABCG8, ATP-binding cassette, sub-family G (WHITE), member 8
ABCG5 and ABCG8 form a complex (G5G8) and are expressed in liver to promote the excretion of cholesterol into bile (http://www.ncbi.nlm.nih.gov/gene/64241) (Li and Wang, 2009). G5G8 influences the development of diet-induced obesity phenotypes (Li and Wang, 2009).

ZFP36L2, ZFP36 ring finger protein-like 2
This nuclear transcription factor regulates the response to growth factors (http://www.ncbi.nlm.nih.gov/gene/678). Dysregulation of ZFP36L2 function may play a role in the pathophysiology of certain types of leukemia (Iwanaga et al., 2011).

ALDH1L1, aldehyde dehydrogenase 1 family, member L1
The encoded protein belongs to the aldehyde dehydrogenase family and is essential for cell growth regulation (http://www.ncbi.nlm.nih.gov/gene/10840). Loss of function or expression of the ALDH1L1 gene is associated with increased cell motility, decreased apoptosis and cancer progression.

ZIC1, Zic family member 1
The ZIC1 gene encodes a transcription factor with zinc finger domains that can transactivate the apolipoprotein E gene (http://www.ncbi.nlm.nih.gov/gene/7545). ZIC1 is linked to the process of neural development. Overexpression of ZIC1 results in inactivation of Shh (sonic hedgehog), PI3K and MAPK signaling pathways, which in turn has been implicated in the process of gastric cancer (Zhong et al., 2012).

FGFRL1, fibroblast growth factor receptor-like 1
FGFRL1 influences mitogenesis and differentiation (http://www.ncbi.nlm.nih.gov/gene/53834). The FGF2 754C/G polymorphism may be closely associated with a risk of developing endometriosis (Kang et al., 2010). This gene stimulates cell proliferation at the ectopic endometriotic site.

KIAA1530, also known as UVSSA, UV-stimulated scaffold protein A
The KIAA1530 gene is important for ubiquitination and dephosphorylation of RNA polymerase II subunits (http://www.ncbi.nlm.nih.gov/gene/57654). UVSSA regulates nucleotide excision repair activity and repairs DNA damage (Schwertman et al., 2013).

ADAMTS16, ADAM metallopeptidase with thrombospondin type 1 motif, 16
This gene encodes a member of the ADAMTS (a disintegrin and metalloproteinase with thrombospondin motifs) protein family (http://www.ncbi.nlm.nih.gov/gene/170690). ADAMTS16 acts as a downstream target of WT1 (Jacobi et al., 2013). This gene has been implicated in the process of branching morphogenesis of the kidneys and regulating blood pressure during urogenital development (Jacobi et al., 2013).

CSF2, colony stimulating factor 2 (granulocyte-macrophage)
This cytokine controls the differentiation and function of granulocytes and macrophages (http://www.ncbi.nlm.nih.gov/gene/1437). Tumor necrosis factor (TNF)-induced overexpression of CSF2 is associated with the etiology of asthma, likely due to stimulation of macrophages and neutrophils during inflammation (Burkhardt et al., 2012).

BTNL2, butyrophilin-like 2 (MHC class II associated)
BTNL2 may be closely associated with the process of two inflammatory diseases, myositis and sarcoidosis (Arnett et al., 2007). This gene down-regulates T cell activation and is implicated in resetting the balance of the immune response in the gut.

SLC22A2, solute carrier family 22 (organic cation transporter), member 2
SLC22A3, solute carrier family 22 (organic cation transporter), member 3
Transporters are essential for elimination of endogenous organic cations, environmental toxins and drugs (http://www.ncbi.nlm.nih.gov/gene/6582). SLC22A2 (expressed in kidney, lung and brain) and SLC22A3 (expressed in skeletal muscle, liver, placenta, kidney and heart) are downstream targets of IGF2R (Burger et al., 2010). They play a role in the detoxification (Burger et al., 2010). SLC22A2 could be a susceptibility gene for aspirin intolerance in asthmatics (Park et al., 2011).

MAGI2, membrane associated guanylate kinase, WW and PDZ domain containing 2
MAGI2 is responsible for recruitment of neurotransmitter receptors such as AMPA- and NMDA-type glutamate receptors (Koide et al., 2012). The MAGI2 protein interacts with atrophin-1 and is involved in dentatorubral and pallidoluysian atrophy (http://www.ncbi.nlm.nih.gov/gene/9863). Schizophrenia is a psychiatric disorder characterized by positive symptoms, negative symptoms, and cognitive impairment. MAGI2 SNPs may confer an excess risk of cognitive impairment in schizophrenic patients (Koide et al., 2012).

PPP1R9A, protein phosphatase 1, regulatory subunit 9A
PPP1R9A is maternally expressed in embryonic skeletal muscle and neuronal tissues (http://www.ncbi.nlm.nih.gov/gene/55607). This protein controls actin cytoskeleton reorganization (Nakabayashi et al., 2004). It is also a neural tissue specific F-actin-binding protein and functions as a key candidate molecule in synaptic formation and function in brain (Nakabayashi et al., 2004). Changes in expression of several genes including PPP1R9A were observed in patients with Huntington disease (Becanovic et al., 2010).

TFPI2, tissue factor pathway inhibitor 2
The TFPI2 protein is a member of the Kunitz-type serine proteinase inhibitor family (http://www.ncbi.nlm.nih.gov/gene/7980). This protein inhibits a variety of serine proteases including factor VIIa/tissue factor, factor Xa, plasmin, trypsin, chymotryspin and plasma kallikrein. This gene has been identified as a tumor suppressor gene in several types of cancer, including endometriosis-associated clear cell carcinoma (Arakawa et al., 2013).

DLX5, distal-less homeobox 5
The DLX5 gene encodes a member of a homeobox transcription factor gene family working in neurogenesis (http://www.ncbi.nlm.nih.gov/gene/1749). DLX5 is a candidate gene for some of the autistic spectrum disorder patients (Nakashima et al., 2010). The encoded protein also plays a role in bone development.

CPA, carboxypeptidase A4
The CPA gene is implicated in the histone hyperacetylation pathway (http://www.ncbi.nlm.nih.gov/gene/51200). CPA has a potential role in cell proliferation and differentiation and is a strong candidate gene for prostate cancer aggressiveness. Changes in CPA cluster expression on human chromosome 7q32-qter increase risk of intrauterine growth restriction associated with Silver-Russell syndrome (Bentley et al., 2003).

KLF14, Kruppel-like factor 14
This intronless gene encodes a member of the Kruppel-like family of transcription factors (http://www.ncbi.nlm.nih.gov/gene/136259). The KLF14 locus is associated with metabolic disease risk (Small et al., 2011). The type 2 diabetes locus on the chromosome 7q32.3 including KLF14 gene acts as a regulator of adipose gene expression (Small et al., 2011).

KCNK9, potassium channel, subfamily K, member 9
KCN functions as a pH-dependent potassium channel, with preferential expression in the brain (http://www.ncbi.nlm.nih.gov/gene/51305). Mutations in this gene were associated with Birk-Barel mental retardation dysmorphism syndrome characterized by mental retardation, hypotonia, and unique dysmorphism with elongated face (Barel et al., 2008). Overexpression of this gene has been observed in several types of human cancer.

KCNQ1DN, potassium channel, subfamily Q, member 1 downstream neighbor
The KCNQ1DN gene is located between CDKN1C (p57, KIP2) and KCNQ1 (KvLQT1) of chromosome 11p15.5 within the WT2 region (Xin et al., 2000). A maternal-specific loss of heterozygosity on the WT2-related chromosome 11p15.5 has been observed in Wilms and other embryonal tumors (Xin et al., 2000). Mouse model of Wilms tumor has been developed using Igf2 biallelic expression and Wt1 knockout. KCNQ1DN existing far from the H19/IGF2 region may play some role in Wilms tumorigenesis along with IGF2.

KCNQ1, potassium channel, subfamily Q, member 1
KCNQ1 within the 11p15.5 imprinted domain has been implicated in the process of congenital growth (Travers et al., 2013). KCNQ1, KCNQ1OT1 and CDKN1C mediate diabetes susceptibility at the KCNQ1 locus (Travers et al., 2013).

OSBPL5, oxysterol binding protein-like 5
The OSBPL5 gene encodes a member of the oxysterol-binding protein (OSBP) family, a group of intracellular lipid receptors that play a key role in the maintenance of cholesterol balance in the body (http://www.ncbi.nlm.nih.gov/gene/114879). A cluster of genes on chromosome 11 (SLC22A18, PHLDA2, NAP1L4, SNORA54, CARS, and OSBPL5) is an important foci involved in alcohol dependence (Edenberg et al., 2010).

SLC22A18, solute carrier family 22, member 18
Alterations or mutations in the SLC22A18 region have been implicated in the process of the Beckwith-Wiedemann syndrome, Wilms tumor, rhabdomyosarcoma, adrenocortical carcinoma, and lung, ovarian, and breast cancer. This protein acts as a transporter of organic cations (Lambertini et al., 2012). SLC22A18 demonstrated the positive correlation with the fetal head circumference (Lambertini et al., 2012).

PHLDA2, pleckstrin homology-like domain, family A, member 2
The PHLDA2 gene is located on chromosome 11p15.5, which harbors a cluster of imprinted genes and is considered to be an important tumor suppressor gene region (http://www.ncbi.nlm.nih.gov/gene/7262). Alterations in this region may be associated with the Beckwith-Wiedemann syndrome, Wilms tumor, rhabdomyosarcoma, adrenocortical carcinoma, and lung, ovarian, and breast cancer. Overexpression of the PHLDA2 gene has strong correlation with lower birth weight phenotype (Ishida et al., 2012).

CDKN1C, cyclin-dependent kinase inhibitor 1C (p57, Kip2)
The encoded protein is a strong G1 cyclin/Cdk-dependent inhibitor and a negative regulator of cell proliferation, suggesting a tumor suppressor candidate (http://www.ncbi.nlm.nih.gov/gene/1028). Mutations in this gene are implicated in sporadic cancers and Beckwith-Wiedemann syndorome. CDKN1C plays a role in endometrial stromal cell differentiation in the process of decidualization (Qian et al., 2005).

H19, imprinted maternally expressed transcript (non-protein coding)
The H19 gene is located in an imprinted region of chromosome 11 near the IGF2 gene (http://www.ncbi.nlm.nih.gov/gene/283120). The H19 gene expresses a non-coding RNA, and functions as a tumor suppressor. H19 and IGF2 are only expressed from the maternally- and paternally-inherited chromosome, respectively. H19 and IGF2 compete for a common enhancer element located in an imprinted domain of chromosome 11. Mutations in these genes are associated with Beckwith-Wiedemann Syndrome and Wilms tumorigenesis.

RBP5, retinol binding protein 5, cellular
Retinol-dependent physiological processes are mediated by retinol-binding proteins (RBPs) and their receptors through regulating cell proliferation and differentiation (Ho et al., 2007). Retinol can prevent potentially malignant disorders. RBP5 down-regulation in hepatocellular carcinoma was associated with aggressive tumor features, suggesting an important role of RPB5 in cancer progression (Ho et al., 2007).

RB1, retinoblastoma 1
The protein encoded by this gene is a negative regulator of the cell cycle and acts as a tumor suppressor gene (http://www.ncbi.nlm.nih.gov/gene/5925). The active form of RB1 protein binds E2F transcription factor 1 (E2F1). Defects in this gene result in the phenotype of childhood cancer retinoblastoma (RB), bladder cancer, and osteogenic sarcoma.

MEG3, maternally expressed 3 (non-protein coding)
The MEG3 gene is a long non-coding RNA (lncRNA) (http://www.ncbi.nlm.nih.gov/gene/55384). The MEG3 locus is frequently deregulated in hepatocellular carcinoma (Anwar et al., 2012). MEG3 interacts with the tumor suppressor p53 and regulates p53 target gene expression. MEG3 inhibits cell proliferation and angiogenesis. Its expression is lost in multiple cancers.
UBE3A, ubiquitin protein ligase E3A
The UBE3A gene encodes an E3 ubiquitin-protein ligase, part of the ubiquitin protein degradation system (http://www.ncbi.nlm.nih.gov/gene/7337). This imprinted gene is maternally expressed in brain. Deletion of this gene causes Angelman Syndrome, characterized by severe motor and intellectual retardation, ataxia, hypotonia, epilepsy, absence of speech, and characteristic facies (Mabb et al., 2011). UBE3A also interacts with high risk subtype HPV16 early oncoprotein E6, resulting in ubiquitination and proteolysis of tumor protein p53.

ATP10A, ATPase, class V, type 10A
The encoded ATP10A protein belongs to the family of P-type cation transport ATPases (http://www.ncbi.nlm.nih.gov/gene/57194). The ATP10A gene maps within the interval of deletion responsible for Angelman syndrome.

NAA60, N(alpha)-acetyltransferase 60, NatF catalytic subunit
NatF is important for normal chromosome segregation (Van Damme et al., 2011).

ZNF597, zinc finger protein 597
ZNF597 is associated with brain development and maintenance (Tanabe et al., 2010).

TCEB3C, transcription elongation factor B polypeptide 3C (elongin A3)
The elongin transcription elongation factor complex enhances the rate of transcription elongation by RNA polymerase II (http://www.ncbi.nlm.nih.gov/gene/162699). The TCEB3C gene encodes elongin subunit A3.

NLRP2, Nucleotide-binding oligomerization domain, Leucine rich Repeat and Pyrin domain containing 2
NLRP proteins, such as NLRP2, are involved in the activation of caspase-1 by Toll-like receptors (http://www.ncbi.nlm.nih.gov/gene/55655). They may be involved in activation of proinflammatory caspases.

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