GENETICS

279 CASES OF IMPRINTED HUMAN LOCI

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igc.otago.ac.nz has a nice list of all imprinted human loci. I have reworked that list and found that most (150) do not have any disease association. Diseases influenced by imprinted marks are

Alcoholism Type II diabetes susceptibility locus Neuroblastoma tumour suppressor locus Bipolar affective disorder Relative hand skill Schizophrenia 4q21/q23 syndrome Asthma Childhood-onset schizophrenia Cornelia de Lange syndrome Cleft palate Body mass index (BMI), obesity Psoriasis IgA deficiency T1D Celiac disease Odour preference Multiple sclerosis Transient neonatal diabetes Acute myeloid leukaemia Silver-Russell syndrome Williams syndrome Autism susceptibility Birk Barel mental retardation dysmorphism syndrome Somatic deletion in ALL 9p22-pter phenotype ABO blood group Obesity and BMI Alzheimer disease pre-eclampsia Silver Russell Syndrome Gingival fibromatosis Beckwith-Wiedemann syndrome (BWS) Birth weight Gilles de la Tourette syndrome Jacobsen syndrome deletions Prader-Willi syndrome (PWS) Angelman syndrome Autism Polycystic kidney disease Type 1 diabetes Psoriatic arthritis Adult height 18q- syndrome Oligodendrogliomas Progressive diaphyseal dysplasia Myotonic dystrophy DiGeorge / velocardiofacial syndrome Schizophrenia linkage Haematopoiesis Longevity Type I diabetes Clotting factor VIII Juvenile idiopathic arthritis Mosaic genome-wide paternal UPD Hypertrophic Cardiomyopathy Spina bifida Psoriasis and Psoriatic arthritis Epilepsy Atrial septal defects Panic disorder Rheumatoid arthritis Crohn disease IMAGE association Post traumatic stress disorder in offspring of Holocaust survivors X inactivation UPD Male transsexuals Turner syndrome – Superior temporal gyrus morphology Klinefelter syndrome Turner syndrome phenotypes Premature ovarian failure associated with fragile X

Affected genes are

DIRAS3 (ARHI, NOEY2) (Provisional data) EPS15 L211 TP73 LRRTM1 U2AF1L1 (U2AF1-RS1, U2AFBPL, Zrsr1) COMMD1 (Murr1) N-MYC SCA7 RASSF1 (RASSF1A) SFRP2 APC SPINK5 clone L59 PRIM2 (PRIM2A) HFE HLA class Ia antigens BRD2 (EMJ1) C6orf66 PLAGL1 (ZAC, LOT1) HYMAI IDDM8 IGF2R (M6PR) (disputed) MAS1 (disputed) SLC22A2 SLC22A3 GRB10 (MEG1) DDC CALCR GNGT1 TFPI2 SGCE (epsilon-sarcoglycan) PEG10 PPP1R9A (neurabin-I) PON1 (Paraoxonase 1) PON3 (paraoxonase 3) PON2 (paraoxonase 2) ASB4 DLX5 (refuted) APS Hypothetical protein DKFZp761N09121 CPA4 MEST (PEG1) MESTIT1 (PEG1-AS) COPG2IT1 (MIT1, CIT1) COPG2 (disputed) KLF14 Autism CNTNAP2 (autism) DLGAP2 KCNK9 IDDM10 RET or other modifier gene CTNNA3 STOX1 INPP5F_V2 AWT1 (WT1 alternative) WT1-AS (provisional) BDNF CD44 ZNF215 (provisional evidence) AMPD3 H19, miR-675 IGF2 IGF2AS, PEG8 INS, insulin TH ASCL2 (HASH2) TSPAN32 (PHEMX, TSSC6) CD81 (TAPA1) TSSC4 TRPM5 (LTRPC5, MTR1) KCNQ1, KvLQT1 KCNQ1OT1 (LIT1, KvLQT1-AS, KvDMR1) KCNQ1DN (BWRT) CDKN1C (p57KIP2) SLC22A18AS (SLC22A1LS, BWR1B, BWSCR1B, ORCTL2S) SLC22A18 (BWR1A, BWSCR1A, HET, IMPT1, ITM, ORCTL2, SLC22A1L, TSSC5) PHLDA2 (TSSC3, IPL, BWR1C) OSBPL5 (OBPH1) MS4A2, FCERI, FCERIB (Fc epsilon RI-beta), DHCR7 SDHD SLC38A4 (NAT3, ATA3) WIF1 DCN RB locus HTR2A (disputed) PHF11 (LOC51131) FLJ13639 SCA8 TGFB3 IG-DMR epimutation syndromes BEGAIN (KIAA1446) DLK1 (PEG9) DLK1 downstream transcripts MEG3 (GTL2) MEG8 DIO3 MKRN3 (ZNF127) NDN MAGEL2 (NDNL1) W89101 (provisional) SNURF, SNRPN, SNORD107 (HBII-436), SNORD64 (HBII-13), SNORD108(HBII-437), SNORD109A (HBII438A), SNORD116@ (PWCR1; HBII-85), SNORD115@ (HBII-52), SNORD109B (HBII438B), UBE3A-AS (incl. IPW,PAR-SN,PAR1,PAR5) UBE3A ATP10A (ATP10C) GABRB3, GABRA5, GABRG3 (conflicting evidence) H73492 (Hs.268983) (provisional) RASGRF1 (GRF1) IL4R (Interleukin 4 receptor) ZNF597 (FLJ33071) GNAL Type II diabetes susceptibility locus Clone L3 IMPACT TCEB3C (Elongin A3) ZNF331 (ZNF463) (provisional evidence) USP29, ZIM3, ZNF264 ITUP1 (MIM1, MIMT1, IMPO1) PEG3, ZIM2 BLCAP (BC10) NNAT (Neuronatin) PSIMCT-1 (MCTS2, MCTS1 pseudogene) HM13 (H13) Differentially methylated gene L3MBTL GNAS1, Gs alpha, NESP55, XLalpha s, SANG (GNAS1-AS) Maternally methylated CpG islands amyloid beta A4 precursor protein, APP clone L88 TCOF1 HLA class II genes HLA-DQ HLA-DR3 HLA-DRB1*15