Wendy P Robinson

List of Publications by Year in Descending Order

174 papers	7,302 citations	46 h-index	79 g-index
212 ext. papers	8,295 ext. citations	4.6 avg, IF	5.85 L-index

#	Paper	IF	Citations
174	Small Non-Coding RNAs in the Human Placenta: Regulatory Roles and Clinical Utility.. *Frontiers in Genetics*, **2022**, 13, 868598	4.5	0
173	Epigenetics in Development **2022**, 97-121		0
172	Are sex differences in cognitive impairment reflected in epigenetic age acceleration metrics?. *Neurobiology of Aging*, **2021**, 109, 192-194	5.6	2
171	Confined placental mosaicism involving multiple de novo copy number variants associated with fetal growth restriction: A case report. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1908-1912	2.5	2
170	Molecular Aspects of Placental Development **2021**, 197-213		
169	A cross-cohort analysis of autosomal DNA methylation sex differences in the term placenta. *Biology of Sex Differences*, **2021**, 12, 38	9.3	3
168	Examining the Vanishing Twin Hypothesis of Neural Tube Defects: Application of an Epigenetic Predictor for Monozygotic Twinning. *Twin Research and Human Genetics*, **2021**, 24, 155-159	2.2	
167	Human placental piwi-interacting RNA transcriptome is characterized by expression from the DLK1-DIO3 imprinted region. *Scientific Reports*, **2021**, 11, 14981	4.9	1
166	Genomic imbalances in the placenta are associated with poor fetal growth. *Molecular Medicine*, **2021**, 27, 3	6.2	5
165	Cell-specific characterization of the placental methylome. *BMC Genomics*, **2021**, 22, 6	4.5	20
164	Sex Differences Are Here to Stay: Relevance to Prenatal Care. *Journal of Clinical Medicine*, **2021**, 10,	5.1	4
163	Profiling the small non-coding RNA transcriptome of the human placenta. *Scientific Data*, **2021**, 8, 166	8.2	4
162	Mistakes Are Common; Should We Worry about Them?. *Trends in Molecular Medicine*, **2021**, 27, 721-722	11.5	0
161	Renpenning syndrome in a female. *American Journal of Medical Genetics, Part A*, **2020**, 182, 498-503	2.5	1
160	Low oxygen enhances trophoblast column growth by potentiating differentiation of the extravillous lineage and promoting LOX activity. *Development (Cambridge)*, **2020**, 147,	6.6	9
159	The significance of the placental genome and methylome in fetal and maternal health. *Human Genetics*, **2020**, 139, 1183-1196	6.3	2
158	Altered levels of placental miR-338-3p and miR-518b are associated with acute chorioamnionitis and IL6 genotype. *Placenta*, **2019**, 82, 42-45	3.4	2

157	Association of a placental Interleukin-6 genetic variant (rs1800796) with DNA methylation, gene expression and risk of acute chorioamnionitis. *BMC Medical Genetics*, **2019**, 20, 36	2.1	10
156	Exome sequencing identified a de novo mutation of PURA gene in a patient with familial Xp22.31 microduplication. *European Journal of Medical Genetics*, **2019**, 62, 103-108	2.6	10
155	Accurate ethnicity prediction from placental DNA methylation data. *Epigenetics and Chromatin*, **2019**, 12, 51	5.8	20
154	A case of intraplacental gestational choriocarcinoma; characterised by the methylation pattern of the early placenta and an absence of driver mutations. *BMC Cancer*, **2019**, 19, 744	4.8	8
153	Placental epigenetic clocks: estimating gestational age using placental DNA methylation levels. *Aging*, **2019**, 11, 4238-4253	5.6	29
152	Considerations when processing and interpreting genomics data of the placenta. *Placenta*, **2019**, 84, 57-62	3.4	12
151	Epigenetic Modifications in the Human Placenta **2019**, 293-311		2
150	Mining DNA methylation alterations towards a classification of placental pathologies. *Human Molecular Genetics*, **2018**, 27, 135-146	5.6	39
149	Utility of DNA methylation to assess placental health. *Placenta*, **2018**, 64 Suppl 1, S23-S28	3.4	10
148	Epigenetic regulation of placental gene expression in transcriptional subtypes of preeclampsia. *Clinical Epigenetics*, **2018**, 10, 28	7.7	41
147	Adjusting for Batch Effects in DNA Methylation Microarray Data, a Lesson Learned. *Frontiers in Genetics*, **2018**, 9, 83	4.5	33
146	No evidence for association of 677C>T and 1298A>C variants with placental DNA methylation. *Clinical Epigenetics*, **2018**, 10, 34	7.7	19
145	DNA methylation profiling of acute chorioamnionitis-associated placentas and fetal membranes: insights into epigenetic variation in spontaneous preterm births. *Epigenetics and Chromatin*, **2018**, 11, 63	5.8	21
144	CHIPS-Child: Testing the developmental programming hypothesis in the offspring of the CHIPS trial. *Pregnancy Hypertension*, **2018**, 14, 15-22	2.6	4
143	IFPA meeting 2016 workshop report II: Placental imaging, placenta and development of other organs, sexual dimorphism in placental function and trophoblast cell lines. *Placenta*, **2017**, 60 Suppl 1, S10-S14	3.4	12
142	Whole exome sequencing of families with 1q21.1 microdeletion or microduplication. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1782-1791	2.5	4
141	An empirically driven data reduction method on the human 450K methylation array to remove tissue specific non-variable CpGs. *Clinical Epigenetics*, **2017**, 9, 11	7.7	22
140	Placentation and Genomic Imprinting **2017**, 159-184		

139	Review: placental biomarkers for assessing fetal health. *Human Molecular Genetics*, **2017**, 26, R237-R245	5.6	22
138	Cell-Free Placental DNA in Maternal Plasma in Relation to Placental Health and Function. *Fetal Diagnosis and Therapy*, **2017**, 41, 258-264	2.4	6
137	Child mortality, hypothalamic-pituitary-adrenal axis activity and cellular aging in mothers. *PLoS ONE* , **2017**, 12, e0177869	3.7	4
136	A de novo mosaic mutation in SPAST with two novel alternative alleles and chromosomal copy number variant in a boy with spastic paraplegia and autism spectrum disorder. *European Journal of Medical Genetics*, **2017**, 60, 548-552	2.6	9
135	Cord blood hematopoietic cells from preterm infants display altered DNA methylation patterns. *Clinical Epigenetics*, **2017**, 9, 39	7.7	16
134	Developmental transcription factor NFIB is a putative target of oncofetal miRNAs and is associated with tumour aggressiveness in lung adenocarcinoma. *Journal of Pathology*, **2016**, 240, 161-72	9.4	29
133	Profiling placental and fetal DNA methylation in human neural tube defects. *Epigenetics and Chromatin*, **2016**, 9, 6	5.8	31
132	Pervasive polymorphic imprinted methylation in the human placenta. *Genome Research*, **2016**, 26, 756-67	9.7	69
131	Number of Children and Telomere Length in Women: A Prospective, Longitudinal Evaluation. *PLoS ONE*, **2016**, 11, e0146424	3.7	32
130	The English 'Teaching Excellence Framework' and Professionalising Teaching and Learning in Research-Intensive Universities: An Exploration of Opportunities, Challenges, Rewards and Values from a Recent Empirical Study. *Foro De Educacion*, **2016**, 14, 151	1.1	9
129	Characterizing the hypomethylated DNA methylation profile of nucleated red blood cells from cord blood. *Epigenomics*, **2016**, 8, 1481-1494	4.4	20
128	Placental telomere length decline with gestational age differs by sex and TERT, DNMT1, and DNMT3A DNA methylation. *Placenta*, **2016**, 48, 26-33	3.4	19
127	Recurrent triploidy due to a failure to complete maternal meiosis II: whole-exome sequencing reveals candidate variants. *Molecular Human Reproduction*, **2015**, 21, 339-46	4.4	30
126	Biallelic mutations in huntington disease: A new case with just one affected parent, review of the literature and terminology. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1152-60	2.5	6
125	ADAM12-directed ectodomain shedding of E-cadherin potentiates trophoblast fusion. *Cell Death and Differentiation*, **2015**, 22, 1970-84	12.7	26
124	The genotypic and phenotypic spectrum of PIGA deficiency. *Orphanet Journal of Rare Diseases*, **2015**, 10, 23	4.2	54
123	Functional consequences of copy number variants in miscarriage. *Molecular Cytogenetics*, **2015**, 8, 6	2	16
122	Genome-wide DNA methylation identifies trophoblast invasion-related genes: Claudin-4 and Fucosyltransferase IV control mobility via altering matrix metalloproteinase activity. *Molecular Human Reproduction*, **2015**, 21, 452-65	4.4	9

121	The human placental methylome. *Cold Spring Harbor Perspectives in Medicine*, **2015**, 5, a023044	5.4	49
120	Noninvasive nucleic acid-based approaches to monitor placental health and predict pregnancy-related complications. *American Journal of Obstetrics and Gynecology*, **2015**, 213, S197-206	6.4	18
119	MG-123 Genomics of early pregnancy loss. *Journal of Medical Genetics*, **2015**, 52, A6.1-A6	5.8	
118	Nucleated red blood cells impact DNA methylation and expression analyses of cord blood hematopoietic cells. *Clinical Epigenetics*, **2015**, 7, 95	7.7	41
117	Placental DNA methylation at term reflects maternal serum levels of INHA and FN1, but not PAPPA, early in pregnancy. *BMC Medical Genetics*, **2015**, 16, 111	2.1	16
116	Defects in fatty acid amide hydrolase 2 in a male with neurologic and psychiatric symptoms. *Orphanet Journal of Rare Diseases*, **2015**, 10, 38	4.2	13
115	Overlapping DNA methylation profile between placentas with trisomy 16 and early-onset preeclampsia. *Placenta*, **2014**, 35, 216-22	3.4	25
114	Variant ATRX syndrome with dysfunction of ATRX and MAGT1 genes. *Human Mutation*, **2014**, 35, 58-62	4.7	6
113	Activation of endocrine-related gene expression in placental choriocarcinoma cell lines following DNA methylation knock-down. *Molecular Human Reproduction*, **2014**, 20, 677-89	4.4	18
112	A cryptic familial rearrangement of 11p15.5, involving both imprinting centers, in a family with a history of short stature. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1587-94	2.5	9
111	Improved reporting of DNA methylation data derived from studies of the human placenta. *Epigenetics*, **2014**, 9, 333-7	5.7	19
110	Quantification of cell-free DNA in normal and complicated pregnancies: overcoming biological and technical issues. *PLoS ONE*, **2014**, 9, e101500	3.7	33
109	Additional annotation enhances potential for biologically-relevant analysis of the Illumina Infinium HumanMethylation450 BeadChip array. *Epigenetics and Chromatin*, **2013**, 6, 4	5.8	349
108	miRNA expression in human lung cancer and fetal lung: a comparative study. *BMC Proceedings*, **2013**, 7,	2.3	78
107	Beckwith-Wiedemann and Silver-Russell syndromes: opposite developmental imbalances in imprinted regulators of placental function and embryonic growth. *Clinical Genetics*, **2013**, 84, 326-34	4	33
106	Hypomethylation of the LEP gene in placenta and elevated maternal leptin concentration in early onset pre-eclampsia. *Molecular and Cellular Endocrinology*, **2013**, 367, 64-73	4.4	53
105	Global analysis of DNA methylation changes during progression of oral cancer. *Oral Oncology*, **2013**, 49, 1033-42	4.4	53
104	Hypoxia alters the epigenetic profile in cultured human placental trophoblasts. *Epigenetics*, **2013**, 8, 192-202	3.7	69

103	DNA methylation profiling of placental villi from karyotypically normal miscarriage and recurrent miscarriage. *American Journal of Pathology*, **2013**, 182, 2276-84	5.8	30
102	Glucose as a fetal nutrient: dynamic regulation of several glucose transporter genes by DNA methylation in the human placenta across gestation. *Journal of Nutritional Biochemistry*, **2013**, 24, 282-8	6.3	41
101	Widespread DNA hypomethylation at gene enhancer regions in placentas associated with early-onset pre-eclampsia. *Molecular Human Reproduction*, **2013**, 19, 697-708	4.4	150
100	X-Chromosome Inactivation **2013**, 63-88		1
99	Maternal NLRP7 and C6orf221 variants are not a common risk factor for androgenetic moles, triploidy and recurrent miscarriage. *Molecular Human Reproduction*, **2013**, 19, 539-44	4.4	17
98	Willing enthusiasts or lame ducks? Issues in teacher professional development policy in England and Wales 1910–1975. *Paedagogica Historica*, **2013**, 49, 345-360	0.2	5
97	The human placenta methylome. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 6037-42	11.5	194
96	Early onset pre-eclampsia is associated with altered DNA methylation of cortisol-signalling and steroidogenic genes in the placenta. *PLoS ONE*, **2013**, 8, e62969	3.7	86
95	Absence of SYCP3 mutations in women with recurrent miscarriage with at least one trisomic miscarriage. *Reproductive BioMedicine Online*, **2012**, 24, 251-3	4	10
94	Prenatal and perinatal environmental influences on the human fetal and placental epigenome. *Clinical Pharmacology and Therapeutics*, **2012**, 92, 716-26	6.1	71
93	DNA methylation changes in whole blood is associated with exposure to the environmental contaminants, mercury, lead, cadmium and bisphenol A, in women undergoing ovarian stimulation for IVF. *Human Reproduction*, **2012**, 27, 1401-10	5.7	127
92	Beckwith-Wiedemann syndrome in sibs discordant for IC2 methylation. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1662-9	2.5	6
91	Response to: "Response to Different measures of 'genome-wide' DNA methylation exhibit unique properties in placental and somatic tissues. *Epigenetics*, **2012**, 7, 965	5.7	2
90	Different measures of "genome-wide" DNA methylation exhibit unique properties in placental and somatic tissues. *Epigenetics*, **2012**, 7, 652-63	5.7	60
89	Patterns of placental development evaluated by X chromosome inactivation profiling provide a basis to evaluate the origin of epigenetic variation. *Human Reproduction*, **2012**, 27, 1745-53	5.7	34
88	Protein kinase profiling in miscarriage: implications for the pathogenesis of trisomic pregnancy. *Journal of Obstetrics and Gynaecology Canada*, **2012**, 34, 1141-1148	1.3	5
87	DNA methylation at H19/IGF2 ICR1 in the placenta of pregnancies conceived by in vitro fertilization and intracytoplasmic sperm injection. *Fertility and Sterility*, **2011**, 95, 2524-6.e1-3	4.8	33
86	Developmental origin of chorionic villus cultures from spontaneous abortion and chorionic villus sampling. *Journal of Obstetrics and Gynaecology Canada*, **2011**, 33, 449-452	1.3	6

85	That great educational experiment the City of London Vacation Course in Education 1922 1938: a forgotten story in the history of teacher professional development. *History of Education*, **2011**, 40, 557-575	93	3
84	Aneuploidy and Polyploidy **2011**, 270-277		
83	The utility of quantitative methylation assays at imprinted genes for the diagnosis of fetal and placental disorders. *Clinical Genetics*, **2011**, 79, 169-75	4	30
82	Review: A high capacity of the human placenta for genetic and epigenetic variation: implications for assessing pregnancy outcome. *Placenta*, **2011**, 32 Suppl 2, S136-41	3.4	37
81	Chromosome-wide DNA methylation analysis predicts human tissue-specific X inactivation. *Human Genetics*, **2011**, 130, 187-201	6.3	95
80	Genome-wide mapping of imprinted differentially methylated regions by DNA methylation profiling of human placentas from triploidies. *Epigenetics and Chromatin*, **2011**, 4, 10	5.8	62
79	Extensive epigenetic reprogramming in human somatic tissues between fetus and adult. *Epigenetics and Chromatin*, **2011**, 4, 7	5.8	51
78	Evidence for widespread changes in promoter methylation profile in human placenta in response to increasing gestational age and environmental/stochastic factors. *BMC Genomics*, **2011**, 12, 529	4.5	142
77	Placenta-specific expression of the interleukin-2 (IL-2) receptor subunit from an endogenous retroviral promoter. *Journal of Biological Chemistry*, **2011**, 286, 35543-35552	5.4	30
76	DNA methylation profiling of human placentas reveals promoter hypomethylation of multiple genes in early-onset preeclampsia. *European Journal of Human Genetics*, **2010**, 18, 1006-12	5.3	164
75	Genetic variation within the hypothalamus-pituitary-ovarian axis in women with recurrent miscarriage. *Human Reproduction*, **2010**, 25, 2664-71	5.7	18
74	Identification of copy number variants in miscarriages from couples with idiopathic recurrent pregnancy loss. *Human Reproduction*, **2010**, 25, 2913-22	5.7	84
73	Fertility and aging: do reproductive-aged Canadian women know what they need to know?. *Fertility and Sterility*, **2010**, 93, 2162-8	4.8	134
72	Decreased placental methylation at the H19/IGF2 imprinting control region is associated with normotensive intrauterine growth restriction but not preeclampsia. *Placenta*, **2010**, 31, 197-202	3.4	136
71	Evaluating DNA methylation and gene expression variability in the human term placenta. *Placenta*, **2010**, 31, 1070-7	3.4	68
70	Pseudohypoparathyroidism type 1a and the GNAS p.R231H mutation: Somatic mosaicism in a mother with two affected sons. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2784-90	2.5	15
69	Methylation profiling in individuals with Russell-Silver syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 347-55	2.5	29
68	Assessing the role of placental trisomy in preeclampsia and intrauterine growth restriction. *Prenatal Diagnosis*, **2010**, 30, 1-8	3.2	37

67	Inactive X chromosome-specific reduction in placental DNA methylation. *Human Molecular Genetics*, **2009**, 18, 3544-52	5.6	60
66	Telomere length and reproductive aging. *Human Reproduction*, **2009**, 24, 1206-11	5.7	78
65	Placental weight in pregnancies with trisomy confined to the placenta. *Journal of Obstetrics and Gynaecology Canada*, **2009**, 31, 605-610	1.3	12
64	Human placental-specific epipolymorphism and its association with adverse pregnancy outcomes. *PLoS ONE*, **2009**, 4, e7389	3.7	54
63	Estrogen receptor alpha gene polymorphisms are associated with idiopathic premature ovarian failure. *Fertility and Sterility*, **2008**, 89, 318-24	4.8	54
62	No difference between wearing a night splint and standing on a tilt table in preventing ankle contracture early after stroke: a randomised trial. *Australian Journal of Physiotherapy*, **2008**, 54, 33-8		23
61	A skewed view of X chromosome inactivation. *Journal of Clinical Investigation*, **2008**, 118, 20-3	15.9	66
60	Chorea as a side effect of gabapentin (Neurontin) in a patient with complex regional pain syndrome Type 1. *Clinical Rheumatology*, **2008**, 27, 389-90	3.9	10
59	MECP2 promoter methylation and X chromosome inactivation in autism. *Autism Research*, **2008**, 1, 169-78	5.1	91
58	Skewed X-chromosome inactivation is associated with primary but not secondary ovarian failure. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 945-51	2.5	10
57	Placental mesenchymal dysplasia associated with fetal overgrowth and mosaic deletion of the maternal copy of 11p15.5. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1752-9	2.5	28
56	Pregnancy and postnatal outcome of mosaic isochromosome 20q. *Prenatal Diagnosis*, **2007**, 27, 143-5	3.2	8
55	Origin and outcome of pregnancies affected by androgenetic/biparental chimerism. *Human Reproduction*, **2007**, 22, 1114-22	5.7	64
54	Toll-like receptor 4 polymorphisms and idiopathic chromosomally normal miscarriage. *Human Reproduction*, **2007**, 22, 440-3	5.7	14
53	Frequency of chromosomal abnormalities in spontaneous abortions derived from intracytoplasmic sperm injection compared with those from in vitro fertilization. *Fertility and Sterility*, **2006**, 85, 236-9	4.8	18
52	Postnatal follow-up of prenatally diagnosed trisomy 16 mosaicism. *Prenatal Diagnosis*, **2006**, 26, 548-58	3.2	45
51	The association between preeclampsia and placental trisomy 16 mosaicism. *Prenatal Diagnosis*, **2006**, 26, 956-61	3.2	30
50	Androgenetic/biparental mosaicism causes placental mesenchymal dysplasia. *Journal of Medical Genetics*, **2006**, 43, 187-92	5.8	128

49	Recurrent trisomy 21: four cases in three generations. *Clinical Genetics*, **2005**, 68, 430-5	4	7
48	FMR1 repeat sizes in the gray zone and high end of the normal range are associated with premature ovarian failure. *Human Genetics*, **2005**, 117, 376-82	6.3	153
47	ICSI and the transmission of X-autosomal translocation: a three-generation evaluation of X;20 translocation: case report. *Human Reproduction*, **2003**, 18, 1377-82	5.7	28
46	Methylation of ZNF261 as an assay for determining X chromosome inactivation patterns **2003**, 120A, 439-41		30
45	Prenatally detected trisomy 4 and 6 mosaicism--cytogenetic results and clinical phenotype. *Prenatal Diagnosis*, **2003**, 23, 128-33	3.2	14
44	Origin of amnion and implications for evaluation of the fetal genotype in cases of mosaicism. *Prenatal Diagnosis*, **2002**, 22, 1076-85	3.2	45
43	Dispermy--origin of diandric triploidy: brief communication. *Human Reproduction*, **2002**, 17, 3037-8	5.7	28
42	Cytogenetic analysis of miscarriages from couples with recurrent miscarriage: a case-control study. *Human Reproduction*, **2002**, 17, 446-51	5.7	320
41	Molecular detection of uniparental disomy. *Methods in Molecular Biology*, **2002**, 204, 291-8	1.4	1
40	Chromosomal Genetic Disease: Numerical Aberrations **2001**,		2
39	Recurrent trisomy 15 in a female carrier of der(15)t(Y;15)(q12;p13). *American Journal of Medical Genetics Part A*, **2001**, 99, 320-4		14
38	Grandmaternal origin of an isochromosome 18p present in two maternal half-sisters. *American Journal of Medical Genetics Part A*, **2001**, 101, 65-9		26
37	Cytogenetic investigation of fetuses and infants conceived through intracytoplasmic sperm injection. *Fertility and Sterility*, **2001**, 76, 1272-5	4.8	24
36	An association between skewed X-chromosome inactivation and abnormal outcome in mosaic trisomy 16 confined predominantly to the placenta. *Clinical Genetics*, **2000**, 58, 436-46	4	20
35	Mechanisms leading to uniparental disomy and their clinical consequences. *BioEssays*, **2000**, 22, 452-9	4.1	235
34	Recurrent trisomy 21 in a couple with a child presenting trisomy 21 mosaicism and maternal uniparental disomy for chromosome 21 in the euploid cell line. *American Journal of Medical Genetics Part A*, **2000**, 94, 35-41		33
33	Somatic segregation errors predominantly contribute to the gain or loss of a paternal chromosome leading to uniparental disomy for chromosome 15. *Clinical Genetics*, **2000**, 57, 349-58	4	46
32	Multipoint genetic mapping with uniparental disomy data. *American Journal of Human Genetics*, **2000**, 67, 851-61	11	6

31	Mechanisms leading to uniparental disomy and their clinical consequences **2000**, 22, 452		3
30	Molecular and clinical correlation study of Williams-Beuren syndrome: No evidence of molecular factors in the deletion region or imprinting affecting clinical outcome **1999**, 86, 34-43		28
29	Extremely skewed X-chromosome inactivation is increased in women with recurrent spontaneous abortion. *American Journal of Human Genetics*, **1999**, 65, 913-7	11	71
28	Partial tetrasomy with triplication of chromosome (5) (p14-p15.33) in a patient with severe multiple congenital anomalies. *American Journal of Medical Genetics Part A*, **1998**, 79, 103-107		31
27	Maternal uniparental disomy of chromosome 1 with no apparent phenotypic effects. *American Journal of Human Genetics*, **1998**, 63, 1216-20	11	54
26	Partial tetrasomy with triplication of chromosome (5) (p14-p15.33) in a patient with severe multiple congenital anomalies **1998**, 79, 103		2
25	Skewed X-chromosome inactivation is common in fetuses or newborns associated with confined placental mosaicism. *American Journal of Human Genetics*, **1997**, 61, 1353-61	11	106
24	XIST expression and X-chromosome inactivation in human preimplantation embryos. *American Journal of Human Genetics*, **1997**, 61, 5-8	11	15
23	Novel case of del(17)(q23.1q23.3) further highlights a recognizable phenotype involving deletions of chromosome (17)(q21q24). *American Journal of Medical Genetics Part A*, **1997**, 71, 275-9		15
22	Maternal uniparental disomy of chromosome 2 and confined placental mosaicism for trisomy 2 in a fetus with intrauterine growth restriction, hypospadias, and oligohydramnios. *Prenatal Diagnosis*, **1997**, 17, 443-50	3.2	49
21	Trisomy First, Translocation Second, Uniparental Disomy and Partial Trisomy Third: A New Mechanism for Complex Chromosomal Aneuploidy. *European Journal of Human Genetics*, **1997**, 5, 308-314	5.3	19
20	Mosaicism most likely accounts for extended survival of trisomy 22. *American Journal of Medical Genetics Part A*, **1996**, 62, 100-1		7
19	Comparison of phenotype in uniparental disomy and deletion Prader-Willi syndrome: sex specific differences. *American Journal of Medical Genetics Part A*, **1996**, 65, 133-6		54
18	Phenotype of maternal UPD(14). *American Journal of Medical Genetics Part A*, **1996**, 66, 89		6
17	Genotype-phenotype correlation in a series of 167 deletion and non-deletion patients with Prader-Willi syndrome. *Human Genetics*, **1995**, 96, 638-43	6.3	132
16	Sex-specific meiotic recombination in the Prader--Willi/Angelman syndrome imprinted region. *Human Molecular Genetics*, **1995**, 4, 801-6	5.6	85
15	Uniparental disomy 7 in Silver-Russell syndrome and primordial growth retardation. *Human Molecular Genetics*, **1995**, 4, 583-7	5.6	254
14	Detection of aberrant DNA methylation in unique Prader-Willi syndrome patients and its diagnostic implications. *Human Molecular Genetics*, **1994**, 3, 893-5	5.6	43

13	Parental origin of the supernumerary chromosome in trisomy 18. *Clinical Genetics*, **1993**, 44, 57-61	4	21
12	Molecular definition of the Prader-Willi syndrome chromosome region and orientation of the SNRPN gene. *Human Molecular Genetics*, **1993**, 2, 1991-4	5.6	35
11	Modification of 15q11-q13 DNA methylation imprints in unique Angelman and Prader-Willi patients. *Human Molecular Genetics*, **1993**, 2, 1377-82	5.6	133
10	Homozygous parent affected sib pair method for detecting disease predisposing variants: application to insulin dependent diabetes mellitus. *Genetic Epidemiology*, **1993**, 10, 273-88	2.6	45
9	Exclusively paternal X chromosomes in a girl with short stature. *Human Genetics*, **1993**, 92, 175-8	6.3	23
8	Deletion breakpoints associated with the Prader-Willi and Angelman syndromes (15q11-q13) are not sites of high homologous recombination. *Human Genetics*, **1993**, 91, 181-4	6.3	11
7	Molecular diagnosis of the Prader-Willi and Angelman syndromes by detection of parent-of-origin specific DNA methylation in 15q11-13. *Human Genetics*, **1992**, 90, 313-5	6.3	160
6	Small nuclear ribonucleoprotein polypeptide N (SNRPN), an expressed gene in the Prader-Willi syndrome critical region. *Nature Genetics*, **1992**, 2, 265-9	36.3	242
5	Clues to IDDM pathogenesis from genetic and serological traits in multiply affected families. *Genetic Epidemiology*, **1989**, 6, 117-22	2.6	3
4	HLA-Bw60 increases susceptibility to ankylosing spondylitis in HLA-B27+ patients. *Arthritis and Rheumatism*, **1989**, 32, 1135-41		138
3	The value of DNA methylation profiling in characterizing preeclampsia and intrauterine growth restriction		1
2	Differences in DNA methylation of white blood cell types at birth and in adulthood reflect postnatal immune maturation and influence accuracy of cell type prediction		3
1	Molecular Aspects of Placental Development1031-1047		