Isabel Karen Temple

List of Publications by Year in Descending Order

151 papers	11,401 citations	54 h-index	105 g-index
158 ext. papers	13,273 ext. citations	8.1 avg, IF	6.8 L-index

#	Paper	IF	Citations
151	A patient with multilocus imprinting disturbance involving hypomethylation at 11p15 and 14q32, and phenotypic features of Beckwith-Wiedemann and Temple syndromes.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	0
150	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences.. *Clinical Epigenetics*, **2022**, 14, 41	7.7	0
149	Experiences of adolescents living with Silver-Russell syndrome. *Archives of Disease in Childhood*, **2021**, 106, 1195-1201	2.2	1
148	Activating mutations in BRAF disrupt the hypothalamo-pituitary axis leading to hypopituitarism in mice and humans. *Nature Communications*, **2021**, 12, 2028	17.4	2
147	Whole-genome analysis as a diagnostic tool for patients referred for diagnosis of Silver-Russell syndrome: a real-world study. *Journal of Medical Genetics*, **2021**,	5.8	4
146	Successful pregnancies in an adult with Meier-Gorlin syndrome harboring biallelic CDT1 variants. *American Journal of Medical Genetics, Part A*, **2021**, 185, 871-876	2.5	1
145	Silver Russell syndrome in a preterm girl with 8q12.1 deletion encompassing PLAG1. *Clinical Dysmorphology*, **2021**, 30, 194-196	0.9	1
144	Phenotype of genetically confirmed Silver-Russell syndrome beyond childhood. *Journal of Medical Genetics*, **2020**, 57, 683-691	5.8	6
143	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. *American Journal of Human Genetics*, **2020**, 106, 596-610	11	26
142	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. *Genome Research*, **2019**, 29, 1057-1066	9.7	20
141	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. *Cell*, **2019**, 177, 32-37	56.2	53
140	Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. *Genetical Research*, **2019**, 101, e3	1.1	9
139	Lived experience of Silver-Russell syndrome: implications for management during childhood and into adulthood. *Archives of Disease in Childhood*, **2019**, 104, 76-82	2.2	7
138	Genetics, molar pregnancies and medieval ideas of monstrous births: the lump of flesh in. *Medical Humanities*, **2019**, 45, 2-9	1.4	1
137	HIST1H1E heterozygous protein-truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2049-2055	2.5	10
136	Pathogenicity and selective constraint on variation near splice sites. *Genome Research*, **2019**, 29, 159-170	9.7	29
135	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. *American Journal of Human Genetics*, **2018**, 102, 175-187	11	108

134	Maternal variants in and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. *Journal of Medical Genetics*, **2018**, 55, 497-504	5.8	66
133	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with constitutive variants. *Wellcome Open Research*, **2018**, 3, 46	4.8	41
132	Growth Hormone Improves Short-Term Growth in Patients with Temple Syndrome. *Hormone Research in Paediatrics*, **2018**, 90, 407-413	3.3	7
131	Prevalence and architecture of de novo mutations in developmental disorders. *Nature*, **2017**, 542, 433-438	38.4	765
130	Hypoinsulinaemic, hypoketotic hypoglycaemia due to mosaic genetic activation of PI3-kinase. *European Journal of Endocrinology*, **2017**, 177, 175-186	6.5	24
129	Analysis of exome data for 4293 trios suggests GPI-anchor biogenesis defects are a rare cause of developmental disorders. *European Journal of Human Genetics*, **2017**, 25, 669-679	5.3	48
128	Human imprinting disorders: Principles, practice, problems and progress. *European Journal of Medical Genetics*, **2017**, 60, 618-626	2.6	35
127	Familial Ebstein Anomaly: Whole Exome Sequencing Identifies Novel Phenotype Associated With. *Circulation: Cardiovascular Genetics*, **2017**, 10,		7
126	Diagnosis and management of Silver-Russell syndrome: first international consensus statement. *Nature Reviews Endocrinology*, **2017**, 13, 105-124	15.2	224
125	Recent Advances in Imprinting Disorders. *Clinical Genetics*, **2017**, 91, 3-13	4	62
124	Mutations in Epigenetic Regulation Genes Are a Major Cause of Overgrowth with Intellectual Disability. *American Journal of Human Genetics*, **2017**, 100, 725-736	11	103
123	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. *Genetics in Medicine*, **2016**, 18, 309-15	8.1	52
122	A Recurrent Mosaic Mutation in SMO, Encoding the Hedgehog Signal Transducer Smoothened, Is the Major Cause of Curry-Jones Syndrome. *American Journal of Human Genetics*, **2016**, 98, 1256-1265	11	53
121	Temple syndrome as a result of isolated hypomethylation of the 14q32 imprinted DLK1/MEG3 region. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 170-5	2.5	22
120	Prenatal molecular testing for Beckwith-Wiedemann and Silver-Russell syndromes: a challenge for molecular analysis and genetic counseling. *European Journal of Human Genetics*, **2016**, 24, 784-93	5.3	34
119	Genetic Analysis of PAX6-Negative Individuals with Aniridia or Gillespie Syndrome. *PLoS ONE*, **2016**, 11, e0153757	3.7	34
118	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. *Human Mutation*, **2016**, 37, 148-54	4.7	31
117	Falling short? The psychosocial impact of living with Russell-Silver syndrome. *Journal of Aesthetic Nursing*, **2016**, 5, 340-342	0.1	2

116	A Restricted Repertoire of De Novo Mutations in ITPR1 Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. *American Journal of Human Genetics*, **2016**, 98, 981-992	11	53
115	Genome-wide DNA methylation analysis of transient neonatal diabetes type 1 patients with mutations in ZFP57. *BMC Medical Genetics*, **2016**, 17, 29	2.1	21
114	The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. *Lancet, The*, **2015**, 386, 957-63	40	186
113	Pallister-Killian syndrome: a study of 22 British patients. *Journal of Medical Genetics*, **2015**, 52, 454-64	5.8	34
112	Role of noninsulin therapies alone or in combination in chromosome 6q24-related transient neonatal diabetes: sulfonylurea improves but does not always normalize insulin secretion. *Diabetes Care*, **2015**, 38, e86-7	14.6	18
111	Beyond the gene roundtable discussion. *Textual Practice*, **2015**, 29, 415-432	0.1	1
110	Very small deletions within the NESP55 gene in pseudohypoparathyroidism type 1b. *European Journal of Human Genetics*, **2015**, 23, 494-9	5.3	18
109	Congenital imprinting disorders: EUCID.net - a network to decipher their aetiology and to improve the diagnostic and clinical care. *Clinical Epigenetics*, **2015**, 7, 23	7.7	19
108	Mutations in NLRP5 are associated with reproductive wastage and multilocus imprinting disorders in humans. *Nature Communications*, **2015**, 6, 8086	17.4	102
107	Large-scale discovery of novel genetic causes of developmental disorders. *Nature*, **2015**, 519, 223-8	50.4	706
106	Heterogeneity of the growth phenotype and birth size in acid-labile subunit (ALS) deficiency. *Journal of Endocrinological Investigation*, **2015**, 38, 407-12	5.2	11
105	A statistical method for single sample analysis of HumanMethylation450 array data: genome-wide methylation analysis of patients with imprinting disorders. *Clinical Epigenetics*, **2015**, 7, 48	7.7	12
104	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. *Clinical Epigenetics*, **2015**, 7, 123	7.7	115
103	Mutations in the DNA methyltransferase gene DNMT3A cause an overgrowth syndrome with intellectual disability. *Nature Genetics*, **2014**, 46, 385-8	36.3	196
102	Genome-wide DNA methylation analysis of patients with imprinting disorders identifies differentially methylated regions associated with novel candidate imprinted genes. *Journal of Medical Genetics*, **2014**, 51, 229-38	5.8	70
101	A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. *European Journal of Medical Genetics*, **2014**, 57, 587-95	2.6	14
100	Identification of novel craniofacial regulatory domains located far upstream of SOX9 and disrupted in Pierre Robin sequence. *Human Mutation*, **2014**, 35, 1011-20	4.7	54
99	A familial disorder of altered DNA-methylation. *Journal of Medical Genetics*, **2014**, 51, 407-12	5.8	22

98	Temple syndrome: improving the recognition of an underdiagnosed chromosome 14 imprinting disorder: an analysis of 51 published cases. *Journal of Medical Genetics*, **2014**, 51, 495-501	5.8	136
97	Clinical utility gene card for: Beckwith-Wiedemann Syndrome. *European Journal of Human Genetics*, **2014**, 22,	5.3	43
96	Clinical utility gene card for: Transient Neonatal Diabetes Mellitus, 6q24-related. *European Journal of Human Genetics*, **2014**, 22,	5.3	18
95	Megalencephaly syndromes: exome pipeline strategies for detecting low-level mosaic mutations. *PLoS ONE*, **2014**, 9, e86940	3.7	17
94	Clinical presentation of 6q24 transient neonatal diabetes mellitus (6q24 TNDM) and genotype-phenotype correlation in an international cohort of patients. *Diabetologia*, **2013**, 56, 758-62	10.3	90
93	Targeted methylation testing of a patient cohort broadens the epigenetic and clinical description of imprinting disorders. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2174-82	2.5	65
92	Mutations in PIK3R1 cause SHORT syndrome. *American Journal of Human Genetics*, **2013**, 93, 158-66	11	125
91	Transient neonatal diabetes, ZFP57, and hypomethylation of multiple imprinted loci: a detailed follow-up. *Diabetes Care*, **2013**, 36, 505-12	14.6	48
90	The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. *Orphanet Journal of Rare Diseases*, **2013**, 8, 63	4.2	47
89	Hepatoblastoma in a child with a paternally-inherited ABCC8 mutation and mosaic paternal uniparental disomy 11p causing focal congenital hyperinsulinism. *European Journal of Medical Genetics*, **2013**, 56, 114-7	2.6	16
88	Elements of morphology: general terms for congenital anomalies. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2726-33	2.5	77
87	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2972-80	2.5	80
86	Evidence for anticipation in Beckwith-Wiedemann syndrome. *European Journal of Human Genetics*, **2013**, 21, 1344-8	5.3	23
85	Familial occurrence of neonatal diabetes with duplications in chromosome 6q24: treatment with sulfonylurea and 40-yr follow-up. *Pediatric Diabetes*, **2012**, 13, 155-62	3.6	15
84	Meier-Gorlin syndrome: growth and secondary sexual development of a microcephalic primordial dwarfism disorder. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2733-42	2.5	34
83	Imprinting Disorders of Early Childhood **2012**, 137-160		1
82	How genetically heterogeneous is Kabuki syndrome?: MLL2 testing in 116 patients, review and analyses of mutation and phenotypic spectrum. *European Journal of Human Genetics*, **2012**, 20, 381-8	5.3	113
81	A primary care specialist genetics service: a cluster-randomised factorial trial. *British Journal of General Practice*, **2012**, 62, e191-7	1.6	8

80	Beckwith-Wiedemann syndrome caused by maternally inherited mutation of an OCT-binding motif in the IGF2/H19-imprinting control region, ICR1. *European Journal of Human Genetics*, **2012**, 20, 240-3	5.3	51
79	Early-onset LBSL: how severe does it get?. *Neuropediatrics*, **2012**, 43, 332-8	1.6	35
78	Meier-Gorlin syndrome genotype-phenotype studies: 35 individuals with pre-replication complex gene mutations and 10 without molecular diagnosis. *European Journal of Human Genetics*, **2012**, 20, 598-606	5.3	76
77	Large de novo deletion of 7p15.1 to 7p12.1 involving the imprinted gene GRB10 associated with a complex phenotype including features of Beckwith Wiedemann syndrome. *European Journal of Medical Genetics*, **2011**, 54, 89-93	2.6	7
76	Deletions and rearrangements of the H19/IGF2 enhancer region in patients with Silver-Russell syndrome and growth retardation. *Journal of Medical Genetics*, **2011**, 48, 308-11	5.8	33
75	Germline mutations in the oncogene EZH2 cause Weaver syndrome and increased human height. *Oncotarget*, **2011**, 2, 1127-33	3.3	110
74	Mutations in the pre-replication complex cause Meier-Gorlin syndrome. *Nature Genetics*, **2011**, 43, 356-9	36.3	186
73	Clinical utility gene card for: Silver-Russell syndrome. *European Journal of Human Genetics*, **2011**, 19,	5.3	11
72	An atypical case of hypomethylation at multiple imprinted loci. *European Journal of Human Genetics* , **2011**, 19, 360-2	5.3	33
71	Methylation analysis of 79 patients with growth restriction reveals novel patterns of methylation change at imprinted loci. *European Journal of Human Genetics*, **2010**, 18, 648-55	5.3	80
70	Coat hanger appearances of the ribs: a useful diagnostic marker of paternal uniparental disomy of chromosome 14. *Archives of Disease in Childhood*, **2010**, 95, 909	2.2	3
69	Persistent Fetal Vasculature and Severe Protein C Deficiency. *Molecular Syndromology*, **2010**, 1, 82-86	1.5	5
68	Epigenotype-phenotype correlations in Silver-Russell syndrome. *Journal of Medical Genetics*, **2010**, 47, 760-8	5.8	98
67	Further refinement of the critical minimal genetic region for the imprinting disorder 6q24 transient neonatal diabetes. *Diabetologia*, **2010**, 53, 2347-51	10.3	27
66	DNA hypomethylation, transient neonatal diabetes, and prune belly sequence in one of two identical twins. *European Journal of Pediatrics*, **2010**, 169, 207-13	4.1	19
65	6q24 transient neonatal diabetes. *Reviews in Endocrine and Metabolic Disorders*, **2010**, 11, 199-204	10.5	59
64	Pancreatic hypoplasia presenting with neonatal diabetes mellitus in association with congenital heart defect and developmental delay. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 340-6	2.5	11
63	Investigation of 90 patients referred for molecular cytogenetic analysis using aCGH uncovers previously unsuspected anomalies of imprinting. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1990-3	2.5	6

62	Transient neonatal diabetes mellitus type 1. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2010**, 154C, 335-42	3.1	81
61	Constitutional haploinsufficiency of tumor suppressor genes in mentally retarded patients with microdeletions in 17p13.1. *Cytogenetic and Genome Research*, **2009**, 125, 1-7	1.9	25
60	Phenotype and genotype in 17 patients with Goltz-Gorlin syndrome. *Journal of Medical Genetics*, **2009**, 46, 716-20	5.8	67
59	Mutations in phospholipase C epsilon 1 are not sufficient to cause diffuse mesangial sclerosis. *Kidney International*, **2009**, 75, 415-9	9.9	32
58	Elements of morphology: standard terminology for the hands and feet. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 93-127	2.5	71
57	Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwith-Wiedemann syndrome. *European Journal of Human Genetics*, **2009**, 17, 611-9	5.3	176
56	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. *Nature Genetics*, **2009**, 41, 359-64	36.3	304
55	Hypomethylation of multiple imprinted loci in individuals with transient neonatal diabetes is associated with mutations in ZFP57. *Nature Genetics*, **2008**, 40, 949-51	36.3	417
54	Clinical characterisation of the multiple maternal hypomethylation syndrome in siblings. *European Journal of Human Genetics*, **2008**, 16, 453-61	5.3	46
53	Mosaic paternal uniparental isodisomy and an ABCC8 gene mutation in a patient with permanent neonatal diabetes and hemihypertrophy. *Diabetes*, **2008**, 57, 255-8	0.9	12
52	Nephrocalcinosis and disordered calcium metabolism in two children with SHORT syndrome. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1296-8	2.5	8
51	Microarray-based DNA methylation analysis of imprinted loci in a patient with transient neonatal diabetes mellitus. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 3227-9	2.5	10
50	Mutations in ATP-sensitive K+ channel genes cause transient neonatal diabetes and permanent diabetes in childhood or adulthood. *Diabetes*, **2007**, 56, 1930-7	0.9	273
49	Isolated imprinting mutation of the DLK1/GTL2 locus associated with a clinical presentation of maternal uniparental disomy of chromosome 14. *Journal of Medical Genetics*, **2007**, 44, 637-40	5.8	54
48	Imprinting in human disease with special reference to transient neonatal diabetes and Beckwith-Wiedemann syndrome. *Endocrine Development*, **2007**, 12, 113-123		25
47	Cowden syndrome and Bannayan Riley Ruvalcaba syndrome represent one condition with variable expression and age-related penetrance: results of a clinical study of PTEN mutation carriers. *Journal of Medical Genetics*, **2007**, 44, 579-85	5.8	128
46	Transient Neonatal Diabetes Mellitus **2007**, 17, 169-171		
45	Feasibility and acceptability of providing nurse counsellor genetics clinics in primary care. *Journal of Advanced Nursing*, **2006**, 53, 591-604	3.1	19

44	Epimutation of the TNDM locus and the Beckwith-Wiedemann syndrome centromeric locus in individuals with transient neonatal diabetes mellitus. *Human Genetics*, **2006**, 119, 179-84	6.3	51
43	A maternal hypomethylation syndrome presenting as transient neonatal diabetes mellitus. *Human Genetics*, **2006**, 120, 262-9	6.3	139
42	Genotype-phenotype associations in Sotos syndrome: an analysis of 266 individuals with NSD1 aberrations. *American Journal of Human Genetics*, **2005**, 77, 193-204	11	218
41	Discriminating power of localized three-dimensional facial morphology. *American Journal of Human Genetics*, **2005**, 77, 999-1010	11	117
40	Clinical and mutational spectrum of Mowat-Wilson syndrome. *European Journal of Medical Genetics*, **2005**, 48, 97-111	2.6	103
39	Evaluation of NSD2 and NSD3 in overgrowth syndromes. *European Journal of Human Genetics*, **2005**, 13, 150-3	5.3	26
38	Kabuki syndrome: new ocular findings but no evidence of 8p22-p23.1 duplications in a clinically defined cohort. *European Journal of Human Genetics*, **2005**, 13, 716-20	5.3	28
37	Bisulphite sequencing of the transient neonatal diabetes mellitus DMR facilitates a novel diagnostic test but reveals no methylation anomalies in patients of unknown aetiology. *Human Genetics*, **2005**, 116, 255-61	6.3	51
36	Monozygous triplets discordant for transient neonatal diabetes mellitus and for imprinting of the TNDM differentially methylated region. *Human Genetics*, **2005**, 117, 398-401	6.3	12
35	Genetic heterogeneity in LEOPARD syndrome: two families with no mutations in PTPN11. *Journal of Human Genetics*, **2005**, 50, 21-25	4.3	22
34	Relapsing diabetes can result from moderately activating mutations in KCNJ11. *Human Molecular Genetics*, **2005**, 14, 925-34	5.6	165
33	The complex nature of constitutional de novo apparently balanced translocations in patients presenting with abnormal phenotypes. *Journal of Medical Genetics*, **2005**, 42, 8-16	5.8	164
32	Macular degeneration associated with a novel Treacher Collins tcof1 mutation and evaluation of this mutation in age related macular degeneration. *British Journal of Ophthalmology*, **2005**, 89, 1063-4	5.5	3
31	Partial NSD1 deletions cause 5% of Sotos syndrome and are readily identifiable by multiplex ligation dependent probe amplification. *Journal of Medical Genetics*, **2005**, 42, e56	5.8	34
30	Permanent neonatal diabetes due to paternal germline mosaicism for an activating mutation of the KCNJ11 Gene encoding the Kir6.2 subunit of the beta-cell potassium adenosine triphosphate channel. *Journal of Clinical Endocrinology and Metabolism*, **2004**, 89, 3932-5	5.6	82
29	Clinical features and molecular analysis of seven British kindreds with hereditary hyperferritinaemia cataract syndrome. *European Journal of Human Genetics*, **2004**, 12, 790-6	5.3	19
28	3D analysis of facial morphology. *American Journal of Medical Genetics Part A*, **2004**, 126A, 339-48		165
27	Identification of mutations in TCOF1: use of molecular analysis in the pre- and postnatal diagnosis of Treacher Collins syndrome. *American Journal of Medical Genetics Part A*, **2004**, 127A, 244-8		34

26	Activating mutations in the gene encoding the ATP-sensitive potassium-channel subunit Kir6.2 and permanent neonatal diabetes. *New England Journal of Medicine*, **2004**, 350, 1838-49	59.2	930
25	A female case of Sedaghatian type spondylometaphyseal dysplasia. *American Journal of Medical Genetics Part A*, **2003**, 118A, 377-81		5
24	NSD1 mutations are the major cause of Sotos syndrome and occur in some cases of Weaver syndrome but are rare in other overgrowth phenotypes. *American Journal of Human Genetics*, **2003**, 72, 132-43	11	221
23	Relaxation of imprinted expression of ZAC and HYMAI in a patient with transient neonatal diabetes mellitus. *Human Genetics*, **2002**, 110, 139-44	6.3	70
22	Duplications of chromosome 11p15 of maternal origin result in a phenotype that includes growth retardation. *Human Genetics*, **2002**, 111, 290-6	6.3	81
21	Neonatal diabetes mellitus. *Pediatric Diabetes*, **2002**, 3, 109-12	3.6	10
20	Transient neonatal diabetes, a disorder of imprinting. *Journal of Medical Genetics*, **2002**, 39, 872-5	5.8	149
19	Twelve novel mutations in the tissue-nonspecific alkaline phosphatase gene (ALPL) in patients with various forms of hypophosphatasia. *Human Mutation*, **2001**, 18, 83-4	4.7	45
18	A survey of TWIST for mutations in craniosynostosis reveals a variable length polyglycine tract in asymptomatic individuals. *Human Mutation*, **2001**, 18, 535-41	4.7	36
17	Maturity onset diabetes of the young (MODY) and early onset Type II diabetes are not caused by loss of imprinting at the transient neonatal diabetes (TNDM) locus. *Diabetologia*, **2001**, 44, 924	10.3	5
16	Skin spots and heart tumors. *Journal of Pediatrics*, **2001**, 139, 901-2	3.6	1
15	Transient neonatal diabetes: widening the understanding of the etiopathogenesis of diabetes. *Diabetes*, **2000**, 49, 1359-66	0.9	214
14	Molecular confirmation of germ line mosaicism for a submicroscopic deletion of chromosome 22q11. *American Journal of Medical Genetics Part A*, **1998**, 78, 103-6		27
13	Atypical chondrodysplasia: a further variant of multiple enchondromatosis with vertebral involvement?. *Pediatric Radiology*, **1998**, 28, 963-6	2.8	2
12	A small intraexonic deletion within the dystrophin gene suggests a possible mechanism of mutagenesis. *Human Genetics*, **1997**, 99, 658-62	6.3	8
11	Masked confirmation of linkage between type 1 congenital vitreous anomaly and COL 2A1 in Stickler syndrome. *Graefes Archive for Clinical and Experimental Ophthalmology*, **1996**, 234, 720-1	3.8	12
10	Unbalanced translocation in a mother and her son in one of two 5;10 translocation families. *American Journal of Medical Genetics Part A*, **1996**, 62, 84-90		15
9	A case of maternal uniparental disomy of chromosome 9 in association with confined placental mosaicism for trisomy 9. *Prenatal Diagnosis*, **1996**, 16, 371-4	3.2	24

8	Further evidence for an imprinted gene for neonatal diabetes localised to chromosome 6q22-q23. *Human Molecular Genetics*, **1996**, 5, 1117-21	5.6	115
7	An imprinted gene(s) for diabetes?. *Nature Genetics*, **1995**, 9, 110-2	36.3	177
6	Clinical features and natural history of Beckwith-Wiedemann syndrome: presentation of 74 new cases. *Clinical Genetics*, **1994**, 46, 168-74	4	247
5	Protelomeric sequences are deleted in cases of short arm inverted duplication of chromosome 8. *American Journal of Medical Genetics Part A*, **1994**, 50, 296-9		20
4	X-linked spastic paraplegia (SPG1), MASA syndrome and X-linked hydrocephalus result from mutations in the L1 gene. *Nature Genetics*, **1994**, 7, 402-7	36.3	377
3	Genetic and clinical heterogeneity of Stickler syndrome. *American Journal of Medical Genetics Part A* , **1991**, 41, 44-8		61
2	Frontonasal dysplasia or craniofrontonasal dysplasia and the Poland anomaly?. *Clinical Genetics*, **1990**, 38, 233-6	4	13
1	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging		2