

# Isabel Karen Temple

## List of Publications by Citations

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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	Activating mutations in the gene encoding the ATP-sensitive potassium-channel subunit Kir6.2 and permanent neonatal diabetes. <i>New England Journal of Medicine</i> , <b>2004</b> , 350, 1838-49	59.2	930
150	Prevalence and architecture of de novo mutations in developmental disorders. <i>Nature</i> , <b>2017</b> , 542, 433-438	38.4	765
149	Large-scale discovery of novel genetic causes of developmental disorders. <i>Nature</i> , <b>2015</b> , 519, 223-8	50.4	706
148	Hypomethylation of multiple imprinted loci in individuals with transient neonatal diabetes is associated with mutations in ZFP57. <i>Nature Genetics</i> , <b>2008</b> , 40, 949-51	36.3	417
147	X-linked spastic paraplegia (SPG1), MASA syndrome and X-linked hydrocephalus result from mutations in the L1 gene. <i>Nature Genetics</i> , <b>1994</b> , 7, 402-7	36.3	377
146	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. <i>Nature Genetics</i> , <b>2009</b> , 41, 359-64	36.3	304
145	Mutations in ATP-sensitive K <sup>+</sup> channel genes cause transient neonatal diabetes and permanent diabetes in childhood or adulthood. <i>Diabetes</i> , <b>2007</b> , 56, 1930-7	0.9	273
144	Clinical features and natural history of Beckwith-Wiedemann syndrome: presentation of 74 new cases. <i>Clinical Genetics</i> , <b>1994</b> , 46, 168-74	4	247
143	Diagnosis and management of Silver-Russell syndrome: first international consensus statement. <i>Nature Reviews Endocrinology</i> , <b>2017</b> , 13, 105-124	15.2	224
142	NSD1 mutations are the major cause of Sotos syndrome and occur in some cases of Weaver syndrome but are rare in other overgrowth phenotypes. <i>American Journal of Human Genetics</i> , <b>2003</b> , 72, 132-43	11	221
141	Genotype-phenotype associations in Sotos syndrome: an analysis of 266 individuals with NSD1 aberrations. <i>American Journal of Human Genetics</i> , <b>2005</b> , 77, 193-204	11	218
140	Transient neonatal diabetes: widening the understanding of the etiopathogenesis of diabetes. <i>Diabetes</i> , <b>2000</b> , 49, 1359-66	0.9	214
139	Mutations in the DNA methyltransferase gene DNMT3A cause an overgrowth syndrome with intellectual disability. <i>Nature Genetics</i> , <b>2014</b> , 46, 385-8	36.3	196
138	The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. <i>Lancet, The</i> , <b>2015</b> , 386, 957-63	40	186
137	Mutations in the pre-replication complex cause Meier-Gorlin syndrome. <i>Nature Genetics</i> , <b>2011</b> , 43, 356-9	36.3	186
136	An imprinted gene(s) for diabetes?. <i>Nature Genetics</i> , <b>1995</b> , 9, 110-2	36.3	177
135	Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwith-Wiedemann syndrome. <i>European Journal of Human Genetics</i> , <b>2009</b> , 17, 611-9	5.3	176

134	3D analysis of facial morphology. <i>American Journal of Medical Genetics Part A</i> , <b>2004</b> , 126A, 339-48		165
133	Relapsing diabetes can result from moderately activating mutations in KCNJ11. <i>Human Molecular Genetics</i> , <b>2005</b> , 14, 925-34	5.6	165
132	The complex nature of constitutional de novo apparently balanced translocations in patients presenting with abnormal phenotypes. <i>Journal of Medical Genetics</i> , <b>2005</b> , 42, 8-16	5.8	164
131	Transient neonatal diabetes, a disorder of imprinting. <i>Journal of Medical Genetics</i> , <b>2002</b> , 39, 872-5	5.8	149
130	A maternal hypomethylation syndrome presenting as transient neonatal diabetes mellitus. <i>Human Genetics</i> , <b>2006</b> , 120, 262-9	6.3	139
129	Temple syndrome: improving the recognition of an underdiagnosed chromosome 14 imprinting disorder: an analysis of 51 published cases. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 495-501	5.8	136
128	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. <i>Journal of Medical Genetics</i> , <b>2007</b> , 44, 579-85	5.8	128
127	Mutations in PIK3R1 cause SHORT syndrome. <i>American Journal of Human Genetics</i> , <b>2013</b> , 93, 158-66	11	125
126	Discriminating power of localized three-dimensional facial morphology. <i>American Journal of Human Genetics</i> , <b>2005</b> , 77, 999-1010	11	117
125	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. <i>Clinical Epigenetics</i> , <b>2015</b> , 7, 123	7.7	115
124	Further evidence for an imprinted gene for neonatal diabetes localised to chromosome 6q22-q23. <i>Human Molecular Genetics</i> , <b>1996</b> , 5, 1117-21	5.6	115
123	How genetically heterogeneous is Kabuki syndrome?: MLL2 testing in 116 patients, review and analyses of mutation and phenotypic spectrum. <i>European Journal of Human Genetics</i> , <b>2012</b> , 20, 381-8	5.3	113
122	Germline mutations in the oncogene EZH2 cause Weaver syndrome and increased human height. <i>Oncotarget</i> , <b>2011</b> , 2, 1127-33	3.3	110
121	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. <i>American Journal of Human Genetics</i> , <b>2018</b> , 102, 175-187	11	108
120	Clinical and mutational spectrum of Mowat-Wilson syndrome. <i>European Journal of Medical Genetics</i> , <b>2005</b> , 48, 97-111	2.6	103
119	Mutations in Epigenetic Regulation Genes Are a Major Cause of Overgrowth with Intellectual Disability. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 725-736	11	103
118	Mutations in NLRP5 are associated with reproductive wastage and multilocus imprinting disorders in humans. <i>Nature Communications</i> , <b>2015</b> , 6, 8086	17.4	102
117	Epigenotype-phenotype correlations in Silver-Russell syndrome. <i>Journal of Medical Genetics</i> , <b>2010</b> , 47, 760-8	5.8	98

116	Clinical presentation of 6q24 transient neonatal diabetes mellitus (6q24 TNDM) and genotype-phenotype correlation in an international cohort of patients. <i>Diabetologia</i> , <b>2013</b> , 56, 758-62	10.3	90
115	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. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2004</b> , 89, 3932-5	5.6	82
114	Transient neonatal diabetes mellitus type 1. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2010</b> , 154C, 335-42	3.1	81
113	Duplications of chromosome 11p15 of maternal origin result in a phenotype that includes growth retardation. <i>Human Genetics</i> , <b>2002</b> , 111, 290-6	6.3	81
112	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 2972-80	2.5	80
111	Methylation analysis of 79 patients with growth restriction reveals novel patterns of methylation change at imprinted loci. <i>European Journal of Human Genetics</i> , <b>2010</b> , 18, 648-55	5.3	80
110	Elements of morphology: general terms for congenital anomalies. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 2726-33	2.5	77
109	Meier-Gorlin syndrome genotype-phenotype studies: 35 individuals with pre-replication complex gene mutations and 10 without molecular diagnosis. <i>European Journal of Human Genetics</i> , <b>2012</b> , 20, 598-606	5.3	76
108	Elements of morphology: standard terminology for the hands and feet. <i>American Journal of Medical Genetics, Part A</i> , <b>2009</b> , 149A, 93-127	2.5	71
107	Genome-wide DNA methylation analysis of patients with imprinting disorders identifies differentially methylated regions associated with novel candidate imprinted genes. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 229-38	5.8	70
106	Relaxation of imprinted expression of ZAC and HYMAI in a patient with transient neonatal diabetes mellitus. <i>Human Genetics</i> , <b>2002</b> , 110, 139-44	6.3	70
105	Phenotype and genotype in 17 patients with Goltz-Gorlin syndrome. <i>Journal of Medical Genetics</i> , <b>2009</b> , 46, 716-20	5.8	67
104	Maternal variants in and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. <i>Journal of Medical Genetics</i> , <b>2018</b> , 55, 497-504	5.8	66
103	Targeted methylation testing of a patient cohort broadens the epigenetic and clinical description of imprinting disorders. <i>American Journal of Medical Genetics, Part A</i> , <b>2013</b> , 161A, 2174-82	2.5	65
102	Recent Advances in Imprinting Disorders. <i>Clinical Genetics</i> , <b>2017</b> , 91, 3-13	4	62
101	Genetic and clinical heterogeneity of Stickler syndrome. <i>American Journal of Medical Genetics Part A</i> , <b>1991</b> , 41, 44-8		61
100	6q24 transient neonatal diabetes. <i>Reviews in Endocrine and Metabolic Disorders</i> , <b>2010</b> , 11, 199-204	10.5	59
99	Identification of novel craniofacial regulatory domains located far upstream of SOX9 and disrupted in Pierre Robin sequence. <i>Human Mutation</i> , <b>2014</b> , 35, 1011-20	4.7	54

98	Isolated imprinting mutation of the DLK1/GTL2 locus associated with a clinical presentation of maternal uniparental disomy of chromosome 14. <i>Journal of Medical Genetics</i> , <b>2007</b> , 44, 637-40	5.8	54
97	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. <i>Cell</i> , <b>2019</b> , 177, 32-37	56.2	53
96	A Recurrent Mosaic Mutation in SMO, Encoding the Hedgehog Signal Transducer Smoothed, Is the Major Cause of Curry-Jones Syndrome. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 1256-1265	11	53
95	A Restricted Repertoire of De Novo Mutations in ITPR1 Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 981-992	11	53
94	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. <i>Genetics in Medicine</i> , <b>2016</b> , 18, 309-15	8.1	52
93	Beckwith-Wiedemann syndrome caused by maternally inherited mutation of an OCT-binding motif in the IGF2/H19-imprinting control region, ICR1. <i>European Journal of Human Genetics</i> , <b>2012</b> , 20, 240-3	5.3	51
92	Epimutation of the TNDM locus and the Beckwith-Wiedemann syndrome centromeric locus in individuals with transient neonatal diabetes mellitus. <i>Human Genetics</i> , <b>2006</b> , 119, 179-84	6.3	51
91	Bisulphite sequencing of the transient neonatal diabetes mellitus DMR facilitates a novel diagnostic test but reveals no methylation anomalies in patients of unknown aetiology. <i>Human Genetics</i> , <b>2005</b> , 116, 255-61	6.3	51
90	Analysis of exome data for 4293 trios suggests GPI-anchor biogenesis defects are a rare cause of developmental disorders. <i>European Journal of Human Genetics</i> , <b>2017</b> , 25, 669-679	5.3	48
89	Transient neonatal diabetes, ZFP57, and hypomethylation of multiple imprinted loci: a detailed follow-up. <i>Diabetes Care</i> , <b>2013</b> , 36, 505-12	14.6	48
88	The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. <i>Orphanet Journal of Rare Diseases</i> , <b>2013</b> , 8, 63	4.2	47
87	Clinical characterisation of the multiple maternal hypomethylation syndrome in siblings. <i>European Journal of Human Genetics</i> , <b>2008</b> , 16, 453-61	5.3	46
86	Twelve novel mutations in the tissue-nonspecific alkaline phosphatase gene (ALPL) in patients with various forms of hypophosphatasia. <i>Human Mutation</i> , <b>2001</b> , 18, 83-4	4.7	45
85	Clinical utility gene card for: Beckwith-Wiedemann Syndrome. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22,	5.3	43
84	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with constitutive variants. <i>Wellcome Open Research</i> , <b>2018</b> , 3, 46	4.8	41
83	A survey of TWIST for mutations in craniosynostosis reveals a variable length polyglycine tract in asymptomatic individuals. <i>Human Mutation</i> , <b>2001</b> , 18, 535-41	4.7	36
82	Human imprinting disorders: Principles, practice, problems and progress. <i>European Journal of Medical Genetics</i> , <b>2017</b> , 60, 618-626	2.6	35
81	Early-onset LBSL: how severe does it get?. <i>Neuropediatrics</i> , <b>2012</b> , 43, 332-8	1.6	35

80	Pallister-Killian syndrome: a study of 22 British patients. <i>Journal of Medical Genetics</i> , <b>2015</b> , 52, 454-64	5.8	34
79	Prenatal molecular testing for Beckwith-Wiedemann and Silver-Russell syndromes: a challenge for molecular analysis and genetic counseling. <i>European Journal of Human Genetics</i> , <b>2016</b> , 24, 784-93	5.3	34
78	Meier-Gorlin syndrome: growth and secondary sexual development of a microcephalic primordial dwarfism disorder. <i>American Journal of Medical Genetics, Part A</i> , <b>2012</b> , 158A, 2733-42	2.5	34
77	Identification of mutations in TCOF1: use of molecular analysis in the pre- and postnatal diagnosis of Treacher Collins syndrome. <i>American Journal of Medical Genetics Part A</i> , <b>2004</b> , 127A, 244-8		34
76	Partial NSD1 deletions cause 5% of Sotos syndrome and are readily identifiable by multiplex ligation dependent probe amplification. <i>Journal of Medical Genetics</i> , <b>2005</b> , 42, e56	5.8	34
75	Genetic Analysis of PAX6-Negative Individuals with Aniridia or Gillespie Syndrome. <i>PLoS ONE</i> , <b>2016</b> , 11, e0153757	3.7	34
74	Deletions and rearrangements of the H19/IGF2 enhancer region in patients with Silver-Russell syndrome and growth retardation. <i>Journal of Medical Genetics</i> , <b>2011</b> , 48, 308-11	5.8	33
73	An atypical case of hypomethylation at multiple imprinted loci. <i>European Journal of Human Genetics</i> , <b>2011</b> , 19, 360-2	5.3	33
72	Mutations in phospholipase C epsilon 1 are not sufficient to cause diffuse mesangial sclerosis. <i>Kidney International</i> , <b>2009</b> , 75, 415-9	9.9	32
71	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. <i>Human Mutation</i> , <b>2016</b> , 37, 148-54	4.7	31
70	Pathogenicity and selective constraint on variation near splice sites. <i>Genome Research</i> , <b>2019</b> , 29, 159-170	9.7	29
69	Kabuki syndrome: new ocular findings but no evidence of 8p22-p23.1 duplications in a clinically defined cohort. <i>European Journal of Human Genetics</i> , <b>2005</b> , 13, 716-20	5.3	28
68	Further refinement of the critical minimal genetic region for the imprinting disorder 6q24 transient neonatal diabetes. <i>Diabetologia</i> , <b>2010</b> , 53, 2347-51	10.3	27
67	Molecular confirmation of germ line mosaicism for a submicroscopic deletion of chromosome 22q11. <i>American Journal of Medical Genetics Part A</i> , <b>1998</b> , 78, 103-6		27
66	Evaluation of NSD2 and NSD3 in overgrowth syndromes. <i>European Journal of Human Genetics</i> , <b>2005</b> , 13, 150-3	5.3	26
65	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. <i>American Journal of Human Genetics</i> , <b>2020</b> , 106, 596-610	11	26
64	Constitutional haploinsufficiency of tumor suppressor genes in mentally retarded patients with microdeletions in 17p13.1. <i>Cytogenetic and Genome Research</i> , <b>2009</b> , 125, 1-7	1.9	25
63	Imprinting in human disease with special reference to transient neonatal diabetes and Beckwith-Wiedemann syndrome. <i>Endocrine Development</i> , <b>2007</b> , 12, 113-123		25

62	Hypoinsulinaemic, hypoketotic hypoglycaemia due to mosaic genetic activation of PI3-kinase. <i>European Journal of Endocrinology</i> , <b>2017</b> , 177, 175-186	6.5	24
61	A case of maternal uniparental disomy of chromosome 9 in association with confined placental mosaicism for trisomy 9. <i>Prenatal Diagnosis</i> , <b>1996</b> , 16, 371-4	3.2	24
60	Evidence for anticipation in Beckwith-Wiedemann syndrome. <i>European Journal of Human Genetics</i> , <b>2013</b> , 21, 1344-8	5.3	23
59	Temple syndrome as a result of isolated hypomethylation of the 14q32 imprinted DLK1/MEG3 region. <i>American Journal of Medical Genetics, Part A</i> , <b>2016</b> , 170A, 170-5	2.5	22
58	A familial disorder of altered DNA-methylation. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 407-12	5.8	22
57	Genetic heterogeneity in LEOPARD syndrome: two families with no mutations in PTPN11. <i>Journal of Human Genetics</i> , <b>2005</b> , 50, 21-25	4.3	22
56	Genome-wide DNA methylation analysis of transient neonatal diabetes type 1 patients with mutations in ZFP57. <i>BMC Medical Genetics</i> , <b>2016</b> , 17, 29	2.1	21
55	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. <i>Genome Research</i> , <b>2019</b> , 29, 1057-1066	9.7	20
54	Protelomeric sequences are deleted in cases of short arm inverted duplication of chromosome 8. <i>American Journal of Medical Genetics Part A</i> , <b>1994</b> , 50, 296-9		20
53	Congenital imprinting disorders: EUCID.net - a network to decipher their aetiology and to improve the diagnostic and clinical care. <i>Clinical Epigenetics</i> , <b>2015</b> , 7, 23	7.7	19
52	DNA hypomethylation, transient neonatal diabetes, and prune belly sequence in one of two identical twins. <i>European Journal of Pediatrics</i> , <b>2010</b> , 169, 207-13	4.1	19
51	Feasibility and acceptability of providing nurse counsellor genetics clinics in primary care. <i>Journal of Advanced Nursing</i> , <b>2006</b> , 53, 591-604	3.1	19
50	Clinical features and molecular analysis of seven British kindreds with hereditary hyperferritinaemia cataract syndrome. <i>European Journal of Human Genetics</i> , <b>2004</b> , 12, 790-6	5.3	19
49	Role of noninsulin therapies alone or in combination in chromosome 6q24-related transient neonatal diabetes: sulfonylurea improves but does not always normalize insulin secretion. <i>Diabetes Care</i> , <b>2015</b> , 38, e86-7	14.6	18
48	Very small deletions within the NESP55 gene in pseudohypoparathyroidism type 1b. <i>European Journal of Human Genetics</i> , <b>2015</b> , 23, 494-9	5.3	18
47	Clinical utility gene card for: Transient Neonatal Diabetes Mellitus, 6q24-related. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22,	5.3	18
46	Megalencephaly syndromes: exome pipeline strategies for detecting low-level mosaic mutations. <i>PLoS ONE</i> , <b>2014</b> , 9, e86940	3.7	17
45	Hepatoblastoma in a child with a paternally-inherited ABCC8 mutation and mosaic paternal uniparental disomy 11p causing focal congenital hyperinsulinism. <i>European Journal of Medical Genetics</i> , <b>2013</b> , 56, 114-7	2.6	16

44	Familial occurrence of neonatal diabetes with duplications in chromosome 6q24: treatment with sulfonylurea and 40-yr follow-up. <i>Pediatric Diabetes</i> , <b>2012</b> , 13, 155-62	3.6	15
43	Unbalanced translocation in a mother and her son in one of two 5;10 translocation families. <i>American Journal of Medical Genetics Part A</i> , <b>1996</b> , 62, 84-90		15
42	A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. <i>European Journal of Medical Genetics</i> , <b>2014</b> , 57, 587-95	2.6	14
41	Frontonasal dysplasia or craniofrontonasal dysplasia and the Poland anomaly?. <i>Clinical Genetics</i> , <b>1990</b> , 38, 233-6	4	13
40	A statistical method for single sample analysis of HumanMethylation450 array data: genome-wide methylation analysis of patients with imprinting disorders. <i>Clinical Epigenetics</i> , <b>2015</b> , 7, 48	7.7	12
39	Mosaic paternal uniparental isodisomy and an ABCC8 gene mutation in a patient with permanent neonatal diabetes and hemihypertrophy. <i>Diabetes</i> , <b>2008</b> , 57, 255-8	0.9	12
38	Monozygous triplets discordant for transient neonatal diabetes mellitus and for imprinting of the TNDM differentially methylated region. <i>Human Genetics</i> , <b>2005</b> , 117, 398-401	6.3	12
37	Masked confirmation of linkage between type 1 congenital vitreous anomaly and COL 2A1 in Stickler syndrome. <i>Graefers Archive for Clinical and Experimental Ophthalmology</i> , <b>1996</b> , 234, 720-1	3.8	12
36	Heterogeneity of the growth phenotype and birth size in acid-labile subunit (ALS) deficiency. <i>Journal of Endocrinological Investigation</i> , <b>2015</b> , 38, 407-12	5.2	11
35	Clinical utility gene card for: Silver-Russell syndrome. <i>European Journal of Human Genetics</i> , <b>2011</b> , 19,	5.3	11
34	Pancreatic hypoplasia presenting with neonatal diabetes mellitus in association with congenital heart defect and developmental delay. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 340-6	2.5	11
33	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. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 2049-2055	2.5	10
32	Microarray-based DNA methylation analysis of imprinted loci in a patient with transient neonatal diabetes mellitus. <i>American Journal of Medical Genetics, Part A</i> , <b>2008</b> , 146A, 3227-9	2.5	10
31	Neonatal diabetes mellitus. <i>Pediatric Diabetes</i> , <b>2002</b> , 3, 109-12	3.6	10
30	Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. <i>Genetical Research</i> , <b>2019</b> , 101, e3	1.1	9
29	A primary care specialist genetics service: a cluster-randomised factorial trial. <i>British Journal of General Practice</i> , <b>2012</b> , 62, e191-7	1.6	8
28	A small intraexonic deletion within the dystrophin gene suggests a possible mechanism of mutagenesis. <i>Human Genetics</i> , <b>1997</b> , 99, 658-62	6.3	8
27	Nephrocalcinosis and disordered calcium metabolism in two children with SHORT syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2008</b> , 146A, 1296-8	2.5	8



26	Lived experience of Silver-Russell syndrome: implications for management during childhood and into adulthood. <i>Archives of Disease in Childhood</i> , <b>2019</b> , 104, 76-82	2.2	7
25	Familial Ebstein Anomaly: Whole Exome Sequencing Identifies Novel Phenotype Associated With. <i>Circulation: Cardiovascular Genetics</i> , <b>2017</b> , 10,		7
24	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. <i>European Journal of Medical Genetics</i> , <b>2011</b> , 54, 89-93	2.6	7
23	Growth Hormone Improves Short-Term Growth in Patients with Temple Syndrome. <i>Hormone Research in Paediatrics</i> , <b>2018</b> , 90, 407-413	3.3	7
22	Investigation of 90 patients referred for molecular cytogenetic analysis using aCGH uncovers previously unsuspected anomalies of imprinting. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 1990-3	2.5	6
21	Phenotype of genetically confirmed Silver-Russell syndrome beyond childhood. <i>Journal of Medical Genetics</i> , <b>2020</b> , 57, 683-691	5.8	6
20	Persistent Fetal Vasculature and Severe Protein C Deficiency. <i>Molecular Syndromology</i> , <b>2010</b> , 1, 82-86	1.5	5
19	A female case of Sedaghatian type spondylometaphyseal dysplasia. <i>American Journal of Medical Genetics Part A</i> , <b>2003</b> , 118A, 377-81		5
18	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. <i>Diabetologia</i> , <b>2001</b> , 44, 924	10.3	5
17	Whole-genome analysis as a diagnostic tool for patients referred for diagnosis of Silver-Russell syndrome: a real-world study. <i>Journal of Medical Genetics</i> , <b>2021</b> ,	5.8	4
16	Coat hanger appearances of the ribs: a useful diagnostic marker of paternal uniparental disomy of chromosome 14. <i>Archives of Disease in Childhood</i> , <b>2010</b> , 95, 909	2.2	3
15	Macular degeneration associated with a novel Treacher Collins tcof1 mutation and evaluation of this mutation in age related macular degeneration. <i>British Journal of Ophthalmology</i> , <b>2005</b> , 89, 1063-4	5.5	3
14	Atypical chondrodysplasia: a further variant of multiple enchondromatosis with vertebral involvement?. <i>Pediatric Radiology</i> , <b>1998</b> , 28, 963-6	2.8	2
13	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging		2
12	Activating mutations in BRAF disrupt the hypothalamo-pituitary axis leading to hypopituitarism in mice and humans. <i>Nature Communications</i> , <b>2021</b> , 12, 2028	17.4	2
11	Falling short? The psychosocial impact of living with Russell-Silver syndrome. <i>Journal of Aesthetic Nursing</i> , <b>2016</b> , 5, 340-342	0.1	2
10	Beyond the gene roundtable discussion. <i>Textual Practice</i> , <b>2015</b> , 29, 415-432	0.1	1
9	Genetics, molar pregnancies and medieval ideas of monstrous births: the lump of flesh in. <i>Medical Humanities</i> , <b>2019</b> , 45, 2-9	1.4	1

8	Imprinting Disorders of Early Childhood <b>2012</b> , 137-160		1
7	Skin spots and heart tumors. <i>Journal of Pediatrics</i> , <b>2001</b> , 139, 901-2	3.6	1
6	Experiences of adolescents living with Silver-Russell syndrome. <i>Archives of Disease in Childhood</i> , <b>2021</b> , 106, 1195-1201	2.2	1
5	Successful pregnancies in an adult with Meier-Gorlin syndrome harboring biallelic CDT1 variants. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 871-876	2.5	1
4	Silver Russell syndrome in a preterm girl with 8q12.1 deletion encompassing PLAG1. <i>Clinical Dysmorphology</i> , <b>2021</b> , 30, 194-196	0.9	1
3	A patient with multilocus imprinting disturbance involving hypomethylation at 11p15 and 14q32, and phenotypic features of Beckwith-Wiedemann and Temple syndromes.. <i>American Journal of Medical Genetics, Part A</i> , <b>2022</b> ,	2.5	0
2	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences.. <i>Clinical Epigenetics</i> , <b>2022</b> , 14, 41	7.7	0
1	Transient Neonatal Diabetes Mellitus <b>2007</b> , 17, 169-171		