# Isabel Karen Temple

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5289000/publications.pdf

Version: 2024-02-01

| 152 | 14,682 | 23544 | 20943 |
|---|---|---|---|
| papers | citations | 58 h-index | 115 g-index |

| 158 | 158 | 158 | 16892 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Prevalence and architecture of de novo mutations in developmental disorders. Nature, 2017, 542, 433-438. | 13.7 | 1,211 |
| 2 | 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, 1838-1849. | 13.9 | 1,077 |
| 3 | Large-scale discovery of novel genetic causes of developmental disorders. Nature, 2015, 519, 223-228. | 13.7 | 998 |
| 4 | Hypomethylation of multiple imprinted loci in individuals with transient neonatal diabetes is associated with mutations in ZFP57. Nature Genetics, 2008, 40, 949-951. | 9.4 | 460 |
| 5 | Xâ€“linked spastic paraplegia (SPG1), MASA syndrome and Xâ€“linked hydrocephalus result from mutations in the L1 gene. Nature Genetics, 1994, 7, 402-407. | 9.4 | 423 |
| 6 | Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364. | 9.4 | 364 |
| 7 | Diagnosis and management of Silverâ€“Russell syndrome: first international consensus statement. Nature Reviews Endocrinology, 2017, 13, 105-124. | 4.3 | 336 |
| 8 | Mutations in ATP-Sensitive K+ Channel Genes Cause Transient Neonatal Diabetes and Permanent Diabetes in Childhood or Adulthood. Diabetes, 2007, 56, 1930-1937. | 0.3 | 320 |
| 9 | Clinical features and natural history of Beckwithâ€Wiedemann syndrome: presentation of 74 new cases. Clinical Genetics, 1994, 46, 168-174. | 1.0 | 303 |
| 10 | Genotype-Phenotype Associations in Sotos Syndrome: An Analysis of 266 Individuals with NSD1 Aberrations. American Journal of Human Genetics, 2005, 77, 193-204. | 2.6 | 298 |
| 11 | Mutations in the DNA methyltransferase gene DNMT3A cause an overgrowth syndrome with intellectual disability. Nature Genetics, 2014, 46, 385-388. | 9.4 | 280 |
| 12 | 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, 132-143. | 2.6 | 273 |
| 13 | The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. Lancet, The, 2015, 386, 957-963. | 6.3 | 250 |
| 14 | Transient neonatal diabetes: widening the understanding of the etiopathogenesis of diabetes. Diabetes, 2000, 49, 1359-1366. | 0.3 | 249 |
| 15 | Mutations in the pre-replication complex cause Meier-Gorlin syndrome. Nature Genetics, 2011, 43, 356-359. | 9.4 | 219 |
| 16 | Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187. | 2.6 | 204 |
| 17 | Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2009, 17, 611-619. | 1.4 | 194 |
| 18 | 3D analysis of facial morphology. American Journal of Medical Genetics Part A, 2004, 126A, 339-348. | 2.4 | 192 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | The complex nature of constitutional de novo apparently balanced translocations in patients presenting with abnormal phenotypes. Journal of Medical Genetics, 2005, 42, 8-16. | 1.5 | 192 |
| 20 | An imprinted gene(s) for diabetes?. Nature Genetics, 1995, 9, 110-112. | 9.4 | 190 |
| 21 | Transient neonatal diabetes, a disorder of imprinting. Journal of Medical Genetics, 2002, 39, 872-875. | 1.5 | 188 |
| 22 | Relapsing diabetes can result from moderately activating mutations in KCNJ11. Human Molecular Genetics, 2005, 14, 925-934. | 1.4 | 184 |
| 23 | 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. | 1.5 | 182 |
| 24 | Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. Clinical Epigenetics, 2015, 7, 123. | 1.8 | 174 |
| 25 | 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, 579-585. | 1.5 | 172 |
| 26 | Mutations in Epigenetic Regulation Genes Are a Major Cause of Overgrowth with Intellectual Disability. American Journal of Human Genetics, 2017, 100, 725-736. | 2.6 | 168 |
| 27 | Mutations in PIK3R1 Cause SHORT Syndrome. American Journal of Human Genetics, 2013, 93, 158-166. | 2.6 | 156 |
| 28 | A maternal hypomethylation syndrome presenting as transient neonatal diabetes mellitus. Human Genetics, 2006, 120, 262-269. | 1.8 | 147 |
| 29 | Germline mutations in the oncogene EZH2 cause Weaver syndrome and increased human height. Oncotarget, 2011, 2, 1127-1133. | 0.8 | 145 |
| 30 | 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, 381-388. | 1.4 | 142 |
| 31 | Further evidence for an imprinted gene for neonatal diabetes localised to chromosome 6q22-q23. Human Molecular Genetics, 1996, 5, 1117-1121. | 1.4 | 134 |
| 32 | Mutations in NLRP5 are associated with reproductive wastage and multilocus imprinting disorders in humans. Nature Communications, 2015, 6, 8086. | 5.8 | 134 |
| 33 | Discriminating Power of Localized Three-Dimensional Facial Morphology. American Journal of Human Genetics, 2005, 77, 999-1010. | 2.6 | 133 |
| 34 | Maternal variants in NLRP and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. Journal of Medical Genetics, 2018, 55, 497-504. | 1.5 | 126 |
| 35 | Epigenotype-phenotype correlations in Silver-Russell syndrome. Journal of Medical Genetics, 2010, 47, 760-768. | 1.5 | 123 |
| 36 | Clinical and Mutational Spectrum of Mowatâ€“Wilson Syndrome. European Journal of Medical Genetics, 2005, 48, 97-111. | 0.7 | 121 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 2972-2980. | 0.7 | 119 |
| 38 | Clinical presentation of 6q24 transient neonatal diabetes mellitus (6q24 TNDM) and genotypeâ€“phenotype correlation in an international cohort of patients. Diabetologia, 2013, 56, 758-762. | 2.9 | 113 |
| 39 | A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37. | 13.5 | 113 |
| 40 | Elements of morphology: General terms for congenital anomalies. American Journal of Medical Genetics, Part A, 2013, 161, 2726-2733. | 0.7 | 101 |
| 41 | Recent Advances in Imprinting Disorders. Clinical Genetics, 2017, 91, 3-13. | 1.0 | 101 |
| 42 | Transient neonatal diabetes mellitus type 1. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 335-342. | 0.7 | 99 |
| 43 | Meierâ€“Gorlin syndrome genotypeâ€“phenotype studies: 35 individuals with pre-replication complex gene mutations and 10 without molecular diagnosis. European Journal of Human Genetics, 2012, 20, 598-606. | 1.4 | 95 |
| 44 | Methylation analysis of 79 patients with growth restriction reveals novel patterns of methylation change at imprinted loci. European Journal of Human Genetics, 2010, 18, 648-655. | 1.4 | 91 |
| 45 | 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, 229-238. | 1.5 | 91 |
| 46 | Elements of morphology: Standard terminology for the hands and feet. American Journal of Medical Genetics, Part A, 2009, 149A, 93-127. | 0.7 | 89 |
| 47 | Duplications of chromosome 11p15 of maternal origin result in a phenotype that includes growth retardation. Human Genetics, 2002, 111, 290-296. | 1.8 | 88 |
| 48 | Permanent Neonatal Diabetes due to Paternal Germline Mosaicism for an Activating Mutation of the KCNJ11 Gene Encoding the Kir6.2 Subunit of the Î²-Cell Potassium Adenosine Triphosphate Channel. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 3932-3935. | 1.8 | 87 |
| 49 | Relaxation of imprinted expression of ZAC and HYMAI in a patient with transient neonatal diabetes mellitus. Human Genetics, 2002, 110, 139-144. | 1.8 | 83 |
| 50 | 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. | 2.6 | 81 |
| 51 | The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46. | 0.9 | 75 |
| 52 | Phenotype and genotype in 17 patients with Goltz-Gorlin syndrome. Journal of Medical Genetics, 2009, 46, 716-720. | 1.5 | 74 |
| 53 | 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. | 2.6 | 70 |
| 54 | Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170. | 2.4 | 70 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | 6q24 transient neonatal diabetes. Reviews in Endocrine and Metabolic Disorders, 2010, 11, 199-204. | 2.6 | 69 |
| 56 | Targeted methylation testing of a patient cohort broadens the epigenetic and clinical description of imprinting disorders. American Journal of Medical Genetics, Part A, 2013, 161, 2174-2182. | 0.7 | 69 |
| 57 | Identification of Novel Craniofacial Regulatory Domains Located far Upstream ofSOX9and Disrupted in Pierre Robin Sequence. Human Mutation, 2014, 35, 1011-1020. | 1.1 | 69 |
| 58 | Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315. | 1.1 | 69 |
| 59 | Genetic and clinical heterogeneity of Stickler syndrome. American Journal of Medical Genetics Part A, 1991, 41, 44-48. | 2.4 | 66 |
| 60 | 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. | 1.4 | 63 |
| 61 | Beckwithâ€“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, 240-243. | 1.4 | 60 |
| 62 | 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. | 1.2 | 60 |
| 63 | 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, 637-640. | 1.5 | 59 |
| 64 | DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610. | 2.6 | 59 |
| 65 | Epimutation of the TNDM locus and the Beckwithâ€“Wiedemann syndrome centromeric locus in individuals with transient neonatal diabetes mellitus. Human Genetics, 2006, 119, 179-184. | 1.8 | 56 |
| 66 | Transient Neonatal Diabetes, ZFP57, and Hypomethylation of Multiple Imprinted Loci. Diabetes Care, 2013, 36, 505-512. | 4.3 | 56 |
| 67 | 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, 255-261. | 1.8 | 54 |
| 68 | Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757. | 1.1 | 54 |
| 69 | Twelve novel mutations in the tissue-nonspecific alkaline phosphatase gene (ALPL) in patients with various forms of hypophosphatasia. Human Mutation, 2001, 18, 83-84. | 1.1 | 53 |
| 70 | Clinical characterisation of the multiple maternal hypomethylation syndrome in siblings. European Journal of Human Genetics, 2008, 16, 453-461. | 1.4 | 51 |
| 71 | Clinical utility gene card for: Beckwithâ€“Wiedemann Syndrome. European Journal of Human Genetics, 2014, 22, 435-435. | 1.4 | 50 |
| 72 | Human imprinting disorders: Principles, practice, problems and progress. European Journal of Medical Genetics, 2017, 60, 618-626. | 0.7 | 49 |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | Identification of mutations inTCOF1: Use of molecular analysis in the pre- and postnatal diagnosis of Treacher Collins syndrome. American Journal of Medical Genetics Part A, 2004, 127A, 244-248. | 2.4 | 48 |
| 74 | Pallister-Killian syndrome: a study of 22 British patients. Journal of Medical Genetics, 2015, 52, 454-464. | 1.5 | 45 |
| 75 | Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154. | 1.1 | 45 |
| 76 | Meierâ€“Gorlin syndrome: Growth and secondary sexual development of a microcephalic primordial dwarfism disorder. American Journal of Medical Genetics, Part A, 2012, 158A, 2733-2742. | 0.7 | 44 |
| 77 | Prenatal molecular testing for Beckwithâ€“Wiedemann and Silverâ€“Russell syndromes: a challenge for molecular analysis and genetic counseling. European Journal of Human Genetics, 2016, 24, 784-793. | 1.4 | 44 |
| 78 | 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-e56. | 1.5 | 42 |
| 79 | Early-Onset LBSL: How Severe Does It Get?. Neuropediatrics, 2012, 43, 332-338. | 0.3 | 41 |
| 80 | A survey ofTWIST for mutations in craniosynostosis reveals a variable length polyglycine tract in asymptomatic individuals. Human Mutation, 2001, 18, 535-541. | 1.1 | 39 |
| 81 | Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. Genome Research, 2019, 29, 1057-1066. | 2.4 | 38 |
| 82 | Mutations in phospholipase C epsilon 1 are not sufficient to cause diffuse mesangial sclerosis. Kidney International, 2009, 75, 415-419. | 2.6 | 35 |
| 83 | Deletions and rearrangements of the H19/IGF2 enhancer region in patients with Silver-Russell syndrome and growth retardation. Journal of Medical Genetics, 2011, 48, 308-311. | 1.5 | 35 |
| 84 | An atypical case of hypomethylation at multiple imprinted loci. European Journal of Human Genetics, 2011, 19, 360-362. | 1.4 | 35 |
| 85 | Evaluation of NSD2 and NSD3 in overgrowth syndromes. European Journal of Human Genetics, 2005, 13, 150-153. | 1.4 | 32 |
| 86 | Constitutional Haploinsufficiency of Tumor Suppressor Genes in Mentally Retarded Patients With Microdeletions in 17p13.1. Cytogenetic and Genome Research, 2009, 125, 1-7. | 0.6 | 32 |
| 87 | Hypoinsulinaemic, hypoketotic hypoglycaemia due to mosaic genetic activation of PI3-kinase. European Journal of Endocrinology, 2017, 177, 175-186. | 1.9 | 32 |
| 88 | 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, 716-720. | 1.4 | 31 |
| 89 | Molecular confirmation of germ line mosaicism for a submicroscopic deletion of chromosome 22q11. , 1998, 78, 103-106. | | 30 |
| 90 | Evidence for anticipation in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2013, 21, 1344-1348. | 1.4 | 30 |

| # | Article | IF | Citations |
|---|---|---|---|
| 91 | Imprinting in Human Disease with Special Reference to Transient Neonatal Diabetes and Beckwith-Wiedemann Syndrome. , 2007, 12, 113-123. | | 29 |
| 92 | Further refinement of the critical minimal genetic region for the imprinting disorder 6q24 transient neonatal diabetes. Diabetologia, 2010, 53, 2347-2351. | 2.9 | 29 |
| 93 | Genome-wide DNA methylation analysis of transient neonatal diabetes type 1 patients with mutations in ZFP57. BMC Medical Genetics, 2016, 17, 29. | 2.1 | 27 |
| 94 | A CASE OF MATERNAL UNIPARENTAL DISOMY OF CHROMOSOME 9 IN ASSOCIATION WITH CONFINED PLACENTAL MOSAICISM FOR TRISOMY 9. , 1996, 16, 371-374. | | 26 |
| 95 | Genetic heterogeneity in LEOPARD syndrome: two families with no mutations in PTPN11. Journal of Human Genetics, 2005, 50, 21-25. | 1.1 | 25 |
| 96 | Feasibility and acceptability of providing nurse counsellor genetics clinics in primary care. Journal of Advanced Nursing, 2006, 53, 591-604. | 1.5 | 25 |
| 97 | 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-e87. | 4.3 | 25 |
| 98 | Temple syndrome as a result of isolated hypomethylation of the 14q32 imprinted DLK1/MEG3 region. American Journal of Medical Genetics, Part A, 2016, 170, 170-175. | 0.7 | 25 |
| 99 | A familial disorder of altered DNA-methylation. Journal of Medical Genetics, 2014, 51, 407-412. | 1.5 | 24 |
| 100 | Congenital imprinting disorders: EUCID.net - a network to decipher their aetiology and to improve the diagnostic and clinical care. Clinical Epigenetics, 2015, 7, 23. | 1.8 | 23 |
| 101 | Protelomeric sequences are deleted in cases of short arm inverted duplication of chromosome 8. American Journal of Medical Genetics Part A, 1994, 50, 296-299. | 2.4 | 21 |
| 102 | DNA hypomethylation, transient neonatal diabetes, and prune belly sequence in one of two identical twins. European Journal of Pediatrics, 2010, 169, 207-213. | 1.3 | 21 |
| 103 | Clinical features and molecular analysis of seven British kindreds with hereditary hyperferritinaemia cataract syndrome. European Journal of Human Genetics, 2004, 12, 790-796. | 1.4 | 20 |
| 104 | Clinical utility gene card for: Transient Neonatal Diabetes Mellitus, 6q24-related. European Journal of Human Genetics, 2014, 22, 1153-1153. | 1.4 | 20 |
| 105 | Very small deletions within the NESP55 gene in pseudohypoparathyroidism type 1b. European Journal of Human Genetics, 2015, 23, 494-499. | 1.4 | 20 |
| 106 | Megalencephaly Syndromes: Exome Pipeline Strategies for Detecting Low-Level Mosaic Mutations. PLoS ONE, 2014, 9, e86940. | 1.1 | 20 |
| 107 | Familial occurrence of neonatal diabetes with duplications in chromosome 6q24: treatment with sulfonylurea and 40-yr follow-up. Pediatric Diabetes, 2012, 13, 155-162. | 1.2 | 19 |
| 108 | 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, 114-117. | 0.7 | 19 |

| # | Article | IF | Citations |
|---|---|---|---|
| 109 | Masked confirmation of linkage between type 1 congenital vitreous anomaly and COL 2A1 in Stickler syndrome. Graefe's Archive for Clinical and Experimental Ophthalmology, 1996, 234, 720-721. | 1.0 | 18 |
| 110 | A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. European Journal of Medical Genetics, 2014, 57, 587-595. | 0.7 | 18 |
| 111 | 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. | 1.8 | 18 |
| 112 | Phenotype of genetically confirmed Silver-Russell syndrome beyond childhood. Journal of Medical Genetics, 2020, 57, 683-691. | 1.5 | 18 |
| 113 | Frontonasal dysplasia or craniofrontonasal dysplasia and the Poland anomaly?. Clinical Genetics, 1990, 38, 233-236. | 1.0 | 17 |
| 114 | Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. Genetical Research, 2019, 101, e3. | 0.3 | 17 |
| 115 | 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. | 2.4 | 16 |
| 116 | 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, 340-346. | 0.7 | 16 |
| 117 | HIST1H1E heterozygous proteinâ€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. | 0.7 | 16 |
| 118 | Mosaic Paternal Uniparental Isodisomy and an ABCC8 Gene Mutation in a Patient With Permanent Neonatal Diabetes and Hemihypertrophy. Diabetes, 2008, 57, 255-258. | 0.3 | 15 |
| 119 | Familial Ebstein Anomaly. Circulation: Cardiovascular Genetics, 2017, 10, . | 5.1 | 14 |
| 120 | Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41. | 1.8 | 14 |
| 121 | Clinical utility gene card for: Silverâ€“Russell syndrome. European Journal of Human Genetics, 2011, 19, 3-3. | 1.4 | 13 |
| 122 | Lived experience of Silver-Russell syndrome: implications for management during childhood and into adulthood. Archives of Disease in Childhood, 2019, 104, 76-82. | 1.0 | 13 |
| 123 | Monozygous triplets discordant for transient neonatal diabetes mellitus and for imprinting of the TNDM differentially methylated region. Human Genetics, 2005, 117, 398-401. | 1.8 | 12 |
| 124 | A primary care specialist genetics service: a cluster-randomised factorial trial. British Journal of General Practice, 2012, 62, e191-e197. | 0.7 | 12 |
| 125 | Heterogeneity of the growth phenotype and birth size in acid-labile subunit (ALS) deficiency. Journal of Endocrinological Investigation, 2015, 38, 407-412. | 1.8 | 12 |
| 126 | ActivatingÂmutations in BRAFÂdisrupt the hypothalamo-pituitary axis leading to hypopituitarism in mice and humans. Nature Communications, 2021, 12, 2028. | 5.8 | 12 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | Neonatal diabetes mellitus. Pediatric Diabetes, 2002, 3, 109-112. | 1.2 | 11 |
| 128 | Microarrayâ€based DNA methylation analysis of imprinted loci in a patient with transient neonatal diabetes mellitus. American Journal of Medical Genetics, Part A, 2008, 146A, 3227-3229. | 0.7 | 11 |
| 129 | Growth Hormone Improves Short-Term Growth in Patients with Temple Syndrome. Hormone Research in Paediatrics, 2018, 90, 407-413. | 0.8 | 10 |
| 130 | A small intraexonic deletion within the dystrophin gene suggests a possible mechanism of mutagenesis. Human Genetics, 1997, 99, 658-662. | 1.8 | 9 |
| 131 | Nephrocalcinosis and disordered calcium metabolism in two children with SHORT syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 1296-1298. | 0.7 | 9 |
| 132 | Observationsï¿½Maturity 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, 924-924. | 2.9 | 8 |
| 133 | Persistent Fetal Vasculature and Severe Protein C Deficiency. Molecular Syndromology, 2010, 1, 82-86. | 0.3 | 8 |
| 134 | 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. | 0.7 | 8 |
| 135 | Whole-genome analysis as a diagnostic tool for patients referred for diagnosis of Silver-Russell syndrome: a real-world study. Journal of Medical Genetics, 2022, 59, 613-622. | 1.5 | 8 |
| 136 | A female case of Sedaghatian type spondylometaphyseal dysplasia. American Journal of Medical Genetics Part A, 2003, 118A, 377-381. | 2.4 | 6 |
| 137 | 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, 1990-1993. | 0.7 | 6 |
| 138 | Experiences of adolescents living with Silver-Russell syndrome. Archives of Disease in Childhood, 2021, 106, 1195-1201. | 1.0 | 6 |
| 139 | A patient with multilocus imprinting disturbance involving hypomethylation at 11p15 and 14q32, and phenotypic features of <scp>Beckwithâ€Wiedemann</scp> and Temple syndromes. American Journal of Medical Genetics, Part A, 2022, , . | 0.7 | 5 |
| 140 | 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, 1063-1064. | 2.1 | 4 |
| 141 | Atypical chondrodysplasia: a further variant of multiple enchondromatosis with vertebral involvement?. Pediatric Radiology, 1998, 28, 963-966. | 1.1 | 3 |
| 142 | Coat hanger appearances of the ribs: a useful diagnostic marker of paternal uniparental disomy of chromosome 14. Archives of Disease in Childhood, 2010, 95, 909-909. | 1.0 | 3 |
| 143 | Successful pregnancies in an adult with <scp>Meierâ€Gorlin</scp> syndrome harboring biallelic <scp>CDT1</scp> variants. American Journal of Medical Genetics, Part A, 2021, 185, 871-876. | 0.7 | 3 |
| 144 | Height and body mass index in molecularly confirmed Silverâ€“Russell syndrome and the longâ€term effects of growth hormone treatment. Clinical Endocrinology, 2022, 97, 284-292. | 1.2 | 3 |

| # | Article | IF | Citations |
|---|---|---|---|
| 145 | Ongoing Challenges in the Diagnosis of 11p15.5-Associated Imprinting Disorders. Molecular Diagnosis and Therapy, 2022, 26, 263-272. | 1.6 | 3 |
| 146 | Falling short? The psychosocial impact of living with Russell-Silver syndrome. Journal of Aesthetic Nursing, 2016, 5, 340-342. | 0.0 | 2 |
| 147 | Silver Russell syndrome in a preterm girl with 8q12.1 deletion encompassing PLAG1. Clinical Dysmorphology, 2021, 30, 194-196. | 0.1 | 2 |
| 148 | Skin spots and heart tumors. Journal of Pediatrics, 2001, 139, 901-902. | 0.9 | 1 |
| 149 | Imprinting Disorders of Early Childhood. , 2012, , 137-160. | | 1 |
| 150 | Beyond the gene roundtable discussion. Textual Practice, 2015, 29, 415-432. | 0.4 | 1 |
| 151 | Genetics, molar pregnancies and medieval ideas of monstrous births: the lump of flesh in The King of Tars. Medical Humanities, 2019, 45, 2-9. | 0.6 | 1 |
| 152 | Transient Neonatal Diabetes Mellitus. , 2007, 17, 169-171. | | 0 |