Leslie D Kulikowski

List of Publications by Year
in descending order

79	830	623188 14	642321 23
papers	citations	h-index	g-index

86	86	86	1553
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Novel FERMT3 and PTPRQ Mutations Associated with Leukocyte Adhesion Deficiency-III and Sensorineural Hearing Loss. Journal of Pediatric Genetics, 2023, 12, 348-351.	0.3	0
2	<scp>Cardiovascular findings in Williamsâ€“Beuren Syndrome</scp>: Experience of a single center with 127 cases. American Journal of Medical Genetics, Part A, 2022, 188, 676-682.	0.7	4
3	Beyond Midline: Diffuse Hemispheric Glioma, H3 K27M-Mutant with Aggressive Behavior. Journal of Neuropathology and Experimental Neurology, 2022, 81, 381-383.	0.9	2
4	The complex search for the cause of gastroschisis. Birth Defects Research, 2022, 114, 1291-1297.	0.8	1
5	Cri-du-Chat Syndrome: Revealing a Familial Atypical Deletion in 5p. Molecular Syndromology, 2022, 13, 527-536.	0.3	0
6	Novel rearrangements between different chromosomes with direct impact on the diagnosis of 5p- syndrome. Clinics, 2022, 77, 100045.	0.6	2
7	Genetic analysis of products of conception. Should we abandon classic karyotyping methodology?. Einstein (Sao Paulo, Brazil), 2021, 19, eAO5945.	0.3	3
8	Fetal gastroschisis: Maternal and fetal methylation profile. Prenatal Diagnosis, 2021, 41, 449-456.	1.1	2
9	Breakpoint delineation in 5pâ€•patients leads to new insights about microcephaly and the typical highâ€pitched cry. Molecular Genetics & Genomic Medicine, 2020, 8, e957.	0.6	5
10	Intragenic variants in the SMN1 gene determine the clinical phenotype in 5q spinal muscular atrophy. Neurology: Genetics, 2020, 6, e505.	0.9	24
11	Comprehensive Genetic Analysis of 128 Candidate Genes in a Cohort With Idiopathic, Severe, or Familial Osteoporosis. Journal of the Endocrine Society, 2020, 4, bvaa148.	0.1	11
12	Lymphoproliferative disorder with polyautoimmunity and hypogammaglobulinemia: An unusual presentation of 22q11.2 deletion syndrome. Clinical Immunology, 2020, 220, 108590.	1.4	1
13	Efficacy of MLPA for detection of Y-chromosome microdeletions in infertile Brazilian patients. Journal of Assisted Reproduction and Genetics, 2020, 37, 1251-1259.	1.2	8
14	Application of Whole-Exome Sequencing in Detecting Copy Number Variants in Patients with Developmental Delay and/or Multiple Congenital Malformations. Journal of Molecular Diagnostics, 2020, 22, 1041-1049.	1.2	9
15	Case of 15q26-qter deletion associated with a Prader-Willi phenotype. European Journal of Medical Genetics, 2020, 63, 103955.	0.7	1
16	Gene expression changes associated with trajectories of psychopathology in a longitudinal cohort of children and adolescents. Translational Psychiatry, 2020, 10, 99.	2.4	3
17	The molecular landscape of osteogenesis imperfecta in a Brazilian tertiary service cohort. Osteoporosis International, 2020, 31, 1341-1352.	1.3	5
18	Clinical features of collagen VI-related dystrophies: A large Brazilian cohort. Clinical Neurology and Neurosurgery, 2020, 192, 105734.	0.6	7

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19	Gene expression profile suggesting immunological dysregulation in two Brazilian Bloom's syndrome cases. Molecular Genetics & Genomic Medicine, 2020, 8, e1133.	0.6	7
20	The Iberian legacy into a young genetic xeroderma pigmentosum cluster in central Brazil. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 2020, 852, 503164.	0.9	2
21	Identifying NAHR mechanism between two distinct Alu elements through breakpoint junction mapping by NGS. Meta Gene, 2020, 24, 100702.	0.3	0
22	Severe brain involvement in 5q spinal muscular atrophy type 0. Annals of Neurology, 2019, 86, 458-462.	2.8	31
23	Reply to â€œGlobal Central Nervous System Atrophy in Spinal Muscular Atrophy Type 0â€: Annals of Neurology, 2019, 86, 803-803.	2.8	0
24	EVALUATION OF GENE EXPRESSION IN EARLY SUBSTANCE ABUSE. European Neuropsychopharmacology, 2019, 29, S884-S885.	0.3	0
25	Mosaic Trisomy 12 Associated with Overgrowth Detected in Fibroblast Cell Lines. Cytogenetic and Genome Research, 2019, 157, 153-157.	0.6	12
26	Global DNA methylation of peripheral blood leukocytes from dogs bearing multicentric non-Hodgkin lymphomas and healthy dogs: A comparative study. PLoS ONE, 2019, 14, e0211898.	1.1	12
27	Deletion of RUNX1 exons 1 and 2 associated with familial platelet disorder with propensity to acute myeloid leukemia. Cancer Genetics, 2018, 222-223, 32-37.	0.2	22
28	Inactivation of AMMECR1 is associated with growth, bone, and heart alterations. Human Mutation, 2018, 39, 281-291.	1.1	15
29	A Multicentric Brazilian Investigative Study of Copy Number Variations in Patients with Congenital Anomalies and Intellectual Disability. Scientific Reports, 2018, 8, 13382.	1.6	1
30	Gestational Tubal Choriocarcinoma Presenting as a Pregnancy of Unknown Location following Ovarian Induction. Case Reports in Obstetrics and Gynecology, 2018, 2018, 1-6.	0.2	2
31	Cytogenomic assessment of the diagnosis of 93 patients with developmental delay and multiple congenital abnormalities: The Brazilian experience. Clinics, 2017, 72, 526-537.	0.6	10
32	Post-mortem cytogenomic investigations in patients with congenital malformations. Experimental and Molecular Pathology, 2016, 101, 116-123.	0.9	5
33	Subtelomeric Copy Number Variations: The Importance of 4p/4q Deletions in Patients with Congenital Anomalies and Developmental Disability. Cytogenetic and Genome Research, 2016, 149, 241-246.	0.6	6
34	Position effect modifying gene expression in a patient with ring chromosome 14. Journal of Applied Genetics, 2016, 57, 183-187.	1.0	12
35	Rare genomic rearrangement in a boy with Williamsâ€“Beuren syndrome associated to XYY syndrome and intriguing behavior. American Journal of Medical Genetics, Part A, 2015, 167, 3197-3203.	0.7	2
36	Cytogenomic delineation and clinical follow-up of 10 Brazilian patients with Pallister-Killian syndrome. Molecular Cytogenetics, 2015, 8, 43.	0.4	10

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37	Attention Deficit Hyperactivity Disorder in the Light of the Epigenetic Paradigm. Frontiers in Psychiatry, 2015, 6, 126.	1.3	49
38	Williams-Beuren Syndrome: A Clinical Study of 55 Brazilian Patients and the Diagnostic Use of MLPA. BioMed Research International, 2015, 2015, 1-6.	0.9	13
39	Terminal 18q deletions are stabilized by neotelomeres. Molecular Cytogenetics, 2015, 8, 32.	0.4	12
40	45,X Karyotype in an Infertile Man: How Is This Possible?. Urologia Internationalis, 2015, 94, 488-490.	0.6	2
41	Duplication 9p and their implication to phenotype. BMC Medical Genetics, 2014, 15, 142.	2.1	32
42	Clinical, cytogenetic, and molecular characterization of six patients with ring chromosomes 22, including one with concomitant 22q11.2 deletion. American Journal of Medical Genetics, Part A, 2014, 164, 1659-1665.	0.7	16
43	Complex structural rearrangement features suggesting chromoanagenesis mechanism in a case of 1p36 deletion syndrome. Molecular Genetics and Genomics, 2014, 289, 1037-1043.	1.0	13
44	NK and B cell deficiency in a MPS type II family with novel mutation in the IDS gene. Clinical Immunology, 2014, 154, 100-104.	1.4	3
45	Trisomy 1q32 and monosomy 11q25 associated with congenital heart defect: cytogenomic delineation and patient fourteen years follow-up. Molecular Cytogenetics, 2014, 7, 57.	0.4	3
46	Complex small supernumerary marker chromosome with a 15q/16p duplication: clinical implications. Molecular Cytogenetics, 2014, 7, 29.	0.4	3
47	Investigation of copy number variation in children with conotruncal heart defects. Arquivos Brasileiros De Cardiologia, 2014, 104, 24-31.	0.3	14
48	A Novel Mutation in HPRT1 Gene Causing Variant Form of Lesch-Nyhan Disease. Pediatric Neurology, 2013, 49, e5-e7.	1.0	0
49	Single-Nucleotide Polymorphism Array-Based Characterization of Ring Chromosome 18. Journal of Pediatrics, 2013, 163, 1174-1178.e3.	0.9	8
50	Disruption of the CREBBP gene and decreased expression of CREB, NFÎºB p65, c-JUN, c-FOS, BCL2 and c-MYC suggest immune dysregulation. Human Immunology, 2013, 74, 911-915.	1.2	6
51	Discrepant outcomes in two Brazilian patients with Bloom syndrome and Wilmsâ€™ tumor: two case reports. Journal of Medical Case Reports, 2013, 7, 284.	0.4	9
52	Ring chromosome 10: report on two patients and review of the literature. Journal of Applied Genetics, 2013, 54, 35-41.	1.0	10
53	Efficacy of two fluorescence in situ hybridization (FISH) probes for diagnosing malignant pleural effusions. Lung Cancer, 2013, 80, 284-288.	0.9	8
54	First Report of a Small Supernumerary der(8;14) Marker Chromosome. Cytogenetic and Genome Research, 2013, 139, 284-288.	0.6	2

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55	Atypical Deletion in Williamsâ€“Beuren Syndrome Critical Region Detected by MLPA in a Patient with Supravalvular Aortic Stenosis and Learning Difficulty. Journal of Genetics and Genomics, 2012, 39, 571-574.	1.7	9
56	Cytogenomic characterization of an unexpected 17.6Mb 9p deletion associated to a 14.8Mb 20p duplication in a dysmorphic patient with multiple congenital anomalies presenting a normal G-banding karyotype. Gene, 2012, 496, 59-62.	1.0	5
57	Wide Clinical Variability in Cat Eye Syndrome Patients: Four Non-Related Patients and Three Patients from the Same Family. Cytogenetic and Genome Research, 2012, 138, 5-10.	0.6	4
58	Role of SNAP29, LZTR1 and P2RXL1 genes on immune regulation in a patient with atypical 0.5Mb deletion in 22q11.2 region. Clinical Immunology, 2012, 145, 55-58.	1.4	6
59	Different Conformation of Two Supernumerary 18p Isochromosomes, One with a Concomitant Partial 18q Trisomy. Cytogenetic and Genome Research, 2012, 138, 1-4.	0.6	3
60	Twenty-year cytogenetic and molecular follow-up of a patient with ring chromosome 15: a case report. Journal of Medical Case Reports, 2012, 6, 283.	0.4	10
61	Copy number variation in Williams-Beuren syndrome: suitable diagnostic strategy for developing countries. BMC Research Notes, 2012, 5, 13.	0.6	19
62	Cytogenetic instability of dental pulp stem cell lines. Journal of Molecular Histology, 2012, 43, 89-94.	1.0	16
63	Mechanisms of ring chromosome formation, ring instability and clinical consequences. BMC Medical Genetics, 2011, 12, 171.	2.1	106
64	Clinical, Cytogenetic and Molecular Study in a Case of r(3) with 3p Deletion and Review of the Literature. Cytogenetic and Genome Research, 2011, 134, 325-330.	0.6	7
65	Subtelomeric rearrangements and copy number variations in people with intellectual disabilities. Journal of Intellectual Disability Research, 2010, 54, 938-942.	1.2	10
66	A rare case of trisomy 15pterâ€q21.2 due to a de novo marker chromosome. American Journal of Medical Genetics, Part A, 2010, 152A, 753-758.	0.7	7
67	Trisomy 16q21â€‰â†’â€‰qter: Sevenâ€year followâ€up of a girl with unusually long survival. American Journal of Medical Genetics, Part A, 2010, 152A, 2074-2078.	0.7	4
68	Cytogenetic and molecular evaluation and 20â€year followâ€up of a patient with ring chromosome 14. American Journal of Medical Genetics, Part A, 2010, 152A, 2865-2869.	0.7	12
69	Cytogenetic molecular delineation of a terminal 18q deletion suggesting neo-telomere formation. European Journal of Medical Genetics, 2010, 53, 404-407.	0.7	5
70	Ring chromosome instability evaluation in six patients with autosomal rings. Genetics and Molecular Research, 2010, 9, 134-143.	0.3	50
71	DeleÃ§Ã£o 22q11.2 em pacientes com defeito cardÃaco conotruncal e fenÃ³tipo da sÃndrome da deleÃ§Ã£o 22q11.2. Arquivos Brasileiros De Cardiologia, 2009, 92, 307-311.	0.3	13
72	Evaluation of clinical checklists for fragile X syndrome screening in Brazilian intellectually disabled males. Journal of Intellectual Disabilities, 2009, 13, 239-248.	1.0	9

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73	Partial 5p monosomy or trisomy in 11 patients from a family with a t(5;15)(p13.3;p12) translocation. Human Genetics, 2008, 124, 387-392.	1.8	14
74	Pure duplication 1q41â€qter: Further delineation of trisomy 1q syndromes. American Journal of Medical Genetics, Part A, 2008, 146A, 2663-2667.	0.7	24
75	Atypical 22q11.2 deletion in a patient with DGS/VCFS spectrum. European Journal of Medical Genetics, 2008, 51, 226-230.	0.7	15
76	Clinical checklists in the selection of mentally retarded males for molecular screening of fragile X syndrome. Genetics and Molecular Biology, 2007, 30, 1047-1050.	0.6	1
77	Deletion 22q11.2: Report of a complex meiotic mechanism of origin. American Journal of Medical Genetics, Part A, 2007, 143A, 1778-1781.	0.7	1
78	Breakpoint mapping in a case of mosaicism with partial monosomy 9p23 â†' pter and partial trisomy 1q41 â†' qter suggests neo-telomere formation in stabilizing the deleted chromosome. American Journal of Medical Genetics, Part A, 2006, 140A, 82-87.	0.7	13
79	Intellectual performance profi le of a sample of children and adolescents from Brazil with 22q11.2 Deletion Syndrome (22q11.2DS) based on the Wechsler Scale. Estudos De Psicologia (Campinas), 0, 36, .	0.8	0