

# Leslie D Kulikowski

## List of Publications by Year in descending order

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79  
papers

830  
citations

623188

14  
h-index

642321

23  
g-index

86  
all docs

86  
docs citations

86  
times ranked

1553  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mechanisms of ring chromosome formation, ring instability and clinical consequences. BMC Medical Genetics, 2011, 12, 171.	2.1	106
2	Ring chromosome instability evaluation in six patients with autosomal rings. Genetics and Molecular Research, 2010, 9, 134-143.	0.3	50
3	Attention Deficit Hyperactivity Disorder in the Light of the Epigenetic Paradigm. Frontiers in Psychiatry, 2015, 6, 126.	1.3	49
4	Duplication 9p and their implication to phenotype. BMC Medical Genetics, 2014, 15, 142.	2.1	32
5	Severe brain involvement in 5q spinal muscular atrophy type 0. Annals of Neurology, 2019, 86, 458-462.	2.8	31
6	Pure duplication 1q41â€”qter: Further delineation of trisomy 1q syndromes. American Journal of Medical Genetics, Part A, 2008, 146A, 2663-2667.	0.7	24
7	Intragenic variants in the <i>SMN1</i> gene determine the clinical phenotype in 5q spinal muscular atrophy. Neurology: Genetics, 2020, 6, e505.	0.9	24
8	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
9	Copy number variation in Williams-Beuren syndrome: suitable diagnostic strategy for developing countries. BMC Research Notes, 2012, 5, 13.	0.6	19
10	Cytogenetic instability of dental pulp stem cell lines. Journal of Molecular Histology, 2012, 43, 89-94.	1.0	16
11	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
12	Atypical 22q11.2 deletion in a patient with DGS/VCFS spectrum. European Journal of Medical Genetics, 2008, 51, 226-230.	0.7	15
13	Inactivation of <i>AMMECR1</i> is associated with growth, bone, and heart alterations. Human Mutation, 2018, 39, 281-291.	1.1	15
14	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
15	Investigation of copy number variation in children with conotruncal heart defects. Arquivos Brasileiros De Cardiologia, 2014, 104, 24-31.	0.3	14
16	Breakpoint mapping in a case of mosaicism with partial monosomy 9p23â€”pter and partial trisomy 1q41â€”qter suggests neo-telomere formation in stabilizing the deleted chromosome. American Journal of Medical Genetics, Part A, 2006, 140A, 82-87.	0.7	13
17	DeleÃ§Ã£o 22q11.2 em pacientes com defeito cardÃ¡cio conotruncal e fenÃ³tipo da sÃndrome da deleÃ§Ã£o 22q11.2. Arquivos Brasileiros De Cardiologia, 2009, 92, 307-311.	0.3	13
18	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

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19	Williams-Beuren Syndrome: A Clinical Study of 55 Brazilian Patients and the Diagnostic Use of MLPA. <i>BioMed Research International</i> , 2015, 2015, 1-6.	0.9	13
20	Cytogenetic and molecular evaluation and 20-year follow-up of a patient with ring chromosome 14. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2865-2869.	0.7	12
21	Terminal 18q deletions are stabilized by neotelomeres. <i>Molecular Cytogenetics</i> , 2015, 8, 32.	0.4	12
22	Position effect modifying gene expression in a patient with ring chromosome 14. <i>Journal of Applied Genetics</i> , 2016, 57, 183-187.	1.0	12
23	Mosaic Trisomy 12 Associated with Overgrowth Detected in Fibroblast Cell Lines. <i>Cytogenetic and Genome Research</i> , 2019, 157, 153-157.	0.6	12
24	Global DNA methylation of peripheral blood leukocytes from dogs bearing multicentric non-Hodgkin lymphomas and healthy dogs: A comparative study. <i>PLoS ONE</i> , 2019, 14, e0211898.	1.1	12
25	Comprehensive Genetic Analysis of 128 Candidate Genes in a Cohort With Idiopathic, Severe, or Familial Osteoporosis. <i>Journal of the Endocrine Society</i> , 2020, 4, bvaa148.	0.1	11
26	Subtelomeric rearrangements and copy number variations in people with intellectual disabilities. <i>Journal of Intellectual Disability Research</i> , 2010, 54, 938-942.	1.2	10
27	Twenty-year cytogenetic and molecular follow-up of a patient with ring chromosome 15: a case report. <i>Journal of Medical Case Reports</i> , 2012, 6, 283.	0.4	10
28	Ring chromosome 10: report on two patients and review of the literature. <i>Journal of Applied Genetics</i> , 2013, 54, 35-41.	1.0	10
29	Cytogenomic delineation and clinical follow-up of 10 Brazilian patients with Pallister-Killian syndrome. <i>Molecular Cytogenetics</i> , 2015, 8, 43.	0.4	10
30	Cytogenomic assessment of the diagnosis of 93 patients with developmental delay and multiple congenital abnormalities: The Brazilian experience. <i>Clinics</i> , 2017, 72, 526-537.	0.6	10
31	Evaluation of clinical checklists for fragile X syndrome screening in Brazilian intellectually disabled males. <i>Journal of Intellectual Disabilities</i> , 2009, 13, 239-248.	1.0	9
32	Atypical Deletion in Williams-Beuren Syndrome Critical Region Detected by MLPA in a Patient with Supravalvular Aortic Stenosis and Learning Difficulty. <i>Journal of Genetics and Genomics</i> , 2012, 39, 571-574.	1.7	9
33	Discrepant outcomes in two Brazilian patients with Bloom syndrome and Wilms tumor: two case reports. <i>Journal of Medical Case Reports</i> , 2013, 7, 284.	0.4	9
34	Application of Whole-Exome Sequencing in Detecting Copy Number Variants in Patients with Developmental Delay and/or Multiple Congenital Malformations. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 1041-1049.	1.2	9
35	Single-Nucleotide Polymorphism Array-Based Characterization of Ring Chromosome 18. <i>Journal of Pediatrics</i> , 2013, 163, 1174-1178.e3.	0.9	8
36	Efficacy of two fluorescence in situ hybridization (FISH) probes for diagnosing malignant pleural effusions. <i>Lung Cancer</i> , 2013, 80, 284-288.	0.9	8

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37	Efficacy of MLPA for detection of Y-chromosome microdeletions in infertile Brazilian patients. <i>Journal of Assisted Reproduction and Genetics</i> , 2020, 37, 1251-1259.	1.2	8
38	A rare case of trisomy 15pterâ€”q21.2 due to a de novo marker chromosome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 753-758.	0.7	7
39	Clinical, Cytogenetic and Molecular Study in a Case of r(3) with 3p Deletion and Review of the Literature. <i>Cytogenetic and Genome Research</i> , 2011, 134, 325-330.	0.6	7
40	Clinical features of collagen VI-related dystrophies: A large Brazilian cohort. <i>Clinical Neurology and Neurosurgery</i> , 2020, 192, 105734.	0.6	7
41	Gene expression profile suggesting immunological dysregulation in two Brazilian Bloom's syndrome cases. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1133.	0.6	7
42	Role of SNAP29, LZTR1 and P2RXL1 genes on immune regulation in a patient with atypical 0.5Mb deletion in 22q11.2 region. <i>Clinical Immunology</i> , 2012, 145, 55-58.	1.4	6
43	Disruption of the CREBBP gene and decreased expression of CREB, NFÎ”B p65, c-JUN, c-FOS, BCL2 and c-MYC suggest immune dysregulation. <i>Human Immunology</i> , 2013, 74, 911-915.	1.2	6
44	Subtelomeric Copy Number Variations: The Importance of 4p/4q Deletions in Patients with Congenital Anomalies and Developmental Disability. <i>Cytogenetic and Genome Research</i> , 2016, 149, 241-246.	0.6	6
45	Cytogenetic molecular delineation of a terminal 18q deletion suggesting neo-telomere formation. <i>European Journal of Medical Genetics</i> , 2010, 53, 404-407.	0.7	5
46	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. <i>Gene</i> , 2012, 496, 59-62.	1.0	5
47	Post-mortem cytogenomic investigations in patients with congenital malformations. <i>Experimental and Molecular Pathology</i> , 2016, 101, 116-123.	0.9	5
48	Breakpoint delineation in 5pâ€”patients leads to new insights about microcephaly and the typical highâ€”pitched cry. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e957.	0.6	5
49	The molecular landscape of osteogenesis imperfecta in a Brazilian tertiary service cohort. <i>Osteoporosis International</i> , 2020, 31, 1341-1352.	1.3	5
50	Trisomy 16q21â€”qter: Sevenâ€”year followâ€”up of a girl with unusually long survival. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2074-2078.	0.7	4
51	Wide Clinical Variability in Cat Eye Syndrome Patients: Four Non-Related Patients and Three Patients from the Same Family. <i>Cytogenetic and Genome Research</i> , 2012, 138, 5-10.	0.6	4
52	<scp>Cardiovascular findings in Williamsâ€”Beuren Syndrome</scp>: Experience of a single center with 127 cases. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 676-682.	0.7	4
53	Different Conformation of Two Supernumerary 18p Isochromosomes, One with a Concomitant Partial 18q Trisomy. <i>Cytogenetic and Genome Research</i> , 2012, 138, 1-4.	0.6	3
54	NK and B cell deficiency in a MPS type II family with novel mutation in the IDS gene. <i>Clinical Immunology</i> , 2014, 154, 100-104.	1.4	3

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55	Trisomy 1q32 and monosomy 11q25 associated with congenital heart defect: cytogenomic delineation and patient fourteen years follow-up. <i>Molecular Cytogenetics</i> , 2014, 7, 57.	0.4	3
56	Complex small supernumerary marker chromosome with a 15q/16p duplication: clinical implications. <i>Molecular Cytogenetics</i> , 2014, 7, 29.	0.4	3
57	Gene expression changes associated with trajectories of psychopathology in a longitudinal cohort of children and adolescents. <i>Translational Psychiatry</i> , 2020, 10, 99.	2.4	3
58	Genetic analysis of products of conception. Should we abandon classic karyotyping methodology?. <i>Einstein (Sao Paulo, Brazil)</i> , 2021, 19, eAO5945.	0.3	3
59	First Report of a Small Supernumerary der(8;14) Marker Chromosome. <i>Cytogenetic and Genome Research</i> , 2013, 139, 284-288.	0.6	2
60	Rare genomic rearrangement in a boy with Williams's Beuren syndrome associated to XYY syndrome and intriguing behavior. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3197-3203.	0.7	2
61	45,X Karyotype in an Infertile Man: How Is This Possible?. <i>Urologia Internationalis</i> , 2015, 94, 488-490.	0.6	2
62	Gestational Tubal Choriocarcinoma Presenting as a Pregnancy of Unknown Location following Ovarian Induction. <i>Case Reports in Obstetrics and Gynecology</i> , 2018, 2018, 1-6.	0.2	2
63	The Iberian legacy into a young genetic xeroderma pigmentosum cluster in central Brazil. <i>Mutation Research - Genetic Toxicology and Environmental Mutagenesis</i> , 2020, 852, 503164.	0.9	2
64	Fetal gastroschisis: Maternal and fetal methylation profile. <i>Prenatal Diagnosis</i> , 2021, 41, 449-456.	1.1	2
65	Beyond Midline: Diffuse Hemispheric Glioma, H3 K27M-Mutant with Aggressive Behavior. <i>Journal of Neuropathology and Experimental Neurology</i> , 2022, 81, 381-383.	0.9	2
66	Novel rearrangements between different chromosomes with direct impact on the diagnosis of 5p-syndrome. <i>Clinics</i> , 2022, 77, 100045.	0.6	2
67	Clinical checklists in the selection of mentally retarded males for molecular screening of fragile X syndrome. <i>Genetics and Molecular Biology</i> , 2007, 30, 1047-1050.	0.6	1
68	Deletion 22q11.2: Report of a complex meiotic mechanism of origin. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1778-1781.	0.7	1
69	A Multicentric Brazilian Investigative Study of Copy Number Variations in Patients with Congenital Anomalies and Intellectual Disability. <i>Scientific Reports</i> , 2018, 8, 13382.	1.6	1
70	Lymphoproliferative disorder with polyautoimmunity and hypogammaglobulinemia: An unusual presentation of 22q11.2 deletion syndrome. <i>Clinical Immunology</i> , 2020, 220, 108590.	1.4	1
71	Case of 15q26-qter deletion associated with a Prader-Willi phenotype. <i>European Journal of Medical Genetics</i> , 2020, 63, 103955.	0.7	1
72	The complex search for the cause of gastroschisis. <i>Birth Defects Research</i> , 2022, 114, 1291-1297.	0.8	1

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73	A Novel Mutation in HPRT1 Gene Causing Variant Form of Lesch-Nyhan Disease. <i>Pediatric Neurology</i> , 2013, 49, e5-e7.	1.0	0
74	Reply to "Global Central Nervous System Atrophy in Spinal Muscular Atrophy Type 0". <i>Annals of Neurology</i> , 2019, 86, 803-803.	2.8	0
75	EVALUATION OF GENE EXPRESSION IN EARLY SUBSTANCE ABUSE. <i>European Neuropsychopharmacology</i> , 2019, 29, S884-S885.	0.3	0
76	Identifying NAHR mechanism between two distinct Alu elements through breakpoint junction mapping by NGS. <i>Meta Gene</i> , 2020, 24, 100702.	0.3	0
77	Novel FERMT3 and PTPRQ Mutations Associated with Leukocyte Adhesion Deficiency-III and Sensorineural Hearing Loss. <i>Journal of Pediatric Genetics</i> , 2023, 12, 348-351.	0.3	0
78	Intellectual performance profile of a sample of children and adolescents from Brazil with 22q11.2 Deletion Syndrome (22q11.2DS) based on the Wechsler Scale. <i>Estudos De Psicologia (Campinas)</i> , 0, 36, .	0.8	0
79	Cri-du-Chat Syndrome: Revealing a Familial Atypical Deletion in 5p. <i>Molecular Syndromology</i> , 2022, 13, 527-536.	0.3	0