

Ian D Krantz

List of Publications by Year in descending order

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Version: 2024-02-01

83
papers

6,527
citations

185998

28
h-index

69108

77
g-index

93
all docs

93
docs citations

93
times ranked

8173
citing authors

#	ARTICLE	IF	CITATIONS
1	Alagille syndrome is caused by mutations in human Jagged1, which encodes a ligand for Notch1. <i>Nature Genetics</i> , 1997, 16, 243-251.	9.4	1,184
2	Mutations in the human Jagged1 gene are responsible for Alagille syndrome. <i>Nature Genetics</i> , 1997, 16, 235-242.	9.4	1,072
3	KILLER/DR5 is a DNA damage-induced p53-regulated death receptor gene. <i>Nature Genetics</i> , 1997, 17, 141-143.	9.4	1,005
4	Features of alagille syndrome in 92 patients: Frequency and relation to prognosis. <i>Hepatology</i> , 1999, 29, 822-829.	3.6	591
5	USP7 Acts as a Molecular Rheostat to Promote WASH-Dependent Endosomal Protein Recycling and Is Mutated in a Human Neurodevelopmental Disorder. <i>Molecular Cell</i> , 2015, 59, 956-969.	4.5	175
6	International electronic health record-derived COVID-19 clinical course profiles: the 4CE consortium. <i>Npj Digital Medicine</i> , 2020, 3, 109.	5.7	128
7	Recommendations for the integration of genomics into clinical practice. <i>Genetics in Medicine</i> , 2016, 18, 1075-1084.	1.1	125
8	A Cohesin-Independent Role for NIPBL at Promoters Provides Insights in CdLS. <i>PLoS Genetics</i> , 2014, 10, e1004153.	1.5	123
9	Germline gain-of-function mutations in AFF4 cause a developmental syndrome functionally linking the super elongation complex and cohesin. <i>Nature Genetics</i> , 2015, 47, 338-344.	9.4	109
10	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. <i>European Journal of Human Genetics</i> , 2015, 23, 1473-1481.	1.4	101
11	Dominant paternal transmission of Cornelia de Lange syndrome: A new case and review of 25 previously reported familial recurrences. <i>American Journal of Medical Genetics Part A</i> , 2001, 104, 267-276.	2.4	91
12	A taxonomy of medical uncertainties in clinical genome sequencing. <i>Genetics in Medicine</i> , 2017, 19, 918-925.	1.1	91
13	Alagille syndrome mutation update: Comprehensive overview of <i>JAG1</i> and <i>NOTCH2</i> mutation frequencies and insight into missense variant classification. <i>Human Mutation</i> , 2019, 40, 2197-2220.	1.1	84
14	Effect of Whole-Genome Sequencing on the Clinical Management of Acutely Ill Infants With Suspected Genetic Disease. <i>JAMA Pediatrics</i> , 2021, 175, 1218.	3.3	83
15	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2019, 104, 1060-1072.	2.6	78
16	Mutation analysis of Jagged1 (JAG1) in Alagille syndrome patients. <i>Human Mutation</i> , 2001, 17, 151-152.	1.1	76
17	Deletions of 20p12 in Alagille syndrome: Frequency and molecular characterization. <i>Am J Hum Genet</i> , 1997, 70, 80-86.		71
18	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. <i>Genetics in Medicine</i> , 2016, 18, 309-315.	1.1	69

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19	Phenotypes and genotypes in individuals with <i>SMC1A</i> variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	0.7	69
20	ESRP1 Mutations Cause Hearing Loss due to Defects in Alternative Splicing that Disrupt Cochlear Development. Developmental Cell, 2017, 43, 318-331.e5.	3.1	68
21	Automated Clinical Exome Reanalysis Reveals Novel Diagnoses. Journal of Molecular Diagnostics, 2019, 21, 38-48.	1.2	68
22	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	2.6	61
23	Missense variants in the chromatin remodeler <i>CHD1</i> are associated with neurodevelopmental disability. Journal of Medical Genetics, 2018, 55, 561-566.	1.5	49
24	Heterozygous Deletion of <i>FOXA2</i> Segregates with Disease in a Family with Heterotaxy, Panhypopituitarism, and Biliary Atresia. Human Mutation, 2015, 36, 631-637.	1.1	43
25	Pathogenic variants in <i>USP7</i> cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	1.1	41
26	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	0.7	40
27	Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431.	1.4	37
28	Outcomes of evaluation and testing of 660 individuals with hearing loss in a pediatric genetics of hearing loss clinic. American Journal of Medical Genetics, Part A, 2016, 170, 2523-2530.	0.7	37
29	De Novo Variants in <i>CNOT1</i> , a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	2.6	37
30	Diagnosing Cornelia de Lange syndrome and related neurodevelopmental disorders using RNA sequencing. Genetics in Medicine, 2020, 22, 927-936.	1.1	34
31	Drosophila Nipped-B Mutants Model Cornelia de Lange Syndrome in Growth and Behavior. PLoS Genetics, 2015, 11, e1005655.	1.5	33
32	International Analysis of Electronic Health Records of Children and Youth Hospitalized With COVID-19 Infection in 6 Countries. JAMA Network Open, 2021, 4, e2112596.	2.8	33
33	The Genomics Research and Innovation Network: creating an interoperable, federated, genomics learning system. Genetics in Medicine, 2020, 22, 371-380.	1.1	30
34	Disruption of cardiac thin filament assembly arising from a mutation in <i>LMOD2</i> : A novel mechanism of neonatal dilated cardiomyopathy. Science Advances, 2019, 5, eaax2066.	4.7	29
35	NIPBL Controls RNA Biogenesis to Prevent Activation of the Stress Kinase PKR. Cell Reports, 2016, 14, 93-102.	2.9	28
36	Novel findings with reassessment of exome data: implications for validation testing and interpretation of genomic data. Genetics in Medicine, 2018, 20, 329-336.	1.1	28

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37	Exclusion of linkage to the CDL1 gene region on chromosome 3q26.3 in some familial cases of Cornelia de Lange syndrome. <i>American Journal of Medical Genetics Part A</i> , 2001, 101, 120-129.	2.4	27
38	AUDIOME: a tiered exome sequencing-based comprehensive gene panel for the diagnosis of heterogeneous nonsyndromic sensorineural hearing loss. <i>Genetics in Medicine</i> , 2018, 20, 1600-1608.	1.1	27
39	NIPBL+/~ haploinsufficiency reveals a constellation of transcriptome disruptions in the pluripotent and cardiac states. <i>Scientific Reports</i> , 2018, 8, 1056.	1.6	26
40	Duplication of chromosome region 4q28.3-qter in monozygotic twins with discordant phenotypes. <i>American Journal of Medical Genetics Part A</i> , 2000, 94, 125-140.	2.4	24
41	Antioxidant treatment ameliorates phenotypic features of SMC1A-mutated Cornelia de Lange syndrome in vitro and in vivo. <i>Human Molecular Genetics</i> , 2018, 27, 3002-3011.	1.4	24
42	Utility and limitations of exome sequencing in the molecular diagnosis of pediatric inherited platelet disorders. <i>American Journal of Hematology</i> , 2018, 93, 8-16.	2.0	22
43	Variable Clinical Manifestations of Xia-Gibbs syndrome: Findings of Consecutively Identified Cases at a Single Children's Hospital. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1890-1896.	0.7	20
44	Clinical and molecular spectrum of CHOPS syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1126-1138.	0.7	20
45	De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. <i>European Journal of Human Genetics</i> , 2019, 27, 1081-1089.	1.4	19
46	Neuronal genes deregulated in Cornelia de Lange Syndrome respond to removal and re-expression of cohesin. <i>Nature Communications</i> , 2021, 12, 2919.	5.8	18
47	Clinical utility of exome sequencing in infantile heart failure. <i>Genetics in Medicine</i> , 2020, 22, 423-426.	1.1	17
48	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. <i>American Journal of Human Genetics</i> , 2020, 106, 830-845.	2.6	17
49	Hyperinsulinemic hypoglycemia in seven patients with de novo <i>NSD1</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 542-551.	0.7	16
50	EP300-related Rubinstein-Taybi syndrome: Highlighted rare phenotypic findings and a genotype-phenotype meta-analysis of 74 patients. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2926-2938.	0.7	16
51	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, 24, 1774-1780.	1.1	16
52	Genome-Wide Expression Analysis in Fibroblast Cell Lines from Proband with Pallister Killian Syndrome. <i>PLoS ONE</i> , 2014, 9, e108853.	1.1	14
53	NKX2.5 mutation identification on exome sequencing in a patient with heterotaxy. <i>European Journal of Medical Genetics</i> , 2014, 57, 558-561.	0.7	13
54	Anticipated responses of early adopter genetic specialists and nongenetic specialists to unsolicited genomic secondary findings. <i>Genetics in Medicine</i> , 2018, 20, 1186-1195.	1.1	11

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55	Alagille syndrome: Chipping away at the tip of the iceberg. American Journal of Medical Genetics Part A, 2002, 112, 160-162.	2.4	10
56	Molecular characterization of HDAC8 deletions in individuals with atypical Cornelia de Lange syndrome. Journal of Human Genetics, 2018, 63, 349-356.	1.1	10
57	Multinational characterization of neurological phenotypes in patients hospitalized with COVID-19. Scientific Reports, 2021, 11, 20238.	1.6	10
58	The Genomics of Congenital Diaphragmatic Hernia: A 10-Year Retrospective Review. Journal of Pediatrics, 2022, 248, 108-113.e2.	0.9	9
59	Genome-first approach to rare EYA4 variants and cardio-auditory phenotypes in adults. Human Genetics, 2021, 140, 957-967.	1.8	7
60	Ciliopathies: Coloring outside of the lines. American Journal of Medical Genetics, Part A, 2021, 185, 687-694.	0.7	7
61	Molecular Diagnostic Outcomes from 700 Cases. Journal of Molecular Diagnostics, 2022, 24, 274-286.	1.2	7
62	International comparisons of laboratory values from the 4CE collaborative to predict COVID-19 mortality. Npj Digital Medicine, 2022, 5, .	5.7	7
63	Audiologic Phenotype and Progression in Pediatric <sc>STRC</sc>-Related Autosomal Recessive Hearing Loss. Laryngoscope, 2021, 131, E2897-E2903.	1.1	6
64	Activating <i>RAC1</i> variants in the switch II region cause a developmental syndrome and alter neuronal morphology. Brain, 2022, 145, 4232-4245.	3.7	6
65	Chromosomal localization, genomic characterization, and mapping to the Noonan syndrome critical region of the human Deltex (DTX1) gene. Human Genetics, 2000, 107, 577-581.	1.8	5
66	Novel microdeletion syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2007, 145C, 323-326.	0.7	5
67	Structural analysis of histone deacetylase 8 mutants associated with Cornelia de Lange Syndrome spectrum disorders. Journal of Structural Biology, 2021, 213, 107681.	1.3	5
68	Diversity, Equity, and Inclusion in The Journal of Pediatrics. Journal of Pediatrics, 2021, 236, 4.	0.9	5
69	Disease-associated <i>c-MYC</i> downregulation in human disorders of transcriptional regulation. Human Molecular Genetics, 2022, 31, 1599-1609.	1.4	5
70	Molecular Mechanisms Contributing to the Etiology of Congenital Diaphragmatic Hernia: A Review and Novel Cases. Journal of Pediatrics, 2022, 246, 251-265.e2.	0.9	4
71	Cornelia de Lange syndrome: Clinical review, diagnostic and scoring systems, and anticipatory guidance Am J Med Genet Part A 143A:1287-1296. American Journal of Medical Genetics, Part A, 2008, 146A, 2713-2713.	0.7	3
72	A de novo <i>SATB2</i> mutation in monozygotic twins with cleft palate, dental anomalies, and developmental delay. American Journal of Medical Genetics, Part A, 2017, 173, 809-812.	0.7	3

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73	Perspective on the Development of a Large-Scale Clinical Data Repository for Pediatric Hearing Research. <i>Ear and Hearing</i> , 2020, 41, 231-238.	1.0	3
74	Expanded non-invasive prenatal diagnostics. <i>Nature Medicine</i> , 2019, 25, 361-362.	15.2	2
75	50 Years Ago in T J P. <i>Journal of Pediatrics</i> , 2021, 236, 291.	0.9	1
76	Cornelia de Lange syndrome and the Cohesin complex: Abstracts from the 9th Biennial Scientific and Educational Virtual Symposium 2020. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1005-1014.	0.7	1
77	Reply to correspondence from Kosztolanyi and Mihes ?Supernumerary digital flexion creases?. <i>American Journal of Medical Genetics Part A</i> , 2003, 121A, 92-92.	2.4	0
78	Back Cover, Volume 40, Issue 12. <i>Human Mutation</i> , 2019, 40, iii.	1.1	0
79	Professor Laird Jackson, M.D., FFACMG (Physician, Scientist, Educator, and Advocate): 1930â€“2019. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 11-12.	0.7	0
80	50 Years Ago in T J P. <i>Journal of Pediatrics</i> , 2021, 233, 211.	0.9	0
81	50 Years Ago in T J P. <i>Journal of Pediatrics</i> , 2021, 239, 49.	0.9	0
82	50 Years Ago in T J P. <i>Journal of Pediatrics</i> , 2022, 241, 195.	0.9	0
83	50 Years Ago in T J P. <i>Journal of Pediatrics</i> , 2022, 241, 61.	0.9	0