Ian D Krantz

List of Publications by Year
in descending order

83 papers	6,527 citations	186265 28 h-index	69250 77 g-index
93 all docs	93 docs citations	93 times ranked	8173 citing authors

#	Article	IF	Citations
1	Disease-associated c-MYC downregulation in human disorders of transcriptional regulation. Human Molecular Genetics, 2022, 31, 1599-1609.	2.9	5
2	Molecular Diagnostic Outcomes from 700 Cases. Journal of Molecular Diagnostics, 2022, 24, 274-286.	2.8	7
3	50 Years Ago in T J P. Journal of Pediatrics, 2022, 241, 195.	1.8	0
4	50 Years Ago in T J P. Journal of Pediatrics, 2022, 241, 61.	1.8	0
5	Activating RAC1 variants in the switch II region cause a developmental syndrome and alter neuronal morphology. Brain, 2022, 145, 4232-4245.	7.6	6
6	Molecular Mechanisms Contributing to the Etiology of Congenital Diaphragmatic Hernia: A Review and Novel Cases. Journal of Pediatrics, 2022, 246, 251-265.e2.	1.8	4
7	Cornelia de Lange syndrome and the Cohesin complex: Abstracts from the 9th Biennial Scientific and Educational Virtual Symposium 2020. American Journal of Medical Genetics, Part A, 2022, 188, 1005-1014.	1.2	1
8	The Genomics of Congenital Diaphragmatic Hernia: A 10-Year Retrospective Review. Journal of Pediatrics, 2022, 248, 108-113.e2.	1.8	9
9	Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780.	2.4	16
10	International comparisons of laboratory values from the 4CE collaborative to predict COVID-19 mortality. Npj Digital Medicine, 2022, 5, .	10.9	7
11	Structural analysis of histone deacetylase 8 mutants associated with Cornelia de Lange Syndrome spectrum disorders. Journal of Structural Biology, 2021, 213, 107681.	2.8	5
12	Genome-first approach to rare EYA4 variants and cardio-auditory phenotypes in adults. Human Genetics, 2021, 140, 957-967.	3.8	7
13	Neuronal genes deregulated in Cornelia de Lange Syndrome respond to removal and re-expression of cohesin. Nature Communications, 2021, 12, 2919.	12.8	18
14	International Analysis of Electronic Health Records of Children and Youth Hospitalized With COVID-19 Infection in 6 Countries. JAMA Network Open, 2021, 4, e2112596.	5.9	33
15	50 Years Ago in T J P. Journal of Pediatrics, 2021, 233, 211.	1.8	0
16	Audiologic Phenotype and Progression in Pediatric <scp>STRCâ€Related</scp> Autosomal Recessive Hearing Loss. Laryngoscope, 2021, 131, E2897-E2903.	2.0	6
17	Diversity, Equity, and Inclusion in The Journal of Pediatrics. Journal of Pediatrics, 2021, 236, 4.	1.8	5
18	Effect of Whole-Genome Sequencing on the Clinical Management of Acutely Ill Infants With Suspected Genetic Disease. JAMA Pediatrics, 2021, 175, 1218.	6.2	83

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19	50 Years Ago in T J P. Journal of Pediatrics, 2021, 236, 291.	1.8	1
20	Ciliopathies: Coloring outside of the lines. American Journal of Medical Genetics, Part A, 2021, 185, 687-694.	1.2	7
21	Multinational characterization of neurological phenotypes in patients hospitalized with COVID-19. Scientific Reports, 2021, 11, 20238.	3.3	10
22	50 Years Ago in T J P. Journal of Pediatrics, 2021, 239, 49.	1.8	0
23	Clinical utility of exome sequencing in infantile heart failure. Genetics in Medicine, 2020, 22, 423-426.	2.4	17
24	The Genomics Research and Innovation Network: creating an interoperable, federated, genomics learning system. Genetics in Medicine, 2020, 22, 371-380.	2.4	30
25	Professor Laird Jackson, M.D., FFACMG (Physician, Scientist, Educator, and Advocate): 1930â€“2019. American Journal of Medical Genetics, Part A, 2020, 182, 11-12.	1.2	0
26	Diagnosing Cornelia de Lange syndrome and related neurodevelopmental disorders using RNA sequencing. Genetics in Medicine, 2020, 22, 927-936.	2.4	34
27	Perspective on the Development of a Large-Scale Clinical Data Repository for Pediatric Hearing Research. Ear and Hearing, 2020, 41, 231-238.	2.1	3
28	EP300 â€related Rubinsteinâ€“Taybi syndrome: Highlighted rare phenotypic findings and a genotypeâ€“phenotype metaâ€analysis of 74 patients. American Journal of Medical Genetics, Part A, 2020, 182, 2926-2938.	1.2	16
29	International electronic health record-derived COVID-19 clinical course profiles: the 4CE consortium. Npj Digital Medicine, 2020, 3, 109.	10.9	128
30	De Novo Variants in CNOT1, 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.	6.2	37
31	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. American Journal of Human Genetics, 2020, 106, 830-845.	6.2	17
32	Alagille syndrome mutation update: Comprehensive overview ofJAG1andNOTCH2mutation frequencies and insight into missense variant classification. Human Mutation, 2019, 40, 2197-2220.	2.5	84
33	Disruption of cardiac thin filament assembly arising from a mutation in LMOD2 : A novel mechanism of neonatal dilated cardiomyopathy. Science Advances, 2019, 5, eaax2066.	10.3	29
34	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	6.2	61
35	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	2.4	41
36	Clinical and molecular spectrum of CHOPS syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1126-1138.	1.2	20

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37	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. American Journal of Human Genetics, 2019, 104, 1060-1072.	6.2	78
38	Expanded non-invasive prenatal diagnostics. Nature Medicine, 2019, 25, 361-362.	30.7	2
39	Hyperinsulinemic hypoglycemia in seven patients with de novo NSD1 mutations. American Journal of Medical Genetics, Part A, 2019, 179, 542-551.	1.2	16
40	De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. European Journal of Human Genetics, 2019, 27, 1081-1089.	2.8	19
41	Back Cover, Volume 40, Issue 12. Human Mutation, 2019, 40, iii.	2.5	0
42	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
43	Automated Clinical Exome Reanalysis Reveals Novel Diagnoses. Journal of Molecular Diagnostics, 2019, 21, 38-48.	2.8	68
44	Anticipated responses of early adopter genetic specialists and nongenetic specialists to unsolicited genomic secondary findings. Genetics in Medicine, 2018, 20, 1186-1195.	2.4	11
45	NIPBL+/â˜ haploinsufficiency reveals a constellation of transcriptome disruptions in the pluripotent and cardiac states. Scientific Reports, 2018, 8, 1056.	3.3	26
46	Molecular characterization of HDAC8 deletions in individuals with atypical Cornelia de Lange syndrome. Journal of Human Genetics, 2018, 63, 349-356.	2.3	10
47	AUDIOME: a tiered exome sequencingâ€“based comprehensive gene panel for the diagnosis of heterogeneous nonsyndromic sensorineural hearing loss. Genetics in Medicine, 2018, 20, 1600-1608.	2.4	27
48	Utility and limitations of exome sequencing in the molecular diagnosis of pediatric inherited platelet disorders. American Journal of Hematology, 2018, 93, 8-16.	4.1	22
49	Novel findings with reassessment of exome data: implications for validation testing and interpretation of genomic data. Genetics in Medicine, 2018, 20, 329-336.	2.4	28
50	Missense variants in the chromatin remodeler CHD1 are associated with neurodevelopmental disability. Journal of Medical Genetics, 2018, 55, 561-566.	3.2	49
51	Variable Clinical Manifestations of Xiaâ€Gibbs syndrome: Findings of Consecutively Identified Cases at a Single Children's Hospital. American Journal of Medical Genetics, Part A, 2018, 176, 1890-1896.	1.2	20
52	Antioxidant treatment ameliorates phenotypic features of SMC1A-mutated Cornelia de Lange syndrome in vitro and in vivo. Human Molecular Genetics, 2018, 27, 3002-3011.	2.9	24
53	A taxonomy of medical uncertainties in clinical genome sequencing. Genetics in Medicine, 2017, 19, 918-925.	2.4	91
54	A de novo SATB2 mutation in monozygotic twins with cleft palate, dental anomalies, and developmental delay. American Journal of Medical Genetics, Part A, 2017, 173, 809-812.	1.2	3

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55	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
56	ESRP1 Mutations Cause Hearing Loss due to Defects in Alternative Splicing that Disrupt Cochlear Development. Developmental Cell, 2017, 43, 318-331.e5.	7.0	68
57	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	2.4	125
58	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.	1.2	37
59	NIPBL Controls RNA Biogenesis to Prevent Activation of the Stress Kinase PKR. Cell Reports, 2016, 14, 93-102.	6.4	28
60	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315.	2.4	69
61	Drosophila Nipped-B Mutants Model Cornelia de Lange Syndrome in Growth and Behavior. PLoS Genetics, 2015, 11, e1005655.	3.5	33
62	Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431.	2.8	37
63	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. European Journal of Human Genetics, 2015, 23, 1473-1481.	2.8	101
64	Germline gain-of-function mutations in AFF4 cause a developmental syndrome functionally linking the super elongation complex and cohesin. Nature Genetics, 2015, 47, 338-344.	21.4	109
65	Heterozygous Deletion ofFOXA2Segregates with Disease in a Family with Heterotaxy, Panhypopituitarism, and Biliary Atresia. Human Mutation, 2015, 36, 631-637.	2.5	43
66	USP7 Acts as a Molecular Rheostat to Promote WASH-Dependent Endosomal Protein Recycling and Is Mutated in a Human Neurodevelopmental Disorder. Molecular Cell, 2015, 59, 956-969.	9.7	175
67	A Cohesin-Independent Role for NIPBL at Promoters Provides Insights in CdLS. PLoS Genetics, 2014, 10, e1004153.	3.5	123
68	NKX2.5 mutation identification on exome sequencing in a patient with heterotaxy. European Journal of Medical Genetics, 2014, 57, 558-561.	1.3	13
69	Genome-Wide Expression Analysis in Fibroblast Cell Lines from Probands with Pallister Killian Syndrome. PLoS ONE, 2014, 9, e108853.	2.5	14
70	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.	1.2	3
71	Novel microdeletion syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2007, 145C, 323-326.	1.6	5
72	Reply to correspondence from Kosztolï¿½nyi and Mï¿½hes ?Supernumerary digital flexion creases?. American Journal of Medical Genetics Part A, 2003, 121A, 92-92.	2.4	0

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73	Alagille syndrome: Chipping away at the tip of the iceberg. American Journal of Medical Genetics Part A, 2002, 112, 160-162.	2.4	10
74	Exclusion of linkage to theCDL1 gene region on chromosome 3q26.3 in some familial cases of Cornelia de Lange syndrome. American Journal of Medical Genetics Part A, 2001, 101, 120-129.	2.4	27
75	Dominant paternal transmission of Cornelia de Lange syndrome: A new case and review of 25 previously reported familial recurrences. American Journal of Medical Genetics Part A, 2001, 104, 267-276.	2.4	91
76	Mutation analysis of Jagged1 (JAG1) in Alagille syndrome patients. Human Mutation, 2001, 17, 151-152.	2.5	76
77	Duplication of chromosome region 4q28.3-qter in monozygotic twins with discordant phenotypes. American Journal of Medical Genetics Part A, 2000, 94, 125-140.	2.4	24
78	Chromosomal localization, genomic characterization, and mapping to the Noonan syndrome critical region of the human Deltex (DTX1) gene. Human Genetics, 2000, 107, 577-581.	3.8	5
79	Features of alagille syndrome in 92 patients: Frequency and relation to prognosis. Hepatology, 1999, 29, 822-829.	7.3	591
80	Mutations in the human Jagged1 gene are responsible for Alagille syndrome. Nature Genetics, 1997, 16, 235-242.	21.4	1,072
81	Alagille syndrome is caused by mutations in human Jagged1, which encodes a ligand for Notch1. Nature Genetics, 1997, 16, 243-251.	21.4	1,184
82	KILLER/DR5 is a DNA damageâ€“inducible p53â€“regulated death receptor gene. Nature Genetics, 1997, 17, 141-143.	21.4	1,005
83	Deletions of 20p12 in Alagille syndrome: Frequency and molecular characterization. , 1997, 70, 80-86.		71