# Ian D Krantz

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8350206/publications.pdf

Version: 2024-02-01

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|---|---|---|---|
| 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 | Disease-associated c-MYC downregulation in human disorders of transcriptional regulation. Human Molecular Genetics, 2022, 31, 1599-1609. | 1.4 | 5 |
| 2 | Molecular Diagnostic Outcomes from 700 Cases. Journal of Molecular Diagnostics, 2022, 24, 274-286. | 1.2 | 7 |
| 3 | 50 Years Ago in T J P. Journal of Pediatrics, 2022, 241, 195. | 0.9 | 0 |
| 4 | 50 Years Ago in T J P. Journal of Pediatrics, 2022, 241, 61. | 0.9 | 0 |
| 5 | Activating RAC1 variants in the switch II region cause a developmental syndrome and alter neuronal morphology. Brain, 2022, 145, 4232-4245. | 3.7 | 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. | 0.9 | 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. | 0.7 | 1 |
| 8 | The Genomics of Congenital Diaphragmatic Hernia: A 10-Year Retrospective Review. Journal of Pediatrics, 2022, 248, 108-113.e2. | 0.9 | 9 |
| 9 | Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1774-1780. | 1.1 | 16 |
| 10 | International comparisons of laboratory values from the 4CE collaborative to predict COVID-19 mortality. Npj Digital Medicine, 2022, 5, . | 5.7 | 7 |
| 11 | 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 |
| 12 | Genome-first approach to rare EYA4 variants and cardio-auditory phenotypes in adults. Human Genetics, 2021, 140, 957-967. | 1.8 | 7 |
| 13 | Neuronal genes deregulated in Cornelia de Lange Syndrome respond to removal and re-expression of cohesin. Nature Communications, 2021, 12, 2919. | 5.8 | 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. | 2.8 | 33 |
| 15 | 50 Years Ago in T J P. Journal of Pediatrics, 2021, 233, 211. | 0.9 | 0 |
| 16 | Audiologic Phenotype and Progression in Pediatric <scp>STRCâ€Related</scp> Autosomal Recessive Hearing Loss. Laryngoscope, 2021, 131, E2897-E2903. | 1.1 | 6 |
| 17 | Diversity, Equity, and Inclusion in The Journal of Pediatrics. Journal of Pediatrics, 2021, 236, 4. | 0.9 | 5 |
| 18 | Effect of Whole-Genome Sequencing on the Clinical Management of Acutely Ill Infants With Suspected Genetic Disease. JAMA Pediatrics, 2021, 175, 1218. | 3.3 | 83 |

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|---|---|---|---|
| 19 | 50 Years Ago in T J P. Journal of Pediatrics, 2021, 236, 291. | 0.9 | 1 |
| 20 | Ciliopathies: Coloring outside of the lines. American Journal of Medical Genetics, Part A, 2021, 185, 687-694. | 0.7 | 7 |
| 21 | Multinational characterization of neurological phenotypes in patients hospitalized with COVID-19. Scientific Reports, 2021, 11, 20238. | 1.6 | 10 |
| 22 | 50 Years Ago in T J P. Journal of Pediatrics, 2021, 239, 49. | 0.9 | 0 |
| 23 | Clinical utility of exome sequencing in infantile heart failure. Genetics in Medicine, 2020, 22, 423-426. | 1.1 | 17 |
| 24 | The Genomics Research and Innovation Network: creating an interoperable, federated, genomics learning system. Genetics in Medicine, 2020, 22, 371-380. | 1.1 | 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. | 0.7 | 0 |
| 26 | Diagnosing Cornelia de Lange syndrome and related neurodevelopmental disorders using RNA sequencing. Genetics in Medicine, 2020, 22, 927-936. | 1.1 | 34 |
| 27 | Perspective on the Development of a Large-Scale Clinical Data Repository for Pediatric Hearing Research. Ear and Hearing, 2020, 41, 231-238. | 1.0 | 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. | 0.7 | 16 |
| 29 | International electronic health record-derived COVID-19 clinical course profiles: the 4CE consortium. Npj Digital Medicine, 2020, 3, 109. | 5.7 | 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. | 2.6 | 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. | 2.6 | 17 |
| 32 | Alagille syndrome mutation update: Comprehensive overview ofJAG1andNOTCH2mutation frequencies and insight into missense variant classification. Human Mutation, 2019, 40, 2197-2220. | 1.1 | 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. | 4.7 | 29 |
| 34 | Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615. | 2.6 | 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. | 1.1 | 41 |
| 36 | Clinical and molecular spectrum of CHOPS syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1126-1138. | 0.7 | 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. | 2.6 | 78 |
| 38 | Expanded non-invasive prenatal diagnostics. Nature Medicine, 2019, 25, 361-362. | 15.2 | 2 |
| 39 | Hyperinsulinemic hypoglycemia in seven patients with de novo NSD1 mutations. American Journal of Medical Genetics, Part A, 2019, 179, 542-551. | 0.7 | 16 |
| 40 | De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. European Journal of Human Genetics, 2019, 27, 1081-1089. | 1.4 | 19 |
| 41 | Back Cover, Volume 40, Issue 12. Human Mutation, 2019, 40, iii. | 1.1 | 0 |
| 42 | Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158. | 0.7 | 40 |
| 43 | Automated Clinical Exome Reanalysis Reveals Novel Diagnoses. Journal of Molecular Diagnostics, 2019, 21, 38-48. | 1.2 | 68 |
| 44 | Anticipated responses of early adopter genetic specialists and nongenetic specialists to unsolicited genomic secondary findings. Genetics in Medicine, 2018, 20, 1186-1195. | 1.1 | 11 |
| 45 | NIPBL+/â˜ haploinsufficiency reveals a constellation of transcriptome disruptions in the pluripotent and cardiac states. Scientific Reports, 2018, 8, 1056. | 1.6 | 26 |
| 46 | Molecular characterization of HDAC8 deletions in individuals with atypical Cornelia de Lange syndrome. Journal of Human Genetics, 2018, 63, 349-356. | 1.1 | 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. | 1.1 | 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. | 2.0 | 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. | 1.1 | 28 |
| 50 | Missense variants in the chromatin remodeler CHD1 are associated with neurodevelopmental disability. Journal of Medical Genetics, 2018, 55, 561-566. | 1.5 | 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. | 0.7 | 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. | 1.4 | 24 |
| 53 | A taxonomy of medical uncertainties in clinical genome sequencing. Genetics in Medicine, 2017, 19, 918-925. | 1.1 | 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. | 0.7 | 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. | 0.7 | 69 |
| 56 | 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 |
| 57 | Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084. | 1.1 | 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. | 0.7 | 37 |
| 59 | NIPBL Controls RNA Biogenesis to Prevent Activation of the Stress Kinase PKR. Cell Reports, 2016, 14, 93-102. | 2.9 | 28 |
| 60 | Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315. | 1.1 | 69 |
| 61 | Drosophila Nipped-B Mutants Model Cornelia de Lange Syndrome in Growth and Behavior. PLoS Genetics, 2015, 11, e1005655. | 1.5 | 33 |
| 62 | Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431. | 1.4 | 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. | 1.4 | 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. | 9.4 | 109 |
| 65 | Heterozygous Deletion ofFOXA2Segregates with Disease in a Family with Heterotaxy, Panhypopituitarism, and Biliary Atresia. Human Mutation, 2015, 36, 631-637. | 1.1 | 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. | 4.5 | 175 |
| 67 | A Cohesin-Independent Role for NIPBL at Promoters Provides Insights in CdLS. PLoS Genetics, 2014, 10, e1004153. | 1.5 | 123 |
| 68 | NKX2.5 mutation identification on exome sequencing in a patient with heterotaxy. European Journal of Medical Genetics, 2014, 57, 558-561. | 0.7 | 13 |
| 69 | Genome-Wide Expression Analysis in Fibroblast Cell Lines from Probands with Pallister Killian Syndrome. PLoS ONE, 2014, 9, e108853. | 1.1 | 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. | 0.7 | 3 |
| 71 | Novel microdeletion syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2007, 145C, 323-326. | 0.7 | 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. | 1.1 | 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. | 1.8 | 5 |
| 79 | Features of alagille syndrome in 92 patients: Frequency and relation to prognosis. Hepatology, 1999, 29, 822-829. | 3.6 | 591 |
| 80 | Mutations in the human Jagged1 gene are responsible for Alagille syndrome. Nature Genetics, 1997, 16, 235-242. | 9.4 | 1,072 |
| 81 | Alagille syndrome is caused by mutations in human Jagged1, which encodes a ligand for Notch1. Nature Genetics, 1997, 16, 243-251. | 9.4 | 1,184 |
| 82 | KILLER/DR5 is a DNA damageâ€“inducible p53â€“regulated death receptor gene. Nature Genetics, 1997, 17, 141-143. | 9.4 | 1,005 |
| 83 | Deletions of 20p12 in Alagille syndrome: Frequency and molecular characterization. , 1997, 70, 80-86. | | 71 |