Neil A Hanchard

List of Publications by Year
in descending order

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78 papers	3,080 citations	172457 29 h-index	182427 51 g-index
82 all docs	82 docs citations	82 times ranked	6432 citing authors

#	Article	IF	Citations
1	Pressure-Mediated Reflection Spectroscopy Criterion Validity as a Biomarker of Fruit and Vegetable Intake: A 2-Site Cross-Sectional Study of 4 Racial or Ethnic Groups. Journal of Nutrition, 2022, 152, 107-116.	2.9	18
2	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate diseaseâ€“associated loci for BAFopathies. Genetics in Medicine, 2022, 24, 364-373.	2.4	12
3	One-carbon metabolism in children with marasmus and kwashiorkor. EBioMedicine, 2022, 75, 103791.	6.1	8
4	A dominant negative variant of RAB5B disrupts maturation of surfactant protein B and surfactant protein C. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	9
5	Genome sequencing reveals novel noncoding variants in <scp>PLA2G6</scp> and <scp>LMNB1</scp> causing progressive neurologic disease. Molecular Genetics & Genomic Medicine, 2022, 10, e1892.	1.2	4
6	Genetics agrees: Africa is thriving in diversity. TheScienceBreaker, 2022, 8, .	0.0	0
7	Polygenic risk scores for CARDINAL study. Nature Genetics, 2022, 54, 527-530.	21.4	5
8	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	8.2	87
9	â€œIronâ€ing out hemophagocytosis through PIEZO1. Cell, 2021, 184, 856-858.	28.9	3
10	Unmapped exome reads implicate a role for Anelloviridae in childhood HIV-1 long-term non-progression. Npj Genomic Medicine, 2021, 6, 24.	3.8	3
11	Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. Genetics in Medicine, 2021, 23, 1465-1473.	2.4	10
12	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
13	Exome Sequencing Reveals a Putative Role for HLA-C*03:02 in Control of HIV-1 in African Pediatric Populations. Frontiers in Genetics, 2021, 12, 720213.	2.3	2
14	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	6.2	18
15	Sex-specific phenotypic effects and evolutionary history of an ancient polymorphic deletion of the human growth hormone receptor. Science Advances, 2021, 7, eabi4476.	10.3	11
16	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
17	High-depth African genomes inform human migration and health. Nature, 2020, 586, 741-748.	27.8	197
18	Mutations in the KIF21B kinesin gene cause neurodevelopmental disorders through imbalanced canonical motor activity. Nature Communications, 2020, 11, 2441.	12.8	37

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19	Biases in arginine codon usage correlate with genetic disease risk. Genetics in Medicine, 2020, 22, 1407-1412.	2.4	7
20	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
21	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	1.5	6
22	Biallelic variants in COX4I1 associated with a novel phenotype resembling Leigh syndrome with developmental regression, intellectual disability, and seizures. American Journal of Medical Genetics, Part A, 2019, 179, 2138-2143.	1.2	11
23	Hydroxyurea-Induced miRNA Expression in Sickle Cell Disease Patients in Africa. Frontiers in Genetics, 2019, 10, 509.	2.3	20
24	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	2.5	19
25	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25.	8.2	22
26	Aberrant DNA methylation as a diagnostic biomarker of diabetic embryopathy. Genetics in Medicine, 2019, 21, 2453-2461.	2.4	8
27	IgG4â€related disease: Association with a rare gene variant expressed in cytotoxic T cells. Molecular Genetics & Genomic Medicine, 2019, 7, e686.	1.2	8
28	Whole genome sequencing reveals novel IGHMBP2 variant leading to unique cryptic spliceâ€site and Charcotâ€Marieâ€Tooth phenotype with early onset symptoms. Molecular Genetics & Genomic Medicine, 2019, 7, e00676.	1.2	18
29	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60.	4.1	18
30	The additional genetic diagnosis of homozygous sickle cell disease in a patient with Waardenburg-Shah syndrome: a case report. Journal of Medical Case Reports, 2019, 13, 10.	0.8	0
31	Edematous severe acute malnutrition is characterized by hypomethylation of DNA. Nature Communications, 2019, 10, 5791.	12.8	23
32	The Sickle Cell Disease Ontology: enabling universal sickle cell-based knowledge representation. Database: the Journal of Biological Databases and Curation, 2019, 2019, .	3.0	14
33	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
34	Genetic architecture of laterality defects revealed by whole exome sequencing. European Journal of Human Genetics, 2019, 27, 563-573.	2.8	44
35	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. American Journal of Human Genetics, 2018, 102, 69-87.	6.2	144
36	Whole-Exome Sequencing Reveals Uncaptured Variation and Distinct Ancestry in the Southern African Population of Botswana. American Journal of Human Genetics, 2018, 102, 731-743.	6.2	38

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37	Wholeâ€exome sequencing of sickle cell disease patients with hyperhemolysis syndrome suggests a role for rare variation in disease predisposition. Transfusion, 2018, 58, 726-735.	1.6	17
38	Phenotypic expansion in <scp>DDX</scp>3X â€“ a common cause of intellectual disability in females. Annals of Clinical and Translational Neurology, 2018, 5, 1277-1285.	3.7	66
39	A locus on chromosome 5 shows African ancestryâ€“limited association with alloimmunization in sickle cell disease. Blood Advances, 2018, 2, 3637-3647.	5.2	18
40	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	6.2	56
41	First case of genetically confirmed CLN3 disease in Chinese with cDNA sequencing revealing pathogenicity of a novel splice site variant. Clinica Chimica Acta, 2018, 486, 151-155.	1.1	12
42	The Collaborative African Genomics Network (CAfGEN): Applying Genomic technologies to probe host factors important to the progression of HIV and HIV-tuberculosis infection in sub-Saharan Africa. AAS Open Research, 2018, 1, 3.	1.5	10
43	The Collaborative African Genomics Network (CAfGEN): Applying Genomic technologies to probe host factors important to the progression of HIV and HIV-tuberculosis infection in sub-Saharan Africa. AAS Open Research, 2018, 1, 3.	1.5	15
44	Structural Variation within the Beta-Globin Gene Cluster Among HbS Haplotype Groups. Blood, 2018, 132, 2365-2365.	1.4	0
45	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
46	Clinical Metabolomics to Segregate Aromatic Amino Acid Decarboxylase Deficiency From Drug-Induced Metabolite Elevations. Pediatric Neurology, 2017, 75, 66-72.	2.1	19
47	Assessment of large copy number variants in patients with apparently isolated congenital leftâ€sided cardiac lesions reveals clinically relevant genomic events. American Journal of Medical Genetics, Part A, 2017, 173, 2176-2188.	1.2	17
48	Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. Genetics in Medicine, 2017, 19, 412-420.	2.4	73
49	Whole exome sequencing in 342 congenital cardiac left sided lesion cases reveals extensive genetic heterogeneity and complex inheritance patterns. Genome Medicine, 2017, 9, 95.	8.2	37
50	The collaborative African genomics network training program: a trainee perspective on training the next generation of African scientists. Genetics in Medicine, 2017, 19, 826-833.	2.4	29
51	Whole-exome sequencing reveals an inherited R566X mutation of the epithelial sodium channel Î²-subunit in a case of early-onset phenotype of Liddle syndrome. Journal of Physical Education and Sports Management, 2016, 2, a001255.	1.2	10
52	Bi-allelic Mutations in PKD1L1 Are Associated with Laterality Defects in Humans. American Journal of Human Genetics, 2016, 99, 886-893.	6.2	57
53	Functional cellular analyses reveal energy metabolism defect and mitochondrial DNA depletion in a case of mitochondrial aconitase deficiency. Molecular Genetics and Metabolism, 2016, 118, 28-34.	1.1	32
54	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98

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55	A genome-wide association study of congenital cardiovascular left-sided lesions shows association with a locus on chromosome 20. Human Molecular Genetics, 2016, 25, 2331-2341.	2.9	31
56	Beta-Globin Gene Haplotypes Among Cameroonians and Review of the Global Distribution: Is There a Case for a Single Sickle Mutation Origin in Africa?. OMICS A Journal of Integrative Biology, 2015, 19, 171-179.	2.0	43
57	Abstract 17616: Recurrent Copy Number Variants are Enriched in Bicuspid Aortic Valve and Affect Cardiac Developmental Genes. Circulation, 2015, 132, .	1.6	0
58	A Genome-Wide Screen for Large-Effect Alloimmunization Susceptibility Loci among Red Blood Cell Transfusion Recipients with Sickle Cell Disease. Transfusion Medicine and Hemotherapy, 2014, 41, 453-461.	1.6	21
59	Wholeâ€Exome Sequencing Reveals GPIHBP1 Mutations in Infantile Colitis With Severe Hypertriglyceridemia. Journal of Pediatric Gastroenterology and Nutrition, 2014, 59, 17-21.	1.8	20
60	Enabling the genomic revolution in Africa. Science, 2014, 344, 1346-1348.	12.6	361
61	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	6.2	43
62	Exploring the utility of wholeâ€exome sequencing as a diagnostic tool in a child withÂatypical episodic muscle weakness. Clinical Genetics, 2013, 83, 457-461.	2.0	27
63	MCTP2 is a dosage-sensitive gene required for cardiac outflow tract development. Human Molecular Genetics, 2013, 22, 4339-4348.	2.9	40
64	A partial MECP2 duplication in a mildly affected adult male: a putative role for the 3' untranslated region in the MECP2 duplication phenotype. BMC Medical Genetics, 2012, 13, 71.	2.1	12
65	Genomic alterations that contribute to the development of isolated and non-isolated congenital diaphragmatic hernia. Journal of Medical Genetics, 2011, 48, 299-307.	3.2	82
66	Deoxyguanosine kinase deficiency presenting as neonatal hemochromatosis. Molecular Genetics and Metabolism, 2011, 103, 262-267.	1.1	33
67	UGT1A1 sequence variants and bilirubin levels in early postnatal life: a quantitative approach. BMC Medical Genetics, 2011, 12, 57.	2.1	10
68	Ethnic differences in F cell levels in Jamaica: a potential tool for identifying new genetic loci controlling fetal haemoglobin. British Journal of Haematology, 2009, 144, 954-960.	2.5	8
69	Rate of change of sickle allele frequency may be influenced by total fertility rate: a sesquicentenary reflection on human microâ€evolution. British Journal of Haematology, 2009, 147, 582-583.	2.5	1
70	Genetic Variation on Chromosome 6 Influences F Cell Levels in Healthy Individuals of African Descent and HbF Levels in Sickle Cell Patients. PLoS ONE, 2009, 4, e4218.	2.5	77
71	Tumor necrosis factor SNP haplotypes are associated with iron deficiency anemia in West African children. Blood, 2008, 112, 4276-4283.	1.4	38
72	Childhood malnutrition is associated with a reduction in the total melanin content of scalp hair. British Journal of Nutrition, 2007, 98, 159-164.	2.3	23

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73	Classical sickle beta-globin haplotypes exhibit a high degree of long-range haplotype similarity in African and Afro-Caribbean populations. BMC Genetics, 2007, 8, 52.	2.7	46
74	Screening for Recently Selected Alleles by Analysis of Human Haplotype Similarity. American Journal of Human Genetics, 2006, 78, 153-159.	6.2	47
75	Implications of inter-population linkage disequilibrium patterns on the approach to a disease association study in the human MHC class III. Immunogenetics, 2006, 58, 465-470.	2.4	10
76	Genetic susceptibility and single-nucleotide polymorphisms. Seminars in Fetal and Neonatal Medicine, 2005, 10, 283-289.	2.3	9
77	Haplotype mapping of the bronchiolitis susceptibility locus near IL8. Human Genetics, 2004, 114, 272-279.	3.8	59
78	Evidence for Extensive Transmission Distortion in the Human Genome. American Journal of Human Genetics, 2004, 74, 62-72.	6.2	111