Andrew H Crosby

List of Publications by Year
in descending order

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124	10,273	66234	34900
papers	citations	42 h-index	98 g-index

129	129	129	12667
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. Nature Genetics, 2001, 29, 465-468.	9.4	1,555
2	Identification of a deletion in plakoglobin in arrhythmogenic right ventricular cardiomyopathy with palmoplantar keratoderma and woolly hair (Naxos disease). Lancet, The, 2000, 355, 2119-2124.	6.3	1,270
3	PTPN11 Mutations in Noonan Syndrome: Molecular Spectrum, Genotype-Phenotype Correlation, and Phenotypic Heterogeneity. American Journal of Human Genetics, 2002, 70, 1555-1563.	2.6	680
4	Hereditary spastic paraplegia: clinical features and pathogenetic mechanisms. Lancet Neurology, The, 2008, 7, 1127-1138.	4.9	481
5	Infantile-onset symptomatic epilepsy syndrome caused by a homozygous loss-of-function mutation of GM3 synthase. Nature Genetics, 2004, 36, 1225-1229.	9.4	359
6	Heterozygous missense mutations in BSCL2 are associated with distal hereditary motor neuropathy and Silver syndrome. Nature Genetics, 2004, 36, 271-276.	9.4	349
7	Novel Mutation in Desmoplakin Causes Arrhythmogenic Left Ventricular Cardiomyopathy. Circulation, 2005, 112, 636-642.	1.6	266
8	Deficiency of terminal ADP-ribose protein glycohydrolase TARG1/C6orf130 in neurodegenerative disease. EMBO Journal, 2013, 32, 1225-1237.	3.5	263
9	SPG20 is mutated in Troyer syndrome, an hereditary spastic paraplegia. Nature Genetics, 2002, 31, 347-348.	9.4	240
10	Alterations in the ankyrin domain of TRPV4 cause congenital distal SMA, scapuloperoneal SMA and HMSN2C. Nature Genetics, 2010, 42, 160-164.	9.4	228
11	The natural history of Noonan syndrome: a long-term follow-up study. Archives of Disease in Childhood, 2006, 92, 128-132.	1.0	218
12	Mutations in FAM20C Are Associated with Lethal Osteosclerotic Bone Dysplasia (Raine Syndrome), Highlighting a Crucial Molecule in Bone Development. American Journal of Human Genetics, 2007, 81, 906-912.	2.6	190
13	Mutation of FA2H underlies a complicated form of hereditary spastic paraplegia (SPG35). Human Mutation, 2010, 31, E1251-E1260.	1.1	174
14	Sequence Alterations within CYP7B1 Implicate Defective Cholesterol Homeostasis in Motor-Neuron Degeneration. American Journal of Human Genetics, 2008, 82, 510-515.	2.6	171
15	Hereditary spastic paraplegia: from diagnosis to emerging therapeutic approaches. Lancet Neurology, The, 2019, 18, 1136-1146.	4.9	171
16	Maspardin Is Mutated in Mast Syndrome, a Complicated Form of Hereditary Spastic Paraplegia Associated with Dementia. American Journal of Human Genetics, 2003, 73, 1147-1156.	2.6	158
17	Mutations in MAP3K1 Cause 46,XY Disorders of Sex Development and Implicate a Common Signal Transduction Pathway in Human Testis Determination. American Journal of Human Genetics, 2010, 87, 898-904.	2.6	155
18	The identification of a conserved domain in both spartin and spastin, mutated in hereditary spastic paraplegia. Genomics, 2003, 81, 437-441.	1.3	128

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19	Is the Transportation Highway the Right Road for Hereditary Spastic Paraplegia?. American Journal of Human Genetics, 2002, 71, 1009-1016.	2.6	119
20	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. Brain, 2013, 136, 3618-3624.	3.7	115
21	Dominant mutations in the cation channel gene transient receptor potential vanilloid 4 cause an unusual spectrum of neuropathies. Brain, 2010, 133, 1798-1809.	3.7	113
22	A partially inactivating mutation in the sodium-dependent lysophosphatidylcholine transporter MFSD2A causes a non-lethal microcephaly syndrome. Nature Genetics, 2015, 47, 814-817.	9.4	108
23	CYP7B1 mutations in pure and complex forms of hereditary spastic paraplegia type 5. Brain, 2009, 132, 1589-1600.	3.7	102
24	Spectrum of DNA variants for non-syndromic deafness in a large cohort from multiple continents. Human Genetics, 2016, 135, 953-961.	1.8	102
25	Mutations in FAM20C also identified in nonâ€lethal osteosclerotic bone dysplasia. Clinical Genetics, 2009, 75, 271-276.	1.0	97
26	Defective Presynaptic Choline Transport Underlies Hereditary Motor Neuropathy. American Journal of Human Genetics, 2012, 91, 1103-1107.	2.6	89
27	The Silver Syndrome Variant of Hereditary Spastic Paraplegia Maps to Chromosome 11q12-q14, with Evidence for Genetic Heterogeneity within This Subtype. American Journal of Human Genetics, 2001, 69, 209-215.	2.6	81
28	A clinical, genetic and biochemical study of SPG7 mutations in hereditary spastic paraplegia. Brain, 2004, 127, 973-980.	3.7	77
29	Hypomorphic PCNA mutation underlies a human DNA repair disorder. Journal of Clinical Investigation, 2014, 124, 3137-3146.	3.9	77
30	Paternal Germline Origin and Sex-Ratio Distortion in Transmission of PTPN11 Mutations in Noonan Syndrome. American Journal of Human Genetics, 2004, 75, 492-497.	2.6	76
31	Defective Mitochondrial mRNA Maturation Is Associated with Spastic Ataxia. American Journal of Human Genetics, 2010, 87, 655-660.	2.6	76
32	A human mitochondrial poly(A) polymerase mutation reveals the complexities of post-transcriptional mitochondrial gene expression. Human Molecular Genetics, 2014, 23, 6345-6355.	1.4	63
33	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952.	3.7	62
34	Recessive nephrocerebellar syndrome on the Galloway-Mowat syndrome spectrum is caused by homozygous protein-truncating mutations ofWDR73. Brain, 2015, 138, 2173-2190.	3.7	60
35	A mutation ofEPT1 (SELENOI)underlies a new disorder of Kennedy pathway phospholipid biosynthesis. Brain, 2017, 140, aww318.	3.7	58
36	Mutation of HERC2 causes developmental delay with Angelman-like features. Journal of Medical Genetics, 2013, 50, 65-73.	1.5	57

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37	Assessing performance of pathogenicity predictors using clinically relevant variant datasets. Journal of Medical Genetics, 2021, 58, 547-555.	1.5	57
38	Analysis of CYP7B1 in non-consanguineous cases of hereditary spastic paraplegia. Neurogenetics, 2009, 10, 97-104.	0.7	54
39	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	1.1	52
40	A novel NIPA1 mutation associated with a pure form of autosomal dominant hereditary spastic paraplegia. Neurogenetics, 2005, 6, 79-84.	0.7	51
41	SLITRK6 mutations cause myopia and deafness in humans and mice. Journal of Clinical Investigation, 2013, 123, 2094-2102.	3.9	50
42	Mapping of the Human Dentin Matrix Acidic Phosphoprotein Gene (DMP1) to the Dentinogenesis Imperfecta Type II Critical Region at Chromosome 4q21. Genomics, 1995, 30, 347-349.	1.3	48
43	Genomic Organization of the Human Osteopontin Gene: Exclusion of the Locus from a Causative Role in the Pathogenesis of Dentinogenesis Imperfecta Type II. Genomics, 1995, 27, 155-160.	1.3	46
44	Absence of PTPN11 mutations in 28 cases of cardiofaciocutaneous (CFC) syndrome. Human Genetics, 2002, 111, 421-427.	1.8	45
45	Loss of PCLO function underlies pontocerebellar hypoplasia type III. Neurology, 2015, 84, 1745-1750.	1.5	45
46	A new locus for autosomal recessive complicated hereditary spastic paraplegia (SPG26) maps to chromosome 12p11.1-12q14. Journal of Medical Genetics, 2005, 42, 80-82.	1.5	43
47	Raine syndrome: An overview. European Journal of Medical Genetics, 2014, 57, 536-542.	0.7	43
48	Troyer syndrome revisited. Journal of Neurology, 2004, 251, 1105-10.	1.8	42
49	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. Genome Research, 2019, 29, 1057-1066.	2.4	38
50	Lipid metabolic pathways converge in motor neuron degenerative diseases. Brain, 2020, 143, 1073-1087.	3.7	36
51	Hereditary deletion of the entire FAM20C gene in a patient with Raine syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 3155-3160.	0.7	35
52	Mutations in KPTN Cause Macrocephaly, Neurodevelopmental Delay, and Seizures. American Journal of Human Genetics, 2014, 94, 87-94.	2.6	35
53	A mutation in NFÎºB interacting protein 1 causes cardiomyopathy and woolly haircoat syndrome of Poll Hereford cattle. Animal Genetics, 2009, 40, 42-46.	0.6	34
54	Mapping of the human and mouse bone sialoprotein and osteopontin loci. Mammalian Genome, 1996, 7, 149-151.	1.0	32

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55	Novel mutations in the pejvakin gene are associated with autosomal recessive nonâ€syndromic hearing loss in Iranian families. Clinical Genetics, 2007, 72, 261-263.	1.0	31
56	Disruption of cellular transport: a common cause of neurodegeneration?. Lancet Neurology, The, 2003, 2, 311-316.	4.9	28
57	Endogenous spartin, mutated in hereditary spastic paraplegia, has a complex subcellular localization suggesting diverse roles in neurons. Experimental Cell Research, 2006, 312, 2764-2777.	1.2	28
58	SPG3A mutation screening in English families with early onset autosomal dominant hereditary spastic paraplegia. Journal of the Neurological Sciences, 2003, 216, 43-45.	0.3	27
59	No association with common Caucasian genotypes in exons 8, 13 and 14 of the human cytoplasmic dynein heavy chain gene (DNCHC1) and familial motor neuron disorders. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2003, 4, 150-157.	1.4	26
60	Genetic heterogeneity in LEOPARD syndrome: two families with no mutations in PTPN11. Journal of Human Genetics, 2005, 50, 21-25.	1.1	25
61	An Amish founder mutation disrupts a PI(3)P-WHAMM-Arp2/3 complexâ€“driven autophagosomal remodeling pathway. Molecular Biology of the Cell, 2017, 28, 2492-2507.	0.9	25
62	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	1.5	25
63	Tissue-Biased Expansion of DNMT3A-Mutant Clones in a Mosaic Individual Is Associated with Conserved Epigenetic Erosion. Cell Stem Cell, 2020, 27, 326-335.e4.	5.2	25
64	Germline CBL mutation associated with a noonanâ€like syndrome with primary lymphedema and teratoma associated with acquired uniparental isodisomy of chromosome 11q23. American Journal of Medical Genetics, Part A, 2014, 164, 1003-1009.	0.7	24
65	Choline transporter mutations in severe congenital myasthenic syndrome disrupt transporter localization. Brain, 2017, 140, 2838-2850.	3.7	24
66	An Amish founder variant consolidates disruption of CEP55 as a cause of hydranencephaly and renal dysplasia. European Journal of Human Genetics, 2019, 27, 657-662.	1.4	24
67	The HERC2 ubiquitin ligase is essential for embryonic development and regulates motor coordination. Oncotarget, 2016, 7, 56083-56106.	0.8	24
68	Complicated Hereditary Spastic Paraplegia with Thin Corpus Callosum (HSPâ€TCC) and Childhood Onset. Neuropediatrics, 2005, 36, 274-278.	0.3	21
69	High carrier frequency of the GJB2 mutation (35delG) in the north of Iran. International Journal of Pediatric Otorhinolaryngology, 2007, 71, 863-867.	0.4	21
70	Further Delineation of the Critical Region for Noonan Syndrome on the Long Arm of Chromosome 12. European Journal of Human Genetics, 1997, 5, 336-337.	1.4	21
71	Mutations in HYAL2, Encoding Hyaluronidase 2, Cause a Syndrome of Orofacial Clefting and Cor Triatriatum Sinister in Humans and Mice. PLoS Genetics, 2017, 13, e1006470.	1.5	20
72	Raine syndrome: a clinical, radiographic and genetic investigation of a case from the Indian subcontinent. Clinical Dysmorphology, 2010, 19, 153-156.	0.1	19

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73	Truncating SLC5A7 mutations underlie a spectrum of dominant hereditary motor neuropathies. Neurology: Genetics, 2018, 4, e222.	0.9	19
74	MNS1 variant associated with situs inversus and male infertility. European Journal of Human Genetics, 2020, 28, 50-55.	1.4	19
75	Genetic screening of Congenital Short Bowel Syndrome patients confirms CLMP as the major gene involved in the recessive form of this disorder. European Journal of Human Genetics, 2016, 24, 1627-1629.	1.4	18
76	Novel Genetic, Clinical, and Pathomechanistic Insights into TFG-Associated Hereditary Spastic Paraplegia. Human Mutation, 2016, 37, 1157-1161.	1.1	17
77	Biallelic PI4KA variants cause neurological, intestinal and immunological disease. Brain, 2021, 144, 3597-3610.	3.7	17
78	TMEM63C mutations cause mitochondrial morphology defects and underlie hereditary spastic paraplegia. Brain, 2022, 145, 3095-3107.	3.7	17
79	Three novel spastin (SPG4) mutations in families with autosomal dominant hereditary spastic paraplegia. Journal of the Neurological Sciences, 2002, 201, 65-69.	0.3	16
80	Novel homozygous missense mutation in GAN associated with Charcot-Marie-Tooth disease type 2 in a large consanguineous family from Israel. BMC Medical Genetics, 2016, 17, 82.	2.1	15
81	A novel homozygous mutation in ALS2 gene in four siblings with infantile-onset ascending hereditary spastic paralysis. European Journal of Medical Genetics, 2014, 57, 275-278.	0.7	14
82	Detailed mapping, mutation analysis, and intragenic polymorphism identification in candidate Noonan syndrome genes MYL2, DCN, EPS8, and RPL6. Journal of Medical Genetics, 2000, 37, 884-886.	1.5	13
83	Complicated hereditary spastic paraplegia with thin corpus callosum: Variation of phenotypic expression over time. Journal of Neurology, 2004, 251, 1285-1287.	1.8	13
84	Lethal Cystic Kidney Disease in Amish Neonates Associated With Homozygous Nonsense Mutation of NPHP3. American Journal of Kidney Diseases, 2009, 53, 790-795.	2.1	13
85	Evidence from a Ghanaian Population of Known African Descent to Support the Proposition That Hemochromatosis Is a Caucasian Disorder. Genetic Testing and Molecular Biomarkers, 1999, 3, 375-377.	1.7	12
86	PCNA dependent cellular activities tolerate dramatic perturbations in PCNA client interactions. DNA Repair, 2017, 50, 22-35.	1.3	12
87	Novel mutations in ALDH1A3 associated with autosomal recessive anophthalmia/microphthalmia, and review of the literature. BMC Medical Genetics, 2018, 19, 160.	2.1	12
88	Homozygous variants in the HEXB and MBOAT7 genes underlie neurological diseases in consanguineous families. BMC Medical Genetics, 2019, 20, 199.	2.1	12
89	Dominant mutations of the Notch ligand Jagged1 cause peripheral neuropathy. Journal of Clinical Investigation, 2020, 130, 1506-1512.	3.9	12
90	A clinical, genetic and candidate gene study of Silver syndrome, a complicated form of hereditary spastic paraplegia. Journal of Neurology, 2004, 251, 1068-74.	1.8	11

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91	No association between SCN9A and monogenic human epilepsy disorders. PLoS Genetics, 2020, 16, e1009161.	1.5	11
92	Homozygosity at chromosome 8qter in individuals affected by mal de Meleda (Meleda disease) originating from the island of Meleda. British Journal of Dermatology, 2001, 144, 731-734.	1.4	10
93	BBS5 and INPP5E mutations associated with ciliopathy disorders in families from Pakistan. Annals of Human Genetics, 2019, 83, 477-482.	0.3	10
94	Delineating the expanding phenotype associated with SCAPER gene mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1665-1671.	0.7	10
95	Dominant mitochondrial membrane protein-associated neurodegeneration (MPAN) variants cluster within a specific C19orf12 isoform. Parkinsonism and Related Disorders, 2021, 82, 84-86.	1.1	10
96	Biallelic variants in TRAPPC10 cause a microcephalic TRAPPopathy disorder in humans and mice. PLoS Genetics, 2022, 18, e1010114.	1.5	10
97	Distal hereditary motor neuropathy with vocal cord paresis: from difficulty in choral singing to a molecular genetic diagnosis. Practical Neurology, 2016, 16, 247-251.	0.5	9
98	Mutations in TYR and OCA2 associated with oculocutaneous albinism in Pakistani families. Meta Gene, 2018, 17, 48-55.	0.3	9
99	A small gene sequencing panel realises a high diagnostic rate in patients with congenital nystagmus following basic phenotyping. Scientific Reports, 2019, 9, 13229.	1.6	9
100	A quantitative LC-MS/MS method for analysis of mitochondrial -specific oxysterol metabolism. Redox Biology, 2020, 36, 101595.	3.9	9
101	Evidence that the Ser192Tyr/Arg402Gln in cis Tyrosinase gene haplotype is a disease-causing allele in oculocutaneous albinism type 1B (OCA1B). Npj Genomic Medicine, 2022, 7, 2.	1.7	9
102	PCNA mutation affects DNA repair not replication. Cell Cycle, 2014, 13, 3157-3158.	1.3	8
103	Identification of novel L2HGDH mutation in a large consanguineous Pakistani family- a case report. BMC Medical Genetics, 2018, 19, 25.	2.1	8
104	Novel nonsense variants in SLURP1 and DSG1 cause palmoplantar keratoderma in Pakistani families. BMC Medical Genetics, 2019, 20, 145.	2.1	8
105	Copy number variation of LINGO1 in familial dystonic tremor. Neurology: Genetics, 2019, 5, e307.	0.9	8
106	Refinement of the locus for distal hereditary motor neuronopathy VII (dHMN-VII) and exclusion of candidate genes. Genome, 2008, 51, 959-962.	0.9	7
107	A biallelic SNIP1 Amish founder variant causes a recognizable neurodevelopmental disorder. PLoS Genetics, 2021, 17, e1009803.	1.5	7
108	Striking phenotypic variability in familial TRPV4â€axonal neuropathy spectrum disorder. American Journal of Medical Genetics, Part A, 2011, 155, 3153-3156.	0.7	6

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109	A homozygous loss-of-function mutation in PTPN14 causes a syndrome of bilateral choanal atresia and early infantile-onset lymphedema. Meta Gene, 2017, 14, 53-58.	0.3	6
110	A large Indian family with rearrangement of chromosome 4p16 and 3p26.3 and divergent clinical presentations. BMC Medical Genetics, 2015, 16, 104.	2.1	5
111	Tyrosinase (TYR) gene sequencing and literature review reveals recurrent mutations and multiple population founder gene mutations as causative of oculocutaneous albinism (OCA) in Pakistani families. Eye, 2019, 33, 1339-1346.	1.1	5
112	Final Exon Frameshift Biallelic PTPN23 Variants Are Associated with Microcephalic Complex Hereditary Spastic Paraplegia. Brain Sciences, 2021, 11, 614.	1.1	5
113	Consolidating biallelic SDHD variants as a cause of mitochondrial complex II deficiency. European Journal of Human Genetics, 2021, 29, 1570-1576.	1.4	3
114	Variants in NIPAL4 and ALOXE3 cause autosomal recessive congenital ichthyosis in Pakistani families. Congenital Anomalies (discontinued), 2020, 60, 149-150.	0.3	2
115	Biochemical phenotype and its relationship to treatment in 16 individuals with PCCB c.1606AÂ>ÂG (p.Asn536Asp) variant propionic acidemia. Molecular Genetics and Metabolism, 2020, 131, 316-324.	0.5	2
116	A Novel Mutation in the OFD1 Gene in a Family with Oral-Facial-Digital Syndrome Type 1: A Case Report. Iranian Journal of Public Health, 2016, 45, 1359-1366.	0.3	2
117	Datasets of whole cell and mitochondrial oxysterols derived from THP-1, SH-SY5Y and human peripheral blood mononuclear cells using targeted metabolomics. Data in Brief, 2020, 33, 106382.	0.5	1
118	Novel mutations in PDE6A and CDHR1 cause retinitis pigmentosa in Pakistani families. International Journal of Ophthalmology, 2021, 14, 1843-1851.	0.5	1
119	CLCC1 c. 75C>A Mutation in Pakistani Derived Retinitis Pigmentosa Families Likely Originated With a Single Founder Mutation 2,000â€“5,000 Years Ago. Frontiers in Genetics, 2022, 13, 804924.	1.1	1
120	DISTAL SPINAL MUSCULAR ATROPHY WITH VOCAL PARESIS: FROM THE WELSH CHOIR TO GENES. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, e2.117-e2.	0.9	0
121	Reply: PRUNE1: a disease-causing gene for secondary microcephaly. Brain, 2017, 140, e62-e62.	3.7	0
122	In Silico analysis of SIGMAR1 gene causing distal hereditary motor neuropathy in a Pakistani family. Gene Reports, 2019, 16, 100445.	0.4	0
123	A recurrent rare intronic variant in CAPN3 alters mRNA splicing and causes autosomal recessive limbâ€girdle muscular dystrophyâ€1 in three Pakistani pedigrees. American Journal of Medical Genetics, Part A, 2021, , .	0.7	0
124	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	1.1	0