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	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	1.1	0
2	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
3	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
4	Biallelic variants in TRAPPC10 cause a microcephalic TRAPPopathy disorder in humans and mice. PLoS Genetics, 2022, 18, e1010114.	1.5	10
5	TMEM63C mutations cause mitochondrial morphology defects and underlie hereditary spastic paraplegia. Brain, 2022, 145, 3095-3107.	3.7	17
6	Assessing performance of pathogenicity predictors using clinically relevant variant datasets. Journal of Medical Genetics, 2021, 58, 547-555.	1.5	57
7	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
8	Consolidating biallelic SDHD variants as a cause of mitochondrial complex II deficiency. European Journal of Human Genetics, 2021, 29, 1570-1576.	1.4	3
9	Final Exon Frameshift Biallelic PTPN23 Variants Are Associated with Microcephalic Complex Hereditary Spastic Paraplegia. Brain Sciences, 2021, 11, 614.	1.1	5
10	Biallelic PI4KA variants cause neurological, intestinal and immunological disease. Brain, 2021, 144, 3597-3610.	3.7	17
11	A biallelic SNIP1 Amish founder variant causes a recognizable neurodevelopmental disorder. PLoS Genetics, 2021, 17, e1009803.	1.5	7
12	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
13	Novel mutations in PDE6A and CDHR1 cause retinitis pigmentosa in Pakistani families. International Journal of Ophthalmology, 2021, 14, 1843-1851.	0.5	1
14	MNS1 variant associated with situs inversus and male infertility. European Journal of Human Genetics, 2020, 28, 50-55.	1.4	19
15	Lipid metabolic pathways converge in motor neuron degenerative diseases. Brain, 2020, 143, 1073-1087.	3.7	36
16	Variants in NIPAL4 and ALOXE3 cause autosomal recessive congenital ichthyosis in Pakistani families. Congenital Anomalies (discontinued), 2020, 60, 149-150.	0.3	2
17	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
18	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

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19	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
20	A quantitative LC-MS/MS method for analysis of mitochondrial -specific oxysterol metabolism. Redox Biology, 2020, 36, 101595.	3.9	9
21	Dominant mutations of the Notch ligand Jagged1 cause peripheral neuropathy. Journal of Clinical Investigation, 2020, 130, 1506-1512.	3.9	12
22	No association between SCN9A and monogenic human epilepsy disorders. PLoS Genetics, 2020, 16, e1009161.	1.5	11
23	Hereditary spastic paraplegia: from diagnosis to emerging therapeutic approaches. Lancet Neurology, The, 2019, 18, 1136-1146.	4.9	171
24	In Silico analysis of SIGMAR1 gene causing distal hereditary motor neuropathy in a Pakistani family. Gene Reports, 2019, 16, 100445.	0.4	0
25	Novel nonsense variants in SLURP1 and DSG1 cause palmoplantar keratoderma in Pakistani families. BMC Medical Genetics, 2019, 20, 145.	2.1	8
26	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
27	BBS5 and INPP5E mutations associated with ciliopathy disorders in families from Pakistan. Annals of Human Genetics, 2019, 83, 477-482.	0.3	10
28	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. Genome Research, 2019, 29, 1057-1066.	2.4	38
29	Delineating the expanding phenotype associated with SCAPER gene mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1665-1671.	0.7	10
30	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
31	Copy number variation of LINGO1 in familial dystonic tremor. Neurology: Genetics, 2019, 5, e307.	0.9	8
32	Homozygous variants in the HEXB and MBOAT7 genes underlie neurological diseases in consanguineous families. BMC Medical Genetics, 2019, 20, 199.	2.1	12
33	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	1.1	52
34	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
35	Truncating SLC5A7 mutations underlie a spectrum of dominant hereditary motor neuropathies. Neurology: Genetics, 2018, 4, e222.	0.9	19
36	Mutations in TYR and OCA2 associated with oculocutaneous albinism in Pakistani families. Meta Gene, 2018, 17, 48-55.	0.3	9

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37	Identification of novel L2HGDH mutation in a large consanguineous Pakistani family- a case report. BMC Medical Genetics, 2018, 19, 25.	2.1	8
38	Novel mutations in ALDH1A3 associated with autosomal recessive anophthalmia/microphthalmia, and review of the literature. BMC Medical Genetics, 2018, 19, 160.	2.1	12
39	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	1.5	25
40	PCNA dependent cellular activities tolerate dramatic perturbations in PCNA client interactions. DNA Repair, 2017, 50, 22-35.	1.3	12
41	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
42	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
43	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952.	3.7	62
44	Choline transporter mutations in severe congenital myasthenic syndrome disrupt transporter localization. Brain, 2017, 140, 2838-2850.	3.7	24
45	Reply: PRUNE1: a disease-causing gene for secondary microcephaly. Brain, 2017, 140, e62-e62.	3.7	0
46	A mutation ofEPT1 (SELENOI)underlies a new disorder of Kennedy pathway phospholipid biosynthesis. Brain, 2017, 140, aww318.	3.7	58
47	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
48	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
49	Novel Genetic, Clinical, and Pathomechanistic Insights into TFG-Associated Hereditary Spastic Paraplegia. Human Mutation, 2016, 37, 1157-1161.	1.1	17
50	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
51	Spectrum of DNA variants for non-syndromic deafness in a large cohort from multiple continents. Human Genetics, 2016, 135, 953-961.	1.8	102
52	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
53	The HERC2 ubiquitin ligase is essential for embryonic development and regulates motor coordination. Oncotarget, 2016, 7, 56083-56106.	0.8	24
54	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

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55	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
56	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
57	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
58	Loss of PCLO function underlies pontocerebellar hypoplasia type III. Neurology, 2015, 84, 1745-1750.	1.5	45
59	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
60	PCNA mutation affects DNA repair not replication. Cell Cycle, 2014, 13, 3157-3158.	1.3	8
61	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
62	Raine syndrome: An overview. European Journal of Medical Genetics, 2014, 57, 536-542.	0.7	43
63	Mutations in KPTN Cause Macrocephaly, Neurodevelopmental Delay, and Seizures. American Journal of Human Genetics, 2014, 94, 87-94.	2.6	35
64	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
65	Hypomorphic PCNA mutation underlies a human DNA repair disorder. Journal of Clinical Investigation, 2014, 124, 3137-3146.	3.9	77
66	Deficiency of terminal ADP-ribose protein glycohydrolase TARG1/C6orf130 in neurodegenerative disease. EMBO Journal, 2013, 32, 1225-1237.	3.5	263
67	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. Brain, 2013, 136, 3618-3624.	3.7	115
68	Mutation of HERC2 causes developmental delay with Angelman-like features. Journal of Medical Genetics, 2013, 50, 65-73.	1.5	57
69	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
70	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
71	SLITRK6 mutations cause myopia and deafness in humans and mice. Journal of Clinical Investigation, 2013, 123, 2094-2102.	3.9	50
72	Defective Presynaptic Choline Transport Underlies Hereditary Motor Neuropathy. American Journal of Human Genetics, 2012, 91, 1103-1107.	2.6	89

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73	Striking phenotypic variability in familial TRPV4â€axonal neuropathy spectrum disorder. American Journal of Medical Genetics, Part A, 2011, 155, 3153-3156.	0.7	6
74	Defective Mitochondrial mRNA Maturation Is Associated with Spastic Ataxia. American Journal of Human Genetics, 2010, 87, 655-660.	2.6	76
75	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
76	Mutation of FA2H underlies a complicated form of hereditary spastic paraplegia (SPG35). Human Mutation, 2010, 31, E1251-E1260.	1.1	174
77	Alterations in the ankyrin domain of TRPV4 cause congenital distal SMA, scapuloperoneal SMA and HMSN2C. Nature Genetics, 2010, 42, 160-164.	9.4	228
78	Raine syndrome: a clinical, radiographic and genetic investigation of a case from the Indian subcontinent. Clinical Dysmorphology, 2010, 19, 153-156.	0.1	19
79	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
80	CYP7B1 mutations in pure and complex forms of hereditary spastic paraplegia type 5. Brain, 2009, 132, 1589-1600.	3.7	102
81	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
82	Analysis of CYP7B1 in non-consanguineous cases of hereditary spastic paraplegia. Neurogenetics, 2009, 10, 97-104.	0.7	54
83	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
84	Mutations in FAM20C also identified in nonâ€lethal osteosclerotic bone dysplasia. Clinical Genetics, 2009, 75, 271-276.	1.0	97
85	Sequence Alterations within CYP7B1 Implicate Defective Cholesterol Homeostasis in Motor-Neuron Degeneration. American Journal of Human Genetics, 2008, 82, 510-515.	2.6	171
86	Hereditary spastic paraplegia: clinical features and pathogenetic mechanisms. Lancet Neurology, The, 2008, 7, 1127-1138.	4.9	481
87	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
88	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
89	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
90	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

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91	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
92	The natural history of Noonan syndrome: a long-term follow-up study. Archives of Disease in Childhood, 2006, 92, 128-132.	1.0	218
93	Genetic heterogeneity in LEOPARD syndrome: two families with no mutations in PTPN11. Journal of Human Genetics, 2005, 50, 21-25.	1.1	25
94	A novel NIPA1 mutation associated with a pure form of autosomal dominant hereditary spastic paraplegia. Neurogenetics, 2005, 6, 79-84.	0.7	51
95	Novel Mutation in Desmoplakin Causes Arrhythmogenic Left Ventricular Cardiomyopathy. Circulation, 2005, 112, 636-642.	1.6	266
96	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
97	Complicated Hereditary Spastic Paraplegia with Thin Corpus Callosum (HSPâ€TCC) and Childhood Onset. Neuropediatrics, 2005, 36, 274-278.	0.3	21
98	A clinical, genetic and biochemical study of SPG7 mutations in hereditary spastic paraplegia. Brain, 2004, 127, 973-980.	3.7	77
99	Heterozygous missense mutations in BSCL2 are associated with distal hereditary motor neuropathy and Silver syndrome. Nature Genetics, 2004, 36, 271-276.	9.4	349
100	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
101	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
102	Troyer syndrome revisited. Journal of Neurology, 2004, 251, 1105-10.	1.8	42
103	Complicated hereditary spastic paraplegia with thin corpus callosum: Variation of phenotypic expression over time. Journal of Neurology, 2004, 251, 1285-1287.	1.8	13
104	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
105	Disruption of cellular transport: a common cause of neurodegeneration?. Lancet Neurology, The, 2003, 2, 311-316.	4.9	28
106	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
107	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
108	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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109	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
110	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
111	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
112	Is the Transportation Highway the Right Road for Hereditary Spastic Paraplegia?. American Journal of Human Genetics, 2002, 71, 1009-1016.	2.6	119
113	SPG20 is mutated in Troyer syndrome, an hereditary spastic paraplegia. Nature Genetics, 2002, 31, 347-348.	9.4	240
114	Absence of PTPN11 mutations in 28 cases of cardiofaciocutaneous (CFC) syndrome. Human Genetics, 2002, 111, 421-427.	1.8	45
115	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
116	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
117	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. Nature Genetics, 2001, 29, 465-468.	9.4	1,555
118	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
119	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
120	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
121	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
122	Mapping of the human and mouse bone sialoprotein and osteopontin loci. Mammalian Genome, 1996, 7, 149-151.	1.0	32
123	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
124	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