Anne M Slavotinek

List of Publications by Year
in descending order

128	3,831	145106	182931
papers	citations	33 h-index	54 g-index

137	137	137	7065
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Preference for secondary findings in prenatal and pediatric exome sequencing. Prenatal Diagnosis, 2022, 42, 753-761.	1.1	11
2	US private payersâ€™ perspectives on insurance coverage for genome sequencing versus exome sequencing: A study by the Clinical Sequencing Evidence-Generating Research Consortium (CSER). Genetics in Medicine, 2022, 24, 238-244.	1.1	6
3	Prenatal presentation of multiple anomalies associated with haploinsufficiency for ARID1A. European Journal of Medical Genetics, 2022, 65, 104407.	0.7	7
4	Predicting genes from phenotypes using human phenotype ontology (HPO) terms. Human Genetics, 2022, 141, 1749-1760.	1.8	1
5	Impact of the <scp>COVID</scp> â€19 pandemic on medical genetics and genomics training: Perspective from clinical trainees. American Journal of Medical Genetics, Part A, 2022, , .	0.7	2
6	Perspectives and preferences regarding genomic secondary findings in underrepresented prenatal and pediatric populations: A mixed-methods approach. Genetics in Medicine, 2022, 24, 1206-1216.	1.1	8
7	Expanding the phenotype of males with OFD1 pathogenic variants-a case report and literature review. European Journal of Medical Genetics, 2022, , 104496.	0.7	2
8	S1P defects cause a new entity of cataract, alopecia, oral mucosal disorder, and psoriasisâ€like syndrome. EMBO Molecular Medicine, 2022, 14, e14904.	3.3	11
9	<scp>Lateâ€onset</scp> Proteus syndrome with cerebriform connective tissue nevus and subsequent development of intraductal papilloma. American Journal of Medical Genetics, Part A, 2022, , .	0.7	0
10	Suleiman-El-Hattab syndrome: a histone modification disorder caused by TASP1 deficiency. Human Molecular Genetics, 2022, 31, 3083-3094.	1.4	3
11	Further description of two patients with biallelic variants in <scp>NADSYN1</scp> in association with cardiac and vertebral anomalies. American Journal of Medical Genetics, Part A, 2022, 188, 2479-2484.	0.7	8
12	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
13	Genetic Testing Leading to Early Identification of Childhood Ocular Manifestations of Usher Syndrome. Laryngoscope, 2021, 131, E2053-E2059.	1.1	10
14	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	2.6	71
15	Further delineation of BCAP31-linked intellectual disability: description of 17 new families with LoF and missense variants. European Journal of Human Genetics, 2021, 29, 1405-1417.	1.4	3
16	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	0.7	34
17	Third case of <scp>Bardetâ€Biedl</scp> syndrome caused by a biallelic variant predicted to affect splicing of <scp>IFT74</scp>. Clinical Genetics, 2021, 100, 93-99.	1.0	12
18	Ensuring best practice in genomics education and evaluation: reporting item standards for education and its evaluation in genomics (RISE2 Genomics). Genetics in Medicine, 2021, 23, 1356-1365.	1.1	17

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19	Pitfalls and challenges in genetic test interpretation: An exploration of genetic professionals experience with interpretation of results. Clinical Genetics, 2021, 99, 638-649.	1.0	15
20	Quantitative analysis of the natural history of prolidase deficiency: description of 17 families and systematic review of published cases. Genetics in Medicine, 2021, 23, 1604-1615.	1.1	10
21	New cases that expand the genotypic and phenotypic spectrum of Congenital NAD Deficiency Disorder. Human Mutation, 2021, 42, 862-876.	1.1	16
22	Genotypeâ€“phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. Genome Medicine, 2021, 13, 90.	3.6	16
23	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	2.6	31
24	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	2.6	17
25	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	0.7	12
26	Application of full-genome analysis to diagnose rare monogenic disorders. Npj Genomic Medicine, 2021, 6, 77.	1.7	22
27	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	2.6	0
28	Clinical and molecular features of 66 patients with musculocontractural Ehlersâ^'Danlos syndrome caused by pathogenic variants in CHST14 (mcEDS-CHST14). Journal of Medical Genetics, 2021, , jmedgenet-2020-107623.	1.5	18
29	The difficulties of broad data sharing in genomic medicine: Empirical evidence from diverse participants in prenatal and pediatric clinical genomics research. Genetics in Medicine, 2021, , .	1.1	5
30	Case Report of Floating-Harbor Syndrome With Bilateral Cleft Lip. Cleft Palate-Craniofacial Journal, 2020, 57, 132-136.	0.5	3
31	Perspectives of US private payers on insurance coverage for pediatric and prenatal exome sequencing: Results of a study from the Program in Prenatal and Pediatric Genomic Sequencing (P3EGS). Genetics in Medicine, 2020, 22, 283-291.	1.1	41
32	Functional characterization of a novel PBX1 de novo missense variant identified in a patient with syndromic congenital heart disease. Human Molecular Genetics, 2020, 29, 1068-1082.	1.4	26
33	A novel truncating variant in ring finger protein 113A (RNF113A) confirms the association of this gene with Xâ€linked trichothiodystrophy. American Journal of Medical Genetics, Part A, 2020, 182, 513-520.	0.7	12
34	The expanding spectrum of NFIB â€associated phenotypes in a diverse patient populationâ€”A report of two new patients. American Journal of Medical Genetics, Part A, 2020, 182, 2959-2963.	0.7	3
35	Going forward in a new world. American Journal of Medical Genetics, Part A, 2020, 182, 1553-1554.	0.7	5
36	Announcing a new manuscript category for the <scp>American Journal of Medical Genetics</scp> Part A: Dispatches from Biotech. American Journal of Medical Genetics, Part A, 2020, 182, 2003-2004.	0.7	1

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37	Exome sequencing in patients with microphthalmia, anophthalmia, and coloboma (MAC) from a consanguineous population. Clinical Genetics, 2020, 98, 499-506.	1.0	7
38	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22
39	Automated syndrome diagnosis by three-dimensional facial imaging. Genetics in Medicine, 2020, 22, 1682-1693.	1.1	47
40	Biallelic variants in the RNA exosome gene EXOSC5 are associated with developmental delays, short stature, cerebellar hypoplasia and motor weakness. Human Molecular Genetics, 2020, 29, 2218-2239.	1.4	19
41	Baraitserâ€“Winter cerebrofrontofacial syndrome: Report of two adult siblings. American Journal of Medical Genetics, Part A, 2020, 182, 1923-1932.	0.7	2
42	A missense variant, p.(Ile269Asn), in MC4R as a secondary finding in a child with BCL11A-related intellectual disability. European Journal of Medical Genetics, 2020, 63, 103969.	0.7	1
43	Novel PXDN biallelic variants in patients with microphthalmia and anterior segment dysgenesis. Journal of Human Genetics, 2020, 65, 487-491.	1.1	5
44	Jumonji domain containing 1C (JMJD1C) sequence variants in seven patients with autism spectrum disorder, intellectual disability and seizures. European Journal of Medical Genetics, 2020, 63, 103850.	0.7	3
45	Modeling Pathogenic Variants in the RNA Exosome. RNA & Disease (Houston, Tex), 2020, 7, .	1.0	1
46	Private payer coverage policies for exome sequencing (ES) in pediatric patients: trends over time and analysis of evidence cited. Genetics in Medicine, 2019, 21, 152-160.	1.1	29
47	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	2.6	43
48	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 631-639.	2.6	42
49	TASP1 mutation in a female with craniofacial anomalies, anterior segment dysgenesis, congenital immunodeficiency and macrocytic anemia. Molecular Genetics & Genomic Medicine, 2019, 7, e818.	0.6	6
50	Developmental and epileptic encephalopathy in two siblings with a novel, homozygous missense variant in SCN1B. American Journal of Medical Genetics, Part A, 2019, 179, 2190-2195.	0.7	10
51	The Genomic Medicine Integrative Research Framework: A Conceptual Framework for Conducting Genomic Medicine Research. American Journal of Human Genetics, 2019, 104, 1088-1096.	2.6	35
52	Introducing in AJMG Part A : Genetic Syndromes in Adults. American Journal of Medical Genetics, Part A, 2019, 179, 1413-1414.	0.7	3
53	Use of PTC124 for nonsense suppression therapy targeting BMP4 nonsense variants in vitro and the bmp4st72 allele in zebrafish. PLoS ONE, 2019, 14, e0212121.	1.1	5
54	NAA10 polyadenylation signal variants cause syndromic microphthalmia. Journal of Medical Genetics, 2019, 56, 444-452.	1.5	28

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55	Biallelic sequence variants in INTS1 in patients with developmental delays, cataracts, and craniofacial anomalies. European Journal of Human Genetics, 2019, 27, 582-593.	1.4	23
56	Genetics of anophthalmia and microphthalmia. PartÂ2: Syndromes associated with anophthalmiaâ€“microphthalmia. Human Genetics, 2019, 138, 831-846.	1.8	47
57	Congenital sodium diarrhea and chorioretinal coloboma with optic disc coloboma in a patient with biallelic SPINT2 mutations, including p.(Tyr163Cys). American Journal of Medical Genetics, Part A, 2018, 176, 997-1000.	0.7	10
58	Natural history and genotypeâ€phenotype correlations in 72 individuals with SATB2â€associated syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 925-935.	0.7	57
59	Variants in members of the cadherinâ€“catenin complex, CDH1 and CTNND1, cause blepharocheilodontic syndrome. European Journal of Human Genetics, 2018, 26, 210-219.	1.4	34
60	Mutation update of transcription factor genesFOXE3,HSF4,MAF, andPITX3causing cataracts and other developmental ocular defects. Human Mutation, 2018, 39, 471-494.	1.1	60
61	Cover Image, Volume 176A, Number 4, April 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	0.7	0
62	A zebrafish model of foxe3 deficiency demonstrates lens and eye defects with dysregulation of key genes involved in cataract formation in humans. Human Genetics, 2018, 137, 315-328.	1.8	26
63	A randomized controlled trial of levodopa in patients with Angelman syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1099-1107.	0.7	18
64	Two patients with FOXF1 mutations with alveolar capillary dysplasia with misalignment of pulmonary veins and other malformations: Two different presentations and outcomes. American Journal of Medical Genetics, Part A, 2018, 176, 2877-2881.	0.7	8
65	Evidenceâ€based assessments of clinical actionability in the context of secondary findings: Updates from ClinGen's Actionability Working Group. Human Mutation, 2018, 39, 1677-1685.	1.1	34
66	The Clinical Sequencing Evidence-Generating Research Consortium: Integrating Genomic Sequencing in Diverse and Medically Underserved Populations. American Journal of Human Genetics, 2018, 103, 319-327.	2.6	122
67	Early inspirations from times gone by. American Journal of Medical Genetics, Part A, 2018, 176, 1797-1798.	0.7	1
68	EXTL3 mutations cause skeletal dysplasia, immune deficiency, and developmental delay. Journal of Experimental Medicine, 2017, 214, 623-637.	4.2	76
69	Inner retinal dystrophy in a patient with biallelic sequence variants in BRAT1. Ophthalmic Genetics, 2017, 38, 559-561.	0.5	6
70	Clinical Report: Warsaw Breakage Syndrome with small radii and fibulae. American Journal of Medical Genetics, Part A, 2017, 173, 3075-3081.	0.7	16
71	De novo, deleterious sequence variants that alter the transcriptional activity of the homeoprotein PBX1 are associated with intellectual disability and pleiotropic developmental defects. Human Molecular Genetics, 2017, 26, 4849-4860.	1.4	42
72	HLX is a candidate gene for a pattern of anomalies associated with congenital diaphragmatic hernia, short bowel, and asplenia. American Journal of Medical Genetics, Part A, 2017, 173, 3070-3074.	0.7	10

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73	A recurrent, non-penetrant sequence variant, p.Arg266Cys in Growth/Differentiation Factor 3 (GDF3) in a female with unilateral anophthalmia and skeletal anomalies. American Journal of Ophthalmology Case Reports, 2017, 7, 102-106.	0.4	4
74	Clinical report: A patient with a late diagnosis of cerebrotendinous xanthomatosis and a response to treatment. American Journal of Medical Genetics, Part A, 2017, 173, 2275-2279.	0.7	10
75	Pierpont syndrome associated with the p.Tyr446Cys missense mutation in TBL1XR1. European Journal of Medical Genetics, 2017, 60, 504-508.	0.7	15
76	The focal facial dermal dysplasias: phenotypic spectrum and molecular genetic heterogeneity. Journal of Medical Genetics, 2017, 54, 585-590.	1.5	10
77	Characterization of a variant of gap junction protein Î±8 identified in a family with hereditary cataract. PLoS ONE, 2017, 12, e0183438.	1.1	6
78	Pregnancy and Birth Outcomes among Women with Idiopathic Thrombocytopenic Purpura. Journal of Pregnancy, 2016, 2016, 1-8.	1.1	23
79	Corneal clouding, cataract, and colobomas with a novel missense mutation in B4GALT7â€"a review of eye anomalies in the linkeropathy syndromes. American Journal of Medical Genetics, Part A, 2016, 170, 2711-2718.	0.7	19
80	An emerging, recognizable facial phenotype in association with mutations in GLIâ€similar 3 (GLIS3). American Journal of Medical Genetics, Part A, 2016, 170, 1918-1923.	0.7	16
81	The Family of Crumbs Genes and Human Disease. Molecular Syndromology, 2016, 7, 274-281.	0.3	29
82	A standardized, evidence-based protocol to assess clinical actionability of genetic disorders associated with genomic variation. Genetics in Medicine, 2016, 18, 1258-1268.	1.1	89
83	Challenges managing endâ€stage renal disease and kidney transplantation in a child with MTFMT mutation and moyamoya disease. Pediatric Transplantation, 2016, 20, 1000-1003.	0.5	5
84	Clinical care models in the era of nextâ€generation sequencing. Molecular Genetics & Genomic Medicine, 2016, 4, 239-242.	0.6	1
85	The phenotype of the musculocontractural type of Ehlersâ€Danlos syndrome due to CHST14 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 103-115.	0.7	53
86	Genetic analysis of consanguineous families presenting with congenital ocular defects. Experimental Eye Research, 2016, 146, 163-171.	1.2	21
87	Expansion of phenotype and genotypic data in CRB2-related syndrome. European Journal of Human Genetics, 2016, 24, 1436-1444.	1.4	36
88	A much needed new journal in the field of Pediatric Genetics. Journal of Pediatric Genetics, 2015, 01, 001-002.	0.3	0
89	Recurrent duplications of 17q12 associated with variable phenotypes. American Journal of Medical Genetics, Part A, 2015, 167, 3038-3045.	0.7	22
90	Novel KIF7 missense substitutions in two patients presenting with multiple malformations and features of acrocallosal syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2767-2776.	0.7	9

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91	Exome sequencing in 32 patients with anophthalmia/microphthalmia and developmental eye defects. Clinical Genetics, 2015, 88, 468-473.	1.0	55
92	Novel de novo heterozygous loss-of-function variants in MED13L and further delineation of the MED13L haploinsufficiency syndrome. European Journal of Human Genetics, 2015, 23, 1499-1504.	1.4	36
93	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183.	2.9	211
94	CRB2 Mutations Produce a Phenotype Resembling Congenital Nephrosis, Finnish Type, with Cerebral Ventriculomegaly and Raised Alpha-Fetoprotein. American Journal of Human Genetics, 2015, 96, 162-169.	2.6	73
95	Recognizable Syndromes in the Newborn Period. Clinics in Perinatology, 2015, 42, 263-280.	0.8	2
96	DLX4is associated with orofacial clefting and abnormal jaw development. Human Molecular Genetics, 2015, 24, 4340-4352.	1.4	36
97	Expanding the phenotypic spectrum of PORCN variants in two males with syndromic microphthalmia. European Journal of Human Genetics, 2015, 23, 551-554.	1.4	24
98	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. Lancet Neurology, The, 2015, 14, 1182-1195.	4.9	74
99	Haploinsufficiency and triploinsensitivity of the same 6p25.1p24.3 region in a family. BMC Medical Genomics, 2015, 8, 38.	0.7	7
100	Novel mutations in PXDN cause microphthalmia and anterior segment dysgenesis. European Journal of Human Genetics, 2015, 23, 337-341.	1.4	42
101	The FgfrL1 receptor is required for development of slow muscle fibers. Developmental Biology, 2014, 394, 228-241.	0.9	25
102	The genetics of common disorders â€“ Congenital diaphragmatic hernia. European Journal of Medical Genetics, 2014, 57, 418-423.	0.7	48
103	ALDH1A3 loss of function causes bilateral anophthalmia/microphthalmia and hypoplasia of the optic nerve and optic chiasm. Human Molecular Genetics, 2013, 22, 3250-3258.	1.4	68
104	Focal facial dermal dysplasia, type IV, is caused by mutations in CYP26C1. Human Molecular Genetics, 2013, 22, 696-703.	1.4	41
105	VAX1mutation associated with microphthalmia, corpus callosum agenesis, and orofacial clefting: The first description of aVAX1phenotype in humans. Human Mutation, 2012, 33, 364-368.	1.1	59
106	Eye development genes and known syndromes. Molecular Genetics and Metabolism, 2011, 104, 448-456.	0.5	96
107	Manitoba-oculo-tricho-anal (MOTA) syndrome is caused by mutations in FREM1. Journal of Medical Genetics, 2011, 48, 375-382.	1.5	60
108	Clinical utility gene card for: Bardetâ€“Biedl syndrome. European Journal of Human Genetics, 2011, 19, 3-4.	1.4	8

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109	A de novo deletion of CALN1 in a male with a bilateral diaphragmatic defect does not definitely cause this malformation. American Journal of Medical Genetics, Part A, 2011, 155, 1196-1201.	0.7	1
110	Expanded spectrum of oculoâ€“auriculoâ€“vertebral spectrum with imperforate anus in a male patient who is negative for SALL1 mutations. Clinical Dysmorphology, 2011, 20, 11-14.	0.1	2
111	A Male with Unilateral Microphthalmia Reveals a Role for TMX3 in Eye Development. PLoS ONE, 2010, 5, e10565.	1.1	34
112	Two novel STRA6 mutations in a patient with anophthalmia and diaphragmatic eventration. American Journal of Medical Genetics, Part A, 2009, 149A, 539-542.	0.7	20
113	Novel FGFR2 deletion in a patient with Beareâ€“Stevensonâ€like syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1814-1817.	0.7	22
114	Sequence variants in the HLX gene at chromosome 1q41â€•1q42 in patients with diaphragmatic hernia. Clinical Genetics, 2009, 75, 429-439.	1.0	30
115	Novel microdeletion syndromes detected by chromosome microarrays. Human Genetics, 2008, 124, 1-17.	1.8	198
116	Mutations in STRA6 Cause a Broad Spectrum of Malformations Including Anophthalmia, Congenital Heart Defects, Diaphragmatic Hernia, Alveolar Capillary Dysplasia, Lung Hypoplasia, and Mental Retardation. American Journal of Human Genetics, 2007, 80, 550-560.	2.6	316
117	Populationâ€based analysis of leftâ€•and rightâ€sided diaphragmatic hernias demonstrates different frequencies of selected additional anomalies. American Journal of Medical Genetics, Part A, 2007, 143A, 3127-3136.	0.7	31
118	Single gene disorders associated with congenital diaphragmatic hernia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2007, 145C, 172-183.	0.7	52
119	Array comparative genomic hybridization in patients with congenital diaphragmatic hernia: mapping of four CDH-critical regions and sequencing of candidate genes at 15q26.1â€“15q26.2. European Journal of Human Genetics, 2006, 14, 999-1008.	1.4	91
120	Marinesco-SjÃ¶gren syndrome in a male with mild dysmorphism. , 2005, 133A, 197-201.		19
121	Craniofacial defects of blastogenesis: Duplication of pituitary with cleft palate and orophgaryngeal tumors. American Journal of Medical Genetics, Part A, 2005, 135A, 13-20.	0.7	31
122	Case reports of oculofaciocardiodental syndrome with unusual dental findings. American Journal of Medical Genetics, Part A, 2005, 136A, 275-277.	0.7	41
123	A family with X-linked anophthalmia: Exclusion ofSOX3 as a candidate gene. American Journal of Medical Genetics, Part A, 2005, 138A, 89-94.	0.7	4
124	Acro-dermato-ungual-lacrimal-tooth (ADULT) syndrome: Report of a child with phenotypic overlap with ulnar-mammary syndrome and a new mutation inTP63. American Journal of Medical Genetics, Part A, 2005, 138A, 146-149.	0.7	23
125	The Genetics of Congenital Diaphragmatic Hernia. Seminars in Perinatology, 2005, 29, 77-85.	1.1	51
126	Fryns syndrome: A review of the phenotype and diagnostic guidelines. American Journal of Medical Genetics Part A, 2004, 124A, 427-433.	2.4	97

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127	Non-syndromic hemihyperplasia in a male and his mother. American Journal of Medical Genetics Part A, 2003, 121A, 47-51.	2.4	2
128	Genetic modifiers in human development and malformation syndromes, including chaperone proteins. Human Molecular Genetics, 2003, 12, 45R-50.	1.4	46