Allyn McConkie-Rosell

List of Publications by Year
in descending order

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58	1,665	257450 24	315739 38
papers	citations	h-index	g-index

62	62	62	2502
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The utility of the traditional medical genetics diagnostic evaluation in the context of next-generation sequencing for undiagnosed genetic disorders. Genetics in Medicine, 2014, 16, 176-182.	2.4	239
2	Genetic Counseling for Fragile X Syndrome: Updated Recommendations of the National Society of Genetic Counselors. Journal of Genetic Counseling, 2005, 14, 249-270.	1.6	103
3	Recommendations from Multi-disciplinary Focus Groups on Cascade Testing and Genetic Counseling for Fragile X-associated Disorders. Journal of Genetic Counseling, 2007, 16, 593-606.	1.6	70
4	â€œFamily Mattersâ€• A Conceptual Framework for Genetic Testing in Children. Journal of Genetic Counseling, 2004, 13, 9-29.	1.6	65
5	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
6	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. Human Molecular Genetics, 2018, 27, 2454-2465.	2.9	54
7	Carrier testing in the fragile X syndrome: Attitudes and opinions of obligate carriers. , 1997, 68, 62-69.		53
8	A window into living with an undiagnosed disease: illness narratives from the Undiagnosed Diseases Network. Orphanet Journal of Rare Diseases, 2017, 12, 71.	2.7	53
9	Psychosocial Profiles of Parents of Children with Undiagnosed Diseases: Managing Well or Just Managing?. Journal of Genetic Counseling, 2018, 27, 935-946.	1.6	49
10	Epigenetics and autism spectrum disorder: A report of an autism case with mutation in H1 linker histone HIST1H1E and literature review. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 426-433.	1.7	48
11	VATER and hydrocephalus: Distinct syndrome?. American Journal of Medical Genetics Part A, 1991, 38, 46-51.	2.4	45
12	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
13	Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. Genetics in Medicine, 2018, 20, 464-469.	2.4	42
14	Parental attitudes regarding carrier testing in children at risk for fragile X syndrome. American Journal of Medical Genetics Part A, 1999, 82, 206-211.	2.4	41
15	Threat to Parental Role: A Possible Mechanism of Altered Self-Concept Related to Carrier Knowledge. Journal of Genetic Counseling, 2000, 9, 285-302.	1.6	39
16	Longitudinal study of the carrier testing process for fragile X syndrome: Perceptions and coping. American Journal of Medical Genetics Part A, 2001, 98, 37-45.	2.4	38
17	Fryns syndrome survivors and neurologic outcome. American Journal of Medical Genetics Part A, 1995, 59, 334-340.	2.4	37
18	Genetic Risk Communication: Experiences of Adolescent Girls and Young Women from Families with Fragile X Syndrome. Journal of Genetic Counseling, 2009, 18, 313-325.	1.6	36

#	Article	IF	Citations
19	Carrier testing in fragile X syndrome: Effect on self-concept. American Journal of Medical Genetics Part A, 2000, 92, 336-342.	2.4	35
20	Living with genetic risk: Effect on adolescent selfâ€concept. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2008, 148C, 56-69.	1.6	35
21	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
22	Genetic Counseling-Stress, Coping, and the Empowerment Perspective. Journal of Genetic Counseling, 1999, 8, 345-357.	1.6	32
23	Carrier testing in fragile X syndrome: When to tell and test. American Journal of Medical Genetics Part A, 2002, 110, 36-44.	2.4	31
24	De novo pathogenic variants in CHAMP1 are associated with global developmental delay, intellectual disability, and dysmorphic facial features. Journal of Physical Education and Sports Management, 2016, 2, a000661.	1.2	31
25	Hemangioma, supraumbilical midline raphÃ©, and coarctation of the aorta with a right aortic arch: Single causal entity?. American Journal of Medical Genetics Part A, 1995, 59, 44-48.	2.4	30
26	Alternative transcripts in variant interpretation: the potential for missed diagnoses and misdiagnoses. Genetics in Medicine, 2020, 22, 1269-1275.	2.4	30
27	Cases from the Undiagnosed Diseases Network: The continued value of counseling skills in a new genomic era. Journal of Genetic Counseling, 2019, 28, 194-201.	1.6	25
28	When to tell and test for genetic carrier status: Perspectives of adolescents and young adults from fragile X families. American Journal of Medical Genetics, Part A, 2009, 149A, 1190-1199.	1.2	23
29	Biallelic mutations in FDXR cause neurodegeneration associated with inflammation. Journal of Human Genetics, 2018, 63, 1211-1222.	2.3	23
30	Characteristics of undiagnosed diseases network applicants: implications for referring providers. BMC Health Services Research, 2018, 18, 652.	2.2	23
31	Interference of nuclear mitochondrial DNA segments in mitochondrial DNA testing resembles biparental transmission of mitochondrial DNA in humans. Genetics in Medicine, 2021, 23, 1514-1521.	2.4	21
32	Prenatal diagnosis of Pompe's disease (type ii glycogenosis) in chorionic villus biopsy using maltose as a substrate. Prenatal Diagnosis, 1992, 12, 169-173.	2.3	19
33	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	2.4	18
34	Prenatal diagnosis of glycogen storage disease type IV using PCR-based DNA mutation analysis. , 1999, 19, 837-839.		15
35	Communication of Genetic Risk Information to Daughters in Families with Fragile X Syndrome: The Parent's Perspective. Journal of Genetic Counseling, 2011, 20, 58-69.	1.6	15
36	Prenatal diagnosis and carrier detection for glycogen storage disease type III using polymorphic DNA markers. , 1998, 18, 61-64.		14

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37	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074.	1.7	14
38	Understanding Adult Participant and Parent Empowerment Prior to Evaluation in the Undiagnosed Diseases Network. Journal of Genetic Counseling, 2018, 27, 1087-1101.	1.6	12
39	Unbalanced translocation 46,xy,â˜15,+der(22)t(15;22)(q13;q11)pat: Case report and review of the literature. American Journal of Medical Genetics Part A, 1992, 44, 24-30.	2.4	11
40	Influence of Genetic Risk Information on Parental Role Identity in Adolescent Girls and Young Women from Families with Fragile X Syndrome. Journal of Genetic Counseling, 2012, 21, 59-71.	1.6	10
41	Genetic Counseling for Fragile X Syndrome: Recommendations of the National Society of Genetic Counselors. Journal of Genetic Counseling, 2000, 9, 303-325.	1.6	7
42	The genome empowerment scale: An assessment of parental empowerment in families with undiagnosed disease. Clinical Genetics, 2019, 96, 521-531.	2.0	7
43	Severe multisystem pathology, metabolic acidosis, mitochondrial dysfunction, and early death associated with an X-linked AIFM1 variant. Journal of Physical Education and Sports Management, 2021, 7, a006081.	1.2	6
44	Medium-chain acyl CoA dehydrogenase deficiency: Its relationship to SIDS and the impact on genetic counseling. Journal of Genetic Counseling, 1993, 2, 17-27.	1.6	4
45	The importance of managing the patient and not the gene: expanded phenotype of GLE1-associated arthrogryposis. Journal of Physical Education and Sports Management, 2017, 3, a002063.	1.2	4
46	Epileptic encephalopathy with features of rapidâ€onset dystonia Parkinsonism and alternating hemiplegia of childhood: a novel combination phenotype associated with ATP1A3 mutation. Epileptic Disorders, 2020, 22, 103-109.	1.3	4
47	Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples. Journal of Genetic Counseling, 2022, 31, 59-70.	1.6	3
48	Editorial Notes. Journal of Genetic Counseling, 2003, 12, 1-3.	1.6	2
49	Parental attitudes regarding carrier testing in children at risk for fragile X syndrome. American Journal of Medical Genetics Part A, 1999, 82, 206-211.	2.4	2
50	Editorial. Journal of Genetic Counseling, 2002, 11, 237-239.	1.6	1
51	Longitudinal study of the carrier testing process for fragile X syndrome: Perceptions and coping. American Journal of Medical Genetics Part A, 2001, 98, 37-45.	2.4	1
52	Genetic Counseling for Fragile X Syndrome: Updated Recommendations of the National Society of Genetic Counselors. Journal of Genetic Counseling, 2005, 14, 249-270.	1.6	1
53	Process in Genetic Counseling: Considerations for Children and Their Families. Issues in Clinical Child Psychology, 2010, , 87-107.	0.2	1
54	Editorial Introduction. Journal of Genetic Counseling, 2002, 11, 1-3.	1.6	0

#	Article	IF	Citations
55	Instructions to Contributors. Journal of Genetic Counseling, 2002, 11, 149-153.	1.6	0
56	Editorial: The JOGC Journey Continues. Journal of Genetic Counseling, 2008, 17, 1-1.	1.6	0
57	INTRODUCTION TO EXPLORING THE EXOME SPECIAL ISSUE. Journal of Genetic Counseling, 2019, 28, 181-181.	1.6	0
58	Men with an FMR1 premutation and their health education needs. Journal of Genetic Counseling, 2021, 30, 1156-1167.	1.6	0