

Beverly S Emanuel

List of Publications by Year in descending order

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105
papers

6,360
citations

87888

38
h-index

71685

76
g-index

112
all docs

112
docs citations

112
times ranked

5766
citing authors

#	ARTICLE	IF	CITATIONS
1	Fusion of a fork head domain gene to PAX3 in the solid tumour alveolar rhabdomyosarcoma. <i>Nature Genetics</i> , 1993, 5, 230-235.	21.4	869
2	Rearrangement of the PAX3 paired box gene in the paediatric solid tumour alveolar rhabdomyosarcoma. <i>Nature Genetics</i> , 1993, 3, 113-117.	21.4	540
3	Chromosome 22-specific low copy repeats and the 22q11.2 deletion syndrome: genomic organization and deletion endpoint analysis. <i>Human Molecular Genetics</i> , 2000, 9, 489-501.	2.9	460
4	Deletions and microdeletions of 22q11.2 in velo-cardio-facial syndrome. <i>American Journal of Medical Genetics Part A</i> , 1992, 44, 261-268.	2.4	387
5	Cognitive and behavior profile of preschool children with chromosome 22q11.2 deletion. <i>American Journal of Medical Genetics Part A</i> , 1999, 85, 127-133.	2.4	263
6	Segmental duplications: an 'expanding' role in genomic instability and disease. <i>Nature Reviews Genetics</i> , 2001, 2, 791-800.	16.3	263
7	Site-specific reciprocal translocation, t(11;22) (q23;q11), in several unrelated families with 3:1 meiotic disjunction. <i>American Journal of Medical Genetics Part A</i> , 1980, 7, 507-521.	2.4	165
8	Cloning a balanced translocation associated with DiGeorge syndrome and identification of a disrupted candidate gene. <i>Nature Genetics</i> , 1995, 10, 269-278.	21.4	152
9	Evolutionarily conserved low copy repeats (LCRs) in 22q11 mediate deletions, duplications, translocations, and genomic instability: An update and literature review. <i>Genetics in Medicine</i> , 2001, 3, 6-13.	2.4	126
10	From microscopes to microarrays: dissecting recurrent chromosomal rearrangements. <i>Nature Reviews Genetics</i> , 2007, 8, 869-883.	16.3	119
11	Juvenile rheumatoid arthritis-like polyarthritis in chromosome 22q11.2 deletion syndrome (digeorge) <i>Tj ETQq1 1 0.784314 rgBT /Over</i> <i>Rheumatism</i> , 1997, 40, 430-436.	6.7	115
12	Clinical, cytogenetic, and pedigree findings in 18 cases of Aicardi syndrome. <i>American Journal of Medical Genetics Part A</i> , 1989, 32, 461-467.	2.4	113
13	What is new with 22q? An update from the 22q and You Center at the Children's Hospital of Philadelphia. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2058-2069.	1.2	106
14	Molecular mechanisms and diagnosis of chromosome 22q11.2 rearrangements. <i>Developmental Disabilities Research Reviews</i> , 2008, 14, 11-18.	2.9	97
15	Molecular genetics of 22q11.2 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2070-2081.	1.2	96
16	Deletions of different segments of the long arm of chromosome 4. <i>American Journal of Medical Genetics Part A</i> , 1981, 8, 73-89.	2.4	93
17	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. <i>Nature Medicine</i> , 2020, 26, 1912-1918.	30.7	90
18	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. <i>Molecular Psychiatry</i> , 2021, 26, 4496-4510.	7.9	87

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19	Chromosomal Translocation t(1;13)(p36;q14) in a Case of Rhabdomyosarcoma. <i>Genes Chromosomes and Cancer</i> , 1991, 3, 483-484.	2.8	85
20	Skeletal anomalies and deformities in patients with deletions of 22q11. , 1997, 72, 210-215.		75
21	Unexpectedly high rate of de novo constitutional t(11;22) translocations in sperm from normal males. <i>Nature Genetics</i> , 2001, 29, 139-140.	21.4	71
22	22q11.2 duplication syndrome: elevated rate of autism spectrum disorder and need for medical screening. <i>Molecular Autism</i> , 2016, 7, 27.	4.9	67
23	Copy-Number Variation of the Glucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. <i>American Journal of Human Genetics</i> , 2015, 96, 753-764.	6.2	62
24	Aberrant Cortical Morphometry in the 22q11.2 Deletion Syndrome. <i>Biological Psychiatry</i> , 2015, 78, 135-143.	1.3	61
25	Patient with a 22q11.2 deletion with no overlap of the minimal DiGeorge syndrome critical region (MDGCR). , 1999, 86, 27-33.		59
26	Mouse and Human CRKL Is Dosage Sensitive for Cardiac Outflow Tract Formation. <i>American Journal of Human Genetics</i> , 2015, 96, 235-244.	6.2	58
27	Velo-cardio-facial syndrome and DiGeorge sequence with meningocele and deletions of the 22q11 region. <i>American Journal of Medical Genetics Part A</i> , 1994, 52, 445-449.	2.4	57
28	DiGeorge anomaly with renal agenesis in infants of mothers with diabetes. <i>American Journal of Medical Genetics Part A</i> , 1993, 47, 1078-1082.	2.4	56
29	Enlarged sylvian fissures in infants with interstitial deletion of chromosome 22q11. <i>American Journal of Medical Genetics Part A</i> , 1997, 74, 538-543.	2.4	56
30	Subthreshold Psychotic Symptoms in 22q11.2 Deletion Syndrome. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2014, 53, 991-1000.e2.	0.5	51
31	Altered white matter microstructure in 22q11.2 deletion syndrome: a multisite diffusion tensor imaging study. <i>Molecular Psychiatry</i> , 2020, 25, 2818-2831.	7.9	50
32	Histone Modifier Genes Alter Conotruncal Heart Phenotypes in 22q11.2 Deletion Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 869-877.	6.2	49
33	Subthreshold Psychosis in 22q11.2 Deletion Syndrome: Multisite Naturalistic Study. <i>Schizophrenia Bulletin</i> , 2017, 43, 1079-1089.	4.3	47
34	Neurocognitive profile in psychotic versus nonpsychotic individuals with 22q11.2 deletion syndrome. <i>European Neuropsychopharmacology</i> , 2016, 26, 1610-1618.	0.7	45
35	The Psychosis Spectrum in 22q11.2 Deletion Syndrome Is Comparable to That of Nondeleted Youths. <i>Biological Psychiatry</i> , 2017, 82, 17-25.	1.3	45
36	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. <i>Human Genetics</i> , 2016, 135, 273-285.	3.8	43

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37	Microphthalmia and chorioretinal lesions in a girl with an Xp22.2-pter deletion and partial 3p trisomy: Clinical observations relevant to aicardi syndrome gene localization. American Journal of Medical Genetics Part A, 1990, 37, 182-186.	2.4	42
38	Structural and Mutational Analysis of a Conserved Gene (DGS1) from the Minimal DiGeorge Syndrome Critical Region. Human Molecular Genetics, 1997, 6, 267-276.	2.9	42
39	Complete Sequence of the 22q11.2 Allele in 1,053 Subjects with 22q11.2 Deletion Syndrome Reveals Modifiers of Conotruncal Heart Defects. American Journal of Human Genetics, 2020, 106, 26-40.	6.2	42
40	Holoprosencephaly: Association with interstitial deletion of 2p and review of the cytogenetic literature. American Journal of Medical Genetics Part A, 1988, 30, 929-938.	2.4	41
41	The 22q11 low copy repeats are characterized by unprecedented size and structural variability. Genome Research, 2019, 29, 1389-1401.	5.5	39
42	Sequence-ready physical map of the mouse Chromosome 16 region with conserved synteny to the human Velocardiofacial syndrome region on 22q11.2. Mammalian Genome, 1999, 10, 438-443.	2.2	37
43	Critical region within 22q11.2 linked to higher rate of autism spectrum disorder. Molecular Autism, 2017, 8, 58.	4.9	37
44	Molecular detection of a Yp/18 translocation in a 45,X holoprosencephalic male. Human Genetics, 1988, 80, 219-223.	3.8	35
45	Congenital heart disease in supernumerary der(22), t(11;22) syndrome. Clinical Genetics, 1986, 29, 269-275.	2.0	35
46	Prenatal detection of Roberts-SC phocomelia syndrome: Report of 2 sibs with characteristic manifestations. American Journal of Medical Genetics Part A, 1989, 32, 390-394.	2.4	34
47	Variance of IQ is partially dependent on deletion type among 1,427 22q11.2 deletion syndrome subjects. American Journal of Medical Genetics, Part A, 2018, 176, 2172-2181.	1.2	33
48	Molecular and cytogenetic analysis of chromosomal arms 2q and 13q in alveolar rhabdomyosarcoma. Genes Chromosomes and Cancer, 1991, 3, 153-161.	2.8	32
49	22q and two: 22q11.2 deletion syndrome and coexisting conditions. American Journal of Medical Genetics, Part A, 2018, 176, 2203-2214.	1.2	30
50	Effects of copy number variations on brain structure and risk for psychiatric illness: Large-scale studies from the ENIGMA working groups on CNVs. Human Brain Mapping, 2022, 43, 300-328.	3.6	30
51	Abnormalities of chromosome 22 in pediatric meningiomas. Genes Chromosomes and Cancer, 1994, 9, 81-87.	2.8	28
52	The Role of mGluR Copy Number Variation in Genetic and Environmental Forms of Syndromic Autism Spectrum Disorder. Scientific Reports, 2016, 6, 19372.	3.3	28
53	Association of Mitochondrial Biogenesis With Variable Penetrance of Schizophrenia. JAMA Psychiatry, 2021, 78, 911.	11.0	25
54	Unbalanced 15;22 translocation in a patient with manifestations of DiGeorge and velocardiofacial syndrome. , 1997, 70, 6-10.		24

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55	Ocular albinism in a male with del (6)(q13-q15): Candidate region for autosomal recessive ocular albinism?. American Journal of Medical Genetics Part A, 1992, 42, 700-705.	2.4	23
56	Tricho-Rhino-Phalangeal syndrome type II (Langer-Giedion) with persistent cloaca and prune belly sequence in a girl with 8q interstitial deletion. American Journal of Medical Genetics Part A, 1992, 44, 790-794.	2.4	23
57	A diagnostic approach to identifying submicroscopic 7p21 deletions in Saethre-Chotzen syndrome: Fluorescence in situ hybridization and dosage-sensitive Southern blot analysis. Genetics in Medicine, 2001, 3, 102-108.	2.4	22
58	Analysis of the t(3;8) of hereditary renal cell carcinoma: a palindrome-mediated translocation. Cancer Genetics, 2014, 207, 133-140.	0.4	22
59	Performance on a computerized neurocognitive battery in 22q11.2 deletion syndrome: A comparison between US and Israeli cohorts. Brain and Cognition, 2016, 106, 33-41.	1.8	22
60	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the <i>GPR98</i> Locus on 5q14.3. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	22
61	Deletion size analysis of 1680 22q11.2DS subjects identifies a new recombination hotspot on chromosome 22q11.2. Human Molecular Genetics, 2018, 27, 1150-1163.	2.9	22
62	Optical mapping of the 22q11.2DS region reveals complex repeat structures and preferred locations for non-allelic homologous recombination (NAHR). Scientific Reports, 2020, 10, 12235.	3.3	20
63	Recurrence rate for de novo 21q21q translocation Down syndrome: A study of 112 families. American Journal of Medical Genetics Part A, 1984, 17, 523-530.	2.4	19
64	Nasal dimple as part of the 22q11.2 deletion syndrome. , 1997, 69, 290-292.		19
65	Attention deficit hyperactivity disorder symptoms as antecedents of later psychotic outcomes in 22q11.2 deletion syndrome. Schizophrenia Research, 2019, 204, 320-325.	2.0	19
66	White matter microstructural deficits in 22q11.2 deletion syndrome. Psychiatry Research - Neuroimaging, 2017, 268, 35-44.	1.8	17
67	Attention Deficit Hyperactivity Disorder Symptoms and Psychosis in 22q11.2 Deletion Syndrome. Schizophrenia Bulletin, 2018, 44, 824-833.	4.3	17
68	Pathogenic variants in CDC45 on the remaining allele in patients with a chromosome 22q11.2 deletion result in a novel autosomal recessive condition. Genetics in Medicine, 2020, 22, 326-335.	2.4	17
69	Negative subthreshold psychotic symptoms distinguish 22q11.2 deletion syndrome from other neurodevelopmental disorders: A two-site study. Schizophrenia Research, 2017, 188, 42-49.	2.0	16
70	Dysregulation of TBX1 dosage in the anterior heart field results in congenital heart disease resembling the 22q11.2 duplication syndrome. Human Molecular Genetics, 2018, 27, 1847-1857.	2.9	16
71	Interstitial deletion of 4(q21q25) in a liveborn male. American Journal of Medical Genetics Part A, 1991, 40, 77-79.	2.4	15
72	Malignant fibrous histiocytoma of the brain in a six-year-old girl. Genes Chromosomes and Cancer, 1992, 4, 309-313.	2.8	15

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73	Classical Noonan syndrome is not associated with deletions of 22q11. <i>American Journal of Medical Genetics Part A</i> , 1995, 56, 94-96.	2.4	15
74	Characterization of CDC45L: a gene in the 22q11.2 deletion region expressed during murine and human development. <i>Mammalian Genome</i> , 1999, 10, 322-326.	2.2	15
75	The dimensional structure of psychopathology in 22q11.2 Deletion Syndrome. <i>Journal of Psychiatric Research</i> , 2017, 92, 124-131.	3.1	13
76	A normative chart for cognitive development in a genetically selected population. <i>Neuropsychopharmacology</i> , 2022, 47, 1379-1386.	5.4	12
77	Breakpoint analysis of the recurrent constitutional t(8;22)(q24.13;q11.21) translocation. <i>Molecular Cytogenetics</i> , 2014, 7, 55.	0.9	11
78	MitoScape: A big-data, machine-learning platform for obtaining mitochondrial DNA from next-generation sequencing data. <i>PLoS Computational Biology</i> , 2021, 17, e1009594.	3.2	11
79	Molecular Cloning and Characterization of the Bovine and Human Tuftelin Genes. <i>Connective Tissue Research</i> , 1998, 39, 13-24.	2.3	10
80	Early language measures associated with later psychosis features in 22q11.2 deletion syndrome. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2020, 183, 392-400.	1.7	10
81	Monoclonal Antibody-Dependent, Cell-Mediated Cytotoxicity against Human Malignant Gliomas. <i>Neurosurgery</i> , 1990, 27, 97-102.	1.1	9
82	Congenital nystagmus in a (46, XX/45,X) mosaic woman from a family with X-linked congenital nystagmus. <i>American Journal of Medical Genetics Part A</i> , 1991, 39, 167-169.	2.4	9
83	Prenatal diagnosis of the derivative chromosome 22 associated with cat eye syndrome by fluorescence in situ hybridization. <i>Prenatal Diagnosis</i> , 1994, 14, 1029-1034.	2.3	8
84	A catalog of hemizygous variation in 127 22q11 deletion patients. <i>Human Genome Variation</i> , 2016, 3, 15065.	0.7	8
85	Olfactory deficits and psychosis-spectrum symptoms in 22q11.2 deletion syndrome. <i>Schizophrenia Research</i> , 2018, 202, 113-119.	2.0	8
86	Relationship between intelligence quotient measures and computerized neurocognitive performance in 22q11.2 deletion syndrome. <i>Brain and Behavior</i> , 2021, 11, e2221.	2.2	8
87	The impact of hypocalcemia on full scale IQ in patients with 22q11.2 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2167-2171.	1.2	7
88	Double strand breaks (DSBs) as indicators of genomic instability in PATRR-mediated translocations. <i>Human Molecular Genetics</i> , 2021, 29, 3872-3881.	2.9	7
89	Cytogenetic and molecular investigation of a balanced Xq13q translocation in a patient with retinoblastoma. <i>American Journal of Medical Genetics Part A</i> , 1992, 42, 771-776.	2.4	6
90	IQ and hemizygosity for the Val ¹⁵⁸ Met functional polymorphism of <i>COMT</i> in 22q11DS. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2016, 171, 1112-1115.	1.7	6

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91	Prenatal diagnosis of mosaicism 46, XX/46, XX, â~21, +(21q21q). Prenatal Diagnosis, 1984, 4, 73-77.	2.3	5
92	Musical auditory processing, cognition, and psychopathology in 22q11.2 deletion syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 765-773.	1.7	5
93	A binational study assessing risk and resilience factors in 22q11.2 deletion syndrome. Journal of Psychiatric Research, 2021, 138, 319-325.	3.1	5
94	A Comprehensive Analysis of Cerebellar Volumes in the 22q11.2 Deletion Syndrome. Biological Psychiatry: Cognitive Neuroscience and Neuroimaging, 2023, 8, 79-90.	1.5	5
95	Frequency of the common fragile site at Xq27.2 under conditions of thymidylate stress: Implications for cytogenetic diagnosis of the fragile-X syndrome. American Journal of Medical Genetics Part A, 1992, 42, 835-838.	2.4	4
96	Disrupted anatomic networks in the 22q11.2 deletion syndrome. NeuroImage: Clinical, 2016, 12, 420-428.	2.7	4
97	Molecular and cytogenetic studies of a patient with philadelphia-negative,BCR-positive chronic myeloid leukemia and t(12;12)(q13;p12). Genes Chromosomes and Cancer, 1990, 1, 284-288.	2.8	3
98	Copy number variations in individuals with conotruncal heart defects reveal some shared developmental pathways irrespective of 22q11.2 deletion status. Birth Defects Research, 2019, 111, 888-905.	1.5	3
99	Commentary on the decision of the American Board of Medical Genetics and Genomics to create a 24-month specialty of Laboratory Genetics and Genomics. Genetics in Medicine, 2017, 19, 294-296.	2.4	2
100	Genome-Wide Association Studies of Conotruncal Heart Defects with Normally Related Great Vessels in the United States. Genes, 2021, 12, 1030.	2.4	1
101	Enlarged sylvian fissures in infants with interstitial deletion of chromosome 22q11. American Journal of Medical Genetics Part A, 1997, 74, 538-543.	2.4	1
102	Altered functional brain dynamics in chromosome 22q11.2 deletion syndrome during facial affect processing. Molecular Psychiatry, 2022, 27, 1158-1166.	7.9	1
103	Expression of two G-6-PD genes in an XX phenotypic male. British Journal of Haematology, 1986, 64, 107-110.	2.5	0
104	Congenital nystagmus in a [46,XX/45,X] Mosaic woman from a family with X-linked congenital nystagmus. American Journal of Medical Genetics Part A, 1992, 43, 897-897.	2.4	0
105	Cover Image, Volume 176A, Number 10, October 2018. , 2018, 176, i-i.		0