Beverly S Emanuel

List of Publications by Year
in descending order

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105 papers	6,360 citations	100601 38 h-index	81351 76 g-index
112 all docs	112 docs citations	112 times ranked	6311 citing authors

#	Article	IF	Citations
1	A Comprehensive Analysis of Cerebellar Volumes in the 22q11.2 Deletion Syndrome. Biological Psychiatry: Cognitive Neuroscience and Neuroimaging, 2023, 8, 79-90.	1.1	5
2	Effects of copy number variations on brain structure and risk for psychiatric illness: Largeâ€scale studies from the<scp>ENIGMA</scp>working groups on<scp>CNVs</scp>. Human Brain Mapping, 2022, 43, 300-328.	1.9	30
3	A normative chart for cognitive development in a genetically selected population. Neuropsychopharmacology, 2022, 47, 1379-1386.	2.8	12
4	Altered functional brain dynamics in chromosome 22q11.2 deletion syndrome during facial affect processing. Molecular Psychiatry, 2022, 27, 1158-1166.	4.1	1
5	Double strand breaks (DSBs) as indicators of genomic instability in PATRR-mediated translocations. Human Molecular Genetics, 2021, 29, 3872-3881.	1.4	7
6	A binational study assessing risk and resilience factors in 22q11.2 deletion syndrome. Journal of Psychiatric Research, 2021, 138, 319-325.	1.5	5
7	Genome-Wide Association Studies of Conotruncal Heart Defects with Normally Related Great Vessels in the United States. Genes, 2021, 12, 1030.	1.0	1
8	Relationship between intelligence quotient measures and computerized neurocognitive performance in 22q11.2 deletion syndrome. Brain and Behavior, 2021, 11, e2221.	1.0	8
9	Association of Mitochondrial Biogenesis With Variable Penetrance of Schizophrenia. JAMA Psychiatry, 2021, 78, 911.	6.0	25
10	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	4.1	87
11	MitoScape: A big-data, machine-learning platform for obtaining mitochondrial DNA from next-generation sequencing data. PLoS Computational Biology, 2021, 17, e1009594.	1.5	11
12	Altered white matter microstructure in 22q11.2 deletion syndrome: a multisite diffusion tensor imaging study. Molecular Psychiatry, 2020, 25, 2818-2831.	4.1	50
13	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.	1.1	17
14	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.	2.6	42
15	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.	1.6	20
16	Early language measures associated with later psychosis features in 22q11.2 deletion syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 392-400.	1.1	10
17	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. Nature Medicine, 2020, 26, 1912-1918.	15.2	90
18	Attention deficit hyperactivity disorder symptoms as antecedents of later psychotic outcomes in 22q11.2 deletion syndrome. Schizophrenia Research, 2019, 204, 320-325.	1.1	19

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19	The 22q11 low copy repeats are characterized by unprecedented size and structural variability. Genome Research, 2019, 29, 1389-1401.	2.4	39
20	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.	0.8	3
21	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.	1.4	16
22	Attention Deficit Hyperactivity Disorder Symptoms and Psychosis in 22q11.2 Deletion Syndrome. Schizophrenia Bulletin, 2018, 44, 824-833.	2.3	17
23	Deletion size analysis of 1680 22q11.2DS subjects identifies a new recombination hotspot on chromosome 22q11.2. Human Molecular Genetics, 2018, 27, 1150-1163.	1.4	22
24	Cover Image, Volume 176A, Number 10, October 2018. , 2018, 176, i-i.		0
25	Musical auditory processing, cognition, and psychopathology in 22q11.2 deletion syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 765-773.	1.1	5
26	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.	0.7	33
27	22q and two: 22q11.2 deletion syndrome and coexisting conditions. American Journal of Medical Genetics, Part A, 2018, 176, 2203-2214.	0.7	30
28	The impact of hypocalcemia on full scale IQ in patients with 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2167-2171.	0.7	7
29	Molecular genetics of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2070-2081.	0.7	96
30	What is new with 22q? An update from the 22q and You Center at the Children's Hospital of Philadelphia. American Journal of Medical Genetics, Part A, 2018, 176, 2058-2069.	0.7	106
31	Olfactory deficits and psychosis-spectrum symptoms in 22q11.2 deletion syndrome. Schizophrenia Research, 2018, 202, 113-119.	1.1	8
32	Negative subthreshold psychotic symptoms distinguish 22q11.2 deletion syndrome from other neurodevelopmental disorders: A two-site study. Schizophrenia Research, 2017, 188, 42-49.	1.1	16
33	The dimensional structure of psychopathology in 22q11.2 Deletion Syndrome. Journal of Psychiatric Research, 2017, 92, 124-131.	1.5	13
34	Subthreshold Psychosis in 22q11.2 Deletion Syndrome: Multisite Naturalistic Study. Schizophrenia Bulletin, 2017, 43, 1079-1089.	2.3	47
35	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the GPR98 Locus on 5q14.3. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	22
36	White matter microstructural deficits in 22q11.2 deletion syndrome. Psychiatry Research - Neuroimaging, 2017, 268, 35-44.	0.9	17

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37	The Psychosis Spectrum in 22q11.2 Deletion Syndrome Is Comparable to That of Nondeleted Youths. Biological Psychiatry, 2017, 82, 17-25.	0.7	45
38	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.	1.1	2
39	Critical region within 22q11.2 linked to higher rate of autism spectrum disorder. Molecular Autism, 2017, 8, 58.	2.6	37
40	Neurocognitive profile in psychotic versus nonpsychotic individuals with 22q11.2 deletion syndrome. European Neuropsychopharmacology, 2016, 26, 1610-1618.	0.3	45
41	The Role of mGluR Copy Number Variation in Genetic and Environmental Forms of Syndromic Autism Spectrum Disorder. Scientific Reports, 2016, 6, 19372.	1.6	28
42	Disrupted anatomic networks in the 22q11.2 deletion syndrome. NeuroImage: Clinical, 2016, 12, 420-428.	1.4	4
43	A catalog of hemizygous variation in 127 22q11 deletion patients. Human Genome Variation, 2016, 3, 15065.	0.4	8
44	IQ and hemizygosity for the Val¹⁵⁸Met functional polymorphism of COMT in 22q11DS. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 1112-1115.	1.1	6
45	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.	0.8	22
46	22q11.2 duplication syndrome: elevated rate of autism spectrum disorder and need for medical screening. Molecular Autism, 2016, 7, 27.	2.6	67
47	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. Human Genetics, 2016, 135, 273-285.	1.8	43
48	Mouse and Human CRKL Is Dosage Sensitive for Cardiac Outflow Tract Formation. American Journal of Human Genetics, 2015, 96, 235-244.	2.6	58
49	Copy-Number Variation of the Glucose Transporter Gene SLC2A3 and Congenital Heart Defects in the 22q11.2 Deletion Syndrome. American Journal of Human Genetics, 2015, 96, 753-764.	2.6	62
50	Aberrant Cortical Morphometry in the 22q11.2 Deletion Syndrome. Biological Psychiatry, 2015, 78, 135-143.	0.7	61
51	Histone Modifier Genes Alter Conotruncal Heart Phenotypes in 22q11.2 Deletion Syndrome. American Journal of Human Genetics, 2015, 97, 869-877.	2.6	49
52	Subthreshold Psychotic Symptoms in 22q11.2 Deletion Syndrome. Journal of the American Academy of Child and Adolescent Psychiatry, 2014, 53, 991-1000.e2.	0.3	51
53	Analysis of the t(3;8) of hereditary renal cell carcinoma: a palindrome-mediated translocation. Cancer Genetics, 2014, 207, 133-140.	0.2	22
54	Breakpoint analysis of the recurrent constitutional t(8;22)(q24.13;q11.21) translocation. Molecular Cytogenetics, 2014, 7, 55.	0.4	11

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55	Molecular mechanisms and diagnosis of chromosome 22q11.2 rearrangements. Developmental Disabilities Research Reviews, 2008, 14, 11-18.	2.9	97
56	From microscopes to microarrays: dissecting recurrent chromosomal rearrangements. Nature Reviews Genetics, 2007, 8, 869-883.	7.7	119
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.	1.1	22
58	Unexpectedly high rate of de novo constitutional t(11;22) translocations in sperm from normal males. Nature Genetics, 2001, 29, 139-140.	9.4	71
59	Segmental duplications: an 'expanding' role in genomic instability and disease. Nature Reviews Genetics, 2001, 2, 791-800.	7.7	263
60	Evolutionarily conserved low copy repeats (LCRs) in 22q11 mediate deletions, duplications, translocations, and genomic instability: An update and literature review. Genetics in Medicine, 2001, 3, 6-13.	1.1	126
61	Chromosome 22-specific low copy repeats and the 22q11.2 deletion syndrome: genomic organization and deletion endpoint analysis. Human Molecular Genetics, 2000, 9, 489-501.	1.4	460
62	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.	1.0	37
63	Characterization of CDC45L: a gene in the 22q11.2 deletion region expressed during murine and human development. Mammalian Genome, 1999, 10, 322-326.	1.0	15
64	Cognitive and behavior profile of preschool children with chromosome 22q11.2 deletion. American Journal of Medical Genetics Part A, 1999, 85, 127-133.	2.4	263
65	Patient with a 22q11.2 deletion with no overlap of the minimal DiGeorge syndrome critical region (MDGCR). , 1999, 86, 27-33.		59
66	Molecular Cloning and Characterization of the Bovine and Human Tuftelin Genes. Connective Tissue Research, 1998, 39, 13-24.	1.1	10
67	Structural and Mutational Analysis of a Conserved Gene (DGSI) from the Minimal DiGeorge Syndrome Critical Region. Human Molecular Genetics, 1997, 6, 267-276.	1.4	42
68	Juvenile rheumatoid arthritis-like polyarthritis in chromosome 22q11.2 deletion syndrome (digeorge) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5 Rheumatism, 1997, 40, 430-436.	6.7	115
69	Nasal dimple as part of the 22q11.2 deletion syndrome. , 1997, 69, 290-292.		19
70	Unbalanced 15;22 translocation in a patient with manifestations of DiGeorge and velocardiofacial syndrome. , 1997, 70, 6-10.		24
71	Enlarged sylvian fissures in infants with interstitial deletion of chromosome 22q11. American Journal of Medical Genetics Part A, 1997, 74, 538-543.	2.4	56
72	Skeletal anomalies and deformities in patients with deletions of 22q11. , 1997, 72, 210-215.		75

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73	Enlarged sylvian fissures in infants with interstitial deletion of chromosome 22q11. , 1997, 74, 538.		1
74	Classical Noonan syndrome is not associated with deletions of 22q11. American Journal of Medical Genetics Part A, 1995, 56, 94-96.	2.4	15
75	Cloning a balanced translocation associated with DiGeorge syndrome and identification of a disrupted candidate gene. Nature Genetics, 1995, 10, 269-278.	9.4	152
76	Prenatal diagnosis of the derivative chromosome 22 associated with cat eye syndrome by fluorescencein situ hybridization. Prenatal Diagnosis, 1994, 14, 1029-1034.	1.1	8
77	Abnormalities of chromosome 22 in pediatric meningiomas. Genes Chromosomes and Cancer, 1994, 9, 81-87.	1.5	28
78	Velo-cardio-facial syndrome and DiGeorge sequence with meningomyelocele and deletions of the 22q11 region. American Journal of Medical Genetics Part A, 1994, 52, 445-449.	2.4	57
79	DiGeorge anomaly with renal agenesis in infants of mothers with diabetes. American Journal of Medical Genetics Part A, 1993, 47, 1078-1082.	2.4	56
80	Rearrangement of the PAX3 paired box gene in the paediatric solid tumour alveolar rhabdomyosarcoma. Nature Genetics, 1993, 3, 113-117.	9.4	540
81	Fusion of a fork head domain gene to PAX3 in the solid tumour alveolar rhabdomyosarcoma. Nature Genetics, 1993, 5, 230-235.	9.4	869
82	Malignant fibrous histiocytoma of the brain in a six-year-old girl. Genes Chromosomes and Cancer, 1992, 4, 309-313.	1.5	15
83	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
84	Cytogenetic and molecular investigation of a balanced Xq13q translocation in a patient with retinoblastoma. American Journal of Medical Genetics Part A, 1992, 42, 771-776.	2.4	6
85	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
86	Congenital nystagmus in a [46,XX/45,X] Mosaic woman from a damily with X-linked congenital nystagmus. American Journal of Medical Genetics Part A, 1992, 43, 897-897.	2.4	0
87	Deletions and microdeletions of 22q11.2 in velo-cardio-facial syndrome. American Journal of Medical Genetics Part A, 1992, 44, 261-268.	2.4	387
88	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
89	Molecular and cytogenetic analysis of chromosomal arms 2q and 13q in alveolar rhabdomyosarcoma. Genes Chromosomes and Cancer, 1991, 3, 153-161.	1.5	32
90	Chromosomal Translocation t(1;13)(p36;q14) in a Case of Rhabdomyosarcoma. Genes Chromosomes and Cancer, 1991, 3, 483-484.	1.5	85

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91	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, 1991, 39, 167-169.	2.4	9
92	Interstitial deletion of 4(q21q25) in a liveborn male. American Journal of Medical Genetics Part A, 1991, 40, 77-79.	2.4	15
93	Monoclonal Antibody-Dependent, Cell-Mediated Cytotoxicity against Human Malignant Gliomas. Neurosurgery, 1990, 27, 97-102.	0.6	9
94	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
95	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.	1.5	3
96	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
97	Clinical, cytogenetic, and pedigree findings in 18 cases of Aicardi syndrome. American Journal of Medical Genetics Part A, 1989, 32, 461-467.	2.4	113
98	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
99	Molecular detection of a Yp/18 translocation in a 45,X holoprosencephalic male. Human Genetics, 1988, 80, 219-223.	1.8	35
100	Expression of two G-6-PD genes in an XX phenotypic male. British Journal of Haematology, 1986, 64, 107-110.	1.2	0
101	Congenital heart disease in supernumerary der(22), t(11;22) syndrome. Clinical Genetics, 1986, 29, 269-275.	1.0	35
102	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
103	Prenatal diagnosis of mosaicism 46, XX/46, XX, â^'21, +t(21q21q). Prenatal Diagnosis, 1984, 4, 73-77.	1.1	5
104	Deletions of different segments of the long arm of chromosome 4. American Journal of Medical Genetics Part A, 1981, 8, 73-89.	2.4	93
105	Site-specific reciprocal translocation, t(11;22) (q23;q11), in several unrelated families with 3:1 meiotic disjunction. American Journal of Medical Genetics Part A, 1980, 7, 507-521.	2.4	165