Elaine H Zackai

List of Publications by Year
in descending order

263 papers	13,363 citations	34105 52 h-index	31849 101 g-index
275 all docs	275 docs citations	275 times ranked	14075 citing authors

#	Article	IF	Citations
1	22q11.2 deletion syndrome. Nature Reviews Disease Primers, 2015, 1, 15071.	30.5	954
2	Psychiatric Disorders From Childhood to Adulthood in 22q11.2 Deletion Syndrome: Results From the International Consortium on Brain and Behavior in 22q11.2 Deletion Syndrome. American Journal of Psychiatry, 2014, 171, 627-639.	7.2	645
3	Mutations in the homeodomain of the human SIX3 gene cause holoprosencephaly. Nature Genetics, 1999, 22, 196-198.	21.4	398
4	Deletions and microdeletions of 22q11.2 in veloâ€cardioâ€facial syndrome. American Journal of Medical Genetics Part A, 1992, 44, 261-268.	2.4	387
5	Mutations in TGIF cause holoprosencephaly and link NODAL signalling to human neural axis determination. Nature Genetics, 2000, 25, 205-208.	21.4	368
6	Identical mutations in three different fibroblast growth factor receptor genes in autosomal dominant craniosynostosis syndromes. Nature Genetics, 1996, 14, 174-176.	21.4	306
7	Growth Charts for Children With Down Syndrome: 1 Month to 18 Years of Age. Pediatrics, 1988, 81, 102-110.	2.1	298
8	Mutations ofARX are associated with striking pleiotropy and consistent genotype-phenotype correlation. Human Mutation, 2004, 23, 147-159.	2.5	293
9	Analysis of 589,306 genomes identifies individuals resilient to severe Mendelian childhood diseases. Nature Biotechnology, 2016, 34, 531-538.	17.5	273
10	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
11	Immunologic features of chromosome 22q11.2 deletion syndrome (DiGeorge) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf 50 342 Td (sy	1.8	240
12	Optic pathway and hypothalamic/chiasmatic gliomas in children younger than age 5 years with a 6-year follow-up. Cancer, 1995, 75, 1051-1059.	4.1	234
13	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
14	Neonatal adrenoleukodystrophy: New cases, biochemical studies, and differentiation from Zellweger and related peroxisomal polydystrophy syndromes. American Journal of Medical Genetics Part A, 1986, 23, 869-901.	2.4	216
15	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
16	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
17	Down syndrome congenital heart disease: A narrowed region and a candidate gene. Genetics in Medicine, 2001, 3, 91-101.	2.4	168
18	Siteâ€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

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19	The 22q11.2 Deletion: Screening, Diagnostic Workup, and Outcome of Results; Report on 181 Patients. Genetic Testing and Molecular Biomarkers, 1997, 1, 99-108.	1.7	150
20	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. American Journal of Human Genetics, 2018, 102, 69-87.	6.2	144
21	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€“Phenotype Correlation. Human Mutation, 2015, 36, 1052-1063.	2.5	143
22	Kabuki syndrome genesKMT2DandKDM6A: functional analyses demonstrate critical roles in craniofacial, heart and brain development. Human Molecular Genetics, 2015, 24, 4443-4453.	2.9	142
23	Aberrant interchromosomal exchanges are the predominant cause of the 22q11.2 deletion. Human Molecular Genetics, 2004, 13, 417-428.	2.9	133
24	T-cell homeostasis in humans with thymic hypoplasia due to chromosome 22q11.2 deletion syndrome. Blood, 2004, 103, 1020-1025.	1.4	119
25	Juvenile rheumatoid arthritisâ€like polyarthritis in chromosome 22q11.2 deletion syndrome (digeorge) Tj ETQq1 1 0.784314 rgBT /Over Rheumatism, 1997, 40, 430-436.	6.7	115
26	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
27	Germline gain-of-function mutations in AFF4 cause a developmental syndrome functionally linking the super elongation complex and cohesin. Nature Genetics, 2015, 47, 338-344.	21.4	109
28	Dominant Mutations in KAT6A Cause Intellectual Disability with Recognizable Syndromic Features. American Journal of Human Genetics, 2015, 96, 507-513.	6.2	107
29	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.	1.2	106
30	Hemizygous mutations in SNAP29 unmask autosomal recessive conditions and contribute to atypical findings in patients with 22q11.2DS. Journal of Medical Genetics, 2013, 50, 80-90.	3.2	104
31	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. European Journal of Human Genetics, 2015, 23, 1473-1481.	2.8	101
32	Oral-facial-digital syndrome type VI (VÃ¡radi syndrome): Further clinical delineation. American Journal of Medical Genetics Part A, 1990, 35, 360-369.	2.4	97
33	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
34	Mutations in CDC45 , Encoding an Essential Component of the Pre-initiation Complex, Cause Meier-Gorlin Syndrome and Craniosynostosis. American Journal of Human Genetics, 2016, 99, 125-138.	6.2	92
35	Mutations in the humanTWIST gene. Human Mutation, 2000, 15, 150-155.	2.5	86
36	Clinical spectrum of individuals with pathogenic N F1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€“phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	2.5	80

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37	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. American Journal of Psychiatry, 2017, 174, 1054-1063.	7.2	77
38	Skeletal anomalies and deformities in patients with deletions of 22q11. , 1997, 72, 210-215.		75
39	Phenotype of the fibroblast growth factor receptor 2 Ser351Cys mutation: Pfeiffer syndrome type III. American Journal of Medical Genetics Part A, 1998, 78, 356-360.	2.4	72
40	Neurodevelopmental outcomes in preschool survivors of the Fontan procedure. Journal of Thoracic and Cardiovascular Surgery, 2014, 147, 1276-1283.e5.	0.8	71
41	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. Clinical Epigenetics, 2019, 11, 64.	4.1	71
42	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
43	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315.	2.4	69
44	Longitudinal Analysis of Lymphocyte Function and Numbers in the First Year of Life in Chromosome 22q11.2 Deletion Syndrome (DiGeorge Syndrome/Velocardiofacial Syndrome). Vaccine Journal, 1999, 6, 906-911.	2.6	68
45	De Novo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap Those Associated with Proximal 1p36 Deletions. American Journal of Human Genetics, 2016, 98, 963-970.	6.2	67
46	22q11.2 duplication syndrome: elevated rate of autism spectrum disorder and need for medical screening. Molecular Autism, 2016, 7, 27.	4.9	67
47	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.	6.2	62
48	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
49	Polytopic anomalies with agenesis of the lower vertebral column. American Journal of Medical Genetics Part A, 1999, 87, 99-114.	2.4	61
50	Aberrant Cortical Morphometry in the 22q11.2 Deletion Syndrome. Biological Psychiatry, 2015, 78, 135-143.	1.3	61
51	22q11.2 Deletion syndrome and obstructive sleep apnea. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 1360-1364.	1.0	58
52	Mouse and Human CRKL Is Dosage Sensitive for Cardiac Outflow Tract Formation. American Journal of Human Genetics, 2015, 96, 235-244.	6.2	58
53	Disrupted auto-regulation of the spliceosomal gene SNRPB causes cerebroâ€“costoâ€“mandibular syndrome. Nature Communications, 2014, 5, 4483.	12.8	57
54	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. American Journal of Human Genetics, 2014, 94, 784-789.	6.2	57

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55	Congenital heart diseases and cardiovascular abnormalities in 22q11.2 deletion syndrome: From wellâ€established knowledge to new frontiers. American Journal of Medical Genetics, Part A, 2018, 176, 2087-2098.	1.2	57
56	Natural history and genotypeâ€phenotype correlations in 72 individuals with SATB2â€associated syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 925-935.	1.2	57
57	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
58	Oculodentodigital dysplasia syndrome associated with abnormal cerebral white matter. American Journal of Medical Genetics Part A, 1991, 41, 18-20.	2.4	55
59	Increasing cumulative exposure to volatile anesthetic agents is associated with poorer neurodevelopmental outcomes in children with hypoplastic left heart syndrome. Journal of Thoracic and Cardiovascular Surgery, 2016, 152, 482-489.	0.8	55
60	Burden of potentially pathologic copy number variants is higher in children with isolated congenital heart disease and significantly impairs covariate-adjusted transplant-free survival. Journal of Thoracic and Cardiovascular Surgery, 2016, 151, 1147-1151.e4.	0.8	55
61	Gainâ€ofâ€function mutations in SMAD4 cause a distinctive repertoire of cardiovascular phenotypes in patients with Myhre syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2617-2631.	1.2	53
62	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	2.4	53
63	Melnick-needles syndrome in males: A lethal multiple congenital anomalies syndrome. American Journal of Medical Genetics Part A, 1987, 27, 159-173.	2.4	52
64	Lateral meningocele syndrome: Three new patients and review of the literature. American Journal of Medical Genetics Part A, 1997, 70, 229-239.	2.4	52
65	Saethre-Chotzen syndrome with familial translocation at chromosome 7p22. American Journal of Medical Genetics Part A, 1993, 47, 637-639.	2.4	51
66	Histone Modifier Genes Alter Conotruncal Heart Phenotypes in 22q11.2 Deletion Syndrome. American Journal of Human Genetics, 2015, 97, 869-877.	6.2	49
67	Association of airway abnormalities with 22q11.2 deletion syndrome. International Journal of Pediatric Otorhinolaryngology, 2017, 96, 11-14.	1.0	49
68	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	6.2	48
69	On lumping and splitting: A fetus with clinical findings of the oral-facial-digital syndrome type VI, the hydrolethalus syndrome, and the Pallister-Hall syndrome. American Journal of Medical Genetics Part A, 1991, 41, 548-556.	2.4	47
70	IntragenicKANSL1mutations and chromosome 17q21.31 deletions: broadening the clinical spectrum and genotypeâ€“phenotype correlations in a large cohort of patients. Journal of Medical Genetics, 2015, 52, 804-814.	3.2	47
71	Subthreshold Psychosis in 22q11.2 Deletion Syndrome: Multisite Naturalistic Study. Schizophrenia Bulletin, 2017, 43, 1079-1089.	4.3	47
72	The 22q11.2 deletion syndrome: Cancer predisposition, platelet abnormalities and cytopenias. American Journal of Medical Genetics, Part A, 2018, 176, 2121-2127.	1.2	47

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73	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154.	2.5	45
74	Neurocognitive profile in psychotic versus nonpsychotic individuals with 22q11.2 deletion syndrome. European Neuropsychopharmacology, 2016, 26, 1610-1618.	0.7	45
75	The Psychosis Spectrum in 22q11.2 Deletion Syndrome Is Comparable to That of Nondeleted Youths. Biological Psychiatry, 2017, 82, 17-25.	1.3	45
76	Elucidating the diagnostic odyssey of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 936-944.	1.2	45
77	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. Human Genetics, 2016, 135, 273-285.	3.8	43
78	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
79	Rates of autism and potential risk factors in children with congenital heart defects. Congenital Heart Disease, 2017, 12, 421-429.	0.2	42
80	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
81	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
82	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
83	Expanding the fetal phenotype: Prenatal sonographic findings and perinatal outcomes in a cohort of patients with a confirmed 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1735-1741.	1.2	41
84	TWIST gene mutation in a patient with radial aplasia and craniosynostosis: Further evidence for heterogeneity of Baller-Gerold syndrome. , 1999, 82, 170-176.		40
85	Mutations inSPECC1L, encoding sperm antigen with calponin homology and coiled-coil domains 1-like, are found in some cases of autosomal dominant Opitz G/BBB syndrome. Journal of Medical Genetics, 2015, 52, 104-110.	3.2	40
86	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	6.2	40
87	De novo variants in Myelin regulatory factor (MYRF) as candidates of a new syndrome of cardiac and urogenital anomalies. American Journal of Medical Genetics, Part A, 2018, 176, 969-972.	1.2	39
88	Craniosynostosis: Another feature of the 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2005, 136A, 358-362.	1.2	38
89	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
90	Cystic kidney disease in Hajdu-Cheney syndrome. American Journal of Medical Genetics Part A, 1995, 56, 25-30.	2.4	37

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91	Critical region within 22q11.2 linked to higher rate of autism spectrum disorder. Molecular Autism, 2017, 8, 58.	4.9	37
92	Mutations in topoisomerase IIÎ² result in a B cell immunodeficiency. Nature Communications, 2019, 10, 3644.	12.8	37
93	Partial duplication 1q: Report of four patients and review of the literature. American Journal of Medical Genetics Part A, 1990, 36, 137-143.	2.4	36
94	Molecular detection of a Yp/18 translocation in a 45,X holoprosencephalic male. Human Genetics, 1988, 80, 219-223.	3.8	35
95	New finding of Schinzel-Giedion syndrome: A case with a malignant sacrococcygeal teratoma. American Journal of Medical Genetics Part A, 1993, 47, 852-856.	2.4	35
96	Congenital heart disease in supernumerary der(22), t(11;22) syndrome. Clinical Genetics, 1986, 29, 269-275.	2.0	35
97	A Prospective Study of Influenza Vaccination and a Comparison of Immunologic Parameters in Children and Adults with Chromosome 22q11.2 Deletion Syndrome (DiGeorge) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf 50 497 Td (S	3.4	34
98	Identification of 22q11.2 Deletion Syndrome via Newborn Screening for Severe Combined Immunodeficiency. Journal of Clinical Immunology, 2017, 37, 476-485.	3.8	35
99	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
100	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.	1.2	34
101	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
102	Ablepharon macrostomia syndrome with associated cutis laxa: Possible localization to 18q. Human Genetics, 1996, 97, 532-536.	3.8	32
103	Neuropathological Findings in Eight Children with Cerebro-oculo-facio-skeletal (COFS) Syndrome. Journal of Neuropathology and Experimental Neurology, 1997, 56, 1147-1157.	1.7	32
104	Further evidence that the Hajdu-Cheney syndrome and the ?serpentine fibula-polycystic kidney syndrome? are a single entity. , 1998, 78, 474-481.		32
105	Blepharo-cheilo-dontic (BCD) syndrome. , 1996, 65, 109-112.		31
106	Bilateral microtia and cleft palate in cousins with Diamond-Blackfan anemia. American Journal of Medical Genetics Part A, 2001, 101, 268-274.	2.4	31
107	Mosaic loss of 15q11q13 in a patient with hypomelanosis of Ito: is there a role for the P gene?. Human Genetics, 1995, 96, 485-9.	3.8	30
108	Further clinical delineation and increased morbidity in males with osteopathia striata with cranial sclerosis: An X-linked disorder?. American Journal of Medical Genetics Part A, 1997, 70, 159-165.	2.4	30

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109	Patient Genotypes Impact Survival After Surgery for Isolated Congenital Heart Disease. Annals of Thoracic Surgery, 2014, 98, 104-111.	1.3	30
110	22q and two: 22q11.2 deletion syndrome and coexisting conditions. American Journal of Medical Genetics, Part A, 2018, 176, 2203-2214.	1.2	30
111	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
112	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.	3.6	30
113	Early ultrasound diagnosis of Neuâ€“Laxova syndrome. Prenatal Diagnosis, 2001, 21, 575-580.	2.3	29
114	Supernumerary inv dup(15) in a patient with Angelman syndrome and a deletion of 15q11-q13. American Journal of Medical Genetics Part A, 1995, 57, 61-65.	2.4	28
115	Kabuki syndrome is not caused by a microdeletion in the DiGeorgeVelocardiofacial chromosomal region within 22q11.2. , 1996, 65, 101-103.		28
116	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
117	Novel findings with reassessment of exome data: implications for validation testing and interpretation of genomic data. Genetics in Medicine, 2018, 20, 329-336.	2.4	28
118	Tracheal cartilaginous sleeves in children with syndromic craniosynostosis. Genetics in Medicine, 2017, 19, 62-68.	2.4	27
119	Disruption of the bloodâ€“brain barrier in 22q11.2 deletion syndrome. Brain, 2021, 144, 1351-1360.	7.6	27
120	Expanding the SPECC1L mutation phenotypic spectrum to include Teebi hypertelorism syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2497-2502.	1.2	26
121	Exome sequencing expands the mechanism of SOX5â€associated intellectual disability: A case presentation with review of soxâ€related disorders. American Journal of Medical Genetics, Part A, 2015, 167, 2548-2554.	1.2	26
122	Utility of genetic evaluation in infants with congenital heart defects admitted to the cardiac intensive care unit. American Journal of Medical Genetics, Part A, 2016, 170, 3090-3097.	1.2	26
123	Thrombocytopenia with absent radius in a boy and his uncle. American Journal of Medical Genetics Part A, 1987, 28, 117-123.	2.4	25
124	Unusual craniofacial dysmorphia due to prenatal alcohol and cocaine exposure. Teratology, 1994, 50, 160-164.	1.6	25
125	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25
126	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. European Journal of Human Genetics, 2020, 28, 1422-1431.	2.8	25

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127	Association of Mitochondrial Biogenesis With Variable Penetrance of Schizophrenia. JAMA Psychiatry, 2021, 78, 911.	11.0	25
128	B cell development in chromosome 22q11.2 deletion syndrome. Clinical Immunology, 2016, 163, 1-9.	3.2	24
129	Phenotypic spectrum associated with SPECC1L pathogenic variants: new families and critical review of the nosology of Teebi, Opitz GBBB, and Baraitser-Winter syndromes. European Journal of Medical Genetics, 2019, 62, 103588.	1.3	24
130	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
131	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
132	Mosaicism for a chromosome 8â€derived minute marker chromosome in a patient with manifestations of trisomy 8 mosaicism. American Journal of Medical Genetics Part A, 1995, 56, 22-24.	2.4	23
133	Phenotype delineation of ZNF462 related syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2075-2082.	1.2	23
134	Diaphragmatic hernia-exomphalos-hypertelorism syndrome: A new case and further evidence of autosomal recessive inheritance. , 1997, 68, 441-444.		22
135	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
136	Boy with bilateral retinoblastoma due to an unusual ring chromosome 13 with activation of a latent centromere. American Journal of Medical Genetics Part A, 2001, 99, 21-28.	2.4	22
137	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
138	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
139	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
140	Anatomic Malformations of the Middle and Inner Ear in 22q11.2 Deletion Syndrome: Case Series and Literature Review. American Journal of Neuroradiology, 2018, 39, 928-934.	2.4	22
141	Cerebroâ€“costoâ€“mandibular syndrome: Clinical, radiological, and genetic findings. American Journal of Medical Genetics, Part A, 2016, 170, 1115-1126.	1.2	21
142	Congenital diaphragmatic hernia in 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 135-142.	1.2	21
143	De novo missense variants in MEIS2 recapitulate the microdeletion phenotype of cardiac and palate abnormalities, developmental delay, intellectual disability and dysmorphic features. American Journal of Medical Genetics, Part A, 2018, 176, 1845-1851.	1.2	21
144	Nonreentrant atrial tachycardia occurs independently of hypertrophic cardiomyopathy in RASopathy patients. American Journal of Medical Genetics, Part A, 2018, 176, 1711-1722.	1.2	21

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145	Activating variants in <scp>PDGFRB</scp> result in a spectrum of disorders responsive to imatinib monotherapy. American Journal of Medical Genetics, Part A, 2020, 182, 1576-1591.	1.2	21
146	Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. Human Molecular Genetics, 2020, 29, 1900-1921.	2.9	21
147	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	2.0	21
148	Aglossia with congenital absence of the mandibular rami and other craniofacial abnormalities. American Journal of Medical Genetics Part A, 1988, 31, 161-166.	2.4	20
149	A human case of SLC35A3â€related skeletal dysplasia. American Journal of Medical Genetics, Part A, 2017, 173, 2758-2762.	1.2	20
150	Variable Clinical Manifestations of Xiaâ€Gibbs syndrome: Findings of Consecutively Identified Cases at a Single Children's Hospital. American Journal of Medical Genetics, Part A, 2018, 176, 1890-1896.	1.2	20
151	PCDH19 -related epilepsy in a male with Klinefelter syndrome: Additional evidence supporting PCDH19 cellular interference disease mechanism. Epilepsy Research, 2018, 145, 89-92.	1.6	20
152	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
153	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
154	Nasal dimple as part of the 22q11.2 deletion syndrome. , 1997, 69, 290-292.		19
155	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
156	Heterotaxia in a fetus with campomelia, cervical lymphocele, polysplenia, and multicystic dysplastic kidneys: Expanding the phenotype of Cumming syndrome. , 1997, 73, 419-424.		18
157	Impairment of different protein domains causes variable clinical presentation within Pitt-Hopkins syndrome and suggests intragenic molecular syndromology of TCF4. European Journal of Medical Genetics, 2017, 60, 565-571.	1.3	18
158	Bi-allelic Loss-of-Function Variants in NUP188 Cause a Recognizable Syndrome Characterized by Neurologic, Ocular, and Cardiac Abnormalities. American Journal of Human Genetics, 2020, 106, 623-631.	6.2	18
159	46, XX, 15p+ documented as dup (17p) by fluorescence in situ hybridization. American Journal of Medical Genetics Part A, 1993, 46, 95-97.	2.4	17
160	Aphallia as part of urorectal septum malformation sequence in an infant of a diabetic mother. American Journal of Medical Genetics Part A, 1999, 82, 363-367.	2.4	17
161	Limb body wall complex, amniotic band sequence, or new syndrome caused by mutation in IQ Motif containing K (IQCK)?. Molecular Genetics & Genomic Medicine, 2015, 3, 424-432.	1.2	17
162	White matter microstructural deficits in 22q11.2 deletion syndrome. Psychiatry Research - Neuroimaging, 2017, 268, 35-44.	1.8	17

#	Article	IF	Citations
163	Attention Deficit Hyperactivity Disorder Symptoms and Psychosis in 22q11.2 Deletion Syndrome. Schizophrenia Bulletin, 2018, 44, 824-833.	4.3	17
164	Neurologic challenges in 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2140-2145.	1.2	17
165	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
166	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
167	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
168	Hyperinsulinemic hypoglycemia in seven patients with de novo NSD1 mutations. American Journal of Medical Genetics, Part A, 2019, 179, 542-551.	1.2	16
169	Anomalies of the genitourinary tract in children with 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 381-385.	1.2	16
170	EP300 â€related Rubinsteinâ€“Taybi syndrome: Highlighted rare phenotypic findings and a genotypeâ€“phenotype metaâ€analysis of 74 patients. American Journal of Medical Genetics, Part A, 2020, 182, 2926-2938.	1.2	16
171	De novo loss-of-function variants in X-linked MED12 are associated with Hardikar syndrome in females. Genetics in Medicine, 2021, 23, 637-644.	2.4	16
172	Understanding the phenotypic spectrum of ASXL â€related disease: Ten cases and a review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 1700-1711.	1.2	16
173	Interstitial deletion of 4(q21q25) in a liveborn male. American Journal of Medical Genetics Part A, 1991, 40, 77-79.	2.4	15
174	Classical Noonan syndrome is not associated with deletions of 22q11. American Journal of Medical Genetics Part A, 1995, 56, 94-96.	2.4	15
175	Autosomal dominant mannose-binding lectin deficiency is associated with worse neurodevelopmental outcomes after cardiac surgery in infants. Journal of Thoracic and Cardiovascular Surgery, 2018, 155, 1139-1147.e2.	0.8	15
176	Phenotypic predictors and final diagnoses in patients referred for RASopathy testing by targeted next-generation sequencing. Genetics in Medicine, 2017, 19, 715-718.	2.4	14
177	Hearing Loss after Cardiac Surgery in Infancy: An Unintended Consequence of Life-Saving Care. Journal of Pediatrics, 2018, 192, 144-151.e1.	1.8	14
178	The dimensional structure of psychopathology in 22q11.2 Deletion Syndrome. Journal of Psychiatric Research, 2017, 92, 124-131.	3.1	13
179	Cytotoxic T-Lymphocyte-Associated Protein 4 Haploinsufficiency-Associated Inflammation Can Occur Independently of T-Cell Hyperproliferation. Frontiers in Immunology, 2018, 9, 1715.	4.8	13
180	Novel variants in CDH2 are associated with a new syndrome including Peters anomaly. Clinical Genetics, 2020, 97, 502-508.	2.0	13

#	Article	IF	Citations
181	Developmental profile in a patient with monosomy 10q and dup(17p) associated with a peripheral neuropathy. , 1996, 61, 377-381.		12
182	<scp>EGFR</scp> mutations cause a lethal syndrome of epithelial dysfunction with progeroid features. Molecular Genetics & Genomic Medicine, 2015, 3, 452-458.	1.2	12
183	Fetal akinesia deformation sequence due to a congenital disorder of glycosylation. American Journal of Medical Genetics, Part A, 2015, 167, 2411-2417.	1.2	12
184	The final demise of Rodriguez lethal acrofacial dysostosis: A case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1063-1068.	1.2	12
185	Management of velopharyngeal dysfunction in patients with 22q11.2 deletion syndrome: A survey of practice patterns. International Journal of Pediatric Otorhinolaryngology, 2019, 116, 43-48.	1.0	12
186	De novo variants in CACNA1E found in patients with intellectual disability, developmental regression and social cognition deficit but no seizures. Molecular Autism, 2021, 12, 69.	4.9	12
187	Frontonasal malformation and cloacal exstrophy: A previously unreported association. , 1996, 61, 75-78.		11
188	Arthritis associated with deletion of 22q11.2: More common than previously suspected. , 1997, 71, 488-488.		11
189	Not Antley-Bixler syndrome. , 1999, 83, 65-66.		11
190	Kabuki syndrome as a cause of nonâ€immune fetal hydrops/ascites. American Journal of Medical Genetics, Part A, 2016, 170, 3333-3337.	1.2	11
191	Early photoreceptor outer segment loss and retinoschisis in Cohen syndrome. Ophthalmic Genetics, 2018, 39, 399-404.	1.2	11
192	Missense Mutations in NKAP Cause a Disorder of Transcriptional Regulation Characterized by Marfanoid Habitus and Cognitive Impairment. American Journal of Human Genetics, 2019, 105, 987-995.	6.2	11
193	Tattonâ€Brownâ€Rahman syndrome: Six individuals with novel features. American Journal of Medical Genetics, Part A, 2020, 182, 673-680.	1.2	11
194	Somatic and germ line mosaicism and mutation origin for a mutation in theL1 gene in a family with X-linked hydrocephalus. , 1998, 75, 200-202.		10
195	Study of carrier frequency of Warsaw breakage syndrome in the Ashkenazi Jewish population and presentation of two cases. American Journal of Medical Genetics, Part A, 2019, 179, 2144-2151.	1.2	10
196	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.	1.7	10
197	Chromatin Modifications in 22q11.2 Deletion Syndrome. Journal of Clinical Immunology, 2021, 41, 1853-1864.	3.8	10
198	Consolidation of the clinical and genetic definition of aSOX4-related neurodevelopmental syndrome. Journal of Medical Genetics, 2022, 59, 1058-1068.	3.2	10

#	Article	IF	Citations
199	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
200	Non-immune hydrops fetalis associated with impaired fetal movement: A case report and review. American Journal of Medical Genetics Part A, 1994, 53, 251-254.	2.4	9
201	Otologic Manifestations of Neurofibromatosis. Laryngoscope, 1994, 104, 663???665.	2.0	9
202	Sibs with cleidocranial dysplasia born to normal parents: Germ line mosaicism?. , 1997, 69, 348-351.		9
203	Expanding the genetic landscape of oralâ€facialâ€digital syndrome with two novel genes. American Journal of Medical Genetics, Part A, 2021, 185, 2409-2416.	1.2	9
204	Bi-allelic variants in the ER quality-control mannosidase gene EDEM3 cause a congenital disorder of glycosylation. American Journal of Human Genetics, 2021, 108, 1342-1349.	6.2	9
205	The Genomics of Congenital Diaphragmatic Hernia: A 10-Year Retrospective Review. Journal of Pediatrics, 2022, 248, 108-113.e2.	1.8	9
206	Genetics etiologies and genotype phenotype correlations in a cohort of individuals with central conducting lymphatic anomaly. European Journal of Human Genetics, 2022, 30, 1022-1028.	2.8	9
207	De novo interstitial deletion of the long arm of chromosome 3:46, XX, del(3) (q25.1q26.1). Clinical Genetics, 1993, 44, 335-337.	2.0	8
208	A catalog of hemizygous variation in 127 22q11 deletion patients. Human Genome Variation, 2016, 3, 15065.	0.7	8
209	Olfactory deficits and psychosis-spectrum symptoms in 22q11.2 deletion syndrome. Schizophrenia Research, 2018, 202, 113-119.	2.0	8
210	Congenital diaphragmatic hernia as a prominent feature of a SPECC1L â€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2919-2925.	1.2	8
211	Relationship between intelligence quotient measures and computerized neurocognitive performance in 22q11.2 deletion syndrome. Brain and Behavior, 2021, 11, e2221.	2.2	8
212	Characteristic calcaneal ossification: an additional early radiographic finding in infants with fibrodysplasia ossificans progressiva. Pediatric Radiology, 2016, 46, 1568-1572.	2.0	7
213	A vascular endothelial growth factor A genetic variant is associated with improved ventricular function and transplant-free survival after surgery for non-syndromic CHD. Cardiology in the Young, 2018, 28, 39-45.	0.8	7
214	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.	1.2	7
215	Clinical variability of TUBB â€associated disorders: Diagnosis through reanalysis. American Journal of Medical Genetics, Part A, 2020, 182, 3035-3039.	1.2	7
216	Lossâ€ofâ€function of Endothelin receptor type A results in Oroâ€Otoâ€Cardiac syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1104-1116.	1.2	7

#	Article	IF	Citations
217	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	2.4	7
218	Cardiac evaluation of patients with 22q11.2 duplication syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 753-758.	1.2	7
219	Molecular Diagnostic Outcomes from 700 Cases. Journal of Molecular Diagnostics, 2022, 24, 274-286.	2.8	7
220	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.	1.7	6
221	CMIP haploinsufficiency in two patients with autism spectrum disorder and coâ€occurring gastrointestinal issues. American Journal of Medical Genetics, Part A, 2017, 173, 2101-2107.	1.2	6
222	A Novel Approach to Dysmorphology to Enhance the Phenotypic Classification of Autism Spectrum Disorder in the Study to Explore Early Development. Journal of Autism and Developmental Disorders, 2019, 49, 2184-2202.	2.7	6
223	Aortic Root Dilation in Patients with 22q11.2 Deletion Syndrome Without Intracardiac Anomalies. Pediatric Cardiology, 2021, 42, 1594-1600.	1.3	6
224	Prenatal diagnosis of mosaicism 46, XX/46, XX, â^'21, +t(21q21q). Prenatal Diagnosis, 1984, 4, 73-77.	2.3	5
225	Primary lymphedema and other lymphatic anomalies are associated with 22q11.2 deletion syndrome. European Journal of Medical Genetics, 2018, 61, 411-415.	1.3	5
226	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
227	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. American Journal of Medical Genetics, Part A, 2019, 179, 1543-1546.	1.2	5
228	A binational study assessing risk and resilience factors in 22q11.2 deletion syndrome. Journal of Psychiatric Research, 2021, 138, 319-325.	3.1	5
229	Clinical Effectiveness of Telemedicine-Based Pediatric Genetics Care. Pediatrics, 2022, 150, .	2.1	5
230	Craniosynostosis: Molecular testing?a necessity for counseling. , 2000, 92, 157-157.		4
231	Detection of mutually exclusive mosaicism in a girl with genotypeâ€phenotype discrepancies. American Journal of Medical Genetics, Part A, 2015, 167, 3091-3095.	1.2	4
232	Compound heterozygote CDK5RAP2 mutations in a Guatemalan/Honduran child with autosomal recessive primary microcephaly, failure to thrive and speech delay. American Journal of Medical Genetics, Part A, 2015, 167, 1414-1417.	1.2	4
233	Disrupted anatomic networks in the 22q11.2 deletion syndrome. NeuroImage: Clinical, 2016, 12, 420-428.	2.7	4
234	Magnetic resonance angiography (MRA) in preoperative planning for patients with 22q11.2 deletion syndrome undergoing craniofacial and otorhinolaryngologic procedures. International Journal of Pediatric Otorhinolaryngology, 2020, 138, 110236.	1.0	4

#	Article	IF	Citations
235	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. Human Genetics, 2021, 140, 1061-1076.	3.8	4
236	A novel <scp>MBTPS2</scp> variant associated with <scp>BRESHECK</scp> syndrome impairs <scp>sterolâ€regulated</scp> transcription and the endoplasmic reticulum stress response. American Journal of Medical Genetics, Part A, 2022, 188, 463-472.	1.2	4
237	Molecular Mechanisms Contributing to the Etiology of Congenital Diaphragmatic Hernia: A Review and Novel Cases. Journal of Pediatrics, 2022, 246, 251-265.e2.	1.8	4
238	Phenotypic modifications of patients with full chromosome aneuploidies and concurrent suspected or confirmed second diagnoses. American Journal of Medical Genetics, Part A, 2015, 167, 2168-2175.	1.2	3
239	10â€yearâ€old female with intragenic KANSL1 mutation, no KANSL1â€related intellectual disability, and preserved verbal intelligence. American Journal of Medical Genetics, Part A, 2017, 173, 762-765.	1.2	3
240	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
241	Congenital polyvalvular disease expands the cardiac phenotype of the RASopathies. American Journal of Medical Genetics, Part A, 2021, 185, 1486-1493.	1.2	3
242	Nonlethal presentations of CYP26B1 â€related skeletal anomalies and multiple synostoses syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2766-2775.	1.2	3
243	Ablepharon macrostomia syndrome with associated cutis laxa: possible localization to 18q. Human Genetics, 1996, 97, 532-536.	3.8	3
244	Further supporting <scp>SMARCC2</scp>â€related neurodevelopmental disorder through exome analysis and reanalysis in two patients. American Journal of Medical Genetics, Part A, 2022, 188, 878-882.	1.2	3
245	Surgical insights and management in patients with the 22q11.2 deletion syndrome. Pediatric Surgery International, 2022, 38, 899-905.	1.4	3
246	Gene location in neurofibromatosis. American Journal of Medical Genetics Part A, 1988, 29, 963-965.	2.4	2
247	Increased Tâ€cell counts in patients with 22q11.2 deletion syndrome who have anxiety. American Journal of Medical Genetics, Part A, 2020, 182, 1815-1818.	1.2	2
248	Hyperinsulinism in an individual with an EP300 variant of Rubinsteinâ€Taybi syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1251-1255.	1.2	2
249	Chromosome 4q28.3q32.3 duplication in a patient with lymphatic malformations, craniosynostosis, and dysmorphic features. Clinical Dysmorphology, 2021, 30, 89-92.	0.3	2
250	Atypical Williams syndrome in an infant with complete atrioventricular canal defect. American Journal of Medical Genetics, Part A, 2015, 167, 3108-3112.	1.2	1
251	Muenke syndrome: Medical and surgical comorbidities and longâ€term management. American Journal of Medical Genetics, Part A, 2019, 179, 1442-1450.	1.2	1
252	Cleft palate morphology, genetic etiology, and risk of mortality in infants with Robin sequence. American Journal of Medical Genetics, Part A, 2021, 185, 3694-3700.	1.2	1

#	Article	IF	Citations
253	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
254	Altered functional brain dynamics in chromosome 22q11.2 deletion syndrome during facial affect processing. Molecular Psychiatry, 2022, 27, 1158-1166.	7.9	1
255	The microdeletion syndromes. , 2005, , .		0
256	Cover Image, Volume 170A, Number 5, May 2016. , 2016, 170, i-i.		0
257	Cover Image, Volume 176A, Number 4, April 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
258	Cover Image, Volume 176A, Number 10, October 2018. , 2018, 176, i-i.		0
259	Mapping the Relationship between Dysmorphology and Cognitive, Behavioral, and Developmental Outcomes in Children with Autism Spectrum Disorder. Autism Research, 2020, 13, 1227-1238.	3.8	0
260	A Case of Prenatally Diagnosed Periventricular Nodular Heterotopia in a Surviving Male Patient with FLNA Mutation. Journal of Pediatric Neurology, 2022, 20, 057-059.	0.2	0
261	Expanding the phenotypic spectrum of Mendelian connective tissue disorders to include prominent kidney phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 3762-3769.	1.2	0
262	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
263	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	2.4	0