Elaine H Zackai

List of Publications by Citations

249 papers	9,601 citations	47 h-index	89 g-index
275 ext. papers	11,611 ext. citations	6.1 avg, IF	5.3 L-index

#	Paper	IF	Citations
249	22q11.2 deletion syndrome. *Nature Reviews Disease Primers*, **2015**, 1, 15071	51.1	492
248	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, 627-39	11.9	472
247	Mutations in the homeodomain of the human SIX3 gene cause holoprosencephaly. *Nature Genetics*, **1999**, 22, 196-8	36.3	360
246	Deletions and microdeletions of 22q11.2 in velo-cardio-facial syndrome. *American Journal of Medical Genetics Part A*, **1992**, 44, 261-8		347
245	Mutations in TGIF cause holoprosencephaly and link NODAL signalling to human neural axis determination. *Nature Genetics*, **2000**, 25, 205-8	36.3	337
244	Identical mutations in three different fibroblast growth factor receptor genes in autosomal dominant craniosynostosis syndromes. *Nature Genetics*, **1996**, 14, 174-6	36.3	270
243	Mutations of ARX are associated with striking pleiotropy and consistent genotype-phenotype correlation. *Human Mutation*, **2004**, 23, 147-59	4.7	259
242	Cognitive and behavior profile of preschool children with chromosome 22q11.2 deletion. *American Journal of Medical Genetics Part A*, **1999**, 85, 127-133		235
241	Immunologic features of chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *Journal of Pediatrics*, **2001**, 139, 715-23	3.6	213
240	Optic pathway and hypothalamic/chiasmatic gliomas in children younger than age 5 years with a 6-year follow-up. *Cancer*, **1995**, 75, 1051-9	6.4	207
239	Analysis of 589,306 genomes identifies individuals resilient to severe Mendelian childhood diseases. *Nature Biotechnology*, **2016**, 34, 531-8	44.5	196
238	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		194
237	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, 507-21		152
236	Down syndrome congenital heart disease: a narrowed region and a candidate gene. *Genetics in Medicine*, **2001**, 3, 91-101	8.1	138
235	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, 343-52	11	136
234	Growth Charts for Children With Down Syndrome: 1 Month to 18 Years of Age. *Pediatrics*, **1988**, 81, 102-110		130
233	The 22q11.2 deletion: screening, diagnostic workup, and outcome of results; report on 181 patients. *Genetic Testing and Molecular Biomarkers*, **1997**, 1, 99-108		124

232	Aberrant interchromosomal exchanges are the predominant cause of the 22q11.2 deletion. *Human Molecular Genetics*, **2004**, 13, 417-28	5.6	122
231	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotype-Phenotype Correlation. *Human Mutation*, **2015**, 36, 1052-63	4.7	112
230	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. *Journal of Medical Genetics*, **2017**, 54, 460-470	5.8	109
229	Reanalysis of Clinical Exome Sequencing Data. *New England Journal of Medicine*, **2019**, 380, 2478-2480	59.2	109
228	T-cell homeostasis in humans with thymic hypoplasia due to chromosome 22q11.2 deletion syndrome. *Blood*, **2004**, 103, 1020-5	2.2	102
227	Juvenile rheumatoid arthritis-like polyarthritis in chromosome 22q11.2 deletion syndrome (DiGeorge anomalad/velocardiofacial syndrome/conotruncal anomaly face syndrome). *Arthritis and Rheumatism*, **1997**, 40, 430-6		101
226	Kabuki syndrome genes KMT2D and KDM6A: functional analyses demonstrate critical roles in craniofacial, heart and brain development. *Human Molecular Genetics*, **2015**, 24, 4443-53	5.6	96
225	Clinical, cytogenetic, and pedigree findings in 18 cases of Aicardi syndrome. *American Journal of Medical Genetics Part A*, **1989**, 32, 461-7		95
224	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844-848. *American Journal of Human Genetics*, **2018**, 102, 69-87	11	93
223	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	5.8	90
222	Oral-facial-digital syndrome type VI (V□adi syndrome): further clinical delineation. *American Journal of Medical Genetics Part A*, **1990**, 35, 360-9		89
221	Deletions of different segments of the long arm of chromosome 4. *American Journal of Medical Genetics Part A*, **1981**, 8, 73-89		88
220	Germline gain-of-function mutations in AFF4 cause a developmental syndrome functionally linking the super elongation complex and cohesin. *Nature Genetics*, **2015**, 47, 338-44	36.3	72
219	Dominant mutations in KAT6A cause intellectual disability with recognizable syndromic features. *American Journal of Human Genetics*, **2015**, 96, 507-13	11	70
218	Mutations in the human TWIST gene. *Human Mutation*, **2000**, 15, 150-5	4.7	70
217	Skeletal anomalies and deformities in patients with deletions of 22q11. *American Journal of Medical Genetics Part A*, **1997**, 72, 210-5		67
216	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, 125-38	11	67
215	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. *European Journal of Human Genetics*, **2015**, 23, 1473-81	5.3	63

214	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, 906-11		63
213	Neurodevelopmental outcomes in preschool survivors of the Fontan procedure. *Journal of Thoracic and Cardiovascular Surgery*, **2014**, 147, 1276-82; discussion 1282-1283.e5	1.5	60
212	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. *American Journal of Psychiatry*, **2017**, 174, 1054-1063	11.9	58
211	Phenotype of the fibroblast growth factor receptor 2 Ser351Cys mutation: Pfeiffer syndrome type III. *American Journal of Medical Genetics Part A*, **1998**, 78, 356-60		58
210	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, 753-64	11	54
209	Polytopic anomalies with agenesis of the lower vertebral column. *American Journal of Medical Genetics Part A*, **1999**, 87, 99-114		54
208	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	2.5	54
207	Aberrant Cortical Morphometry in the 22q11.2 Deletion Syndrome. *Biological Psychiatry*, **2015**, 78, 135-43	7.9	53
206	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. *Genetics in Medicine*, **2016**, 18, 309-15	8.1	52
205	Enlarged sylvian fissures in infants with interstitial deletion of chromosome 22q11. *American Journal of Medical Genetics Part A*, **1997**, 74, 538-543		50
204	Mouse and human CRKL is dosage sensitive for cardiac outflow tract formation. *American Journal of Human Genetics*, **2015**, 96, 235-44	11	47
203	Clinical spectrum of individuals with pathogenic NF1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotype-phenotype study in neurofibromatosis type 1. *Human Mutation*, **2020**, 41, 299-315	4.7	47
202	Oculodentodigital dysplasia syndrome associated with abnormal cerebral white matter. *American Journal of Medical Genetics Part A*, **1991**, 41, 18-20		46
201	Melnick-Needles syndrome in males: a lethal multiple congenital anomalies syndrome. *American Journal of Medical Genetics Part A*, **1987**, 27, 159-73		46
200	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotype-phenotype correlation. *Genetics in Medicine*, **2019**, 21, 867-876	8.1	43
199	22q11.2 duplication syndrome: elevated rate of autism spectrum disorder and need for medical screening. *Molecular Autism*, **2016**, 7, 27	6.5	42
198	Lateral meningocele syndrome: Three new patients and review of the literature. *American Journal of Medical Genetics Part A*, **1997**, 70, 229-239		42
197	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	11	42

196	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, 548-56		41
195	Saethre-Chotzen syndrome with familial translocation at chromosome 7p22. *American Journal of Medical Genetics Part A*, **1993**, 47, 637-9		41
194	Histone Modifier Genes Alter Conotruncal Heart Phenotypes in 22q11.2 Deletion Syndrome. *American Journal of Human Genetics*, **2015**, 97, 869-77	11	40
193	De novo truncating mutations in AHDC1 in individuals with syndromic expressive language delay, hypotonia, and sleep apnea. *American Journal of Human Genetics*, **2014**, 94, 784-9	11	40
192	Disrupted auto-regulation of the spliceosomal gene SNRPB causes cerebro-costo-mandibular syndrome. *Nature Communications*, **2014**, 5, 4483	17.4	40
191	22q11.2 Deletion syndrome and obstructive sleep apnea. *International Journal of Pediatric Otorhinolaryngology*, **2014**, 78, 1360-4	1.7	38
190	Natural history and genotype-phenotype correlations in 72 individuals with SATB2-associated syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 925-935	2.5	37
189	Holoprosencephaly: association with interstitial deletion of 2p and review of the cytogenetic literature. *American Journal of Medical Genetics Part A*, **1988**, 30, 929-38		37
188	Association of airway abnormalities with 22q11.2 deletion syndrome. *International Journal of Pediatric Otorhinolaryngology*, **2017**, 96, 11-14	1.7	36
187	Partial duplication 1q: report of four patients and review of the literature. *American Journal of Medical Genetics Part A*, **1990**, 36, 137-43		36
186	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, 1147-51.e4	1.5	35
185	TWIST gene mutation in a patient with radial aplasia and craniosynostosis: further evidence for heterogeneity of Baller-Gerold syndrome. *American Journal of Medical Genetics Part A*, **1999**, 82, 170-6		35
184	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, 482-9	1.5	35
183	The 22q11.2 deletion syndrome: Cancer predisposition, platelet abnormalities and cytopenias. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2121-2127	2.5	34
182	Molecular detection of a Yp/18 translocation in a 45,X holoprosencephalic male. *Human Genetics*, **1988**, 80, 219-23	6.3	34
181	The Psychosis Spectrum in 22q11.2 Deletion Syndrome Is Comparable to That of Nondeleted Youths. *Biological Psychiatry*, **2017**, 82, 17-25	7.9	33
180	Cystic kidney disease in Hajdu-Cheney syndrome. *American Journal of Medical Genetics Part A*, **1995**, 56, 25-30		33
179	Subthreshold Psychosis in 22q11.2 Deletion Syndrome: Multisite Naturalistic Study. *Schizophrenia Bulletin*, **2017**, 43, 1079-1089	1.3	32

178	Neurocognitive profile in psychotic versus nonpsychotic individuals with 22q11.2 deletion syndrome. *European Neuropsychopharmacology*, **2016**, 26, 1610-8	1.2	31
177	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. *Human Genetics*, **2016**, 135, 273-85	6.3	31
176	Congenital heart disease in supernumerary der(22),t(11;22) syndrome. *Clinical Genetics*, **1986**, 29, 269-754		31
175	Craniosynostosis: another feature of the 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2005**, 136A, 358-62	2.5	31
174	New finding of Schinzel-Giedion syndrome: a case with a malignant sacrococcygeal teratoma. *American Journal of Medical Genetics Part A*, **1993**, 47, 852-6		31
173	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. *Human Mutation*, **2016**, 37, 148-54	4.7	31
172	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. *Clinical Epigenetics*, **2019**, 11, 64	7.7	29
171	Congenital heart diseases and cardiovascular abnormalities in 22q11.2 deletion syndrome: From well-established knowledge to new frontiers. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2087-2098	2.5	29
170	A prospective study of influenza vaccination and a comparison of immunologic parameters in children and adults with chromosome 22q11.2 deletion syndrome (digeorge syndrome/velocardiofacial syndrome). *Journal of Clinical Immunology*, **2011**, 31, 927-35	5.7	29
169	Further evidence that the Hajdu-Cheney syndrome and the "serpentine fibula-polycystic kidney syndrome" are a single entity. *American Journal of Medical Genetics Part A*, **1998**, 78, 474-81		29
168	Gain-of-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, 2617-31	2.5	29
167	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		28
166	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	2.5	27
165	Neuropathological findings in eight children with cerebro-oculo-facio-skeletal (COFS) syndrome. *Journal of Neuropathology and Experimental Neurology*, **1997**, 56, 1147-57	3.1	27
164	Bilateral microtia and cleft palate in cousins with Diamond-Blackfan anemia. *American Journal of Medical Genetics Part A*, **2001**, 101, 268-74		27
163	Elucidating the diagnostic odyssey of 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 936-944	2.5	26
162	Mutations in SPECC1L, 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, 104-10	5.8	26
161	Ablepharon macrostomia syndrome with associated cutis laxa: possible localization to 18q. *Human Genetics*, **1996**, 97, 532-6	6.3	26

160	Blepharo-cheilo-dontic (BCD) syndrome. *American Journal of Medical Genetics Part A*, **1996**, 65, 109-12		26
159	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, 61-5		26
158	Prenatal detection of Roberts-SC phocomelia syndrome: report of 2 sibs with characteristic manifestations. *American Journal of Medical Genetics Part A*, **1989**, 32, 390-4		26
157	Identification of 22q11.2 Deletion Syndrome via Newborn Screening for Severe Combined Immunodeficiency. *Journal of Clinical Immunology*, **2017**, 37, 476-485	5.7	25
156	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	2.5	24
155	Mutations in topoisomerase IIβ result in a B cell immunodeficiency. *Nature Communications*, **2019**, 10, 3644	17.4	24
154	Tracheal cartilaginous sleeves in children with syndromic craniosynostosis. *Genetics in Medicine*, **2017**, 19, 62-68	8.1	24
153	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	6.3	24
152	Unusual craniofacial dysmorphia due to prenatal alcohol and cocaine exposure. *Teratology*, **1994**, 50, 160-4		24
151	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	11	24
150	Intragenic KANSL1 mutations and chromosome 17q21.31 deletions: broadening the clinical spectrum and genotype-phenotype correlations in a large cohort of patients. *Journal of Medical Genetics*, **2015**, 52, 804-14	5.8	23
149	Kabuki syndrome is not caused by a microdeletion in the DiGeorge/velocardiofacial chromosomal region within 22q 11.2. *American Journal of Medical Genetics Part A*, **1996**, 65, 101-3		23
148	Patient genotypes impact survival after surgery for isolated congenital heart disease. *Annals of Thoracic Surgery*, **2014**, 98, 104-10; discussion 110-1	2.7	22
147	Thrombocytopenia with absent radius in a boy and his uncle. *American Journal of Medical Genetics Part A*, **1987**, 28, 117-23		22
146	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotype-phenotype correlations, and molecular basis. *Genetics in Medicine*, **2020**, 22, 389-397	8.1	22
145	Rates of autism and potential risk factors in children with congenital heart defects. *Congenital Heart Disease*, **2017**, 12, 421-429	3.1	21
144	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	11	21
143	Expanding the SPECC1L mutation phenotypic spectrum to include Teebi hypertelorism syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2497-502	2.5	21

142	Mosaicism for a chromosome 8-derived minute marker chromosome in a patient with manifestations of trisomy 8 mosaicism. *American Journal of Medical Genetics Part A*, **1995**, 56, 22-4		21
141	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, 700-5		21
140	Novel findings with reassessment of exome data: implications for validation testing and interpretation of genomic data. *Genetics in Medicine*, **2018**, 20, 329-336	8.1	20
139	The Role of mGluR Copy Number Variation in Genetic and Environmental Forms of Syndromic Autism Spectrum Disorder. *Scientific Reports*, **2016**, 6, 19372	4.9	20
138	B cell development in chromosome 22q11.2 deletion syndrome. *Clinical Immunology*, **2016**, 163, 1-9	9	20
137	Diaphragmatic hernia-exomphalos-hypertelorism syndrome: a new case and further evidence of autosomal recessive inheritance. *American Journal of Medical Genetics Part A*, **1997**, 68, 441-4		19
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, 21-8		19
135	Recurrence rate for de novo 21q21q translocation Down syndrome: a study of 112 families. *American Journal of Medical Genetics Part A*, **1984**, 17, 523-30		19
134	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Signaling. *Biological Psychiatry*, **2020**, 87, 100-112	7.9	19
133	A dyadic approach to the delineation of diagnostic entities in clinical genomics. *American Journal of Human Genetics*, **2021**, 108, 8-15	11	19
132	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. *American Journal of Human Genetics*, **2018**, 103, 752-768	11	19
131	Critical region within 22q11.2 linked to higher rate of autism spectrum disorder. *Molecular Autism*, **2017**, 8, 58	6.5	18
130	Deletion size analysis of 1680 22q11.2DS subjects identifies a new recombination hotspot on chromosome 22q11.2. *Human Molecular Genetics*, **2018**, 27, 1150-1163	5.6	18
129	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, 102-8	8.1	18
128	Early ultrasound diagnosis of Neu-Laxova syndrome. *Prenatal Diagnosis*, **2001**, 21, 575-80	3.2	18
127	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. *Brain*, **2020**, 143, 55-68	11.2	18
126	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	2.5	18
125	Congenital diaphragmatic hernia in 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 135-142	2.5	17

124	Exome sequencing expands the mechanism of SOX5-associated intellectual disability: A case presentation with review of sox-related disorders. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2548-54	2.5	17
123	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	2.5	17
122	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	11	17
121	Nasal dimple as part of the 22q11.2 deletion syndrome **1997**, 69, 290-292		16
120	Heterotaxia in a fetus with campomelia, cervical lymphocele, polysplenia, and multicystic dysplastic kidneys: expanding the phenotype of Cumming syndrome. *American Journal of Medical Genetics Part A*, **1997**, 73, 419-24		16
119	A human case of SLC35A3-related skeletal dysplasia. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2758-2762	2.5	15
118	Interstitial deletion of 4(q21q25) in a liveborn male. *American Journal of Medical Genetics Part A*, **1991**, 40, 77-9		15
117	46,XX,15p+ documented as dup (17p) by fluorescence in situ hybridization. *American Journal of Medical Genetics Part A*, **1993**, 46, 95-7		15
116	Aglossia with congenital absence of the mandibular rami and other craniofacial abnormalities. *American Journal of Medical Genetics Part A*, **1988**, 4, 161-6		15
115	22q and two: 22q11.2 deletion syndrome and coexisting conditions. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2203-2214	2.5	15
114	Variable Clinical Manifestations of Xia-Gibbs syndrome: Findings of Consecutively Identified Cases at a Single Children's Hospital. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1890-1896	2.5	15
113	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the Locus on 5q14.3. *Circulation: Cardiovascular Genetics*, **2017**, 10,		14
112	Attention Deficit Hyperactivity Disorder Symptoms and Psychosis in 22q11.2 Deletion Syndrome. *Schizophrenia Bulletin*, **2018**, 44, 824-833	1.3	14
111	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	2.7	14
110	White matter microstructural deficits in 22q11.2 deletion syndrome. *Psychiatry Research - Neuroimaging*, **2017**, 268, 35-44	2.9	14
109	Classical Noonan syndrome is not associated with deletions of 22q11. *American Journal of Medical Genetics Part A*, **1995**, 56, 94-6		14
108	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, 790-4		14
107	Negative subthreshold psychotic symptoms distinguish 22q11.2 deletion syndrome from other neurodevelopmental disorders: A two-site study. *Schizophrenia Research*, **2017**, 188, 42-49	3.6	13

106	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, 424-32	2.3	13
105	Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. *Human Molecular Genetics*, **2020**, 29, 1900-1921	5.6	12
104	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	5.6	12
103	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	4.4	12
102	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	2.5	12
101	Nonreentrant atrial tachycardia occurs independently of hypertrophic cardiomyopathy in RASopathy patients. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1711-1722	2.5	12
100	Phenotypic predictors and final diagnoses in patients referred for RASopathy testing by targeted next-generation sequencing. *Genetics in Medicine*, **2017**, 19, 715-718	8.1	12
99	Developmental profile in a patient with monosomy 10q and dup(17p) associated with a peripheral neuropathy. *American Journal of Medical Genetics Part A*, **1996**, 61, 377-81		12
98	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6,	14.3	12
97	Cerebro-costo-mandibular syndrome: Clinical, radiological, and genetic findings. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1115-26	2.5	12
96	Neurologic challenges in 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2140-2145	2.5	12
95	PCDH19-related epilepsy in a male with Klinefelter syndrome: Additional evidence supporting PCDH19 cellular interference disease mechanism. *Epilepsy Research*, **2018**, 145, 89-92	3	12
94	Activating variants in PDGFRB result in a spectrum of disorders responsive to imatinib monotherapy. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1576-1591	2.5	11
93	Attention deficit hyperactivity disorder symptoms as antecedents of later psychotic outcomes in 22q11.2 deletion syndrome. *Schizophrenia Research*, **2019**, 204, 320-325	3.6	11
92	Phenotype delineation of ZNF462 related syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2075-2082	2.5	11
91	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	2.6	11
90	Arthritis associated with deletion of 22q11.2: More common than previously suspected **1997**, 71, 488-488		11
89	The dimensional structure of psychopathology in 22q11.2 Deletion Syndrome. *Journal of Psychiatric Research*, **2017**, 92, 124-131	5.2	10

88	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	5.3	10
87	Early photoreceptor outer segment loss and retinoschisis in Cohen syndrome. *Ophthalmic Genetics*, **2018**, 39, 399-404	1.2	10
86	Cytotoxic T-Lymphocyte-Associated Protein 4 Haploinsufficiency-Associated Inflammation Can Occur Independently of T-Cell Hyperproliferation. *Frontiers in Immunology*, **2018**, 9, 1715	8.4	10
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