Elaine H Zackai

List of Publications by Year in Descending Order

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	Surgical insights and management in patients with the 22q11.2 deletion syndrome.. *Pediatric Surgery International*, **2022**, 38, 899	2.1	0
248	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	6.5	0
247	Chromosome 4q28.3q32.3 duplication in a patient with lymphatic malformations, craniosynostosis, and dysmorphic features. *Clinical Dysmorphology*, **2021**, 30, 89-92	0.9	1
246	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1649-1665	2.5	0
245	Understanding the phenotypic spectrum of ASXL-related disease: Ten cases and a review of the literature. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1700-1711	2.5	5
244	Congenital polyvalvular disease expands the cardiac phenotype of the RASopathies. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1486-1493	2.5	1
243	Disruption of the blood-brain barrier in 22q11.2 deletion syndrome. *Brain*, **2021**, 144, 1351-1360	11.2	7
242	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. *Human Genetics*, **2021**, 140, 1061-1076	6.3	1
241	ANKRD11 variants: KBG syndrome and beyond. *Clinical Genetics*, **2021**, 100, 187-200	4	4
240	A binational study assessing risk and resilience factors in 22q11.2 deletion syndrome. *Journal of Psychiatric Research*, **2021**, 138, 319-325	5.2	3
239	Aortic Root Dilation in Patients with 22q11.2 Deletion Syndrome Without Intracardiac Anomalies. *Pediatric Cardiology*, **2021**, 42, 1594-1600	2.1	1
238	Nonlethal presentations of CYP26B1-related skeletal anomalies and multiple synostoses syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2766-2775	2.5	0
237	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. *Genetics in Medicine*, **2021**, 23, 1952-1960	8.1	1
236	Expanding the genetic landscape of oral-facial-digital syndrome with two novel genes. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2409-2416	2.5	1
235	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	2.5	
234	De novo loss-of-function variants in X-linked MED12 are associated with Hardikar syndrome in females. *Genetics in Medicine*, **2021**, 23, 637-644	8.1	7
233	A dyadic approach to the delineation of diagnostic entities in clinical genomics. *American Journal of Human Genetics*, **2021**, 108, 8-15	11	19

232	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*, **2021**,	5.9	6
231	Relationship between intelligence quotient measures and computerized neurocognitive performance in 22q11.2 deletion syndrome. *Brain and Behavior*, **2021**, 11, e2221	3.4	2
230	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	11	1
229	Association of Mitochondrial Biogenesis With Variable Penetrance of Schizophrenia. *JAMA Psychiatry*, **2021**, 78, 911-921	14.5	3
228	Chromatin Modifications in 22q11.2 Deletion Syndrome. *Journal of Clinical Immunology*, **2021**, 41, 1853-1864	3.7	1
227	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	2.5	
226	Response to Hamosh et´al. *American Journal of Human Genetics*, **2021**, 108, 1809-1810	11	
225	Hyperinsulinism in an individual with an EP300 variant of Rubinstein-Taybi syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1251-1255	2.5	0
224	Cardiac evaluation of patients with 22q11.2 duplication syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 753-758	2.5	1
223	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. *Genetics in Medicine*, **2020**, 22, 1338-1347	8.1	9
222	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
221	Mapping the Relationship between Dysmorphology and Cognitive, Behavioral, and Developmental Outcomes in Children with Autism Spectrum Disorder. *Autism Research*, **2020**, 13, 1227-1238	5.1	
220	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
219	Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. *Human Molecular Genetics*, **2020**, 29, 1900-1921	5.6	12
218	Loss-of-function of Endothelin receptor type A results in Oro-Oto-Cardiac syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1104-1116	2.5	2
217	Tatton-Brown-Rahman syndrome: Six individuals with novel features. *American Journal of Medical Genetics, Part A*, **2020**, 182, 673-680	2.5	3
216	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	11	5
215	Novel variants in CDH2 are associated with a new syndrome including Peters anomaly. *Clinical Genetics*, **2020**, 97, 502-508	4	6

214	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
213	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
212	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. *Brain*, **2020**, 143, 55-68	11.2	18
211	Clinical variability of TUBB-associated disorders: Diagnosis through reanalysis. *American Journal of Medical Genetics, Part A*, **2020**, 182, 3035-3039	2.5	1
210	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	1.7	2
209	EP300-related Rubinstein-Taybi syndrome: Highlighted rare phenotypic findings and a genotype-phenotype meta-analysis of 74 patients. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2926-2938	2.5	3
208	Congenital diaphragmatic hernia as a prominent feature of a SPECC1L-related syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2919-2925	2.5	2
207	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6,	14.3	12
206	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	4.9	10
205	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	3.5	3
204	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
203	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
202	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	8.1	10
201	Increased T-cell counts in patients with 22q11.2 deletion syndrome who have anxiety. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1815-1818	2.5	1
200	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1543-1546	2.5	3
199	Reanalysis of Clinical Exome Sequencing Data. *New England Journal of Medicine*, **2019**, 380, 2478-2480	59.2	109
198	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	2.9	2
197	Muenke syndrome: Medical and surgical comorbidities and long-term management. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1442-1450	2.5	

196	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. *Clinical Epigenetics*, **2019**, 11, 64	7.7	29
195	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	2.5	6
194	Hyperinsulinemic hypoglycemia in seven patients with de novo NSD1 mutations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 542-551	2.5	5
193	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
192	Mutations in topoisomerase II result in a B cell immunodeficiency. *Nature Communications*, **2019**, 10, 3644	17.4	24
191	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	2.5	6
190	Phenotype delineation of ZNF462 related syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2075-2082	2.5	11
189	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	11	3
188	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
187	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	4.6	3
186	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
185	Anomalies of the genitourinary tract in children with 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 381-385	2.5	6
184	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	1.7	7
183	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	2.6	10
182	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
181	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
180	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
179	Elucidating the diagnostic odyssey of 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 936-944	2.5	26

178	Early photoreceptor outer segment loss and retinoschisis in Cohen syndrome. *Ophthalmic Genetics*, **2018**, 39, 399-404	1.2	10
177	Primary lymphedema and other lymphatic anomalies are associated with 22q11.2 deletion syndrome. *European Journal of Medical Genetics*, **2018**, 61, 411-415	2.6	4
176	Attention Deficit Hyperactivity Disorder Symptoms and Psychosis in 22q11.2 Deletion Syndrome. *Schizophrenia Bulletin*, **2018**, 44, 824-833	1.3	14
175	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
174	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
173	Hearing Loss after Cardiac Surgery in Infancy: An Unintended Consequence of Life-Saving Care. *Journal of Pediatrics*, **2018**, 192, 144-151.e1	3.6	9
172	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	1.5	7
171	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
170	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
169	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
168	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
167	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
166	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
165	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
164	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
163	Olfactory deficits and psychosis-spectrum symptoms in 22q11.2 deletion syndrome. *Schizophrenia Research*, **2018**, 202, 113-119	3.6	7
162	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
161	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	1	6

160	Musical auditory processing, cognition, and psychopathology in 22q11.2 deletion syndrome. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2018**, 177, 765-773	3.5	2
159	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
158	22q and two: 22q11.2 deletion syndrome and coexisting conditions. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2203-2214	2.5	15
157	Neurologic challenges in 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2140-2145	2.5	12
156	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	2.5	2
155	What is new with 22q? An update from the 22q and You Center at the Children Hospital of Philadelphia. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2058-2069	2.5	54
154	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. *American Journal of Human Genetics*, **2018**, 103, 752-768	11	19
153	Variable Clinical Manifestations of Xia-Gibbs syndrome: Findings of Consecutively Identified Cases at a Single Children Hospital. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1890-1896	2.5	15
152	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
151	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
150	10-year-old female with intragenic KANSL1 mutation, no KANSL1-related intellectual disability, and preserved verbal intelligence. *American Journal of Medical Genetics, Part A*, **2017**, 173, 762-765	2.5	3
149	Association of airway abnormalities with 22q11.2 deletion syndrome. *International Journal of Pediatric Otorhinolaryngology*, **2017**, 96, 11-14	1.7	36
148	The dimensional structure of psychopathology in 22q11.2 Deletion Syndrome. *Journal of Psychiatric Research*, **2017**, 92, 124-131	5.2	10
147	CMIP haploinsufficiency in two patients with autism spectrum disorder and co-occurring gastrointestinal issues. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2101-2107	2.5	4
146	Subthreshold Psychosis in 22q11.2 Deletion Syndrome: Multisite Naturalistic Study. *Schizophrenia Bulletin*, **2017**, 43, 1079-1089	1.3	32
145	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. *Journal of Medical Genetics*, **2017**, 54, 460-470	5.8	109
144	Rates of autism and potential risk factors in children with congenital heart defects. *Congenital Heart Disease*, **2017**, 12, 421-429	3.1	21
143	Nested Inversion Polymorphisms Predispose Chromosome 22q11.2 to Meiotic Rearrangements. *American Journal of Human Genetics*, **2017**, 101, 616-622	11	6

142	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
141	Critical region within 22q11.2 linked to higher rate of autism spectrum disorder. *Molecular Autism*, **2017**, 8, 58	6.5	18
140	White matter microstructural deficits in 22q11.2 deletion syndrome. *Psychiatry Research - Neuroimaging*, **2017**, 268, 35-44	2.9	14
139	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
138	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
137	A human case of SLC35A3-related skeletal dysplasia. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2758-2762	2.5	15
136	Identification of 22q11.2 Deletion Syndrome via Newborn Screening for Severe Combined Immunodeficiency. *Journal of Clinical Immunology*, **2017**, 37, 476-485	5.7	25
135	The Psychosis Spectrum in 22q11.2 Deletion Syndrome Is Comparable to That of Nondeleted Youths. *Biological Psychiatry*, **2017**, 82, 17-25	7.9	33
134	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
133	Tracheal cartilaginous sleeves in children with syndromic craniosynostosis. *Genetics in Medicine*, **2017**, 19, 62-68	8.1	24
132	Congenital diaphragmatic hernia in 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 135-142	2.5	17
131	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. *Genetics in Medicine*, **2016**, 18, 309-15	8.1	52
130	Neurocognitive profile in psychotic versus nonpsychotic individuals with 22q11.2 deletion syndrome. *European Neuropsychopharmacology*, **2016**, 26, 1610-8	1.2	31
129	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
128	Disrupted anatomic networks in the 22q11.2 deletion syndrome. *NeuroImage: Clinical*, **2016**, 12, 420-8	5.3	3
127	A catalog of hemizygous variation in 127 22q11 deletion patients. *Human Genome Variation*, **2016**, 3, 15065	1.8	8
126	IQ and hemizygosity for the Val Met functional polymorphism of COMT in 22q11DS. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2016**, 171, 1112-1115	3.5	5
125	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

124	22q11.2 duplication syndrome: elevated rate of autism spectrum disorder and need for medical screening. *Molecular Autism*, **2016**, 7, 27	6.5	42
123	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
122	B cell development in chromosome 22q11.2 deletion syndrome. *Clinical Immunology*, **2016**, 163, 1-9	9	20
121	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. *Human Genetics*, **2016**, 135, 273-85	6.3	31
120	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
119	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
118	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. *Human Mutation*, **2016**, 37, 148-54	4.7	31
117	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
116	Analysis of 589,306 genomes identifies individuals resilient to severe Mendelian childhood diseases. *Nature Biotechnology*, **2016**, 34, 531-8	44.5	196
115	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
114	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
113	Kabuki syndrome as a cause of non-immune fetal hydrops/ascites. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3333-3337	2.5	10
112	Characteristic calcaneal ossification: an additional early radiographic finding in infants with fibrodysplasia ossificans progressiva. *Pediatric Radiology*, **2016**, 46, 1568-72	2.8	6
111	Cerebro-costo-mandibular syndrome: Clinical, radiological, and genetic findings. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1115-26	2.5	12
110	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
109	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
108	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
107	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

106	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
105	Aberrant Cortical Morphometry in the 22q11.2 Deletion Syndrome. *Biological Psychiatry*, **2015**, 78, 135-43	7.9	53
104	Histone Modifier Genes Alter Conotruncal Heart Phenotypes in 22q11.2 Deletion Syndrome. *American Journal of Human Genetics*, **2015**, 97, 869-77	11	40
103	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
102	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
101	EGFR mutations cause a lethal syndrome of epithelial dysfunction with progeroid features. *Molecular Genetics & Genomic Medicine*, **2015**, 3, 452-8	2.3	8
100	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
99	22q11.2 deletion syndrome. *Nature Reviews Disease Primers*, **2015**, 1, 15071	51.1	492
98	Atypical Williams syndrome in an infant with complete atrioventricular canal defect. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3108-12	2.5	1
97	Phenotypic modifications of patients with full chromosome aneuploidies and concurrent suspected or confirmed second diagnoses. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2168-75	2.5	3
96	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
95	Detection of mutually exclusive mosaicism in a girl with genotype-phenotype discrepancies. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3091-5	2.5	3
94	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, 1414-7	2.5	4
93	Fetal akinesia deformation sequence due to a congenital disorder of glycosylation. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2411-7	2.5	10
92	Dominant mutations in KAT6A cause intellectual disability with recognizable syndromic features. *American Journal of Human Genetics*, **2015**, 96, 507-13	11	70
91	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
90	Mouse and human CRKL is dosage sensitive for cardiac outflow tract formation. *American Journal of Human Genetics*, **2015**, 96, 235-44	11	47
89	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

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