Donna M Mcdonald-Mcginn

List of Publications by Citations

305 papers	17,471 citations	67 h-index	121 g-index
319 ext. papers	19,970 ext. citations	5.5 avg, IF	5.98 L-index

#	Paper	IF	Citations
305	Mutations in IRF6 cause Van der Woude and popliteal pterygium syndromes. *Nature Genetics*, **2002**, 32, 285-9	36.3	657
304	22q11.2 deletion syndrome. *Nature Reviews Disease Primers*, **2015**, 1, 15071	51.1	492
303	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
302	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. *Nature Genetics*, **2010**, 42, 203-9	36.3	461
301	Frequency of 22q11 deletions in patients with conotruncal defects. *Journal of the American College of Cardiology*, **1998**, 32, 492-8	15.1	457
300	Chromosome 22-specific low copy repeats and the 22q11.2 deletion syndrome: genomic organization and deletion endpoint analysis. *Human Molecular Genetics*, **2000**, 9, 489-501	5.6	394
299	Practical guidelines for managing patients with 22q11.2 deletion syndrome. *Journal of Pediatrics*, **2011**, 159, 332-9.e1	3.6	381
298	Prevalence of 22q11 microdeletions in DiGeorge and velocardiofacial syndromes: implications for genetic counselling and prenatal diagnosis. *Journal of Medical Genetics*, **1993**, 30, 813-7	5.8	379
297	Deletions and microdeletions of 22q11.2 in velo-cardio-facial syndrome. *American Journal of Medical Genetics Part A*, **1992**, 44, 261-8		347
296	Genomic and genic deletions of the FOX gene cluster on 16q24.1 and inactivating mutations of FOXF1 cause alveolar capillary dysplasia and other malformations. *American Journal of Human Genetics*, **2009**, 84, 780-91	11	328
295	Chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *Medicine (United States)*, **2011**, 90, 1-18	1.8	296
294	A unique point mutation in the fibroblast growth factor receptor 3 gene (FGFR3) defines a new craniosynostosis syndrome. *American Journal of Human Genetics*, **1997**, 60, 555-64	11	256
293	Psychoeducational profile of the 22q11.2 microdeletion: A complex pattern. *Journal of Pediatrics*, **1999**, 134, 193-8	3.6	251
292	The Philadelphia story: the 22q11.2 deletion: report on 250 patients. *Genetic Counseling*, **1999**, 10, 11-24		247
291	Cognitive and behavior profile of preschool children with chromosome 22q11.2 deletion. *American Journal of Medical Genetics Part A*, **1999**, 85, 127-133		235
290	Phenotype of the 22q11.2 deletion in individuals identified through an affected relative: cast a wide FISHing net!. *Genetics in Medicine*, **2001**, 3, 23-9	8.1	228
289	Immunologic features of chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *Journal of Pediatrics*, **2001**, 139, 715-23	3.6	213

288	Association of chromosome 22q11 deletion with isolated anomalies of aortic arch laterality and branching. *Journal of the American College of Cardiology*, **2001**, 37, 2114-9	15.1	204
287	Genomic screening of fibroblast growth-factor receptor 2 reveals a wide spectrum of mutations in patients with syndromic craniosynostosis. *American Journal of Human Genetics*, **2002**, 70, 472-86	11	203
286	Neuropsychological profile of children and adolescents with the 22q11.2 microdeletion. *Genetics in Medicine*, **2001**, 3, 34-9	8.1	199
285	De novo alu-element insertions in FGFR2 identify a distinct pathological basis for Apert syndrome. *American Journal of Human Genetics*, **1999**, 64, 446-61	11	198
284	Further delineation of deletion 1p36 syndrome in 60 patients: a recognizable phenotype and common cause of developmental delay and mental retardation. *Pediatrics*, **2008**, 121, 404-10	7.4	197
283	Autism spectrum disorders and symptoms in children with molecularly confirmed 22q11.2 deletion syndrome. *Journal of Autism and Developmental Disorders*, **2005**, 35, 461-70	4.6	186
282	The neurocognitive phenotype of the 22q11.2 deletion syndrome: selective deficit in visual-spatial memory. *Journal of Clinical and Experimental Neuropsychology*, **2001**, 23, 447-64	2.1	178
281	Microdeletions of chromosomal region 22q11 in patients with congenital conotruncal cardiac defects. *Journal of Medical Genetics*, **1993**, 30, 807-12	5.8	176
280	Volumetric, connective, and morphologic changes in the brains of children with chromosome 22q11.2 deletion syndrome: an integrative study. *NeuroImage*, **2005**, 25, 169-80	7.9	169
279	Apolipoprotein E genotype and neurodevelopmental sequelae of infant cardiac surgery. *Journal of Thoracic and Cardiovascular Surgery*, **2003**, 126, 1736-45	1.5	169
278	Practical guidelines for managing adults with 22q11.2 deletion syndrome. *Genetics in Medicine*, **2015**, 17, 599-609	8.1	154
277	Cognitive decline preceding the onset of psychosis in patients with 22q11.2 deletion syndrome. *JAMA Psychiatry*, **2015**, 72, 377-85	14.5	139
276	Autosomal dominant "Opitz" GBBB syndrome due to a 22q11.2 deletion. *American Journal of Medical Genetics Part A*, **1995**, 59, 103-13		131
275	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
274	Aberrant interchromosomal exchanges are the predominant cause of the 22q11.2 deletion. *Human Molecular Genetics*, **2004**, 13, 417-28	5.6	122
273	Identification of a patient with Bernard-Soulier syndrome and a deletion in the DiGeorge/velo-cardio-facial chromosomal region in 22q11.2. *Human Molecular Genetics*, **1995**, 4, 763-6	5.6	121
272	Mutation analysis of TBX1 in non-deleted patients with features of DGS/VCFS or isolated cardiovascular defects. *Journal of Medical Genetics*, **2001**, 38, E45	5.8	120
271	Five additional Costello syndrome patients with rhabdomyosarcoma: proposal for a tumor screening protocol. *American Journal of Medical Genetics Part A*, **2002**, 108, 80-7		115

270	Polymicrogyria and deletion 22q11.2 syndrome: window to the etiology of a common cortical malformation. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2416-25	2.5	113
269	The changing epidemiologic spectrum of single-suture synostoses. *Plastic and Reconstructive Surgery*, **2008**, 122, 527-533	2.7	111
268	Effects of a functional COMT polymorphism on prefrontal cognitive function in patients with 22q11.2 deletion syndrome. *American Journal of Psychiatry*, **2004**, 161, 1700-2	11.9	110
267	Adults with genetic syndromes and cardiovascular abnormalities: clinical history and management. *Genetics in Medicine*, **2008**, 10, 469-94	8.1	106
266	Dysphagia in children with a 22q11.2 deletion: unusual pattern found on modified barium swallow. *Journal of Pediatrics*, **2000**, 137, 158-64	3.6	105
265	Developmental trajectories in 22q11.2 deletion. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2015**, 169, 172-81	3.1	102
264	T-cell homeostasis in humans with thymic hypoplasia due to chromosome 22q11.2 deletion syndrome. *Blood*, **2004**, 103, 1020-5	2.2	102
263	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
262	Otolaryngologic manifestations of the 22q11.2 deletion syndrome. *JAMA Otolaryngology*, **2002**, 128, 1408-12		97
261	Genotype-phenotype correlation for nucleotide substitutions in the IgII-IgIII linker of FGFR2. *Human Molecular Genetics*, **1997**, 6, 137-43	5.6	93
260	Lack of correlation between impaired T cell production, immunodeficiency, and other phenotypic features in chromosome 22q11.2 deletion syndromes. *Clinical Immunology and Immunopathology*, **1998**, 86, 141-6		92
259	Misalignment of pulmonary veins with alveolar capillary dysplasia: affected siblings and variable phenotypic expression. *Journal of Pediatrics*, **1994**, 124, 125-8	3.6	91
258	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
257	CHARGE (coloboma, heart defect, atresia choanae, retarded growth and development, genital hypoplasia, ear anomalies/deafness) syndrome and chromosome 22q11.2 deletion syndrome: a comparison of immunologic and nonimmunologic phenotypic features. *Pediatrics*, **2009**, 123, e871-7	7.4	86
256	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. *New England Journal of Medicine*, **2017**, 376, 742-754	59.2	83
255	Muenke syndrome (FGFR3-related craniosynostosis): expansion of the phenotype and review of the literature. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 3204-15	2.5	83
254	A second mutation in the type II procollagen gene (COL2AI) causing stickler syndrome (arthro-ophthalmopathy) is also a premature termination codon. *American Journal of Human Genetics*, **1993**, 52, 39-45	11	81
253	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. *Genetics in Medicine*, **2018**, 20, 1175-1185	8.1	80

252	Clinical experience with single-nucleotide polymorphism-based non-invasive prenatal screening for 22q11.2 deletion syndrome. *Ultrasound in Obstetrics and Gynecology*, **2016**, 47, 177-83	5.8	80
251	Clinical spectrum of SIX3-associated mutations in holoprosencephaly: correlation between genotype, phenotype and function. *Journal of Medical Genetics*, **2009**, 46, 389-98	5.8	80
250	Identification of a previously unrecognized microdeletion syndrome of 16q11.2q12.2. *Clinical Genetics*, **2008**, 74, 469-75	4	80
249	Surgical Airway Management in Pierre Robin Sequence: Is There a Role for Tongue-Lip Adhesion?. *Cleft Palate-Craniofacial Journal*, **2003**, 40, 13-18	1.9	79
248	Psychiatric disorders in 22q11.2 deletion syndrome are prevalent but undertreated. *Psychological Medicine*, **2014**, 44, 1267-77	6.9	78
247	Genomic and Genic Deletions of the FOX Gene Cluster on 16q24.1 and Inactivating Mutations of FOXF1 Cause Alveolar Capillary Dysplasia and Other Malformations. *American Journal of Human Genetics*, **2009**, 85, 537	11	78
246	Identification of a genetic cause for isolated unilateral coronal synostosis: a unique mutation in the fibroblast growth factor receptor 3. *Journal of Pediatrics*, **1998**, 132, 714-6	3.6	78
245	Consistent chromosome abnormalities identify novel polymicrogyria loci in 1p36.3, 2p16.1-p23.1, 4q21.21-q22.1, 6q26-q27, and 21q2. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1637-54	2.5	76
244	The relationship of postoperative electrographic seizures to neurodevelopmental outcome at 1 year of age after neonatal and infant cardiac surgery. *Journal of Thoracic and Cardiovascular Surgery* , **2006**, 131, 181-9	1.5	75
243	Communication issues in 22q11.2 deletion syndrome: children at risk. *Genetics in Medicine*, **2001**, 3, 67-71	8.1	74
242	Safety of live viral vaccines in patients with chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *Pediatrics*, **2003**, 112, e325	7.4	71
241	Alterations in midline cortical thickness and gyrification patterns mapped in children with 22q11.2 deletions. *Cerebral Cortex*, **2009**, 19, 115-26	5.1	69
240	Fibroblast growth factor homologous factor 2 (FHF2): gene structure, expression and mapping to the BFLS-like patient. *Human Genetics*, **1999**, 104, 56-63	6.3	69
239	Is cardiac diagnosis a predictor of neurodevelopmental outcome after cardiac surgery in infancy?. *Journal of Thoracic and Cardiovascular Surgery*, **2010**, 140, 1230-7	1.5	68
238	22q11.2 deletion syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 879-888	2.5	67
237	Skeletal anomalies and deformities in patients with deletions of 22q11. *American Journal of Medical Genetics Part A*, **1997**, 72, 210-5		67
236	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
235	Cognitive and behavior profile of preschool children with chromosome 22q11.2 deletion. *American Journal of Medical Genetics Part A*, **1999**, 85, 127-33		67

234	Neurocognitive development in 22q11.2 deletion syndrome: comparison with youth having developmental delay and medical comorbidities. *Molecular Psychiatry*, **2014**, 19, 1205-11	15.1	66
233	Maladaptive conflict monitoring as evidence for executive dysfunction in children with chromosome 22q11.2 deletion syndrome. *Developmental Science*, **2005**, 8, 36-43	4.5	66
232	Growth hormone deficiency in patients with 22q11.2 deletion: expanding the phenotype. *Pediatrics*, **1998**, 101, 929-32	7.4	66
231	Cerebellar atrophy in a patient with velocardiofacial syndrome. *Journal of Medical Genetics*, **1995**, 32, 561-3	5.8	66
230	Thrombocytopenia in patients with chromosome 22q11.2 deletion syndrome. *Journal of Pediatrics*, **2003**, 143, 277-8	3.6	64
229	A multilevel analysis of cognitive dysfunction and psychopathology associated with chromosome 22q11.2 deletion syndrome in children. *Development and Psychopathology*, **2005**, 17, 753-84	4.3	64
228	Large-scale mapping of cortical alterations in 22q11.2 deletion syndrome: Convergence with idiopathic psychosis and effects of deletion size. *Molecular Psychiatry*, **2020**, 25, 1822-1834	15.1	64
227	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
226	Increased prevalence of immunoglobulin A deficiency in patients with the chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *Vaccine Journal*, **1998**, 5, 415-7		63
225	Immune abnormalities are a frequent manifestation of Kabuki syndrome. *American Journal of Medical Genetics, Part A*, **2005**, 135, 278-81	2.5	61
224	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
223	Overlapping numerical cognition impairments in children with chromosome 22q11.2 deletion or Turner syndromes. *Neuropsychologia*, **2008**, 46, 82-94	3.2	60
222	Identification of familial and de novo microduplications of 22q11.21-q11.23 distal to the 22q11.21 microdeletion syndrome region. *Human Molecular Genetics*, **2009**, 18, 1377-83	5.6	59
221	Taking advantage of early diagnosis: preschool children with the 22q11.2 deletion. *Genetics in Medicine*, **2001**, 3, 40-4	8.1	59
220	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
219	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
218	Genetic counseling for the 22q11.2 deletion. *Developmental Disabilities Research Reviews*, **2008**, 14, 69-74		57
217	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

216	Aortic root dilation in patients with 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 939-42	2.5	54
215	Autosomal dominant inheritance of infantile myofibromatosis. *American Journal of Medical Genetics Part A*, **2004**, 126A, 261-6		54
214	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
213	Aberrant Cortical Morphometry in the 22q11.2 Deletion Syndrome. *Biological Psychiatry*, **2015**, 78, 135-43	7.9	53
212	Tracheal anomalies in Pfeiffer syndrome. *JAMA Otolaryngology*, **2004**, 130, 1298-302		52
211	Communication disorders in the 22Q11.2 microdeletion syndrome. *Journal of Communication Disorders*, **2000**, 33, 187-203; quiz 203-4	1.9	52
210	Velopharyngeal anatomy in 22q11.2 deletion syndrome: a three-dimensional cephalometric analysis. *Cleft Palate-Craniofacial Journal*, **2006**, 43, 446-56	1.9	51
209	Enlarged sylvian fissures in infants with interstitial deletion of chromosome 22q11. *American Journal of Medical Genetics Part A*, **1997**, 74, 538-543		50
208	Specific cerebellar reductions in children with chromosome 22q11.2 deletion syndrome. *Neuroscience Letters*, **2006**, 399, 245-8	3.3	49
207	The 22q11.2 deletion in African-American patients: an underdiagnosed population?. *American Journal of Medical Genetics, Part A*, **2005**, 134, 242-6	2.5	49
206	A neurogenetic model for the study of schizophrenia spectrum disorders: the International 22q11.2 Deletion Syndrome Brain Behavior Consortium. *Molecular Psychiatry*, **2017**, 22, 1664-1672	15.1	48
205	Genotype and cardiovascular phenotype correlations with TBX1 in 1,022 velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. *Human Mutation*, **2011**, 32, 1278-89	4.7	48
204	Disruption of the clathrin heavy chain-like gene (CLTCL) associated with features of DGS/VCFS: a balanced (21;22)(p12;q11) translocation. *Human Molecular Genetics*, **1997**, 6, 357-67	5.6	48
203	Mouse and human CRKL is dosage sensitive for cardiac outflow tract formation. *American Journal of Human Genetics*, **2015**, 96, 235-44	11	47
202	The natural history of patients treated for FGFR3-associated (Muenke-type) craniosynostosis. *Plastic and Reconstructive Surgery*, **2008**, 121, 919-931	2.7	46
201	Malignancy in chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *American Journal of Medical Genetics, Part A*, **2006**, 140, 906-9	2.5	46
200	Increased prevalence of unprovoked seizures in patients with a 22q11.2 deletion. *American Journal of Medical Genetics Part A*, **2004**, 129A, 29-34		46
199	Graves' disease in patients with 22q11.2 deletion. *Journal of Pediatrics*, **2001**, 139, 892-5	3.6	46

198	Oculodentodigital dysplasia syndrome associated with abnormal cerebral white matter. *American Journal of Medical Genetics Part A*, **1991**, 41, 18-20		46
197	Molecular genetics of 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2070-2081	2.5	46
196	Radiographic study of the upper cervical spine in the 22q11.2 deletion syndrome. *Journal of Bone and Joint Surgery - Series A*, **2004**, 86, 1751-60	5.6	45
195	Three patients with oculo-auriculo-vertebral spectrum and microdeletion 22q11.2. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2860-4	2.5	44
194	Cognitive development in VCFS. *Progress in Pediatric Cardiology*, **2002**, 15, 109-117	0.4	44
193	Coloboma and other ophthalmologic anomalies in Kabuki syndrome: distinction from charge association. *American Journal of Medical Genetics Part A*, **2003**, 123A, 249-52		44
192	Corpus callosum morphology and ventricular size in chromosome 22q11.2 deletion syndrome. *Brain Research*, **2007**, 1131, 197-210	3.7	43
191	22q11.2 duplication syndrome: elevated rate of autism spectrum disorder and need for medical screening. *Molecular Autism*, **2016**, 7, 27	6.5	42
190	Allergies in patients with chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome) and patients with chronic granulomatous disease. *Pediatric Allergy and Immunology*, **2005**, 16, 226-30	4.2	42
189	Chromosomal and cardiovascular anomalies associated with congenital laryngeal web. *International Journal of Pediatric Otorhinolaryngology*, **2002**, 66, 23-27	1.7	41
188	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
187	Saethre-Chotzen syndrome with familial translocation at chromosome 7p22. *American Journal of Medical Genetics Part A*, **1993**, 47, 637-9		41
186	Histone Modifier Genes Alter Conotruncal Heart Phenotypes in 22q11.2 Deletion Syndrome. *American Journal of Human Genetics*, **2015**, 97, 869-77	11	40
185	Autoimmune disorders in Kabuki syndrome **2005**, 132A, 260-2		40
184	Genitourinary Malformations in Chromosome 22q11.2 Deletion. *Journal of Urology*, **2002**, 168, 2564-2565	2.5	40
183	More Clinical Overlap between 22q11.2 Deletion Syndrome and CHARGE Syndrome than Often Anticipated. *Molecular Syndromology*, **2013**, 4, 235-45	1.5	39
182	Enhanced maternal origin of the 22q11.2 deletion in velocardiofacial and DiGeorge syndromes. *American Journal of Human Genetics*, **2013**, 92, 439-47	11	39
181	Ocular findings in the chromosome 22q11.2 deletion syndrome. *Journal of AAPOS*, **2007**, 11, 179-82	1.3	39

180	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. *Molecular Psychiatry*, **2021**, 26, 4496-4510	15.1	39
179	22q11.2 Deletion syndrome and obstructive sleep apnea. *International Journal of Pediatric Otorhinolaryngology*, **2014**, 78, 1360-4	1.7	38
178	A 35-year experience with syndromic cleft palate repair: operative outcomes and long-term speech function. *Annals of Plastic Surgery*, **2014**, 73 Suppl 2, S130-5	1.7	37
177	Association of airway abnormalities with 22q11.2 deletion syndrome. *International Journal of Pediatric Otorhinolaryngology*, **2017**, 96, 11-14	1.7	36
176	Subthreshold psychotic symptoms in 22q11.2 deletion syndrome. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2014**, 53, 991-1000.e2	7.2	36
175	Altered white matter microstructure in 22q11.2 deletion syndrome: a multisite diffusion tensor imaging study. *Molecular Psychiatry*, **2020**, 25, 2818-2831	15.1	36
174	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. *Nature Medicine*, **2020**, 26, 1912-1918	50.5	35
173	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
172	Affective disorders and other psychiatric diagnoses in children and adolescents with 22q11.2 Deletion Syndrome. *Journal of Affective Disorders*, **2009**, 119, 177-80	6.6	35
171	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
170	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
169	The Psychosis Spectrum in 22q11.2 Deletion Syndrome Is Comparable to That of Nondeleted Youths. *Biological Psychiatry*, **2017**, 82, 17-25	7.9	33
168	Effects of COMT genotype on behavioral symptomatology in the 22q11.2 Deletion Syndrome. *Child Neuropsychology*, **2005**, 11, 109-17	2.7	33
167	Subthreshold Psychosis in 22q11.2 Deletion Syndrome: Multisite Naturalistic Study. *Schizophrenia Bulletin*, **2017**, 43, 1079-1089	1.3	32
166	CD4(+) CD25(+) T-cell production in healthy humans and in patients with thymic hypoplasia. *Vaccine Journal*, **2002**, 9, 1129-31		32
165	Neurocognitive profile in psychotic versus nonpsychotic individuals with 22q11.2 deletion syndrome. *European Neuropsychopharmacology*, **2016**, 26, 1610-8	1.2	31
164	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. *Human Genetics*, **2016**, 135, 273-85	6.3	31
163	Computerized neurocognitive profile in young people with 22q11.2 deletion syndrome compared to youths with schizophrenia and at-risk for psychosis. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2012**, 159B, 87-93	3.5	31

162	The natural history of patients treated for TWIST1-confirmed Saethre-Chotzen syndrome. *Plastic and Reconstructive Surgery*, **2009**, 124, 2085-2095	2.7	31
161	Craniosynostosis: another feature of the 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2005**, 136A, 358-62	2.5	31
160	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
159	Primary amenorrhea and absent uterus in the 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2016-8	2.5	30
158	Regional brain abnormalities in 22q11.2 deletion syndrome: association with cognitive abilities and behavioral symptoms. *Neurocase*, **2004**, 10, 198-206	0.8	30
157	Toriello-Carey syndrome: delineation and review. *American Journal of Medical Genetics Part A*, **2003**, 123A, 84-90		30
156	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
155	Syndrome-specific growth charts for 22q11.2 deletion syndrome in Caucasian children. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2665-71	2.5	29
154	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
153	Metopic craniosynostosis due to mutations in GLI3: A novel association. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1654-60	2.5	29
152	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
151	Sclerocornea associated with the chromosome 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 904-9	2.5	28
150	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
149	Bilateral microtia and cleft palate in cousins with Diamond-Blackfan anemia. *American Journal of Medical Genetics Part A*, **2001**, 101, 268-74		27
148	Elucidating the diagnostic odyssey of 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 936-944	2.5	26
147	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
146	Asymmetric crying facies in the 22q11.2 deletion syndrome: implications for future screening. *Clinical Pediatrics*, **2013**, 52, 1144-8	1.2	26
145	Ocular phenotype correlations in patients with TWIST versus FGFR3 genetic mutations. *Journal of AAPOS*, **2006**, 10, 435-44	1.3	26

144	Identification of 22q11.2 Deletion Syndrome via Newborn Screening for Severe Combined Immunodeficiency. *Journal of Clinical Immunology*, **2017**, 37, 476-485	5.7	25
143	Mapping Subcortical Brain Alterations in 22q11.2 Deletion Syndrome: Effects of Deletion Size and Convergence With Idiopathic Neuropsychiatric Illness. *American Journal of Psychiatry*, **2020**, 177, 589-600	11.9	24
142	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
141	Tracheal cartilaginous sleeves in children with syndromic craniosynostosis. *Genetics in Medicine*, **2017**, 19, 62-68	8.1	24
140	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
139	PEMapper and PECaller provide a simplified approach to whole-genome sequencing. *Proceedings of the National Academy of Sciences of the United States of America*, **2017**, 114, E1923-E1932	11.5	23
138	Detection of a 22q11.2 deletion in cardiac patients suggests a risk for velopharyngeal incompetence. *Pediatrics*, **1997**, 99, E9	7.4	23
137	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
136	A de novo 8.8-Mb deletion of 21q21.1-q21.3 in an autistic male with a complex rearrangement involving chromosomes 6, 10, and 21. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 196-202	2.5	22
135	Rates of autism and potential risk factors in children with congenital heart defects. *Congenital Heart Disease*, **2017**, 12, 421-429	3.1	21
134	The 22q11 low copy repeats are characterized by unprecedented size and structural variability. *Genome Research*, **2019**, 29, 1389-1401	9.7	21
133	Incidental radiologic findings in the 22q11.2 deletion syndrome. *American Journal of Neuroradiology* , **2014**, 35, 2186-91	4.4	21
132	Evaluation of potential modifiers of the cardiac phenotype in the 22q11.2 deletion syndrome. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2009**, 85, 125-9		21
131	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
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, 700-5		21
129	Speech-Language Disorders in 22q11.2 Deletion Syndrome: Best Practices for Diagnosis and Management. *American Journal of Speech-Language Pathology*, **2019**, 28, 984-999	3.1	21
128	A Genetics-First Approach to Dissecting the Heterogeneity of Autism: Phenotypic Comparison of Autism Risk Copy Number Variants. *American Journal of Psychiatry*, **2021**, 178, 77-86	11.9	21
127	The benefits and limitations of cell-free DNA screening for 22q11.2 deletion syndrome. *Prenatal Diagnosis*, **2017**, 37, 53-60	3.2	20

126	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
125	Emergent, remitted and persistent psychosis-spectrum symptoms in 22q11.2 deletion syndrome. *Translational Psychiatry*, **2017**, 7, e1180	8.6	19
124	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
123	Ocular abnormalities in Apert syndrome: genotype/phenotype correlations with fibroblast growth factor receptor type 2 mutations. *Journal of AAPOS*, **2006**, 10, 521-7	1.3	19
122	Molecular cytogenetic characterization of multiple intrachromosomal rearrangements of chromosome 2q in a patient with Waardenburg© syndrome and other congenital defects. *Clinical Genetics*, **2004**, 66, 46-52	4	19
121	Independent de novo 22q11.2 deletions in first cousins with DiGeorge/velocardiofacial syndrome. *American Journal of Medical Genetics Part A*, **2004**, 124A, 313-7		19
120	Developmental presentation of 22q11.2 deletion (DiGeorge/velocardiofacial syndrome). *Journal of Developmental and Behavioral Pediatrics*, **1998**, 19, 342-5	2.4	19
119	Critical region within 22q11.2 linked to higher rate of autism spectrum disorder. *Molecular Autism*, **2017**, 8, 58	6.5	18
118	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
117	CHARGE-like presentation, craniosynostosis and mild Mowat-Wilson Syndrome diagnosed by recognition of the distinctive facial gestalt in a cohort of 28 new cases. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2557-66	2.5	18
116	Contribution of congenital heart disease to neuropsychiatric outcome in school-age children with 22q11.2 deletion syndrome. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2014**, 165B, 137-47	3.5	18
115	Evaluation of potential modifiers of the palatal phenotype in the 22q11.2 deletion syndrome. *Cleft Palate-Craniofacial Journal*, **2006**, 43, 435-41	1.9	18
114	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
113	Impact of psychiatric comorbidity and cognitive deficit on function in 22q11.2 deletion syndrome. *Journal of Clinical Psychiatry*, **2015**, 76, e1262-70	4.6	18
112	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
111	Congenital diaphragmatic hernia in 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 135-142	2.5	17
110	Parathyroid hormone reserve in 22q11.2 deletion syndrome. *Genetics in Medicine*, **2008**, 10, 224-8	8.1	17
109	Perioperative risk factors in patients with 22q11.2 deletion syndrome requiring surgery for velopharyngeal dysfunction. *Cleft Palate-Craniofacial Journal*, **2015**, 52, 183-91	1.9	16

108	Diagnosis of 22q11.2 deletion syndrome and artemis deficiency in two children with T-B-NK+ immunodeficiency. *Journal of Clinical Immunology*, **2012**, 32, 1141-4	5.7	16
107	Expanding the spectrum of microdeletion 4q21 syndrome: a partial phenotype with incomplete deletion of the minimal critical region and a new association with cleft palate and Pierre Robin sequence. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2327-33	2.5	16
106	Nasal dimple as part of the 22q11.2 deletion syndrome **1997**, 69, 290-292		16
105	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
104	A human case of SLC35A3-related skeletal dysplasia. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2758-2762	2.5	15
103	Congenital heart defects in oculodentodigital dysplasia: Report of two cases. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3150-4	2.5	15
102	What© in a name? The 22q11.2 deletion **1997**, 72, 247-247		15
101	Autosomal dominant "Opitz" GBBB syndrome due to a 22q11.2 deletion. *American Journal of Medical Genetics Part A*, **1996**, 64, 525-6		15
100	Interstitial deletion of 4(q21q25) in a liveborn male. *American Journal of Medical Genetics Part A*, **1991**, 40, 77-9		15
99	46,XX,15p+ documented as dup (17p) by fluorescence in situ hybridization. *American Journal of Medical Genetics Part A*, **1993**, 46, 95-7		15
98	22q and two: 22q11.2 deletion syndrome and coexisting conditions. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2203-2214	2.5	15
97	Attention Deficit Hyperactivity Disorder Symptoms and Psychosis in 22q11.2 Deletion Syndrome. *Schizophrenia Bulletin*, **2018**, 44, 824-833	1.3	14
96	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
95	White matter microstructural deficits in 22q11.2 deletion syndrome. *Psychiatry Research - Neuroimaging*, **2017**, 268, 35-44	2.9	14
94	Prenatal genetic testing with chromosomal microarray analysis identifies major risk variants for schizophrenia and other later-onset disorders. *American Journal of Psychiatry*, **2013**, 170, 1498	11.9	14
93	Ablepharon-Macrostomia syndrome--extension of the phenotype. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 3060-2	2.5	14
92	A 3.1-Mb microdeletion of 3p21.31 associated with cortical blindness, cleft lip, CNS abnormalities, and developmental delay. *European Journal of Medical Genetics*, **2009**, 52, 265-8	2.6	14
91	Long-term follow-up of ocular findings in children with Stickler© syndrome. *American Journal of Ophthalmology*, **1996**, 122, 727-8	4.9	14

90	Classical Noonan syndrome is not associated with deletions of 22q11. *American Journal of Medical Genetics Part A*, **1995**, 56, 94-6		14
89	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
88	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
87	Craniosynostosis with tracheal sleeve: a patient with Pfeiffer syndrome, tracheal sleeve and additional malformations in whom an FGFR2 mutation was found. *Clinical Dysmorphology*, **2003**, 12, 209	0.9	13
86	Neuropsychiatric expression and catatonia in 22q11.2 deletion syndrome: An overview and case series. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2146-2159	2.5	13
85	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
84	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
83	Cardiac magnetic resonance imaging for accurate diagnosis of aortic arch anomalies in patients with 22q11.2 deletion. *American Journal of Cardiology*, **2005**, 96, 1726-30	3	12
82	PCR assay confirms diagnosis in syndrome with variably expressed phenotype: mutation detection in Stickler syndrome. *Journal of Medical Genetics*, **1996**, 33, 678-81	5.8	12
81	Velopharyngeal Anatomy in 22q11.2 Deletion Syndrome: A Three-Dimensional Cephalometric Analysis. *Cleft Palate-Craniofacial Journal*, **2006**, 43, 446	1.9	12
80	Neurologic challenges in 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2140-2145	2.5	12
79	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
78	A candidate gene approach to identify modifiers of the palatal phenotype in 22q11.2 deletion syndrome patients. *International Journal of Pediatric Otorhinolaryngology*, **2013**, 77, 123-7	1.7	11
77	New Pattern of Sutural Synostosis Associated With TWIST Gene Mutation and Saethre-Chotzen Syndrome: Peace Sign Synostosis. *Journal of Craniofacial Surgery*, **2015**, 26, 1564-7	1.2	11
76	Arthritis associated with deletion of 22q11.2: More common than previously suspected **1997**, 71, 488-488		11
75	Cleft lip and palate repair in Hay-Wells/ankyloblepharon-ectodermal dysplasia-clefting syndrome. *Cleft Palate-Craniofacial Journal*, **2007**, 44, 335-9	1.9	11
74	Scoliosis in association with the 22q11.2 deletion syndrome: an observational study. *Archives of Disease in Childhood*, **2019**, 104, 19-24	2.2	11
73	Orthopaedic manifestations within the 22q11.2 Deletion syndrome: A systematic review. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2104-2120	2.5	11

72	Genitourinary malformations in chromosome 22q11.2 deletion. *Journal of Urology*, **2002**, 168, 2564-5	2.5	11
71	Effect of congenital heart disease on neurodevelopmental outcomes within multiple-gestation births. *Journal of Thoracic and Cardiovascular Surgery*, **2005**, 130, 1511-6	1.5	10
70	Aphallia as part of urorectal septum malformation sequence in an infant of a diabetic mother **1999**, 82, 363-367		10
69	Frontonasal malformation and cloacal exstrophy: a previously unreported association. *American Journal of Medical Genetics Part A*, **1996**, 61, 75-8		10
68	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
67	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
66	Beare-Stevenson syndrome: two new patients, including a novel finding of tracheal cartilaginous sleeve. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 852-7	2.5	9
65	Effect of congenital heart disease on 4-year neurodevelopment within multiple-gestation births. *Journal of Thoracic and Cardiovascular Surgery*, **2017**, 154, 273-281.e2	1.5	8
64	A catalog of hemizygous variation in 127 22q11 deletion patients. *Human Genome Variation*, **2016**, 3, 15065	1.8	8
63	De novo interstitial deletion of the long arm of chromosome 3: 46,XX,del(3)(q25.1q26.1). *Clinical Genetics*, **1993**, 44, 335-7	4	8
62	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, 167-9		8
61	Evaluation of Potential Modifiers of the Palatal Phenotype in the 22q11.2 Deletion Syndrome. *Cleft Palate-Craniofacial Journal*, **2006**, 43, 435	1.9	8
60	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
59	Olfactory deficits and psychosis-spectrum symptoms in 22q11.2 deletion syndrome. *Schizophrenia Research*, **2018**, 202, 113-119	3.6	7
58	Pediatric healthcare costs for patients with 22q11.2 deletion syndrome. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 631-638	2.3	7
57	Disruption of the blood-brain barrier in 22q11.2 deletion syndrome. *Brain*, **2021**, 144, 1351-1360	11.2	7
56	Ocular findings associated with chromosome 22q11.2 duplication. *Journal of AAPOS*, **2016**, 20, 278-80	1.3	7
55	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

54	Nested Inversion Polymorphisms Predispose Chromosome 22q11.2 to Meiotic Rearrangements. *American Journal of Human Genetics*, **2017**, 101, 616-622	11	6
53	Ablepharon macrostomia syndrome: A distinct genetic entity clinically related to the group of FRAS-FREM complex disorders. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3012-7	2.5	6
52	Sibs with cleidocranial dysplasia born to normal parents: germ line mosaicism?. *American Journal of Medical Genetics Part A*, **1997**, 69, 348-51		6
51	Hemifacial microsomia in a newborn with hypoplastic skin lesions, an eyelid skin tag, and microphthalmia: an unusual presentation of Delleman syndrome. *Clinical Dysmorphology*, **1998**, 7, 279-83	0.9	6
50	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
49	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
48	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
47	T-cell lymphopenia in 22q11.2 deletion syndrome: Relationship to cardiac disease. *Journal of Allergy and Clinical Immunology: in Practice*, **2018**, 6, 690-691	5.4	5
46	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
45	Deletion 22q11.2 (Velo-Cardio-Facial Syndrome/Digeorge Syndrome) **2010**, 263-284		5
44	The 22q11 low copy repeats are characterized by unprecedented size and structure variability		5
43	The Clinical Utility of Flexion-extension Cervical Spine MRI in 22q11.2 Deletion Syndrome. *Journal of Pediatric Orthopaedics*, **2019**, 39, e674-e679	2.4	5
42	Association of hypocalcemia with congenital heart disease in 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2099-2103	2.5	5
41	What's in a name? The 22q11.2 deletion. *American Journal of Medical Genetics Part A*, **1997**, 72, 247-9		5
40	Palatal evaluation and treatment in 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1184-1195	2.5	4
39	Defining Risk of Postoperative Obstructive Sleep Apnea in Patients With 22q11.2DS Undergoing Pharyngeal Flap Surgery for Velopharyngeal Dysfunction Using Polysomnographic Evaluation. *Cleft Palate-Craniofacial Journal*, **2020**, 57, 808-818	1.9	4
38	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
37	Late-onset partial complex seizures secondary to cortical dysplasia in a patient with maternal vitamin K deficient embryopathy: comments on the article by Toriello et al. [2013] and first report of the natural history. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2396-8	2.5	4

36	A normative chart for cognitive development in a genetically selected population. *Neuropsychopharmacology*, **2021**,	8.7	4
35	Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. *Human Molecular Genetics*, **2019**, 28, 3724-3733	5.6	4
34	Prioritizing Genetic Contributors to Cortical Alterations in 22q11.2 Deletion Syndrome Using Imaging Transcriptomics. *Cerebral Cortex*, **2021**, 31, 3285-3298	5.1	4
33	Poverty and Risk of Cleft Lip and Palate: An Analysis of United States Birth Data.. *Plastic and Reconstructive Surgery*, **2022**, 149, 169-182	2.7	4
32	Orofacial Manifestations of Stickler Syndrome: An Analysis of Speech Outcome and Facial Growth After Cleft Palate Repair. *Annals of Plastic Surgery*, **2020**, 84, 665-671	1.7	3
31	The role of 22q11.2 deletion syndrome in the relationship between congenital heart disease and scoliosis. *Spine Journal*, **2020**, 20, 956-963	4	3
30	Crouzon with Acanthosis Nigricans and Odontogenic Tumors: A Rare Form of Syndromic Craniosynostosis. *Cleft Palate-Craniofacial Journal*, **2018**, 55, 296-300	1.9	3
29	Disrupted anatomic networks in the 22q11.2 deletion syndrome. *NeuroImage: Clinical*, **2016**, 12, 420-8	5.3	3
28	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
27	A binational study assessing risk and resilience factors in 22q11.2 deletion syndrome. *Journal of Psychiatric Research*, **2021**, 138, 319-325	5.2	3
26	Club foot in association with the 22q11.2 deletion syndrome: An observational study. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2135-2139	2.5	3
25	Association of Mitochondrial Biogenesis With Variable Penetrance of Schizophrenia. *JAMA Psychiatry*, **2021**, 78, 911-921	14.5	3
24	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
23	The 22q11.2 deletion syndrome as a model for idiopathic scoliosis - A hypothesis. *Medical Hypotheses*, **2019**, 127, 57-62	3.8	2
22	Alternative diffusion anisotropy measures for the investigation of white matter alterations in 22q11.2 deletion syndrome **2018**,		2
21	Genitourinary Malformations in Chromosome 22q11.2 Deletion. *Journal of Urology*, **2002**, 2564-2565	2.5	2
20	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
19	Typical 22q11.2 deletion syndrome appears to confer a reduced risk of schwannoma. *Genetics in Medicine*, **2021**, 23, 1779-1782	8.1	2

18	Pathways to understanding psychosis through rare - 22q11.2DS - and common variants. *Current Opinion in Genetics and Development*, **2021**, 68, 35-40	4.9	2
17	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
16	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
15	Relationship between intelligence quotient measures and computerized neurocognitive performance in 22q11.2 deletion syndrome. *Brain and Behavior*, **2021**, 11, e2221	3.4	2
14	22q11.2 deletion - a tiny piece leading to a big picture. *Nature Reviews Disease Primers*, **2020**, 6, 33	51.1	1
13	Aortic Root Dilation in Patients with 22q11.2 Deletion Syndrome Without Intracardiac Anomalies. *Pediatric Cardiology*, **2021**, 42, 1594-1600	2.1	1
12	Enlarged sylvian fissures in infants with interstitial deletion of chromosome 22q11 **1997**, 74, 538		1
11	Cardiac evaluation of patients with 22q11.2 duplication syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 753-758	2.5	1
10	Genetics of Common Congenital Syndromes of the Head and Neck **2014**, 1-22		1
9	Genes To Mental Health (G2MH): A Framework to Map the Combined Effects of Rare and Common Variants on Dimensions of Cognition and Psychopathology.. *American Journal of Psychiatry*, **2022**, 179, 189-203	11.9	1
8	Surgical insights and management in patients with the 22q11.2 deletion syndrome.. *Pediatric Surgery International*, **2022**, 38, 899	2.1	0
7	Muenke syndrome: Medical and surgical comorbidities and long-term management. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1442-1450	2.5	
6	DiGeorge Syndrome: A Serendipitous Discovery **2014**, 229-240		
5	Craniosynostosis with tracheal sleeve: a patient with Pfeiffer syndrome, tracheal sleeve and additional malformations in whom an FGFR2 mutation was found. *Clinical Dysmorphology*, **2003**, 12, 209-209	0.8	
4	DELETION 22q11.2 (VELO-CARDIO-FACIAL SYNDROME/DIGEORGE SYNDROME) **2021**, 291-316		
3	Inter-rater reliability of subthreshold psychotic symptoms in individuals with 22q11.2 deletion syndrome. *Journal of Neurodevelopmental Disorders*, **2021**, 13, 23	4.6	
2	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	
1	Chromosome 22q11 copy number variants and single ventricle CHD.. *Cardiology in the Young*, **2022**, 1-5	1	