Donna M Mcdonald-Mcginn

List of Publications by Year in Descending Order

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	Chromosome 22q11 copy number variants and single ventricle CHD.. *Cardiology in the Young*, **2022**, 1-5	1	
304	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
303	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
302	Surgical insights and management in patients with the 22q11.2 deletion syndrome.. *Pediatric Surgery International*, **2022**, 38, 899	2.1	0
301	DELETION 22q11.2 (VELO-CARDIO-FACIAL SYNDROME/DIGEORGE SYNDROME) **2021**, 291-316		
300	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. *Molecular Psychiatry*, **2021**, 26, 4496-4510	15.1	39
299	A normative chart for cognitive development in a genetically selected population. *Neuropsychopharmacology*, **2021**,	8.7	4
298	Disruption of the blood-brain barrier in 22q11.2 deletion syndrome. *Brain*, **2021**, 144, 1351-1360	11.2	7
297	Typical 22q11.2 deletion syndrome appears to confer a reduced risk of schwannoma. *Genetics in Medicine*, **2021**, 23, 1779-1782	8.1	2
296	A binational study assessing risk and resilience factors in 22q11.2 deletion syndrome. *Journal of Psychiatric Research*, **2021**, 138, 319-325	5.2	3
295	Aortic Root Dilation in Patients with 22q11.2 Deletion Syndrome Without Intracardiac Anomalies. *Pediatric Cardiology*, **2021**, 42, 1594-1600	2.1	1
294	Pathways to understanding psychosis through rare - 22q11.2DS - and common variants. *Current Opinion in Genetics and Development*, **2021**, 68, 35-40	4.9	2
293	Inter-rater reliability of subthreshold psychotic symptoms in individuals with 22q11.2 deletion syndrome. *Journal of Neurodevelopmental Disorders*, **2021**, 13, 23	4.6	
292	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	
291	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
290	Prioritizing Genetic Contributors to Cortical Alterations in 22q11.2 Deletion Syndrome Using Imaging Transcriptomics. *Cerebral Cortex*, **2021**, 31, 3285-3298	5.1	4
289	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

288	Relationship between intelligence quotient measures and computerized neurocognitive performance in 22q11.2 deletion syndrome. *Brain and Behavior*, **2021**, 11, e2221	3.4	2
287	Association of Mitochondrial Biogenesis With Variable Penetrance of Schizophrenia. *JAMA Psychiatry*, **2021**, 78, 911-921	14.5	3
286	Cardiac evaluation of patients with 22q11.2 duplication syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 753-758	2.5	1
285	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
284	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
283	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
282	The role of 22q11.2 deletion syndrome in the relationship between congenital heart disease and scoliosis. *Spine Journal*, **2020**, 20, 956-963	4	3
281	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
280	22q11.2 deletion - a tiny piece leading to a big picture. *Nature Reviews Disease Primers*, **2020**, 6, 33	51.1	1
279	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
278	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
277	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
276	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
275	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
274	Altered white matter microstructure in 22q11.2 deletion syndrome: a multisite diffusion tensor imaging study. *Molecular Psychiatry*, **2020**, 25, 2818-2831	15.1	36
273	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
272	The 22q11 low copy repeats are characterized by unprecedented size and structural variability. *Genome Research*, **2019**, 29, 1389-1401	9.7	21
271	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	2.8	2

270	Muenke syndrome: Medical and surgical comorbidities and long-term management. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1442-1450	2.5	
269	The 22q11.2 deletion syndrome as a model for idiopathic scoliosis - A hypothesis. *Medical Hypotheses*, **2019**, 127, 57-62	3.8	2
268	Palatal evaluation and treatment in 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1184-1195	2.5	4
267	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
266	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
265	Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. *Human Molecular Genetics*, **2019**, 28, 3724-3733	5.6	4
264	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
263	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
262	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
261	Scoliosis in association with the 22q11.2 deletion syndrome: an observational study. *Archives of Disease in Childhood*, **2019**, 104, 19-24	2.2	11
260	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. *Genetics in Medicine*, **2018**, 20, 1175-1185	8.1	80
259	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
258	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
257	Elucidating the diagnostic odyssey of 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 936-944	2.5	26
256	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
255	Attention Deficit Hyperactivity Disorder Symptoms and Psychosis in 22q11.2 Deletion Syndrome. *Schizophrenia Bulletin*, **2018**, 44, 824-833	1.3	14
254	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
253	Crouzon with Acanthosis Nigricans and Odontogenic Tumors: A Rare Form of Syndromic Craniosynostosis. *Cleft Palate-Craniofacial Journal*, **2018**, 55, 296-300	1.9	3

252	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
251	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
250	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
249	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
248	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
247	Olfactory deficits and psychosis-spectrum symptoms in 22q11.2 deletion syndrome. *Schizophrenia Research*, **2018**, 202, 113-119	3.6	7
246	Alternative diffusion anisotropy measures for the investigation of white matter alterations in 22q11.2 deletion syndrome **2018**,		2
245	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
244	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
243	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
242	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
241	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
240	22q and two: 22q11.2 deletion syndrome and coexisting conditions. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2203-2214	2.5	15
239	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
238	Neurologic challenges in 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2140-2145	2.5	12
237	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
236	Molecular genetics of 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2070-2081	2.5	46
235	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

234	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
233	The benefits and limitations of cell-free DNA screening for 22q11.2 deletion syndrome. *Prenatal Diagnosis*, **2017**, 37, 53-60	3.2	20
232	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
231	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. *New England Journal of Medicine*, **2017**, 376, 742-754	59.2	83
230	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
229	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
228	Association of airway abnormalities with 22q11.2 deletion syndrome. *International Journal of Pediatric Otorhinolaryngology*, **2017**, 96, 11-14	1.7	36
227	Subthreshold Psychosis in 22q11.2 Deletion Syndrome: Multisite Naturalistic Study. *Schizophrenia Bulletin*, **2017**, 43, 1079-1089	1.3	32
226	22q11.2 deletion syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 879-888	2.5	67
225	Rates of autism and potential risk factors in children with congenital heart defects. *Congenital Heart Disease*, **2017**, 12, 421-429	3.1	21
224	Nested Inversion Polymorphisms Predispose Chromosome 22q11.2 to Meiotic Rearrangements. *American Journal of Human Genetics*, **2017**, 101, 616-622	11	6
223	Critical region within 22q11.2 linked to higher rate of autism spectrum disorder. *Molecular Autism*, **2017**, 8, 58	6.5	18
222	White matter microstructural deficits in 22q11.2 deletion syndrome. *Psychiatry Research - Neuroimaging*, **2017**, 268, 35-44	2.9	14
221	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
220	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
219	Emergent, remitted and persistent psychosis-spectrum symptoms in 22q11.2 deletion syndrome. *Translational Psychiatry*, **2017**, 7, e1180	8.6	19
218	A human case of SLC35A3-related skeletal dysplasia. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2758-2762	2.5	15
217	Pediatric healthcare costs for patients with 22q11.2 deletion syndrome. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 631-638	2.3	7

216	Identification of 22q11.2 Deletion Syndrome via Newborn Screening for Severe Combined Immunodeficiency. *Journal of Clinical Immunology*, **2017**, 37, 476-485	5.7	25
215	The Psychosis Spectrum in 22q11.2 Deletion Syndrome Is Comparable to That of Nondeleted Youths. *Biological Psychiatry*, **2017**, 82, 17-25	7.9	33
214	Tracheal cartilaginous sleeves in children with syndromic craniosynostosis. *Genetics in Medicine*, **2017**, 19, 62-68	8.1	24
213	Congenital diaphragmatic hernia in 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 135-142	2.5	17
212	Neurocognitive profile in psychotic versus nonpsychotic individuals with 22q11.2 deletion syndrome. *European Neuropsychopharmacology*, **2016**, 26, 1610-8	1.2	31
211	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
210	Disrupted anatomic networks in the 22q11.2 deletion syndrome. *NeuroImage: Clinical*, **2016**, 12, 420-8	5.3	3
209	A catalog of hemizygous variation in 127 22q11 deletion patients. *Human Genome Variation*, **2016**, 3, 15065	1.8	8
208	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
207	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
206	22q11.2 duplication syndrome: elevated rate of autism spectrum disorder and need for medical screening. *Molecular Autism*, **2016**, 7, 27	6.5	42
205	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
204	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
203	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. *Human Genetics*, **2016**, 135, 273-85	6.3	31
202	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
201	Ocular findings associated with chromosome 22q11.2 duplication. *Journal of AAPOS*, **2016**, 20, 278-80	1.3	7
200	Practical guidelines for managing adults with 22q11.2 deletion syndrome. *Genetics in Medicine*, **2015**, 17, 599-609	8.1	154
199	Cognitive decline preceding the onset of psychosis in patients with 22q11.2 deletion syndrome. *JAMA Psychiatry*, **2015**, 72, 377-85	14.5	139

198	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
197	Aberrant Cortical Morphometry in the 22q11.2 Deletion Syndrome. *Biological Psychiatry*, **2015**, 78, 135-43	7.9	53
196	Histone Modifier Genes Alter Conotruncal Heart Phenotypes in 22q11.2 Deletion Syndrome. *American Journal of Human Genetics*, **2015**, 97, 869-77	11	40
195	22q11.2 deletion syndrome. *Nature Reviews Disease Primers*, **2015**, 1, 15071	51.1	492
194	Developmental trajectories in 22q11.2 deletion. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2015**, 169, 172-81	3.1	102
193	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
192	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
191	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
190	Mouse and human CRKL is dosage sensitive for cardiac outflow tract formation. *American Journal of Human Genetics*, **2015**, 96, 235-44	11	47
189	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
188	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
187	Incidental radiologic findings in the 22q11.2 deletion syndrome. *American Journal of Neuroradiology* , **2014**, 35, 2186-91	4.4	21
186	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
185	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
184	22q11.2 Deletion syndrome and obstructive sleep apnea. *International Journal of Pediatric Otorhinolaryngology*, **2014**, 78, 1360-4	1.7	38
183	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
182	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
181	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

180	DiGeorge Syndrome: A Serendipitous Discovery **2014**, 229-240		
179	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
178	Psychiatric disorders in 22q11.2 deletion syndrome are prevalent but undertreated. *Psychological Medicine*, **2014**, 44, 1267-77	6.9	78
177	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
176	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
175	Genetics of Common Congenital Syndromes of the Head and Neck **2014**, 1-22		1
174	More Clinical Overlap between 22q11.2 Deletion Syndrome and CHARGE Syndrome than Often Anticipated. *Molecular Syndromology*, **2013**, 4, 235-45	1.5	39
173	Enhanced maternal origin of the 22q11.2 deletion in velocardiofacial and DiGeorge syndromes. *American Journal of Human Genetics*, **2013**, 92, 439-47	11	39
172	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
171	Congenital heart defects in oculodentodigital dysplasia: Report of two cases. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3150-4	2.5	15
170	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
169	Asymmetric crying facies in the 22q11.2 deletion syndrome: implications for future screening. *Clinical Pediatrics*, **2013**, 52, 1144-8	1.2	26
168	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
167	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
166	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
165	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
164	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
163	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

162	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
161	Practical guidelines for managing patients with 22q11.2 deletion syndrome. *Journal of Pediatrics*, **2011**, 159, 332-9.e1	3.6	381
160	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
159	Ablepharon-Macrostomia syndrome--extension of the phenotype. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 3060-2	2.5	14
158	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
157	Chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *Medicine (United States)*, **2011**, 90, 1-18	1.8	296
156	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. *Nature Genetics*, **2010**, 42, 203-9	36.3	461
155	Deletion 22q11.2 (Velo-Cardio-Facial Syndrome/Digeorge Syndrome) **2010**, 263-284		5
154	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
153	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
152	Metopic craniosynostosis due to mutations in GLI3: A novel association. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1654-60	2.5	29
151	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
150	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
149	Alterations in midline cortical thickness and gyrification patterns mapped in children with 22q11.2 deletions. *Cerebral Cortex*, **2009**, 19, 115-26	5.1	69
148	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
147	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
146	Aortic root dilation in patients with 22q11.2 deletion syndrome. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 939-42	2.5	54
145	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

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