

# Practical guidelines for managing adults with 22q11.2 de

Genetics in Medicine

17, 599-609

DOI: [10.1038/gim.2014.175](https://doi.org/10.1038/gim.2014.175)

Citation Report

#	ARTICLE	IF	CITATIONS
1	22q11.2 deletion syndrome. Nature Reviews Disease Primers, 2015, 1, 15071.	18.1	954
2	Whole-Genome Sequencing Suggests Schizophrenia Risk Mechanisms in Humans with 22q11.2 Deletion Syndrome. G3: Genes, Genomes, Genetics, 2015, 5, 2453-2461.	0.8	43
3	Monogenic and chromosomal causes of isolated speech and language impairment. Journal of Medical Genetics, 2015, 52, 719-729.	1.5	17
4	Comparative mapping of the 22q11.2 deletion region and the potential of simple model organisms. Journal of Neurodevelopmental Disorders, 2015, 7, 18.	1.5	90
5	Burden Analysis of Rare Microdeletions Suggests a Strong Impact of Neurodevelopmental Genes in Genetic Generalised Epilepsies. PLoS Genetics, 2015, 11, e1005226.	1.5	91
6	Response to clozapine in a clinically identifiable subtype of schizophrenia. British Journal of Psychiatry, 2015, 206, 484-491.	1.7	61
7	Amniotic Band Syndrome, Perinatal Hospice, and Palliative Care versus Active Management. Case Reports in Obstetrics and Gynecology, 2016, 2016, 1-4.	0.2	2
8	The importance of understanding cognitive trajectories. Current Opinion in Psychiatry, 2016, 29, 133-137.	3.1	27
10	The importance of copy number variation in congenital heart disease. Npj Genomic Medicine, 2016, 1, 16031.	1.7	62
11	Two patients with chromosome 22q11.2 deletion presenting with childhood obesity and hyperphagia. European Journal of Medical Genetics, 2016, 59, 401-403.	0.7	5
12	Cardiac sympathetic activity in 22q11.2 deletion syndrome. International Journal of Cardiology, 2016, 212, 346-351.	0.8	1
13	Cerebral microbleeds: A new presenting feature of chromosome 22q11.2 deletion syndrome. Journal of the Neurological Sciences, 2016, 368, 300-303.	0.3	4
14	Communication of Psychiatric Risk in 22q11.2 Deletion Syndrome: A Pilot Project. Journal of Genetic Counseling, 2016, 25, 6-17.	0.9	9
15	Deletions at 22q11.2 in idiopathic Parkinson's disease: a combined analysis of genome-wide association data. Lancet Neurology, The, 2016, 15, 585-596.	4.9	77
16	Early-onset Parkinson disease leading to diagnosis of 22q11.2 deletion syndrome. Parkinsonism and Related Disorders, 2016, 25, 110-111.	1.1	9
17	Fetal growth and gestational factors as predictors of schizophrenia in 22q11.2 deletion syndrome. Genetics in Medicine, 2016, 18, 350-355.	1.1	29
18	Brain Calcification and Movement Disorders. Current Neurology and Neuroscience Reports, 2017, 17, 2.	2.0	18
19	Association of airway abnormalities with 22q11.2 deletion syndrome. International Journal of Pediatric Otorhinolaryngology, 2017, 96, 11-14.	0.4	49

#	ARTICLE	IF	CITATIONS
20	Hypothyroidism associated with parathyroid disorders. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2017, 31, 161-173.	2.2	8
21	Update on the 22q11.2 deletion syndrome and its relevance to schizophrenia. <i>Current Opinion in Psychiatry</i> , 2017, 30, 191-196.	3.1	77
22	Hyposmia, symptoms of rapid eye movement sleep behavior disorder, and parkinsonian motor signs suggest prodromal neurodegeneration in 22q11 deletion syndrome. <i>NeuroReport</i> , 2017, 28, 677-681.	0.6	10
23	Burden and impact of congenital syndromes and comorbidities among adults with congenital heart disease. <i>International Journal of Cardiology</i> , 2017, 240, 159-164.	0.8	16
24	Incidence of the 22q11.2 deletion in a large cohort of miscarriage samples. <i>Molecular Cytogenetics</i> , 2017, 10, 6.	0.4	27
25	Neuroimaging and clinical features in adults with a 22q11.2 deletion at risk of Parkinson's disease. <i>Brain</i> , 2017, 140, 1371-1383.	3.7	41
26	22q11.2 deletion syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 879-888.	0.7	103
28	Copy Number Variation in Syndromic Forms of Psychiatric Illness: The Emerging Value of Clinical Genetic Testing in Psychiatry. <i>American Journal of Psychiatry</i> , 2017, 174, 1036-1050.	4.0	16
29	Psychosis Beyond the 22q11.2 Deletion: Do Additional Genetic Factors Play a Role?. <i>American Journal of Psychiatry</i> , 2017, 174, 1027-1029.	4.0	6
30	Diagnosis and Management of Noncardiac Complications in Adults With Congenital Heart Disease: A Scientific Statement From the American Heart Association. <i>Circulation</i> , 2017, 136, e348-e392.	1.6	147
31	Could I, should I? Parenting aspirations and personal considerations of five young women with 22q11.2 deletion syndrome. <i>Journal of Intellectual and Developmental Disability</i> , 2017, 42, 364-374.	1.1	5
32	Genomic Disorders in Psychiatry—What Does the Clinician Need to Know?. <i>Current Psychiatry Reports</i> , 2017, 19, 82.	2.1	36
33	Familial disorders of parathyroid glands. <i>Diagnostic Histopathology</i> , 2017, 23, 359-365.	0.2	0
34	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. <i>American Journal of Psychiatry</i> , 2017, 174, 1054-1063.	4.0	77
35	Pediatric healthcare costs for patients with 22q11.2 deletion syndrome. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2017, 5, 631-638.	0.6	12
36	Treatment of Comorbid Bipolar Disorder Improves Disabilities and Neuropsychological Functioning in DiGeorge Syndrome. <i>Journal of Clinical Psychopharmacology</i> , 2017, 37, 736-738.	0.7	3
37	Chromosomal Microarray Testing for Children With Unexplained Neurodevelopmental Disorders. <i>JAMA - Journal of the American Medical Association</i> , 2017, 317, 2545.	3.8	17
38	Del(22q11.2) Syndrome. , 2017, , 761-772.		0

#	ARTICLE	IF	CITATIONS
39	Camptodactyly and the 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 515-518.	0.7	4
40	Obesity in adults with 22q11.2 deletion syndrome. Genetics in Medicine, 2017, 19, 204-208.	1.1	60
41	“You don’t know until you get there” The positive and negative “lived” experience of parenting an adult child with 22q11.2 deletion syndrome.. Health Psychology, 2017, 36, 45-54.	1.3	19
42	Neuropsychiatric aspects of 22q11.2 deletion syndrome: considerations in the prenatal setting. Prenatal Diagnosis, 2017, 37, 61-69.	1.1	13
43	Impact of IQ on the diagnostic yield of chromosomal microarray in a community sample of adults with schizophrenia. Genome Medicine, 2017, 9, 105.	3.6	30
44	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.	0.7	57
45	Elucidating the diagnostic odyssey of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 936-944.	0.7	45
46	Client-centred clinical genetic diagnostics. Advances in Mental Health and Intellectual Disabilities, 2018, 12, 1-10.	0.7	3
47	Non-pharmacological treatment of psychiatric disorders in individuals with 22q11.2 deletion syndrome; a systematic review. American Journal of Medical Genetics, Part A, 2018, 176, 1742-1747.	0.7	12
48	Neurodevelopmental outcome in 22q11.2 deletion syndrome and management. American Journal of Medical Genetics, Part A, 2018, 176, 2160-2166.	0.7	29
49	DiGeorge syndrome. Wiener Klinische Wochenschrift, 2018, 130, 283-287.	1.0	16
50	The clue in the face: An unusual first episode psychosis presentation. Australian and New Zealand Journal of Psychiatry, 2018, 52, 293-294.	1.3	1
51	Higher adaptive functioning and lower rate of psychotic comorbidity in married versus unmarried individuals with 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2365-2374.	0.7	3
52	Recurrent Copy Number Variants Associated with Syndromic Short Stature of Unknown Cause. Hormone Research in Paediatrics, 2018, 89, 13-21.	0.8	29
53	Cardiovascular Diseases of Genetic Etiology and Implications for the Pregnant Woman. Indian Journal of Cardiovascular Disease in Women WINCARS, 2018, 03, 149-154.	0.1	0
54	Childhood Executive Functioning Predicts Young Adult Outcomes in 22q11.2 Deletion Syndrome. Journal of the International Neuropsychological Society, 2018, 24, 905-916.	1.2	11
55	Periventricular nodular heterotopia in 22q11.2 deletion and frontal lobe migration. Annals of Clinical and Translational Neurology, 2018, 5, 1314-1322.	1.7	11
56	Defective Vagal Innervation in Murine Tbx1 Mutant Hearts. Journal of Cardiovascular Development and Disease, 2018, 5, 49.	0.8	5

#	ARTICLE	IF	CITATIONS
57	Psycho-social problems in patients with 22q11.2 deletion syndrome according to subjective evaluation by parents. <i>Family Medicine and Primary Care Review</i> , 2018, 20, 117-123.	0.1	3
58	22q11.2 deletion syndrome: A tiny piece leading to a big picture. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2055-2057.	0.7	15
59	Adult Head and Neck Health Care Needs for Individuals with Complex Chronic Conditions of Childhood. <i>Medical Clinics of North America</i> , 2018, 102, 1055-1061.	1.1	2
60	Hypoparathyroidism due to 22Q11 Deletion Syndrome Presenting as Acute Cardiomyopathy. <i>AACE Clinical Case Reports</i> , 2018, 4, e467-e471.	0.4	0
61	Understanding the pediatric psychiatric phenotype of 22q11.2 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2182-2191.	0.7	51
62	Neuropsychiatric expression and catatonia in 22q11.2 deletion syndrome: An overview and case series. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2146-2159.	0.7	25
63	Accurate diagnosis of spinal muscular atrophy and 22q11.2 deletion syndrome using limited deoxynucleotide triphosphates and high-resolution melting. <i>BMC Genomics</i> , 2018, 19, 485.	1.2	7
64	Typical features of Parkinson disease and diagnostic challenges with microdeletion 22q11.2. <i>Neurology</i> , 2018, 90, e2059-e2067.	1.5	35
65	Education and employment trajectories from childhood to adulthood in individuals with 22q11.2 deletion syndrome. <i>European Child and Adolescent Psychiatry</i> , 2019, 28, 31-42.	2.8	21
66	Neurobiological perspective of 22q11.2 deletion syndrome. <i>Lancet Psychiatry</i> , 2019, 6, 951-960.	3.7	70
67	A rare mosaic 22q11.2 microdeletion identified in a Chinese family with recurrent fetal conotruncal defects. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e847.	0.6	7
68	Glutamatergic function in a genetic high-risk group for psychosis: A proton magnetic resonance spectroscopy study in individuals with 22q11.2 deletion. <i>European Neuropsychopharmacology</i> , 2019, 29, 1333-1342.	0.3	8
69	Adverse effects of antipsychotic medication in patients with 22q11.2 deletion syndrome: A systematic review. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2292-2306.	0.7	12
70	Introducing in AJMG Part A : Genetic Syndromes in Adults. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1413-1414.	0.7	3
71	Lower cortisol levels and attenuated cortisol reactivity to daily-life stressors in adults with 22q11.2 deletion syndrome. <i>Psychoneuroendocrinology</i> , 2019, 106, 85-94.	1.3	18
72	Epilepsy and seizures in young people with 22q11.2 deletion syndrome: Prevalence and links with other neurodevelopmental disorders. <i>Epilepsia</i> , 2019, 60, 818-829.	2.6	37
73	All-cause mortality and survival in adults with 22q11.2 deletion syndrome. <i>Genetics in Medicine</i> , 2019, 21, 2328-2335.	1.1	44
74	Increased mortality and morbidity in patients with chronic hypoparathyroidism: A population-based study. <i>Clinical Endocrinology</i> , 2019, 90, 285-292.	1.2	58

#	ARTICLE	IF	CITATIONS
75	22q11.2 Deletion Syndromeâ€“Associated Parkinson's Disease. <i>Movement Disorders Clinical Practice</i> , 2019, 6, 11-16.	0.8	22
76	Effectiveness and side effects of psychopharmacotherapy in individuals with 22q11.2 deletion syndrome with comorbid psychiatric disorders: a systematic review. <i>European Child and Adolescent Psychiatry</i> , 2020, 29, 1035-1048.	2.8	18
77	A genetic model for multimorbidity in young adults. <i>Genetics in Medicine</i> , 2020, 22, 132-141.	1.1	29
78	Participatory development of a patientâ€“clinician communication tool to enhance healthcare transitions for young people with 22q11.2. <i>Irish Journal of Medical Science</i> , 2020, 189, 761-769.	0.8	5
79	The Feasibility and Outcomes of Genetic Testing for Autism and Neurodevelopmental Disorders on an Inpatient Child and Adolescent Psychiatry Service. <i>Autism Research</i> , 2020, 13, 1450-1464.	2.1	6
80	Impact of Interdisciplinary Team Care for Children With 22q11.2 Deletion Syndrome. <i>Cleft Palate-Craniofacial Journal</i> , 2020, 57, 1362-1369.	0.5	9
81	Cognitive behavioral therapy in 22q11.2 deletion syndrome: A case study of two young adults with an anxiety disorder. <i>Journal of Intellectual Disabilities</i> , 2021, 25, 695-704.	1.0	5
82	Consequences of 22q11.2 Microdeletion on the Genome, Individual and Population Levels. <i>Genes</i> , 2020, 11, 977.	1.0	6
83	22q11.2 microdeletion and increased risk for type 2 diabetes. <i>EClinicalMedicine</i> , 2020, 26, 100528.	3.2	15
84	<scp>Ageâ€“Related</scp> Parkinsonian Signs in Microdeletion 22q11.2. <i>Movement Disorders</i> , 2020, 35, 1239-1245.	2.2	4
85	Personalized medical information card for adults with 22q11.2 deletion syndrome: An initiative to improve communication between patients and healthcare providers. <i>Journal of Applied Research in Intellectual Disabilities</i> , 2020, 33, 1534-1540.	1.3	2
86	High Rates of Genetic Diagnosis in Psychiatric Patients with and without Neurodevelopmental Disorders: Toward Improved Genetic Diagnosis in Psychiatric Populations. <i>Canadian Journal of Psychiatry</i> , 2020, 65, 865-873.	0.9	0
87	Relationship between parent-reported gastrointestinal symptoms, sleep problems, autism spectrum disorder symptoms, and behavior problems in children and adolescents with 22q11.2 deletion syndrome. <i>Research in Developmental Disabilities</i> , 2020, 104, 103698.	1.2	10
88	Access to Multidisciplinary Care for Patients With 22q11.2 Deletion Syndrome. <i>Journal of Craniofacial Surgery</i> , 2020, 31, 428-431.	0.3	3
89	The Genetics and Epigenetics of 22q11.2 Deletion Syndrome. <i>Frontiers in Genetics</i> , 2019, 10, 1365.	1.1	64
90	Inborn errors of thymic stromal cell development and function. <i>Seminars in Immunopathology</i> , 2021, 43, 85-100.	2.8	19
91	Medical and dental characteristics of children with chromosome 22q11.2 deletion syndrome at the Royal Children's Hospital, Melbourne. <i>International Journal of Paediatric Dentistry</i> , 2021, 31, 682-690.	1.0	4
92	22q11.2 Deletion Syndrome. , 2021, , 163-194.		0

#	ARTICLE	IF	CITATIONS
93	Biological Basis of Branchial Arch Diseases. , 2021, , 149-174.		0
95	22q11.2 deletion syndrome: 20 years of experience from two pediatric immunology units and review of clues for diagnosis and disease management. <i>Allergologia Et Immunopathologia</i> , 2021, 49, 95-100.	1.0	5
97	Chromosome 22q11.21 and 11p15.4 microdeletions confirmed by high-throughput sequencing analysis in one patient with asymmetric cry syndrome: Case report and review of the literature. <i>Clinical Case Reports (discontinued)</i> , 2021, 9, e04072.	0.2	1
98	A normative chart for cognitive development in a genetically selected population. <i>Neuropsychopharmacology</i> , 2022, 47, 1379-1386.	2.8	12
99	Population Genomic Screening for Genetic Etiologies of Neurodevelopmental/Psychiatric Disorders Demonstrates Personal Utility and Positive Participant Responses. <i>Journal of Personalized Medicine</i> , 2021, 11, 365.	1.1	6
100	Consensus document on optimal management of patients with common arterial trunk. <i>Cardiology in the Young</i> , 2021, 31, 915-939.	0.4	1
101	Abnormal spirometry in adults with 22q11.2 microdeletion and congenital heart disease. <i>International Journal of Cardiology Congenital Heart Disease</i> , 2021, 3, 100085.	0.2	2
102	What can clinical immunology learn from inborn errors of epigenetic regulators?. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, 1602-1618.	1.5	8
103	Late diagnosed DiGeorge syndrome in a 44-year-old female: a rare cause for recurrent syncope in adulthood—a case report. <i>European Heart Journal - Case Reports</i> , 2021, 5, ytab166.	0.3	2
104	Consensus document on optimal management of patients with common arterial trunk. <i>European Journal of Cardio-thoracic Surgery</i> , 2021, 60, 7-33.	0.6	7
105	Age-Related Improvements in Executive Functions and Focal Attention in 22q11.2 Deletion Syndrome Vary Across Domain and Task. <i>Journal of the International Neuropsychological Society</i> , 2021, , 1-14.	1.2	4
106	Clinical evaluation of patients with a neuropsychiatric risk copy number variant. <i>Current Opinion in Genetics and Development</i> , 2021, 68, 26-34.	1.5	12
107	Novel treatments for autism spectrum disorder based on genomics and systems biology. , 2022, 230, 107939.		19
108	Resilience and quality of life in young adults with a 22q11.2 deletion syndrome: a patient's perspective. <i>European Child and Adolescent Psychiatry</i> , 2022, 31, 1885-1894.	2.8	5
109	Camptodactyly and DiGeorge syndrome: A rare hand anomaly. <i>JPRAS Open</i> , 2021, 28, 126-130.	0.4	0
110	Distinct immune trajectories in patients with chromosome 22q11.2 deletion syndrome and immune-mediated diseases. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 445-450.	1.5	15
111	Sexual knowledge and behaviour in 22q11.2 deletion syndrome, a complex care condition. <i>Journal of Applied Research in Intellectual Disabilities</i> , 2022, 35, 966-975.	1.3	4
112	Numerical and Structural Chromosomal Abnormalities Associated with Immunodeficiency. <i>Cytology and Genetics</i> , 2021, 55, 340-349.	0.2	0

#	ARTICLE	IF	CITATIONS
113	An ethical analysis of divergent clinical approaches to the application of genetic testing for autism and schizophrenia. <i>Human Genetics</i> , 2021, , 1.	1.8	11
114	How does genetic variation modify ND-CNV phenotypes?. <i>Trends in Genetics</i> , 2022, 38, 140-151.	2.9	11
115	Identification of a DNA Methylation Episignature in the 22q11.2 Deletion Syndrome. <i>International Journal of Molecular Sciences</i> , 2021, 22, 8611.	1.8	15
116	Predominance of Psychiatric Manifestations in an Adult Man With a Recent Diagnosis of 22q11.2 Deletion Syndrome. <i>Psychiatric Annals</i> , 2021, 51, 394-396.	0.1	0
117	Medical, welfare, and educational challenges and psychological distress in parents caring for an individual with 22q11.2 deletion syndrome: A cross-sectional survey in Japan. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 37-45.	0.7	7
118	A Rare Etiology of Hypocalcemic Seizures in Adulthood: Clues to Diagnosis from Facial Dysmorphism. <i>Neurology India</i> , 2021, 69, 161.	0.2	2
119	22q11.2 Deletion Syndrome. , 2021, , 154-163.		2
120	Neuroradiographic findings in 22q11.2 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2158-2165.	0.7	23
121	Psychosis and ASD. , 2019, , 51-65.		5
122	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. <i>Molecular Psychiatry</i> , 2021, 26, 4496-4510.	4.1	87
123	Speech-Language Disorders in 22q11.2 Deletion Syndrome: Best Practices for Diagnosis and Management. <i>American Journal of Speech-Language Pathology</i> , 2019, 28, 984-999.	0.9	45
124	Psychosocial Risks and Management for Children and Adolescents With 22q11.2 Deletion Syndrome. <i>Perspectives of the ASHA Special Interest Groups</i> , 2019, 4, 633-640.	0.4	2
125	Parkinson's disease with hypocalcaemia: adult presentation of 22q11.2 deletion syndrome. <i>BMJ Case Reports</i> , 2018, 2018, bcr-2017-223751.	0.2	3
126	Whole-genome sequencing suggests mechanisms for 22q11.2 deletion-associated Parkinson's disease. <i>PLoS ONE</i> , 2017, 12, e0173944.	1.1	17
127	Prenatal screening of DiGeorge (22q11.2 deletion) syndrome by abnormalities of the great arteries among Thai pregnant women. <i>Obstetrics and Gynecology Science</i> , 2020, 63, 330-336.	0.6	3
128	Delayed diagnosis of 22q11 deletion syndrome due to late onset hypocalcemia in a 11-year-old girl with imperforated anus. <i>Annals of Pediatric Endocrinology and Metabolism</i> , 2017, 22, 133.	0.8	7
129	Del(22q11.2) Syndrome. , 2016, , 1-13.		0
130	Hypocalcemia due to 22q11.2 deletion syndrome diagnosed in adulthood. <i>Endocrinology, Diabetes and Metabolism Case Reports</i> , 2018, 2018, .	0.2	7



#	ARTICLE	IF	CITATIONS
131	Chromosome 22q11.2 deletion: world definition criteria, standards for diagnosis and monitoring. ZdorovÉ1e Rebenka, 2018, 13, 106-114.	0.0	0
134	22q11.2 Deletion Syndrome. , 2019, , 241-252.		1
135	Family Members with Congenital Heart Disease and Hypogammaglobulinemia. , 2019, , 689-693.		0
136	Communication and Feeding/Swallowing Disorders in 22q11.2 Deletion Syndrome: A Primer for the Pediatric Speech-Language Pathologist. Perspectives of the ASHA Special Interest Groups, 2019, 4, 624-632.	0.4	0
138	How diseases became "genetic". Ciencia E Saude Coletiva, 2019, 24, 3607-3617.	0.1	1
139	Intellectual and developmental disabilities. , 2020, , 249-259.		1
141	The Treatment of the Dually Diagnosed: Intellectual Disability and Severe Psychopathology. Autism and Child Psychopathology Series, 2020, , 475-504.	0.1	0
142	THE CLINICAL CASE REPORT OF DI GIORGI SYNDROME IN TWINS. Neonatology Surgery and Perinatal Medicine, 2020, 10, 79-84.	0.0	0
144	Medical manifestations and healthcare utilization among adult MyCode participants with neurodevelopmental psychiatric copy number variants. Genetics in Medicine, 2021, , .	1.1	2
145	What Every Internist-Endocrinologist Should Know about Rare Genetic Syndromes in Order to Prevent Needless Diagnostics, Missed Diagnoses and Medical Complications: Five Years of "Internal Medicine for Rare Genetic Syndromes"™. Journal of Clinical Medicine, 2021, 10, 5457.	1.0	7
146	Impact of Equine-Assisted Interventions on Heart Rate Variability in Two Participants with 22q11.2 Deletion Syndrome: A Pilot Study. Children, 2021, 8, 1073.	0.6	1
147	Neurological manifestation of 22q11.2 deletion syndrome. Neurological Sciences, 2022, 43, 1695-1700.	0.9	15
148	Psychopathology in adults with copy number variants. Psychological Medicine, 2023, 53, 3142-3149.	2.7	6
149	The COVID-19 pandemic's impact on worry and medical disruptions reported by individuals with chromosome 22q11.2 copy number variants and their caregivers. Journal of Intellectual Disability Research, 2022, 66, 313-322.	1.2	1
150	Collaboration of perioperative management in an adult patient with 22 q 11.2 deletion syndrome: A case report. Clinical Case Reports (discontinued), 2022, 10, e05489.	0.2	0
151	An overview of the trajectory of Brazilian individuals with 22q11.2 deletion syndrome until diagnosis. Orphanet Journal of Rare Diseases, 2022, 17, 67.	1.2	5
152	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.	4.0	29
153	22q11.2 Deletion Syndrome " A series of patients with midline skull base defects. Otolaryngology Case Reports, 2022, 23, 100429.	0.0	0

#	ARTICLE	IF	CITATIONS
155	An examination of adaptive behavior and functional outcomes in adults with 22q11.2 deletion syndrome: A parental perspective. American Journal of Medical Genetics, Part A, 2022, 188, 1040-1047.	0.7	1
156	Isolated idiopathic hypoparathyroidism that developed in adulthood: a case report. SeĀenovskij Vestnik, 2021, 12, 68-75.	0.3	0
157	Designing rare disease care pathways in the Republic of Ireland: a co-operative model. Orphanet Journal of Rare Diseases, 2022, 17, 162.	1.2	4
159	Late maternal diagnosis of DiGeorge syndrome with congenital hypoparathyroidism following antenatal detection of the same 22q11.2 microdeletion syndrome in the fetus. BMJ Case Reports, 2022, 15, e250350.	0.2	0
160	22q11.2 Deletion Syndrome: Impact of Genetics in the Treatment of Conotruncal Heart Defects. Children, 2022, 9, 772.	0.6	7
161	Gastrointestinal Features of 22q11.2 Deletion Syndrome Include Chronic Motility Problems From Childhood to Adulthood. Journal of Pediatric Gastroenterology and Nutrition, 2022, 75, e8-e14.	0.9	3
162	Infectious Complications of DiGeorge Syndrome in the Setting of Malignancy. Cureus, 2022, , .	0.2	0
163	Evaluation of rotavirus vaccine administration among a 22q11.2DS patient population. Allergy, Asthma and Clinical Immunology, 2022, 18, .	0.9	0
164	Psychological Outcomes and Interventions for Individuals With Congenital Heart Disease: A Scientific Statement From the American Heart Association. Circulation: Cardiovascular Quality and Outcomes, 2022, 15, .	0.9	27
165	22q11.2 deletion syndrome: Setting the stage. , 2022, , 2-32.		2
166	The immune system in 22q11.2 deletion syndrome. , 2022, , 102-120.		0
167	General management principles for 22q11.2 deletion syndrome. , 2022, , 420-457.		0
168	Deletion Syndrome 22q11.2: A Systematic Review. Children, 2022, 9, 1168.	0.6	14
169	Congenital heart disease and cardiovascular abnormalities associated with 22q11.2 deletion syndrome. , 2022, , 78-100.		0
170	Reproduction, prenatal screening, and diagnosis in 22q11.2 deletion syndrome. , 2022, , 370-386.		0
171	Gastroenterological manifestations associated with 22q11.2 deletion syndrome. , 2022, , 182-209.		0
172	Neurodevelopmental outcome, developmental trajectories, and management in 22q11.2 deletion syndrome. , 2022, , 270-284.		0
173	Psychiatric profile in children and youth with 22q11.2 deletion syndrome. , 2022, , 302-321.		0

#	ARTICLE	IF	CITATIONS
174	Mental health in adults with 22q11.2 deletion syndrome. , 2022, , 322-337.		0
175	Healthcare transitions for adolescents and adults with 22q11.2 deletion syndrome. , 2022, , 350-369.		0
176	The Unique Experience of a New Multidisciplinary Program for 22q Deletion and Duplication Syndromes in a Community Hospital in Florida: A Reaffirmation That Multidisciplinary Care Is Essential for Best Outcomes in These Patients. <i>Genes</i> , 2022, 13, 1949.	1.0	2
177	Cardiac function in adolescents and young adults with 22q11.2 deletion syndrome without congenital heart disease. <i>European Journal of Medical Genetics</i> , 2023, 66, 104651.	0.7	0
178	Hypoparathyroidism and late-onset hypogonadism in an adult male with familial 22q11.2 deletion syndrome: a case report with 3-year follow-up and review of the literature. <i>BMC Endocrine Disorders</i> , 2022, 22, .	0.9	1
179	Reproductive Outcomes in Adults with 22q11.2 Deletion Syndrome. <i>Genes</i> , 2022, 13, 2126.	1.0	5
180	Clinical management of psychosis in 22q11.2 deletion syndrome. <i>Journal of Psychiatry and Neuroscience</i> , 2022, 47, E391-E392.	1.4	2
181	Adult Height, 22q11.2 Deletion Extent, and Short Stature in 22q11.2 Deletion Syndrome. <i>Genes</i> , 2022, 13, 2038.	1.0	1
182	Clinical genetics of schizophrenia and related neuropsychiatric disorders. <i>Psychiatry Research</i> , 2023, 319, 114992.	1.7	1
183	Parathyroid Gland. , 2022, , 51-97.		0
184	Increased Prevalence of Rare Copy Number Variants in Treatment-Resistant Psychosis. <i>Schizophrenia Bulletin</i> , 2023, 49, 881-892.	2.3	6
185	Clinical Risk Factors for Aortic Root Dilation in Patients with 22q11.2 Deletion Syndrome: A Longitudinal Single-Center Study. <i>Genes</i> , 2022, 13, 2334.	1.0	1
186	NRG1 knockdown rescues PV interneuron GABAergic maturation deficits and schizophrenia behaviors in fetal growth restriction mice. <i>Cell Death Discovery</i> , 2022, 8, .	2.0	2
187	Prenatal Screening and Diagnostic Considerations for 22q11.2 Microdeletions. <i>Genes</i> , 2023, 14, 160.	1.0	12
188	Clinical Practice Guidelines for the Immunological Management of Chromosome 22q11.2 Deletion Syndrome and Other Defects in Thymic Development. <i>Journal of Clinical Immunology</i> , 2023, 43, 247-270.	2.0	5
189	Sphincter Pharyngoplasty for Velopharyngeal Dysfunction: Impact of 22q11.2 Deletion Syndrome. <i>Laryngoscope</i> , 0, , .	1.1	0
190	Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome. <i>Genetics in Medicine</i> , 2023, 25, 100344.	1.1	16
191	Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome. <i>Genetics in Medicine</i> , 2023, 25, 100338.	1.1	20

#	ARTICLE	IF	CITATIONS
192	Educational challenges for 22q11.2 deletion syndrome in <scp>Japan</scp>: Findings from a mixed methods survey. Journal of Applied Research in Intellectual Disabilities, 2023, 36, 558-570.	1.3	0
193	Relationship between high trait anxiety in 22q11.2 deletion syndrome and the difficulties in medical, welfare, and educational services. , 2023, 2, .		1
194	Chromosome Microarray Analysis for the Investigation of Deletions in Pediatric Movement Disorders: A Systematic Review of the Literature. Movement Disorders Clinical Practice, 2023, 10, 547-557.	0.8	0
195	Positive cfDNA screening results for 22q11.2 deletion syndromeâ€”Clinical and laboratory considerations. Frontiers in Genetics, 0, 14, .	1.1	2
196	Congenital Athymia: Unmet Needs and Practical Guidance. Therapeutics and Clinical Risk Management, 0, Volume 19, 239-254.	0.9	4
198	Digeorge syndrome, schizophrenia, intellectual disability and borderline personality disorder: A case report. Clinical Medicine Insights Psychiatry, 2023, 14, 117955732311685.	0.4	0
205	Maternal Secondary Genomic Findings Detected by Fetal Genetic Testing. , 2023, , 333-355.		0
214	Diagnosis and Classification of Branchial Arch Diseases. , 2023, , 91-115.		0
219	Psychiatric Cases: Psychosis and Learning Disabilities. , 2024, , 493-494.		0