Elaine H Zackai

List of Publications by Year
in descending order

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263 papers	13,363 citations	34105 52 h-index	31849 101 g-index
275 all docs	275 docs citations	275 times ranked	14075 citing authors

#	Article	IF	Citations
1	Effects of copy number variations on brain structure and risk for psychiatric illness: Largeâ€scale studies from the<scp>ENIGMA</scp>working groups on<scp>CNVs</scp>. Human Brain Mapping, 2022, 43, 300-328.	3.6	30
2	A Case of Prenatally Diagnosed Periventricular Nodular Heterotopia in a Surviving Male Patient with FLNA Mutation. Journal of Pediatric Neurology, 2022, 20, 057-059.	0.2	0
3	Altered functional brain dynamics in chromosome 22q11.2 deletion syndrome during facial affect processing. Molecular Psychiatry, 2022, 27, 1158-1166.	7.9	1
4	A novel <scp>MBTPS2</scp> variant associated with <scp>BRESHECK</scp> syndrome impairs <scp>sterolâ€regulated</scp> transcription and the endoplasmic reticulum stress response. American Journal of Medical Genetics, Part A, 2022, 188, 463-472.	1.2	4
5	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	2.4	0
6	Molecular Diagnostic Outcomes from 700 Cases. Journal of Molecular Diagnostics, 2022, 24, 274-286.	2.8	7
7	Consolidation of the clinical and genetic definition of aSOX4-related neurodevelopmental syndrome. Journal of Medical Genetics, 2022, 59, 1058-1068.	3.2	10
8	Molecular Mechanisms Contributing to the Etiology of Congenital Diaphragmatic Hernia: A Review and Novel Cases. Journal of Pediatrics, 2022, 246, 251-265.e2.	1.8	4
9	Further supporting <scp>SMARCC2</scp>â€related neurodevelopmental disorder through exome analysis and reanalysis in two patients. American Journal of Medical Genetics, Part A, 2022, 188, 878-882.	1.2	3
10	Surgical insights and management in patients with the 22q11.2 deletion syndrome. Pediatric Surgery International, 2022, 38, 899-905.	1.4	3
11	The Genomics of Congenital Diaphragmatic Hernia: A 10-Year Retrospective Review. Journal of Pediatrics, 2022, 248, 108-113.e2.	1.8	9
12	Genetics etiologies and genotype phenotype correlations in a cohort of individuals with central conducting lymphatic anomaly. European Journal of Human Genetics, 2022, 30, 1022-1028.	2.8	9
13	Clinical Effectiveness of Telemedicine-Based Pediatric Genetics Care. Pediatrics, 2022, 150, .	2.1	5
14	De novo loss-of-function variants in X-linked MED12 are associated with Hardikar syndrome in females. Genetics in Medicine, 2021, 23, 637-644.	2.4	16
15	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
16	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
17	Understanding the phenotypic spectrum of ASXL â€related disease: Ten cases and a review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 1700-1711.	1.2	16
18	Congenital polyvalvular disease expands the cardiac phenotype of the RASopathies. American Journal of Medical Genetics, Part A, 2021, 185, 1486-1493.	1.2	3

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19	Disruption of the bloodâ€“brain barrier in 22q11.2 deletion syndrome. Brain, 2021, 144, 1351-1360.	7.6	27
20	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. Human Genetics, 2021, 140, 1061-1076.	3.8	4
21	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	2.0	21
22	A binational study assessing risk and resilience factors in 22q11.2 deletion syndrome. Journal of Psychiatric Research, 2021, 138, 319-325.	3.1	5
23	Aortic Root Dilation in Patients with 22q11.2 Deletion Syndrome Without Intracardiac Anomalies. Pediatric Cardiology, 2021, 42, 1594-1600.	1.3	6
24	Nonlethal presentations of CYP26B1 â€related skeletal anomalies and multiple synostoses syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2766-2775.	1.2	3
25	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	2.4	7
26	Expanding the genetic landscape of oralâ€facialâ€digital syndrome with two novel genes. American Journal of Medical Genetics, Part A, 2021, 185, 2409-2416.	1.2	9
27	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.	1.2	1
28	Relationship between intelligence quotient measures and computerized neurocognitive performance in 22q11.2 deletion syndrome. Brain and Behavior, 2021, 11, e2221.	2.2	8
29	Bi-allelic variants in the ER quality-control mannosidase gene EDEM3 cause a congenital disorder of glycosylation. American Journal of Human Genetics, 2021, 108, 1342-1349.	6.2	9
30	Association of Mitochondrial Biogenesis With Variable Penetrance of Schizophrenia. JAMA Psychiatry, 2021, 78, 911.	11.0	25
31	Chromatin Modifications in 22q11.2 Deletion Syndrome. Journal of Clinical Immunology, 2021, 41, 1853-1864.	3.8	10
32	Expanding the phenotypic spectrum of Mendelian connective tissue disorders to include prominent kidney phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 3762-3769.	1.2	0
33	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
34	Hyperinsulinism in an individual with an EP300 variant of Rubinsteinâ€Taybi syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1251-1255.	1.2	2
35	Cardiac evaluation of patients with 22q11.2 duplication syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 753-758.	1.2	7
36	Chromosome 4q28.3q32.3 duplication in a patient with lymphatic malformations, craniosynostosis, and dysmorphic features. Clinical Dysmorphology, 2021, 30, 89-92.	0.3	2

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37	De novo variants in CACNA1E found in patients with intellectual disability, developmental regression and social cognition deficit but no seizures. Molecular Autism, 2021, 12, 69.	4.9	12
38	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	2.4	53
39	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
40	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.	2.4	17
41	Novel variants in CDH2 are associated with a new syndrome including Peters anomaly. Clinical Genetics, 2020, 97, 502-508.	2.0	13
42	Clinical spectrum of individuals with pathogenic N F1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€“phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	2.5	80
43	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.	6.2	42
44	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
45	Clinical variability of TUBB â€associated disorders: Diagnosis through reanalysis. American Journal of Medical Genetics, Part A, 2020, 182, 3035-3039.	1.2	7
46	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.	1.0	4
47	EP300 â€related Rubinsteinâ€“Taybi syndrome: Highlighted rare phenotypic findings and a genotypeâ€“phenotype metaâ€analysis of 74 patients. American Journal of Medical Genetics, Part A, 2020, 182, 2926-2938.	1.2	16
48	Congenital diaphragmatic hernia as a prominent feature of a SPECC1L â€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2919-2925.	1.2	8
49	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
50	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.	3.3	20
51	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.	1.7	10
52	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25
53	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. European Journal of Human Genetics, 2020, 28, 1422-1431.	2.8	25
54	Mapping the Relationship between Dysmorphology and Cognitive, Behavioral, and Developmental Outcomes in Children with Autism Spectrum Disorder. Autism Research, 2020, 13, 1227-1238.	3.8	0

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55	Activating variants in <scp>PDGFRB</scp> result in a spectrum of disorders responsive to imatinib monotherapy. American Journal of Medical Genetics, Part A, 2020, 182, 1576-1591.	1.2	21
56	Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. Human Molecular Genetics, 2020, 29, 1900-1921.	2.9	21
57	Lossâ€ofâ€function of Endothelin receptor type A results in Oroâ€Otoâ€Cardiac syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1104-1116.	1.2	7
58	Tattonâ€Brownâ€Rahman syndrome: Six individuals with novel features. American Journal of Medical Genetics, Part A, 2020, 182, 673-680.	1.2	11
59	Bi-allelic Loss-of-Function Variants in NUP188 Cause a Recognizable Syndrome Characterized by Neurologic, Ocular, and Cardiac Abnormalities. American Journal of Human Genetics, 2020, 106, 623-631.	6.2	18
60	Increased Tâ€cell counts in patients with 22q11.2 deletion syndrome who have anxiety. American Journal of Medical Genetics, Part A, 2020, 182, 1815-1818.	1.2	2
61	Attention deficit hyperactivity disorder symptoms as antecedents of later psychotic outcomes in 22q11.2 deletion syndrome. Schizophrenia Research, 2019, 204, 320-325.	2.0	19
62	Mutations in topoisomerase IIÎ² result in a B cell immunodeficiency. Nature Communications, 2019, 10, 3644.	12.8	37
63	Study of carrier frequency of Warsaw breakage syndrome in the Ashkenazi Jewish population and presentation of two cases. American Journal of Medical Genetics, Part A, 2019, 179, 2144-2151.	1.2	10
64	Phenotype delineation of ZNF462 related syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2075-2082.	1.2	23
65	Missense Mutations in NKAP Cause a Disorder of Transcriptional Regulation Characterized by Marfanoid Habitus and Cognitive Impairment. American Journal of Human Genetics, 2019, 105, 987-995.	6.2	11
66	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. American Journal of Medical Genetics, Part A, 2019, 179, 1543-1546.	1.2	5
67	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
68	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.	1.5	3
69	Muenke syndrome: Medical and surgical comorbidities and longâ€term management. American Journal of Medical Genetics, Part A, 2019, 179, 1442-1450.	1.2	1
70	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. Clinical Epigenetics, 2019, 11, 64.	4.1	71
71	The final demise of Rodriguez lethal acrofacial dysostosis: A case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1063-1068.	1.2	12
72	Hyperinsulinemic hypoglycemia in seven patients with de novo NSD1 mutations. American Journal of Medical Genetics, Part A, 2019, 179, 542-551.	1.2	16

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73	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
74	A Novel Approach to Dysmorphology to Enhance the Phenotypic Classification of Autism Spectrum Disorder in the Study to Explore Early Development. Journal of Autism and Developmental Disorders, 2019, 49, 2184-2202.	2.7	6
75	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
76	Anomalies of the genitourinary tract in children with 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 381-385.	1.2	16
77	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.	1.0	12
78	Phenotypic spectrum associated with SPECC1L pathogenic variants: new families and critical review of the nosology of Teebi, Opitz GBBB, and Baraitser-Winter syndromes. European Journal of Medical Genetics, 2019, 62, 103588.	1.3	24
79	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.	2.9	16
80	De novo variants in Myelin regulatory factor (MYRF) as candidates of a new syndrome of cardiac and urogenital anomalies. American Journal of Medical Genetics, Part A, 2018, 176, 969-972.	1.2	39
81	Congenital heart diseases and cardiovascular abnormalities in 22q11.2 deletion syndrome: From wellâ€established knowledge to new frontiers. American Journal of Medical Genetics, Part A, 2018, 176, 2087-2098.	1.2	57
82	Elucidating the diagnostic odyssey of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 936-944.	1.2	45
83	Early photoreceptor outer segment loss and retinoschisis in Cohen syndrome. Ophthalmic Genetics, 2018, 39, 399-404.	1.2	11
84	Primary lymphedema and other lymphatic anomalies are associated with 22q11.2 deletion syndrome. European Journal of Medical Genetics, 2018, 61, 411-415.	1.3	5
85	Attention Deficit Hyperactivity Disorder Symptoms and Psychosis in 22q11.2 Deletion Syndrome. Schizophrenia Bulletin, 2018, 44, 824-833.	4.3	17
86	Deletion size analysis of 1680 22q11.2DS subjects identifies a new recombination hotspot on chromosome 22q11.2. Human Molecular Genetics, 2018, 27, 1150-1163.	2.9	22
87	Natural history and genotypeâ€phenotype correlations in 72 individuals with SATB2â€associated syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 925-935.	1.2	57
88	Hearing Loss after Cardiac Surgery in Infancy: An Unintended Consequence of Life-Saving Care. Journal of Pediatrics, 2018, 192, 144-151.e1.	1.8	14
89	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.	0.8	15
90	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. American Journal of Human Genetics, 2018, 102, 69-87.	6.2	144

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91	Cover Image, Volume 176A, Number 4, April 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
92	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.	2.4	22
93	Novel findings with reassessment of exome data: implications for validation testing and interpretation of genomic data. Genetics in Medicine, 2018, 20, 329-336.	2.4	28
94	The 22q11.2 deletion syndrome: Cancer predisposition, platelet abnormalities and cytopenias. American Journal of Medical Genetics, Part A, 2018, 176, 2121-2127.	1.2	47
95	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.	0.8	7
96	Cover Image, Volume 176A, Number 10, October 2018. , 2018, 176, i-i.		0
97	Musical auditory processing, cognition, and psychopathology in 22q11.2 deletion syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 765-773.	1.7	5
98	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.	1.2	33
99	22q and two: 22q11.2 deletion syndrome and coexisting conditions. American Journal of Medical Genetics, Part A, 2018, 176, 2203-2214.	1.2	30
100	Neurologic challenges in 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2140-2145.	1.2	17
101	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.	1.2	7
102	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.	1.2	106
103	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	6.2	40
104	Variable Clinical Manifestations of Xiaâ€Gibbs syndrome: Findings of Consecutively Identified Cases at a Single Children's Hospital. American Journal of Medical Genetics, Part A, 2018, 176, 1890-1896.	1.2	20
105	PCDH19 -related epilepsy in a male with Klinefelter syndrome: Additional evidence supporting PCDH19 cellular interference disease mechanism. Epilepsy Research, 2018, 145, 89-92.	1.6	20
106	De novo missense variants in MEIS2 recapitulate the microdeletion phenotype of cardiac and palate abnormalities, developmental delay, intellectual disability and dysmorphic features. American Journal of Medical Genetics, Part A, 2018, 176, 1845-1851.	1.2	21
107	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.	1.2	41
108	Nonreentrant atrial tachycardia occurs independently of hypertrophic cardiomyopathy in RASopathy patients. American Journal of Medical Genetics, Part A, 2018, 176, 1711-1722.	1.2	21

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109	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	6.2	48
110	Olfactory deficits and psychosis-spectrum symptoms in 22q11.2 deletion syndrome. Schizophrenia Research, 2018, 202, 113-119.	2.0	8
111	Cytotoxic T-Lymphocyte-Associated Protein 4 Haploinsufficiency-Associated Inflammation Can Occur Independently of T-Cell Hyperproliferation. Frontiers in Immunology, 2018, 9, 1715.	4.8	13
112	Negative subthreshold psychotic symptoms distinguish 22q11.2 deletion syndrome from other neurodevelopmental disorders: A two-site study. Schizophrenia Research, 2017, 188, 42-49.	2.0	16
113	10â€yearâ€old female with intragenic KANSL1 mutation, no KANSL1â€related intellectual disability, and preserved verbal intelligence. American Journal of Medical Genetics, Part A, 2017, 173, 762-765.	1.2	3
114	Association of airway abnormalities with 22q11.2 deletion syndrome. International Journal of Pediatric Otorhinolaryngology, 2017, 96, 11-14.	1.0	49
115	The dimensional structure of psychopathology in 22q11.2 Deletion Syndrome. Journal of Psychiatric Research, 2017, 92, 124-131.	3.1	13
116	CMIP haploinsufficiency in two patients with autism spectrum disorder and coâ€occurring gastrointestinal issues. American Journal of Medical Genetics, Part A, 2017, 173, 2101-2107.	1.2	6
117	Subthreshold Psychosis in 22q11.2 Deletion Syndrome: Multisite Naturalistic Study. Schizophrenia Bulletin, 2017, 43, 1079-1089.	4.3	47
118	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
119	Rates of autism and potential risk factors in children with congenital heart defects. Congenital Heart Disease, 2017, 12, 421-429.	0.2	42
120	Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the GPR98 Locus on 5q14.3. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	22
121	White matter microstructural deficits in 22q11.2 deletion syndrome. Psychiatry Research - Neuroimaging, 2017, 268, 35-44.	1.8	17
122	Impairment of different protein domains causes variable clinical presentation within Pitt-Hopkins syndrome and suggests intragenic molecular syndromology of TCF4. European Journal of Medical Genetics, 2017, 60, 565-571.	1.3	18
123	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. American Journal of Psychiatry, 2017, 174, 1054-1063.	7.2	77
124	A human case of SLC35A3â€related skeletal dysplasia. American Journal of Medical Genetics, Part A, 2017, 173, 2758-2762.	1.2	20
125	Identification of 22q11.2 Deletion Syndrome via Newborn Screening for Severe Combined Immunodeficiency. Journal of Clinical Immunology, 2017, 37, 476-485.	3.8	35
126	The Psychosis Spectrum in 22q11.2 Deletion Syndrome Is Comparable to That of Nondeleted Youths. Biological Psychiatry, 2017, 82, 17-25.	1.3	45

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127	Phenotypic predictors and final diagnoses in patients referred for RASopathy testing by targeted next-generation sequencing. Genetics in Medicine, 2017, 19, 715-718.	2.4	14
128	Tracheal cartilaginous sleeves in children with syndromic craniosynostosis. Genetics in Medicine, 2017, 19, 62-68.	2.4	27
129	Congenital diaphragmatic hernia in 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 135-142.	1.2	21
130	Critical region within 22q11.2 linked to higher rate of autism spectrum disorder. Molecular Autism, 2017, 8, 58.	4.9	37
131	Gainâ€ofâ€function mutations in SMAD4 cause a distinctive repertoire of cardiovascular phenotypes in patients with Myhre syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2617-2631.	1.2	53
132	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, 125-138.	6.2	92
133	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154.	2.5	45
134	De Novo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap Those Associated with Proximal 1p36 Deletions. American Journal of Human Genetics, 2016, 98, 963-970.	6.2	67
135	Analysis of 589,306 genomes identifies individuals resilient to severe Mendelian childhood diseases. Nature Biotechnology, 2016, 34, 531-538.	17.5	273
136	Increasing cumulative exposure to volatile anesthetic agents is associated with poorer neurodevelopmental outcomes in children with hypoplastic left heart syndrome. Journal of Thoracic and Cardiovascular Surgery, 2016, 152, 482-489.	0.8	55
137	Utility of genetic evaluation in infants with congenital heart defects admitted to the cardiac intensive care unit. American Journal of Medical Genetics, Part A, 2016, 170, 3090-3097.	1.2	26
138	Kabuki syndrome as a cause of nonâ€immune fetal hydrops/ascites. American Journal of Medical Genetics, Part A, 2016, 170, 3333-3337.	1.2	11
139	Characteristic calcaneal ossification: an additional early radiographic finding in infants with fibrodysplasia ossificans progressiva. Pediatric Radiology, 2016, 46, 1568-1572.	2.0	7
140	Cover Image, Volume 170A, Number 5, May 2016. , 2016, 170, i-i.		0
141	Cerebroâ€“costoâ€“mandibular syndrome: Clinical, radiological, and genetic findings. American Journal of Medical Genetics, Part A, 2016, 170, 1115-1126.	1.2	21
142	Neurocognitive profile in psychotic versus nonpsychotic individuals with 22q11.2 deletion syndrome. European Neuropsychopharmacology, 2016, 26, 1610-1618.	0.7	45
143	The Role of mGluR Copy Number Variation in Genetic and Environmental Forms of Syndromic Autism Spectrum Disorder. Scientific Reports, 2016, 6, 19372.	3.3	28
144	Disrupted anatomic networks in the 22q11.2 deletion syndrome. NeuroImage: Clinical, 2016, 12, 420-428.	2.7	4

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145	A catalog of hemizygous variation in 127 22q11 deletion patients. Human Genome Variation, 2016, 3, 15065.	0.7	8
146	IQ and hemizygosity for the Val¹⁵⁸Met functional polymorphism of COMT in 22q11DS. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 1112-1115.	1.7	6
147	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.	1.8	22
148	22q11.2 duplication syndrome: elevated rate of autism spectrum disorder and need for medical screening. Molecular Autism, 2016, 7, 27.	4.9	67
149	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, 1147-1151.e4.	0.8	55
150	B cell development in chromosome 22q11.2 deletion syndrome. Clinical Immunology, 2016, 163, 1-9.	3.2	24
151	Rare copy number variants and congenital heart defects in the 22q11.2 deletion syndrome. Human Genetics, 2016, 135, 273-285.	3.8	43
152	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315.	2.4	69
153	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€“Phenotype Correlation. Human Mutation, 2015, 36, 1052-1063.	2.5	143
154	Limb body wall complex, amniotic band sequence, or new syndrome caused by mutation in IQ Motif containing K (IQCK)?. Molecular Genetics & Genomic Medicine, 2015, 3, 424-432.	1.2	17
155	<scp>EGFR</scp> mutations cause a lethal syndrome of epithelial dysfunction with progeroid features. Molecular Genetics & Genomic Medicine, 2015, 3, 452-458.	1.2	12
156	Expanding the SPECC1L mutation phenotypic spectrum to include Teebi hypertelorism syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2497-2502.	1.2	26
157	22q11.2 deletion syndrome. Nature Reviews Disease Primers, 2015, 1, 15071.	30.5	954
158	Atypical Williams syndrome in an infant with complete atrioventricular canal defect. American Journal of Medical Genetics, Part A, 2015, 167, 3108-3112.	1.2	1
159	Phenotypic modifications of patients with full chromosome aneuploidies and concurrent suspected or confirmed second diagnoses. American Journal of Medical Genetics, Part A, 2015, 167, 2168-2175.	1.2	3
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