Merlin Butler

List of Publications by Year
in descending order

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311 papers	15,314 citations	20817 60 h-index	24982 109 g-index
317 all docs	317 docs citations	317 times ranked	10263 citing authors

#	Article	IF	Citations
1	Prader-Willi Syndrome: Consensus Diagnostic Criteria. Pediatrics, 1993, 91, 398-402.	2.1	1,057
2	Genetic imprinting suggested by maternal heterodisomy in non-deletion Prader-Willi syndrome. Nature, 1989, 342, 281-285.	27.8	852
3	Subset of individuals with autism spectrum disorders and extreme macrocephaly associated with germline PTEN tumour suppressor gene mutations. Journal of Medical Genetics, 2005, 42, 318-321.	3.2	673
4	Praderâ€Willi syndrome: Current understanding of cause and diagnosis. American Journal of Medical Genetics Part A, 1990, 35, 319-332.	2.4	548
5	Prader-Willi syndrome: a review of clinical, genetic, and endocrine findings. Journal of Endocrinological Investigation, 2015, 38, 1249-1263.	3.3	429
6	Clinical and cytogenetic survey of 39 individuals with Prader-Labhart-Willi syndrome. American Journal of Medical Genetics Part A, 1986, 23, 793-809.	2.4	325
7	Nutritional phases in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1040-1049.	1.2	325
8	Prader-Willi syndrome: consensus diagnostic criteria. Pediatrics, 1993, 91, 398-402.	2.1	286
9	Praderâ€“Willi syndrome: clinical genetics, cytogenetics and molecular biology. Expert Reviews in Molecular Medicine, 2005, 7, 1-20.	3.9	276
10	Imprinting-Mutation Mechanisms in Prader-Willi Syndrome. American Journal of Human Genetics, 1999, 64, 397-413.	6.2	262
11	The Frequency of Uniparental Disomy in Prader-Willi Syndrome. New England Journal of Medicine, 1992, 326, 1599-1607.	27.0	257
12	Behavioral Differences Among Subjects With Prader-Willi Syndrome and Type I or Type II Deletion and Maternal Disomy. Pediatrics, 2004, 113, 565-573.	2.1	251
13	Microdeletion/microduplication of proximal 15q11.2 between BP1 and BP2: a susceptibility region for neurological dysfunction including developmental and language delay. Human Genetics, 2011, 130, 517-528.	3.8	219
14	Prader-Willi Syndrome: Obesity due to Genomic Imprinting. Current Genomics, 2011, 12, 204-215.	1.6	211
15	Prader-Willi Syndrome - Clinical Genetics, Diagnosis and Treatment Approaches: An Update. Current Pediatric Reviews, 2019, 15, 207-244.	0.8	203
16	Genomic imprinting disorders in humans: a mini-review. Journal of Assisted Reproduction and Genetics, 2009, 26, 477-486.	2.5	185
17	Intellectual characteristics of Prader-Willi syndrome: comparison of genetic subtypes. Journal of Intellectual Disability Research, 2000, 44, 25-30.	2.0	165
18	An infant with deletion of the distal long arm of chromosome 15 (q26.1â†'qter) and loss of insulinâ€like growth factor 1 receptor gene. American Journal of Medical Genetics Part A, 1991, 38, 74-79.	2.4	160

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19	The 15q11.2 BP1â€“BP2 Microdeletion Syndrome: A Review. International Journal of Molecular Sciences, 2015, 16, 4068-4082.	4.1	160
20	Neural Mechanisms Underlying Hyperphagia in Praderâ€Willi Syndrome. Obesity, 2006, 14, 1028-1037.	3.0	155
21	Specific Genetic Diseases at Risk for Sedation/Anesthesia Complications. Anesthesia and Analgesia, 2000, 91, 837-855.	2.2	144
22	Energy expenditure and physical activity in Praderâ€“Willi syndrome: Comparison with obese subjects. American Journal of Medical Genetics, Part A, 2007, 143A, 449-459.	1.2	138
23	Deficiency in prohormone convertase PC1 impairs prohormone processing in Prader-Willi syndrome. Journal of Clinical Investigation, 2016, 127, 293-305.	8.2	120
24	Causes of death in Prader-Willi syndrome: Prader-Willi Syndrome Association (USA) 40-year mortality survey. Genetics in Medicine, 2017, 19, 635-642.	2.4	117
25	Molecular genetic classification in Prader-Willi syndrome: a multisite cohort study. Journal of Medical Genetics, 2019, 56, 149-153.	3.2	112
26	Expression of 4 Genes Between Chromosome 15 Breakpoints 1 and 2 and Behavioral Outcomes in Prader-Willi Syndrome. Pediatrics, 2006, 118, e1276-e1283.	2.1	103
27	Comparison of X-chromosome inactivation patterns in multiple tissues from human females. Journal of Medical Genetics, 2008, 45, 309-313.	3.2	101
28	Paracentric inversions in humans: A review of 446 paracentric inversions with presentation of 120 new cases. American Journal of Medical Genetics Part A, 1995, 55, 171-187.	2.4	98
29	Umbilical cord blood banking: an update. Journal of Assisted Reproduction and Genetics, 2011, 28, 669-676.	2.5	97
30	Importance of reward and prefrontal circuitry in hunger and satiety: Praderâ€“Willi syndrome vs simple obesity. International Journal of Obesity, 2012, 36, 638-647.	3.4	97
31	Clinical Assessment, Genetics, and Treatment Approaches in Autism Spectrum Disorder (ASD). International Journal of Molecular Sciences, 2020, 21, 4726.	4.1	97
32	Prader-Willi Syndrome. , 2000, 10, 3S-16S.		96
33	Insulin resistance and obesityâ€related factors in Praderâ€“Willi syndrome: Comparison with obese subjects. Clinical Genetics, 2005, 67, 230-239.	2.0	94
34	Chromosomal microarray analysis of consecutive individuals with autism spectrum disorders or learning disability presenting for genetic services. Gene, 2014, 535, 70-78.	2.2	94
35	Gastric Rupture and Necrosis in Praderâ€Willi Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2007, 45, 272-274.	1.8	93
36	Hypopigmentation: a common feature of Prader-Labhart-Willi syndrome. American Journal of Human Genetics, 1989, 45, 140-6.	6.2	93

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37	X chromosome gene expression in human tissues: Male and female comparisons. Genomics, 2006, 88, 675-681.	2.9	92
38	Maladaptive behaviors and risk factors among the genetic subtypes of Prader-Willi syndrome. American Journal of Medical Genetics, Part A, 2005, 136A, 140-145.	1.2	91
39	Parental origin of chromosome 15 deletion in Prader-Willi syndrome. Lancet, The, 1983, 1, 1285-6.	13.7	91
40	Oxytocin treatment in children with Praderâ€“Willi syndrome: A doubleâ€blind, placeboâ€controlled, crossover study. American Journal of Medical Genetics, Part A, 2017, 173, 1243-1250.	1.2	90
41	Effects of <scp>MetAP2</scp> inhibition on hyperphagia and body weight in Praderâ€“Willi syndrome: A randomized, doubleâ€blind, placeboâ€controlled trial. Diabetes, Obesity and Metabolism, 2017, 19, 1751-1761.	4.4	88
42	Clinical and molecular studies in fragile X patients with a Prader-Willi-like phenotype.. Journal of Medical Genetics, 1993, 30, 761-766.	3.2	86
43	Hypopigmentation in the Prader-Willi syndrome correlates withP gene deletion but not with haplotype of the hemizygousP allele. , 1997, 71, 57-62.		85
44	Standards for selected anthropometric measurements in Prader-Willi syndrome. Pediatrics, 1991, 88, 853-60.	2.1	82
45	Clinical and genetic aspects of the 15q11.2 BP1â€“BP2 microdeletion disorder. Journal of Intellectual Disability Research, 2017, 61, 568-579.	2.0	81
46	Whole Exome Sequencing in Females with Autism Implicates Novel and Candidate Genes. International Journal of Molecular Sciences, 2015, 16, 1312-1335.	4.1	77
47	Array comparative genomic hybridization (aCGH) analysis in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 854-860.	1.2	76
48	Appetitive behavior, compulsivity, and neurochemistry in Prader-Willi syndrome. Mental Retardation and Developmental Disabilities Research Reviews, 2000, 6, 125-130.	3.6	75
49	Breakage in the SNRPN locus in a balanced 46,XY,t(15;19) Prader-Willi syndrome patient. Human Molecular Genetics, 1996, 5, 517-524.	2.9	73
50	Plasma cytokine levels in children with autistic disorder and unrelated siblings. International Journal of Developmental Neuroscience, 2012, 30, 121-127.	1.6	72
51	Whole genome microarray analysis of gene expression in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 430-442.	1.2	71
52	Laparoscopic sleeve gastrectomy in children and adolescents with Prader-Willi syndrome: a matched-control study. Surgery for Obesity and Related Diseases, 2016, 12, 100-110.	1.2	71
53	An anthropometric study of 38 individuals with Prader-Labhart-Willi syndrome. American Journal of Medical Genetics Part A, 1987, 26, 445-455.	2.4	70
54	The relationship between compulsive behaviour and academic achievement across the three genetic subtypes of Prader?Willi syndrome. Journal of Intellectual Disability Research, 2007, 51, 478-487.	2.0	69

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55	GeneAnalytics Pathway Analysis and Genetic Overlap among Autism Spectrum Disorder, Bipolar Disorder and Schizophrenia. International Journal of Molecular Sciences, 2017, 18, 527.	4.1	67
56	Decreased bone mineral density in Prader-Willi syndrome: Comparison with obese subjects. American Journal of Medical Genetics Part A, 2001, 103, 216-222.	2.4	65
57	Rare FMR1 gene mutations causing fragile X syndrome: A review. American Journal of Medical Genetics, Part A, 2018, 176, 11-18.	1.2	65
58	Telomerase activity and oncogenesis in giant cell tumor of bone. Cancer, 1995, 75, 1094-1099.	4.1	64
59	Prader-Willi Syndrome: Clinical Genetics and Diagnostic Aspects with Treatment Approaches. Current Pediatric Reviews, 2016, 12, 136-166.	0.8	64
60	Two patients with ring chromosome 15 syndrome. American Journal of Medical Genetics Part A, 1988, 29, 149-154.	2.4	63
61	Refining the 22q11.2 deletion breakpoints in DiGeorge syndrome by aCGH. Cytogenetic and Genome Research, 2009, 124, 113-120.	1.1	63
62	Do some patients with seckel syndrome have hematological problems and/or chromosome breakage?. American Journal of Medical Genetics Part A, 1987, 27, 645-649.	2.4	62
63	Global DNA promoter methylation in frontal cortex of alcoholics and controls. Gene, 2012, 498, 5-12.	2.2	61
64	Elevated plasma oxytocin levels in children with Praderâ€“Willi syndrome compared with healthy unrelated siblings. American Journal of Medical Genetics, Part A, 2016, 170, 594-601.	1.2	58
65	Management of obesity in Praderâ€“Willi syndrome. Nature Clinical Practice Endocrinology and Metabolism, 2006, 2, 592-593.	2.8	57
66	Genetics and Mitochondrial Abnormalities in Autism Spectrum Disorders:A Review. Current Genomics, 2011, 12, 322-332.	1.6	57
67	Single Gene and Syndromic Causes of Obesity: Illustrative Examples. Progress in Molecular Biology and Translational Science, 2016, 140, 1-45.	1.7	57
68	Comparison of leptin protein levels in Prader-Willi syndrome and control individuals. American Journal of Medical Genetics Part A, 1998, 75, 7-12.	2.4	56
69	Deaths due to choking in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 484-487.	1.2	56
70	Growth Charts for Non-Growth Hormone Treated Prader-Willi Syndrome. Pediatrics, 2015, 135, e126-e135.	2.1	56
71	Chromosomal Microarray Analysis of Consecutive Individuals with Autism Spectrum Disorders Using an Ultra-High Resolution Chromosomal Microarray Optimized for Neurodevelopmental Disorders. International Journal of Molecular Sciences, 2016, 17, 2070.	4.1	56
72	Methylation-Specific Multiplex Ligation-Dependent Probe Amplification Analysis of Subjects with Chromosome 15 Abnormalities. Genetic Testing and Molecular Biomarkers, 2007, 11, 467-476.	1.7	55

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73	Risk Factors for Self-Injury, Aggression, and Stereotyped Behavior Among Young Children At Risk for Intellectual and Developmental Disabilities. American Journal on Intellectual and Developmental Disabilities, 2014, 119, 351-370.	1.6	55
74	A clinical case report and literature review of the 3q29 microdeletion syndrome. Clinical Dysmorphology, 2015, 24, 89-94.	0.3	55
75	Microarray analysis of gene/transcript expression in Prader-Willi syndrome: deletion versus UPD. Journal of Medical Genetics, 2003, 40, 568-574.	3.2	54
76	Brief Report: Non-Random X Chromosome Inactivation in Females with Autism. Journal of Autism and Developmental Disorders, 2005, 35, 675-681.	2.7	53
77	Growth Standards of Infants With Prader-Willi Syndrome. Pediatrics, 2011, 127, 687-695.	2.1	53
78	High-Resolution Chromosome Ideogram Representation of Currently Recognized Genes for Autism Spectrum Disorders. International Journal of Molecular Sciences, 2015, 16, 6464-6495.	4.1	51
79	A 15â€item checklist for screening mentally retarded males for the fragile X syndrome. Clinical Genetics, 1991, 39, 347-354.	2.0	50
80	Contributing factors of mortality in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 196-205.	1.2	50
81	Thyroid function studies in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 488-492.	1.2	49
82	Survival trends from the Praderâ€“Willi Syndrome Association (USA) 40-year mortality survey. Genetics in Medicine, 2018, 20, 24-30.	2.4	49
83	Plasma Peptide YY and Ghrelin Levels in Infants and Children with Prader-Willi Syndrome. Journal of Pediatric Endocrinology and Metabolism, 2004, 17, 1177-84.	0.9	46
84	The neuroanatomy of genetic subtype differences in Praderâ€“Willi syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 243-253.	1.7	46
85	Imprinting disorders in humans: a review. Current Opinion in Pediatrics, 2020, 32, 719-729.	2.0	46
86	Plasma obestatin and ghrelin levels in subjects with Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 415-421.	1.2	45
87	Effects of growth hormone treatment in adults with Praderâ€“Willi syndrome. Growth Hormone and IGF Research, 2013, 23, 81-87.	1.1	44
88	Is gestation in Prader-Willi syndrome affected by the genetic subtype?. Journal of Assisted Reproduction and Genetics, 2009, 26, 461-466.	2.5	43
89	The 15q11.2 BP1-BP2 Microdeletion (Burnsideâ€“Butler) Syndrome: In Silico Analyses of the Four Coding Genes Reveal Functional Associations with Neurodevelopmental Disorders. International Journal of Molecular Sciences, 2020, 21, 3296.	4.1	41
90	Antley-Bixler syndrome: report of a patient and review of literature. Clinical Genetics, 2008, 46, 372-376.	2.0	39

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91	Genetics of hypertension. Current status. Journal Medical Libanais, 2010, 58, 175-8.	0.0	39
92	Anthropometric comparison of mentally retarded males with and without the fragile X syndrome. American Journal of Medical Genetics Part A, 1991, 38, 260-268.	2.4	38
93	Coenzyme Q10 levels in Prader-Willi syndrome: Comparison with obese and non-obese subjects. American Journal of Medical Genetics Part A, 2003, 119A, 168-171.	2.4	38
94	A 5-year-old white girl with Prader-Willi syndrome and a submicroscopic deletion of chromosome 15q11q13. , 1996, 65, 137-141.		37
95	Telomerase activity in skeletal sarcomas. Annals of Surgical Oncology, 1998, 5, 627-634.	1.5	37
96	Methylation-Specific Multiplex Ligation-Dependent Probe Amplification and Identification of Deletion Genetic Subtypes in Prader-Willi Syndrome. Genetic Testing and Molecular Biomarkers, 2012, 16, 178-186.	0.7	37
97	Coding and noncoding expression patterns associated with rare obesity-related disorders: Prader–Willi and Alström syndromes. Advances in Genomics and Genetics, 2015, 2015, 53.	0.8	37
98	Growth Charts for Prader-Willi Syndrome During Growth Hormone Treatment. Clinical Pediatrics, 2016, 55, 957-974.	0.8	37
99	Functional analysis of schizophrenia genes using GeneAnalytics program and integrated databases. Gene, 2018, 641, 25-34.	2.2	37
100	Acrodysostosis: Report of a 13-year-old boy with review of literature and metacarphphalangeal pattern profile analysis. American Journal of Medical Genetics Part A, 1988, 30, 971-980.	2.4	36
101	Praderâ€“Willi syndrome and earlyâ€onset morbid obesity NIH rare disease consortium: A review of natural history study. American Journal of Medical Genetics, Part A, 2018, 176, 368-375.	1.2	34
102	Clinical Findings and Natural History of Prader-Willi Syndrome. , 2006, , 3-48.		34
103	Characterization of Obesity in the Prader-Labhart-Willi Syndrome: Fatness Patterning. Medical Anthropology Quarterly, 1989, 3, 294-305.	1.4	33
104	Metabolic profiling in Prader-Willi syndrome and nonsyndromic obesity: sex differences and the role of growth hormone. Clinical Endocrinology, 2015, 83, 797-805.	2.4	33
105	Transcranial direct current stimulation reduces foodâ€craving and measures of hyperphagia behavior in participants with Praderâ€Willi syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 266-275.	1.7	32
106	Over-expression of the miRNA cluster at chromosome 14q32 in the alcoholic brain correlates with suppression of predicted target mRNA required for oligodendrocyte proliferation. Gene, 2013, 526, 356-363.	2.2	31
107	Methylation PCR analysis of Prader-Willi syndrome, Angelman syndrome, and control subjects. American Journal of Medical Genetics Part A, 1998, 80, 263-265.	2.4	30
108	Microarray analysis of gene/transcript expression in Angelman syndrome: deletion versus UPD. Genomics, 2005, 85, 85-91.	2.9	30

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109	Multiplex Immunoassay of Plasma Cytokine Levels in Men with Alcoholism and the Relationship to Psychiatric Assessments. International Journal of Molecular Sciences, 2016, 17, 472.	4.1	30
110	Metacarpophalangeal pattern profile analysis in sotos syndrome: A follow-up report on 34 subjects. American Journal of Medical Genetics Part A, 1988, 29, 143-147.	2.4	29
111	ANKRD11 gene deletion in a 17-year-old male. Clinical Dysmorphology, 2011, 20, 170-171.	0.3	29
112	Deletion of <i>TOP3B</i> Is Associated with Cognitive Impairment and Facial Dysmorphism. Cytogenetic and Genome Research, 2016, 150, 106-111.	1.1	29
113	A postnatal role for embryonic myosin revealed by MYH3 mutations that alter TGFÎ² signaling and cause autosomal dominant spondylocarpotarsal synostosis. Scientific Reports, 2017, 7, 41803.	3.3	29
114	Prader-Willi syndrome and atypical submicroscopic 15q11-q13 deletions with or without imprinting defects. European Journal of Medical Genetics, 2016, 59, 584-589.	1.3	28
115	Metacarpophalangeal pattern profile analysis in Sotos syndrome. American Journal of Medical Genetics Part A, 1985, 20, 625-629.	2.4	27
116	Screen for MAOA mutations in target human groups. American Journal of Medical Genetics Part A, 1999, 88, 25-28.	2.4	27
117	Clinically relevant known and candidate genes for obesity and their overlap with human infertility and reproduction. Journal of Assisted Reproduction and Genetics, 2015, 32, 495-508.	2.5	27
118	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
119	PATERNAL HYDROCARBON EXPOSURE IN PRADER-WILLI SYNDROME. Lancet, The, 1987, 330, 1458.	13.7	26
120	Microsatellite instability in sarcomas. Annals of Surgical Oncology, 1998, 5, 356-360.	1.5	26
121	X-chromosome inactivation patterns in females with Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 469-475.	1.2	26
122	IL1RAPL1 gene deletion as a cause of X-linked intellectual disability and dysmorphic features. European Journal of Medical Genetics, 2012, 55, 32-36.	1.3	26
123	The High Direct Medical Costs of Prader-Willi Syndrome. Journal of Pediatrics, 2016, 175, 137-143.	1.8	26
124	Clinical Observations and Treatment Approaches for Scoliosis in Praderâ€“Willi Syndrome. Genes, 2020, 11, 260.	2.4	26
125	Do some patients with fragile X syndrome have precocious puberty?. American Journal of Medical Genetics Part A, 1988, 31, 779-781.	2.4	25
126	Multicentre study of maternal and neonatal outcomes in individuals with Prader-Willi syndrome. Journal of Medical Genetics, 2018, 55, 594-598.	3.2	25

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127	Early Diagnosis in Praderâ€“Willi Syndrome Reduces Obesity and Associated Co-Morbidities. Genes, 2019, 10, 898.	2.4	25
128	Craniofacial variation and growth in the Prader-Labhart-Willi syndrome. American Journal of Physical Anthropology, 1987, 74, 459-464.	2.1	24
129	Praderâ€”Willi Syndrome: Genetics and Behavior. Peabody Journal of Education, 1996, 71, 187-212.	1.3	24
130	Microsatellite instability in sacral chordoma. , 2000, 73, 100-103.		24
131	Currently recognized genes for schizophrenia: Highâ€resolution chromosome ideogram representation. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 181-202.	1.7	24
132	Praderâ€Willi syndrome genetic subtypes and clinical neuropsychiatric diagnoses in residential care adults. Clinical Genetics, 2018, 93, 622-631.	2.0	24
133	Tobacco and cannabis use in college students are predicted by sexâ€dimorphic interactions between <scp>MAOA</scp> genotype and child abuse. CNS Neuroscience and Therapeutics, 2019, 25, 101-111.	3.9	24
134	Occupational hydrocarbon exposure among fathers of Prader-Willi syndrome patients with and without deletions of 15q. American Journal of Human Genetics, 1989, 44, 806-10.	6.2	24
135	Genetic variants of the human obesity (OB) gene in subjects with and without Praderâ€”Willi syndrome: comparison with body mass index and weight. Clinical Genetics, 1998, 54, 385-393.	2.0	23
136	Anthropometric and craniofacial patterns in mentally retarded males with emphasis on the fragile X syndrome. Clinical Genetics, 1993, 44, 129-138.	2.0	23
137	Impact of genetic subtypes of Praderâ€“Willi syndrome with growth hormone therapy on intelligence and body mass index. American Journal of Medical Genetics, Part A, 2019, 179, 1826-1835.	1.2	23
138	GeneAnalytics Pathways and Profiling of Shared Autism and Cancer Genes. International Journal of Molecular Sciences, 2019, 20, 1166.	4.1	23
139	Diphenylbutylpiperidine Antipsychotic Drugs Inhibit Prolactin Receptor Signaling to Reduce Growth of Pancreatic Ductal Adenocarcinoma in Mice. Gastroenterology, 2020, 158, 1433-1449.e27.	1.3	23
140	Chromosome 15 Imprinting Disorders: Genetic Laboratory Methodology and Approaches. Frontiers in Pediatrics, 2020, 8, 154.	1.9	23
141	Metacarpophalangeal pattern profile analysis in clinical genetics: An applied anthropometric method. American Journal of Physical Anthropology, 1986, 70, 195-201.	2.1	22
142	A 26-month-old child with Marden-Walker syndrome and pyloric stenosis. American Journal of Medical Genetics Part A, 1987, 26, 915-919.	2.4	22
143	The developing role of anthropologists in medical genetics: Anthropometric assessment of the Praderâ€Labhartâ€Willi syndrome as an illustration. Medical Anthropology: Cross Cultural Studies in Health and Illness, 1989, 10, 247-253.	1.2	22
144	Familial double pericentric inversion of chromosome 5 with some features of cri-du-chat syndrome. Human Genetics, 1996, 97, 802-807.	3.8	22

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145	Assessment and Treatment in Autism Spectrum Disorders: A Focus on Genetics and Psychiatry. Autism Research & Treatment, 2012, 2012, 1-11.	0.5	22
146	High-resolution chromosome ideogram representation of recognized genes for bipolar disorder. Gene, 2016, 586, 136-147.	2.2	22
147	Benefits and limitations of prenatal screening for Praderâ€“Willi syndrome. Prenatal Diagnosis, 2017, 37, 81-94.	2.3	22
148	Parent-of-Origin Effects in 15q11.2 BP1-BP2 Microdeletion (Burnside-Butler) Syndrome. International Journal of Molecular Sciences, 2019, 20, 1459.	4.1	22
149	Decreased bone mineral density in Prader-Willi syndrome: comparison with obese subjects. American Journal of Medical Genetics Part A, 2001, 103, 216-22.	2.4	22
150	Metacarpophalangeal pattern profile analysis in Robinow syndrome. American Journal of Medical Genetics Part A, 1987, 27, 219-223.	2.4	21
151	Double-blind, randomized placebo-controlled clinical trial of benfotiamine for severe alcohol dependence. Drug and Alcohol Dependence, 2013, 133, 562-570.	3.2	21
152	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. Pediatric Neurology, 2021, 123, 30-37.	2.1	21
153	RESTING METABOLIC RATE IN PRADER-WILLI SYNDROME. Dysmorphology and Clinical Genetics, 1990, 4, 27-32.	1.0	21
154	Linkage analysis in a large kindred with autosomal dominant transmission of polyglandular autoimmune disease type II (Schmidt syndrome). American Journal of Medical Genetics Part A, 1984, 18, 61-65.	2.4	20
155	Photoanthropometric study of craniofacial traits of individuals with Prader-Willi syndrome. American Journal of Medical Genetics Part A, 1995, 58, 38-45.	2.4	20
156	Molecular diagnosis of Prader-Willi syndrome: Comparison of cytogenetic and molecular genetic data including parent of origin dependent methylation DNA patterns. American Journal of Medical Genetics Part A, 1996, 61, 188-190.	2.4	20
157	Klinefelter and trisomy X syndromes in patients with Prader-Willi syndrome and uniparental maternal disomy of chromosome 15â€”A coincidence?. American Journal of Medical Genetics Part A, 1997, 72, 111-114.	2.4	20
158	Prader-Willi syndrome: Are there population differences?. Clinical Genetics, 2008, 22, 292-294.	2.0	20
159	Frequency of Praderâ€“Willi syndrome in births conceived via assisted reproductive technology. Genetics in Medicine, 2014, 16, 164-169.	2.4	20
160	Pharmacogenetics Informed Decision Making in Adolescent Psychiatric Treatment: A Clinical Case Report. International Journal of Molecular Sciences, 2015, 16, 4416-4428.	4.1	20
161	High resolution chromosome analysis and fluorescence in situ hybridization in patients referred for Prader-Willi or Angelman syndrome. American Journal of Medical Genetics Part A, 1995, 56, 420-422.	2.4	19
162	C-reactive protein levels in subjects with Prader-Willi syndrome and obesity. Genetics in Medicine, 2006, 8, 243-248.	2.4	19

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163	High plasma neurotensin levels in children with Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1773-1778.	1.2	19
164	Loxapine Add-on for Adolescents and Adults with Autism Spectrum Disorders and Irritability. Journal of Child and Adolescent Psychopharmacology, 2015, 25, 150-159.	1.3	19
165	Pharmacogenetics and Psychiatric Care: A Review and Commentary. Journal of Mental Health and Clinical Psychology, 2018, 2, 17-24.	0.5	19
166	Autistic and dysmorphic features associated with a submicroscopic 2q33.3â€“q34 interstitial deletion detected by array comparative genomic hybridization. American Journal of Medical Genetics, Part A, 2008, 146A, 521-524.	1.2	18
167	Magnesium Supplement and the 15q11.2 BP1â€“BP2 Microdeletion (Burnsideâ€“Butler) Syndrome: A Potential Treatment?. International Journal of Molecular Sciences, 2019, 20, 2914.	4.1	18
168	Mutation in TNXB gene causes moderate to severe Ehlers-Danlos syndrome. World Journal of Medical Genetics, 2016, 6, 17.	1.0	18
169	Methylation PCR analysis of Prader-Willi syndrome, Angelman syndrome, and control subjects. American Journal of Medical Genetics Part A, 1998, 80, 263-5.	2.4	18
170	Standards for selected anthropometric measurements in males with the fragile X syndrome. Pediatrics, 1992, 89, 1059-62.	2.1	18
171	BIRTH SEASONALITY IN PRADER-WILLI SYNDROME. Lancet, The, 1985, 326, 828-829.	13.7	17
172	Preliminary communication: Photoanthropometric analysis of individuals with the fragile X syndrome. American Journal of Medical Genetics Part A, 1988, 30, 165-168.	2.4	17
173	Metacarpophalangeal pattern profile analysis in fragile X syndrome. American Journal of Medical Genetics Part A, 1988, 31, 767-773.	2.4	17
174	Molecular cytogenetic analysis of patients with holoprosencephaly and structural rearrangements of 7q. American Journal of Medical Genetics Part A, 1998, 76, 51-57.	2.4	17
175	Partial Deletion of Chromosome 1p31.1 Including only the Neuronal Growth Regulator 1 Gene in Two Siblings. Journal of Pediatric Genetics, 2015, 04, 023-028.	0.7	17
176	Further phenotypic expansion of 15q11.2 BP1-BP2 microdeletion (Burnside-Butler) syndrome. Journal of Pediatric Genetics, 2015, 03, 041-044.	0.7	17
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