David Amor

List of Publications by Year
in descending order

272 papers	14,785 citations	28736 57 h-index	27587 110 g-index
282 all docs	282 docs citations	282 times ranked	23854 citing authors

#	Article	IF	Citations
1	The Cost of Raising Individuals with Fragile X or Chromosome 15 Imprinting Disorders in Australia. Journal of Autism and Developmental Disorders, 2023, 53, 1682-1692.	1.7	4
2	Non-Invasive Prenatal Testing for â€œNon-Medicalâ€•Traits: Ensuring Consistency in Ethical Decision-Making. American Journal of Bioethics, 2023, 23, 3-20.	0.5	21
3	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.0	42
4	Feasibility of Screening for Chromosome 15 Imprinting Disorders in 16â€¯579 Newborns by Using a Novel Genomic Workflow. JAMA Network Open, 2022, 5, e2141911.	2.8	14
5	A framework for reporting secondary and incidental findings in prenatal sequencing: When and for whom?. Prenatal Diagnosis, 2022, 42, 697-704.	1.1	10
6	Reproductive function in men conceived with inÂvitro fertilization and intracytoplasmic sperm injection. Fertility and Sterility, 2022, 117, 727-737.	0.5	7
7	Selfâ€•reported impact of developmental stuttering across the lifespan. Developmental Medicine and Child Neurology, 2022, 64, 1297-1306.	1.1	7
8	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	2.6	16
9	Heterozygous <scp>PNPT1</scp> Variants Cause Spinocerebellar Ataxia Type 25. Annals of Neurology, 2022, 92, 122-137.	2.8	8
10	Social motivation a relative strength in DYRK1A syndrome on a background of significant speech and language impairments. European Journal of Human Genetics, 2022, 30, 800-811.	1.4	13
11	ISPD 2021 debate â€•All in vitro fertilization cycles should involve preâ€•implantation genetic testing to improve fetal health and pregnancy outcomes. Prenatal Diagnosis, 2022, , .	1.1	2
12	Does being conceived by assisted reproductive technology influence adult quality of life?. Human Fertility, 2022, , 1-7.	0.7	2
13	Setting Preconception Care Priorities in Australia Using a Delphi Technique. Seminars in Reproductive Medicine, 2022, 40, 214-226.	0.5	2
14	Gene selection for the Australian Reproductive Genetic Carrier Screening Project (â€œMackenzieâ€™s) Tj ETQq0 0 0 rgBT /Overlock 10	1.4	60
15	Firstâ€•trimester maternal serum biomarkers and the risk of cerebral palsy. Developmental Medicine and Child Neurology, 2021, 63, 183-189.	1.1	5
16	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	2.2	33
17	Speech and language phenotype in Phelan-McDermid (22q13.3) syndrome. European Journal of Human Genetics, 2021, 29, 564-574.	1.4	14
18	Factors influencing medical practitioner participation in population carrier screening for cystic fibrosis. Australian and New Zealand Journal of Obstetrics and Gynaecology, 2021, 61, 232-238.	0.4	3

#	Article	IF	Citations
19	Lymphedema distichiasis syndrome may be caused by <scp>FOXC2</scp> promoterâ€enhancer dissociation and disruption of a topological associated domain. American Journal of Medical Genetics, Part A, 2021, 185, 150-156.	0.7	4
20	Child health after preimplantation genetic testing. Reproductive BioMedicine Online, 2021, 42, 609-619.	1.1	3
21	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
22	ALG13 Xâ€linked intellectual disability: New variants, glycosylation analysis, and expanded phenotypes. Journal of Inherited Metabolic Disease, 2021, 44, 1001-1012.	1.7	9
23	Speech and language deficits are central to SETBP1 haploinsufficiency disorder. European Journal of Human Genetics, 2021, 29, 1216-1225.	1.4	26
24	Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. Genetics in Medicine, 2021, 23, 1234-1245.	1.1	6
25	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	2.6	19
26	Personal utility of genomic sequencing for infants with congenital deafness. American Journal of Medical Genetics, Part A, 2021, 185, 3634-3643.	0.7	6
27	Expanding the clinical and radiological phenotypes of leukoencephalopathy due to biallelic <scp>HMBS</scp> mutations. American Journal of Medical Genetics, Part A, 2021, 185, 2941-2950.	0.7	2
28	Genome sequencing in congenital cataracts improves diagnostic yield. Human Mutation, 2021, 42, 1173-1183.	1.1	10
29	Severe speech impairment is a distinguishing feature of FOXP1â€related disorder. Developmental Medicine and Child Neurology, 2021, 63, 1417-1426.	1.1	24
30	Principles of Genomic Newborn Screening Programs. JAMA Network Open, 2021, 4, e2114336.	2.8	39
31	Second trimester maternal serum biomarkers and the risk of cerebral palsy. Prenatal Diagnosis, 2021, 41, 1101-1110.	1.1	0
32	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. Lancet Oncology, The, 2021, 22, 1014-1022.	5.1	58
33	A systematic review of brain MRI findings in monogenic disorders strongly associated with autism spectrum disorder. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2021, 62, 1339-1352.	3.1	6
34	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. British Journal of Cancer, 2021, 125, 1135-1145.	2.9	9
35	Paternal retraction of a fragile X allele to normal size, showing normal function over two generations. American Journal of Medical Genetics, Part A, 2021, , .	0.7	1
36	Exome Sequencing for Isolated Congenital Hearing Loss: A Costâ€Effectiveness Analysis. Laryngoscope, 2021, 131, E2371-E2377.	1.1	5

#	Article	IF	Citations
37	Study protocol: childhood outcomes of fetal genomic variants: the PrenatAL Microarray (PALM) cohort study. BMC Pediatrics, 2021, 21, 447.	0.7	0
38	American Heart Association ideal cardiovascular health score and subclinical atherosclerosis in 22â€“35-year-old adults conceived with and without assisted reproductive technologies. Human Reproduction, 2020, 35, 232-239.	0.4	16
39	Exome sequencing in infants with congenital hearing impairment: a population-based cohort study. European Journal of Human Genetics, 2020, 28, 587-596.	1.4	38
40	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. Genetics in Medicine, 2020, 22, 797-802.	1.1	15
41	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	9.4	96
42	Health and fertility of ICSI-conceived young men: study protocol. Human Reproduction Open, 2020, 2020, hoaa042.	2.3	6
43	FMR1 mRNA from full mutation alleles is associated with ABC-CFX scores in males with fragile X syndrome. Scientific Reports, 2020, 10, 11701.	1.6	11
44	Growth Trajectories in Genetic Subtypes of Praderâ€“Willi Syndrome. Genes, 2020, 11, 736.	1.0	5
45	Relationships between UBE3A and SNORD116 expression and features of autism in chromosome 15 imprinting disorders. Translational Psychiatry, 2020, 10, 362.	2.4	14
46	DNA Methylation at Birth Predicts Intellectual Functioning and Autism Features in Children with Fragile X Syndrome. International Journal of Molecular Sciences, 2020, 21, 7735.	1.8	10
47	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	2.6	39
48	Successful treatment of lathosterolosis: A rare defect in cholesterol biosynthesisâ€”A case report and review of literature. JIMD Reports, 2020, 56, 14-19.	0.7	7
49	Evaluating systematic reanalysis of clinical genomic data in rare disease from single center experience and literature review. Molecular Genetics & Genomic Medicine, 2020, 8, e1508.	0.6	44
50	Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. American Heart Journal, 2020, 225, 108-119.	1.2	25
51	Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System. JAMA - Journal of the American Medical Association, 2020, 323, 2503.	3.8	160
52	Expanding the genotypic and phenotypic spectrum of severe serine biosynthesis disorders. Human Mutation, 2020, 41, 1615-1628.	1.1	14
53	Revealing hidden genetic diagnoses in the ocular anterior segment disorders. Genetics in Medicine, 2020, 22, 1623-1632.	1.1	31
54	Genetics and pediatric hospital admissions, 1985 to 2017. Genetics in Medicine, 2020, 22, 1777-1785.	1.1	13

#	Article	IF	Citations
55	Exome sequencing in newborns with congenital deafness as a model for genomic newborn screening: the Baby Beyond Hearing project. Genetics in Medicine, 2020, 22, 937-944.	1.1	22
56	Current controversies in prenatal diagnosis 2: The 59 genes ACMG recommends reporting as secondary findings when sequencing postnatally should be reported when detected on fetal (and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.1	18
57	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	1.1	14
58	Severe childhood speech disorder. Neurology, 2020, 94, e2148-e2167.	1.5	68
59	Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System. Obstetrical and Gynecological Survey, 2020, 75, 662-664.	0.2	7
60	Psychosocial impact of undergoing prostate cancer screening for men with <scp>BRCA</scp>1 or <scp>BRCA</scp>2 mutations. BJU International, 2019, 123, 284-292.	1.3	9
61	Epigenetics of fragile X syndrome and fragile Xâ€related disorders. Developmental Medicine and Child Neurology, 2019, 61, 121-127.	1.1	43
62	HIST1H1E heterozygous proteinâ€truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. American Journal of Medical Genetics, Part A, 2019, 179, 2049-2055.	0.7	16
63	Expansion of phenotype of DDX3X syndrome: six new cases. Clinical Dysmorphology, 2019, 28, 169-174.	0.1	26
64	IREB2-associated neurodegeneration. Brain, 2019, 142, e40-e40.	3.7	19
65	Childhood Hearing Australasian Medical Professionals network: Consensus guidelines on investigation and clinical management of childhood hearing loss. Journal of Paediatrics and Child Health, 2019, 55, 1013-1022.	0.4	12
66	Significantly Elevated FMR1 mRNA and Mosaicism for Methylated Premutation and Full Mutation Alleles in Two Brothers with Autism Features Referred for Fragile X Testing. International Journal of Molecular Sciences, 2019, 20, 3907.	1.8	12
67	Assisted reproductive technologies are associated with limited epigenetic variation at birth that largely resolves by adulthood. Nature Communications, 2019, 10, 3922.	5.8	94
68	Inhibition of Upf2-Dependent Nonsense-Mediated Decay Leads to Behavioral and Neurophysiological Abnormalities by Activating the Immune Response. Neuron, 2019, 104, 665-679.e8.	3.8	43
69	Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. American Journal of Human Genetics, 2019, 105, 151-165.	2.6	170
70	Incomplete silencing of full mutation alleles in males with fragile X syndrome is associated with autistic features. Molecular Autism, 2019, 10, 21.	2.6	20
71	Health of adults aged 22 to 35Âyears conceived by assisted reproductive technology. Fertility and Sterility, 2019, 112, 130-139.	0.5	49
72	Exploring the speech and language of individuals with nonâ€syndromic submucous cleft palate: a preliminary report. International Journal of Language and Communication Disorders, 2019, 54, 767-778.	0.7	7

#	Article	IF	Citations
73	Recessive variants in ZNF142 cause a complex neurodevelopmental disorder with intellectual disability, speech impairment, seizures, and dystonia. Genetics in Medicine, 2019, 21, 2532-2542.	1.1	17
74	Genetic or Other Causation Should Not Change the Clinical Diagnosis of Cerebral Palsy. Journal of Child Neurology, 2019, 34, 472-476.	0.7	82
75	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	2.6	23
76	A novel approach to offering additional genomic findingsâ€"A protocol to test a twoâ€step approach in the healthcare system. Journal of Genetic Counseling, 2019, 28, 388-397.	0.9	14
77	The incidence, prevalence and clinical features of MECP2 duplication syndrome in Australian children. Journal of Paediatrics and Child Health, 2019, 55, 1315-1322.	0.4	28
78	Speech and language in children with Klinefelter syndrome. Journal of Communication Disorders, 2019, 78, 84-96.	0.8	23
79	Population genomic screening of all young adults in a health-care system: a cost-effectiveness analysis. Genetics in Medicine, 2019, 21, 1958-1968.	1.1	63
80	Diagnostic and service impact of genomic testing technologies in a neonatal intensive care unit. Journal of Paediatrics and Child Health, 2019, 55, 1309-1314.	0.4	11
81	Fragile Females: Case Series of Epilepsy in Girls With FMR1 Disruption. Pediatrics, 2019, 144, .	1.0	5
82	Genetic Analysis of Patients Who Experienced Awareness with Recall while under General Anesthesia. Anesthesiology, 2019, 131, 974-982.	1.3	9
83	Clinical Spectrum and Functional Consequences Associated with Bi-Allelic Pathogenic PNPT1 Variants. Journal of Clinical Medicine, 2019, 8, 2020.	1.0	16
84	Abnormally Methylated FMR1 in Absence of a Detectable Full Mutation in a U.S.A Patient Cohort Referred for Fragile X Testing. Scientific Reports, 2019, 9, 15315.	1.6	9
85	Intellectual functioning and behavioural features associated with mosaicism in fragile X syndrome. Journal of Neurodevelopmental Disorders, 2019, 11, 41.	1.5	26
86	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	1.1	127
87	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	2.6	711
88	DNA methylation analysis for screening and diagnostic testing in neurodevelopmental disorders. Essays in Biochemistry, 2019, 63, 785-795.	2.1	9
89	Heritable DNA methylation marks associated with susceptibility to breast cancer. Nature Communications, 2018, 9, 867.	5.8	76
90	Deep phenotyping of speech and language skills in individuals with 16p11.2 deletion. European Journal of Human Genetics, 2018, 26, 676-686.	1.4	58

#	Article	IF	Citations
91	Offering pregnant women different levels of genetic information from prenatal chromosome microarray: a prospective study. European Journal of Human Genetics, 2018, 26, 485-494.	1.4	19
92	The novel RAF1 mutation p.(Gly361Ala) located outside the kinase domain of the CR3 region in two patients with Noonan syndrome, including one with a rare brain tumor. American Journal of Medical Genetics, Part A, 2018, 176, 470-476.	0.7	17
93	Epigenome-wide analysis in newborn blood spots from monozygotic twins discordant for cerebral palsy reveals consistent regional differences in DNA methylation. Clinical Epigenetics, 2018, 10, 25.	1.8	47
94	Intragenic DNA methylation in buccal epithelial cells and intellectual functioning in a paediatric cohort of males with fragile X. Scientific Reports, 2018, 8, 3644.	1.6	17
95	FMR1 allele size distribution in 35,000 males and females: a comparison of developmental delay and general population cohorts. Genetics in Medicine, 2018, 20, 1627-1634.	1.1	23
96	Letter regarding the article â€œExtending the phenotype of recurrent rearrangements of 16p11.2: Deletions in mentally retarded patients without autism and in normal individuals (Bijlsma et al., 2009) Tj ETQq0 0 0 rgBT /Overlock 10 T Journal of Medical Genetics, 2018, 61, 48-49.	0.7	0
97	Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: outcomes of 12,000 tests. Genetics in Medicine, 2018, 20, 513-523.	1.1	80
98	Early speech development in Koolen de Vries syndrome limited by oral praxis and hypotonia. European Journal of Human Genetics, 2018, 26, 75-84.	1.4	30
99	A craniosynostosis massively parallel sequencing panel study in 309 Australian and New Zealand patients: findings and recommendations. Genetics in Medicine, 2018, 20, 1061-1068.	1.1	37
100	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	2.6	184
101	Investigating the child with intellectual disability. Journal of Paediatrics and Child Health, 2018, 54, 1154-1158.	0.4	12
102	Characterization of speech and language phenotype in children with NRXN1 deletions. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 700-708.	1.1	10
103	Genetic variation affecting DNA methylation and the human imprinting disorder, Beckwith-Wiedemann syndrome. Clinical Epigenetics, 2018, 10, 114.	1.8	10
104	Variants in <scp>ACTG</scp>2 underlie a substantial number of Australasian patients with primary chronic intestinal pseudoâ€obstruction. Neurogastroenterology and Motility, 2018, 30, e13371.	1.6	23
105	Natural History of Vanishing White Matter. Annals of Neurology, 2018, 84, 274-288.	2.8	69
106	Attitudes of sperm, egg and embryo donors and recipients towards genetic information and screening of donors. Reproductive Health, 2018, 15, 26.	1.2	10
107	Exploring autism symptoms in an Australian cohort of patients with Prader-Willi and Angelman syndromes. Journal of Neurodevelopmental Disorders, 2018, 10, 24.	1.5	26
108	Functional analysis of a hypomorphic allele shows that MMP14 catalytic activity is the prime determinant of the Winchester syndrome phenotype. Human Molecular Genetics, 2018, 27, 2775-2788.	1.4	25

#	Article	IF	Citations
109	Autosomal Reciprocal Translocations. , 2018, , .		1
110	Chromosome Analysis. , 2018, , .		0
111	Chromosome Instability Syndromes. , 2018, , .		0
112	Robertsonian Translocations. , 2018, , .		1
113	Sex Chromosome Translocations. , 2018, , .		0
114	Elements of Medical Cytogenetics. , 2018, , .		1
115	The Origins and Consequences of Chromosome Pathology. , 2018, , .		1
116	Normal Chromosomal Variation. , 2018, , .		0
117	Autosomal Ring Chromosomes. , 2018, , .		0
118	Inversions. , 2018, , .		0
119	Preimplantation Genetic Diagnosis. , 2018, , .		0
120	Prenatal Testing Procedures. , 2018, , .		0
121	Down Syndrome, Other Full Aneuploidies, Polyploidy, and the Influence of Parental Age. , 2018, , .		0
122	Autosomal Structural Rearrangements. , 2018, , .		0
123	Insertions. , 2018, , .		0
124	Deriving and Using A Risk Figure. , 2018, , .		0
125	Complex Chromosomal Rearrangements. , 2018, , .		0
126	Reproductive Failure. , 2018, , .		0

#	Article	IF	Citations
127	Centromere Fissions, Complementary Isochromosomes, Telomeric Fusions, Balancing Supernumerary Chromosomes, Neocentromeres, Jumping Translocations, and Chromothripsis. , 2018, , .		0
128	Sex Chromosome Aneuploidy and Structural Rearrangement. , 2018, , .		0
129	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. Nature Genetics, 2017, 49, 511-514.	9.4	69
130	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	1.8	66
131	Clinical utility gene card for: 16p12.2 microdeletion. European Journal of Human Genetics, 2017, 25, 271-271.	1.4	4
132	Diagnostic Impact and Cost-effectiveness of Whole-Exome Sequencing for Ambulant Children With Suspected Monogenic Conditions. JAMA Pediatrics, 2017, 171, 855.	3.3	252
133	Neuropathology of childhoodâ€onset basal ganglia degeneration caused by mutation of VAC14. Annals of Clinical and Translational Neurology, 2017, 4, 859-864.	1.7	17
134	Health outcomes of school-aged children conceived using donor sperm. Reproductive BioMedicine Online, 2017, 35, 445-452.	1.1	3
135	Germline Loss-of-Function Mutations in EPHB4 Cause a Second Form of Capillary Malformation-Arteriovenous Malformation (CM-AVM2) Deregulating RAS-MAPK Signaling. Circulation, 2017, 136, 1037-1048.	1.6	204
136	A protocol for whole-exome sequencing in newborns with congenital deafness: a prospective population-based cohort. BMJ Paediatrics Open, 2017, 1, e000119.	0.6	16
137	Alcohol consumption in a general antenatal population and child neurodevelopment at 2 years. Journal of Epidemiology and Community Health, 2017, 71, 990-998.	2.0	18
138	Clinical review of 24â€“35Âyear olds conceived with and without in vitro fertilization: study protocol. Reproductive Health, 2017, 14, 117.	1.2	14
139	Pseudodiastrophic dysplasia: Two cases delineating and expanding the pre and postnatal phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 1363-1366.	0.7	5
140	Heterozygous mutations in HSD17B4 cause juvenile peroxisomal D-bifunctional protein deficiency. Neurology: Genetics, 2016, 2, e114.	0.9	18
141	ALPK3-deficient cardiomyocytes generated from patient-derived induced pluripotent stem cells and mutant human embryonic stem cells display abnormal calcium handling and establish that ALPK3 deficiency underlies familial cardiomyopathy. European Heart Journal, 2016, 37, 2586-2590.	1.0	49
142	Familial cortical dysplasia caused by mutation in the mammalian target of rapamycin regulator NPRL3. Annals of Neurology, 2016, 79, 132-137.	2.8	116
143	Metalloprotease SPRTN/DVC1 Orchestrates Replication-Coupled DNA-Protein Crosslink Repair. Molecular Cell, 2016, 64, 704-719.	4.5	193
144	â€œI'm Healthy, It's Not Going To Be Meâ€: Exploring experiences of carriers identified through a population reproductive genetic carrier screening panel in Australia. American Journal of Medical Genetics, Part A, 2016, 170, 2052-2059.	0.7	20

#	Article	IF	Citations
145	A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. Genetics in Medicine, 2016, 18, 1090-1096.	1.1	332
146	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. American Journal of Human Genetics, 2016, 98, 579-587.	2.6	88
147	Identification of Males with Cryptic Fragile X Alleles by Methylation-Specific Quantitative Melt Analysis. Clinical Chemistry, 2016, 62, 343-352.	1.5	28
148	Loss of RMI2 Increases Genome Instability and Causes a Bloom-Like Syndrome. PLoS Genetics, 2016, 12, e1006483.	1.5	47
149	Detection of skewed X-chromosome inactivation in Fragile X syndrome and X chromosome aneuploidy using quantitative melt analysis. Expert Reviews in Molecular Medicine, 2015, 17, e13.	1.6	12
150	Complete callosal agenesis, pontocerebellar hypoplasia, and axonal neuropathy due to AMPD2 loss. Neurology: Genetics, 2015, 1, e16.	0.9	29
151	YPEL1 overexpression in early avian craniofacial mesenchyme causes mandibular dysmorphogenesis by upâ€regulating apoptosis. Developmental Dynamics, 2015, 244, 1022-1030.	0.8	10
152	Hemispheric cortical dysplasia secondary to a mosaic somatic mutation in MTOR. Neurology, 2015, 84, 2029-2032.	1.5	64
153	Familial cortical dysplasia type <scp>IIA</scp> caused by a germline mutation in <scp>DEPDC</scp>5. Annals of Clinical and Translational Neurology, 2015, 2, 575-580.	1.7	95
154	Mutations in SEC24D, Encoding a Component of the COPII Machinery, Cause a Syndromic Form of Osteogenesis Imperfecta. American Journal of Human Genetics, 2015, 96, 432-439.	2.6	143
155	Future of whole genome sequencing. Journal of Paediatrics and Child Health, 2015, 51, 251-254.	0.4	10
156	Preferences for results from genomic microarrays: comparing parents and health care providers. Clinical Genetics, 2015, 87, 21-29.	1.0	18
157	Populationâ€based genetic carrier screening for cystic fibrosis in Victoria. Medical Journal of Australia, 2014, 200, 205-206.	0.8	5
158	New ZMPSTE24 (FACE1) mutations in patients affected with restrictive dermopathy or related progeroid syndromes and mutation update. European Journal of Human Genetics, 2014, 22, 1002-1011.	1.4	51
159	Identification of a Novel RNF213 Variant in a Family with Heterogeneous Intracerebral Vasculopathy. International Journal of Stroke, 2014, 9, E26-E27.	2.9	9
160	Mutations in RAB39B Cause X-Linked Intellectual Disability and Early-Onset Parkinson Disease with Î±-Synuclein Pathology. American Journal of Human Genetics, 2014, 95, 729-735.	2.6	207
161	Mutations in SH3PXD2B cause Borrone dermato-cardio-skeletal syndrome. European Journal of Human Genetics, 2014, 22, 741-747.	1.4	30
162	Implementation of written consent for newborn screening in <scp>V</scp>ictoria, <scp>A</scp>ustralia. Journal of Paediatrics and Child Health, 2014, 50, 399-404.	0.4	14

#	Article	IF	Citations
163	Methylation Analysis in Newborn Screening for Fragile X Syndrome. JAMA Neurology, 2014, 71, 800.	4.5	4
164	A qualitative exploration of mothersâ€™ and fathersâ€™ experiences of having a child with Klinefelter syndrome and the process of reaching this diagnosis. European Journal of Human Genetics, 2014, 22, 18-24.	1.4	32
165	Prenatally detected de novo apparently balanced chromosomal rearrangements: the effect on maternal worry, family functioning and intent of disclosure. Prenatal Diagnosis, 2014, 34, 598-604.	1.1	1
166	Targeted Prostate Cancer Screening in BRCA1 and BRCA2 Mutation Carriers: Results from the Initial Screening Round of the IMPACT Study. European Urology, 2014, 66, 489-499.	0.9	195
167	Early Detection of Fragile X Syndrome: Applications of a Novel Approach for Improved Quantitative Methylation Analysis in Venous Blood and Newborn Blood Spots. Clinical Chemistry, 2014, 60, 963-973.	1.5	43
168	Comparing indicators of health and development of singleton young adults conceived with and without assisted reproductive technology. Fertility and Sterility, 2014, 101, 1055-1063.	0.5	46
169	Availability of treatment drives decisions of genetic health professionals about disclosure of incidental findings. European Journal of Human Genetics, 2014, 22, 1225-1228.	1.4	13
170	Mutations in SPRTN cause early onset hepatocellular carcinoma, genomic instability and progeroid features. Nature Genetics, 2014, 46, 1239-1244.	9.4	165
171	Expanding the phenotypic spectrum of ARID1B-mediated disorders and identification of altered cell-cycle dynamics due to ARID1B haploinsufficiency. Orphanet Journal of Rare Diseases, 2014, 9, 43.	1.2	16
172	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900.	1.4	120
173	Telomere Length Shows No Association with BRCA1 and BRCA2 Mutation Status. PLoS ONE, 2014, 9, e86659.	1.1	10
174	Health and development of ART conceived young adults: a study protocol for the follow-up of a cohort. Reproductive Health, 2013, 10, 15.	1.2	11
175	Meeting the challenge of interpreting highâ€resolution single nucleotide polymorphism array data in prenatal diagnosis: does increased diagnostic power outweigh the dilemma of rare variants?. BJOG: an International Journal of Obstetrics and Gynaecology, 2013, 120, 594-606.	1.1	34
176	Clinical Comparison of Overlapping Deletions of 19p13.3. American Journal of Medical Genetics, Part A, 2013, 161, 1110-1116.	0.7	13
177	Fertility in <scp>T</scp>urner syndrome. Clinical Endocrinology, 2013, 79, 606-614.	1.2	39
178	Considerations for Reporting Genome Results to Patients. Journal of Paediatrics and Child Health, 2013, 49, 82-82.	0.4	0
179	Genotype and clinical care correlations in craniosynostosis: Findings from a cohort of 630 Australian and New Zealand patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 259-270.	0.7	44
180	Fragile Xâ€“related element 2 methylation analysis may provide a suitable option for inclusion of fragile X syndrome and/or sex chromosome aneuploidy into newborn screening: a technical validation study. Genetics in Medicine, 2013, 15, 290-298.	1.1	19

#	Article	IF	Citations
181	Cascade carrier testing after a child is diagnosed with cystic fibrosis through newborn screening: investigating why most relatives do not have testing. Genetics in Medicine, 2013, 15, 533-540.	1.1	22
182	Relationships between age and epi-genotype of the FMR1 exon 1/intron 1 boundary are consistent with non-random X-chromosome inactivation in FM individuals, with the selection for the unmethylated state being most significant between birth and puberty. Human Molecular Genetics, 2013, 22, 1516-1524.	1.4	22
183	RASA1Mutations and Associated Phenotypes in 68 Families with Capillary Malformation-Arteriovenous Malformation. Human Mutation, 2013, 34, 1632-1641.	1.1	221
184	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 2972-2980.	0.7	119
185	Authors' response to: Meeting the challenge of interpreting highâ€resolution single nucleotide polymorphism array data: does increased diagnostic power outweigh the dilemma of rare variants. BJOG: an International Journal of Obstetrics and Gynaecology, 2013, 120, 1297-1297.	1.1	1
186	Longâ€term health and development of children diagnosed prenatally with a de novo apparently balanced chromosomal rearrangement. Prenatal Diagnosis, 2013, 33, 831-838.	1.1	9
187	Anaplastic oligodendroglioma in an adolescent with Lynch syndrome. Pediatric Blood and Cancer, 2013, 60, E13-5.	0.8	8
188	An exploration of genetic health professionals' experience with direct-to-consumer genetic testing in their clinical practice. European Journal of Human Genetics, 2012, 20, 825-830.	1.4	48
189	Further clinical and molecular delineation of the 15q24 microdeletion syndrome. Journal of Medical Genetics, 2012, 49, 110-118.	1.5	40
190	Questionable pathogenicity of FOXG1 duplication. European Journal of Human Genetics, 2012, 20, 595-596.	1.4	14
191	14q12 and severe Rett-like phenotypes: new clinical insights and physical mapping of FOXG1-regulatory elements. European Journal of Human Genetics, 2012, 20, 1216-1223.	1.4	48
192	Fragile X Mental Retardation 1 (FMR1) Intron 1 Methylation in Blood Predicts Verbal Cognitive Impairment in Female Carriers of Expanded FMR1 Alleles: Evidence from a Pilot Study. Clinical Chemistry, 2012, 58, 590-598.	1.5	37
193	Dominant missense mutations in ABCC9 cause CantÃº syndrome. Nature Genetics, 2012, 44, 793-796.	9.4	184
194	Fragile Xâ€associated tremor/ataxia phenotype in a male carrier of unmethylated full mutation in the FMR1 gene. Clinical Genetics, 2012, 82, 88-92.	1.0	66
195	Genetic Counselling. , 2012, , 203-214.		1
196	FMR1 Intron 1 Methylation Predicts FMRP Expression in Blood of Female Carriers of Expanded FMR1 Alleles. Journal of Molecular Diagnostics, 2011, 13, 528-536.	1.2	36
197	Reducing the exome search space for Mendelian diseases using genetic linkage analysis of exome genotypes. Genome Biology, 2011, 12, R85.	13.9	72
198	Human and Mouse Mutations in WDR35 Cause Short-Rib Polydactyly Syndromes Due to Abnormal Ciliogenesis. American Journal of Human Genetics, 2011, 88, 508-515.	2.6	122

#	Article	IF	Citations
199	Germline mutations in the oncogene EZH2 cause Weaver syndrome and increased human height. Oncotarget, 2011, 2, 1127-1133.	0.8	145
200	A case for cystic fibrosis carrier testing in the general population. Medical Journal of Australia, 2011, 194, 208-209.	0.8	17
201	Adverse Obstetric and Perinatal Outcomes in Subfertile Women Conceiving Without Assisted Reproductive Technologies. Obstetrical and Gynecological Survey, 2011, 66, 203-204.	0.2	0
202	Lambdoid synostosis and craniofacial dysmorphism with normal intellect: A novel syndrome?. , 2011, 155, 233-234.		1
203	Phenotypic variability of distal 22q11.2 copy number abnormalities. American Journal of Medical Genetics, Part A, 2011, 155, 1623-1633.	0.7	59
204	Extending the scope of diagnostic chromosome analysis: Detection of single gene defects using high-resolution SNP microarrays. Human Mutation, 2011, 32, 1500-1506.	1.1	41
205	Pathogenic aberrations revealed exclusively by single nucleotide polymorphism (SNP) genotyping data in 5000 samples tested by molecular karyotyping. Journal of Medical Genetics, 2011, 48, 831-839.	1.5	71
206	Looking downstream: a review of the literature on physical and psychosocial health outcomes in adolescents and young adults who were conceived by ART. Human Reproduction, 2011, 26, 1209-1219.	0.4	54
207	Mutations in TRPV4 cause an inherited arthropathy of hands and feet. Nature Genetics, 2011, 43, 1142-1146.	9.4	134
208	Molecular analysis expands the spectrum of phenotypes associated with GLI3 mutations. Human Mutation, 2010, 31, 1142-1154.	1.1	111
209	Uptake of prenatal diagnostic testing and the effectiveness of prenatal screening for Down syndrome. Prenatal Diagnosis, 2010, 30, 522-530.	1.1	24
210	Uptake of carrier testing in families after cystic fibrosis diagnosis through newborn screening. European Journal of Human Genetics, 2010, 18, 1084-1089.	1.4	36
211	Developmental perspectives on copy number abnormalities of the 22q11.2 region. Clinical Genetics, 2010, 78, 201-218.	1.0	11
212	A Genotype-First Approach for the Molecular and Clinical Characterization of Uncommon De Novo Microdeletion of 20q13.33. PLoS ONE, 2010, 5, e12462.	1.1	19
213	Using population-based data to predict the impact of introducing noninvasive prenatal diagnosis for Down syndrome. Genetics in Medicine, 2010, 12, 298-303.	1.1	23
214	Increased risk of blastogenesis birth defects, arising in the first 4 weeks of pregnancy, after assisted reproductive technologies. Human Reproduction, 2010, 25, 59-65.	0.4	103
215	Methylation analysis of fragile X-related epigenetic elements may provide a suitable newborn screening test for fragile X syndrome. Genetics in Medicine, 2010, 12, 595-595.	1.1	11
216	De novo 325 kb microdeletion in chromosome band 10q25.3 including ATRNL1 in a boy with cognitive impairment, autism and dysmorphic features. European Journal of Medical Genetics, 2010, 53, 337-339.	0.7	19

#	Article	IF	Citations
217	Adverse obstetric and perinatal outcomes in subfertile women conceiving without assisted reproductive technologies. Fertility and Sterility, 2010, 94, 2674-2679.	0.5	134
218	Russellâ€“Silver syndrome due to paternal H19/IGF2 hypomethylation in a patient conceived using intracytoplasmic sperm injection. Reproductive BioMedicine Online, 2010, 20, 843-847.	1.1	25
219	Further molecular and clinical delineation of co-locating 17p13.3 microdeletions and microduplications that show distinctive phenotypes. Journal of Medical Genetics, 2010, 47, 299-311.	1.5	137
220	Nonâ€invasive prenatal diagnosis â€" toward a new horizon. Medical Journal of Australia, 2009, 191, 414-414.	0.8	2
221	Pallisterâ€“Killian syndrome caused by mosaicism for a supernumerary ring chromosome 12p. American Journal of Medical Genetics, Part A, 2009, 149A, 505-509.	0.7	16
222	Familial upper eyelid coloboma with ipsilateral anterior hairline abnormality: Two new reports of MOTA syndrome. , 2009, 149A, 767-769.		10
223	Functional disomy of proximal Xp causes a distinct phenotype comprising early hypotonia, hypertelorism, small hands and feet, ear abnormalities, myopia and cognitive impairment. American Journal of Medical Genetics, Part A, 2009, 149A, 1763-1767.	0.7	4
224	Imprinting, Small Babies and Assisted Reproduction: Genomic imprinting, small babies and assisted reproduction. European Journal of Human Genetics, 2009, 17, 1-2.	1.4	8
225	Extending the phenotype of recurrent rearrangements of 16p11.2: Deletions in mentally retarded patients without autism and in normal individuals. European Journal of Medical Genetics, 2009, 52, 77-87.	0.7	226
226	4.45ÂMb microduplication in chromosome band 14q12 including FOXG1 in a girl with refractory epilepsy and intellectual impairment. European Journal of Medical Genetics, 2009, 52, 440-442.	0.7	55
227	Registry- and Clinic-Based Analyses of Birth Defects and Syndromes Associated with Cleft Lip/Palate in Victoria, Australia. Cleft Palate-Craniofacial Journal, 2009, 46, 583-587.	0.5	7
228	Pregnancies conceived using assisted reproductive technologies (ART) have low levels of pregnancy-associated plasma protein-A (PAPP-A) leading to a high rate of false-positive results in first trimester screening for Down syndrome. Human Reproduction, 2009, 24, 1330-1338.	0.4	96
229	Identification of 34 novel and 56 knownFOXL2mutations in patients with blepharophimosis syndrome. Human Mutation, 2008, 29, E205-E219.	1.1	42
230	Spondylocostal dysostosis in a pregnancy complicated by confined placental mosaicism for tetrasomy 9p. American Journal of Medical Genetics, Part A, 2008, 146A, 1972-1976.	0.7	4
231	Detection of cryptic pathogenic copy number variations and constitutional loss of heterozygosity using high resolution SNP microarray analysis in 117 patients referred for cytogenetic analysis and impact on clinical practice. Journal of Medical Genetics, 2008, 46, 123-131.	1.5	61
232	A review of known imprinting syndromes and their association with assisted reproduction technologies. Human Reproduction, 2008, 23, 2826-2834.	0.4	226
233	PGD gender selection for non-Mendelian disorders with unequal sex incidence. Human Reproduction, 2008, 23, 729-734.	0.4	19
234	Increased genetic counseling support improves communication of genetic information in families. Genetics in Medicine, 2008, 10, 167-172.	1.1	94

#	Article	IF	Citations
235	Obesity, hypothyroidism, craniosynostosis, cardiac hypertrophy, colitis, and developmental delay: A novel syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 114-118.	0.7	3
236	Two siblings with 46,XY DSD, congenital adrenal hypoplasia, aniridia, craniofacial, and skeletal abnormalities and intrauterine growth retardation: A new syndrome?. American Journal of Medical Genetics, Part A, 2007, 143A, 2085-2088.	0.7	7
237	Keipert syndrome (Nasodigitoacoustic syndrome) is Xâ€linked and maps to Xq22.2â€“Xq28. American Journal of Medical Genetics, Part A, 2007, 143A, 2236-2241.	0.7	9
238	Juvenile papillomatosis of the breast associated with neurofibromatosis 1. Pediatric Blood and Cancer, 2007, 49, 363-364.	0.8	7
239	Mutations in smooth muscle Î±-actin (ACTA2) lead to thoracic aortic aneurysms and dissections. Nature Genetics, 2007, 39, 1488-1493.	9.4	767
240	Genome-wide association study identifies novel breast cancer susceptibility loci. Nature, 2007, 447, 1087-1093.	13.7	2,165
241	Recurrence risk in Autism Spectrum Disorder: A study of parental knowledge. Journal of Paediatrics and Child Health, 2007, 43, 752-754.	0.4	12
242	Health and developmental outcome of children following prenatal diagnosis of confined placental mosaicism. Prenatal Diagnosis, 2006, 26, 443-448.	1.1	34
243	Tumour surveillance in Beckwithâ€“Wiedemann syndrome and hemihyperplasia: A critical review of the evidence and suggested guidelines for local practice. Journal of Paediatrics and Child Health, 2006, 42, 486-490.	0.4	118
244	Deletion at 14q22-23 indicates a contiguous gene syndrome comprising anophthalmia, pituitary hypoplasia, and ear anomalies. American Journal of Medical Genetics, Part A, 2006, 140A, 1711-1718.	0.7	89
245	DLX3 mutation associated with autosomal dominant amelogenesis imperfecta with taurodontism. American Journal of Medical Genetics, Part A, 2005, 133A, 138-141.	0.7	119
246	Mosaic monosomy of a neocentric ring chromosome maps brachyphalangy and growth hormone deficiency to 13q31.1-13q32.3. American Journal of Medical Genetics, Part A, 2005, 133A, 151-157.	0.7	21
247	Human centromere repositioning "in progress". Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 6542-6547.	3.3	192
248	Building the centromere: from foundation proteins to 3D organization. Trends in Cell Biology, 2004, 14, 359-368.	3.6	154
249	Chromosome size and origin as determinants of the level of CENP-A incorporation into human centromeres. Chromosome Research, 2004, 12, 805-815.	1.0	54
250	Growth, behavior, and clinical findings in 27 patients with Kabuki (Niikawa-Kuroki) syndrome. American Journal of Medical Genetics Part A, 2004, 127A, 118-127.	2.4	71
251	De novo mutations in the mitochondrialND3 gene as a cause of infantile mitochondrial encephalopathy and complex I deficiency. Annals of Neurology, 2004, 55, 58-64.	2.8	164
252	Beckwith-Wiedemann Syndrome and IVF: A Case-Control Study. American Journal of Human Genetics, 2004, 75, 526-528.	2.6	348

#	Article	IF	Citations
253	Morbid obesity and hyperphagia in the WAGR syndrome. Clinical Dysmorphology, 2002, 11, 73-74.	0.1	18
254	Neocentromeres: Role in Human Disease, Evolution, and Centromere Study. American Journal of Human Genetics, 2002, 71, 695-714.	2.6	246
255	Familial digital arthropathy-brachydactyly. American Journal of Medical Genetics Part A, 2002, 108, 235-240.	2.4	10
256	Prenatal cortical hyperostosis (Caffey disease). Pediatric Radiology, 2002, 32, 694-694.	1.1	4
257	Situs inversus totalis and congenital hypoglossia. Clinical Dysmorphology, 2001, 10, 47-50.	0.1	26
258	New variant of familial cerebellar ataxia with hypergonadotropic hypogonadism and sensorineural deafness. American Journal of Medical Genetics Part A, 2001, 99, 29-33.	2.4	33
259	Intermediate form of ablepharon-macrostomia syndrome with CNS abnormalities. American Journal of Medical Genetics Part A, 2001, 103, 252-254.	2.4	9
260	Parental mosaicism of JAG1 mutations in families with Alagille syndrome. European Journal of Human Genetics, 2001, 9, 209-216.	1.4	61
261	Genetic Factors in Athetoid Cerebral Palsy. Journal of Child Neurology, 2001, 16, 793-797.	0.7	12
262	Familial cancers. An overview. Australian Family Physician, 2001, 30, 937-45.	0.5	0
263	Gene therapy. Principles and potential applications. Australian Family Physician, 2001, 30, 953-8.	0.5	0
264	Pseudotrisomy 13 syndrome in siblings. Clinical Dysmorphology, 2000, 9, 115-118.	0.1	5
265	New case of Cole-Carpenter syndrome. , 2000, 92, 273-277.		23
266	Polymicrogyria associated with scalp and limb defects: Variant of Adams-Oliver syndrome. American Journal of Medical Genetics Part A, 2000, 93, 328-334.	2.4	41
267	Autosomal dominant inheritance of scapuloiliac dysostosis. American Journal of Medical Genetics Part A, 2000, 95, 507-509.	2.4	5
268	Analysis of CDKN1C in Beckwith Wiedemann Syndrome. Human Mutation, 2000, 15, 497-508.	1.1	20
269	Encephalocraniocutaneous lipomatosis (Fishman syndrome): A rare neurocutaneous syndrome. Journal of Paediatrics and Child Health, 2000, 36, 603-605.	0.4	34
270	46,XX/46,XY at amniocentesis in a fetus with true hermaphroditism. Journal of Medical Genetics, 1999, 36, 866-9.	1.5	9

#	Article	IF	Citations
271	High frequency of t(12;21) in childhood acute lymphoblastic leukemia detected by RT-PCR. Pathology, 1998, 30, 381-385.	0.3	25
272	Mefloquine resistant P. vivax malaria in PNG. Medical Journal of Australia, 1992, 156, 883-883.	0.8	5