David R Fitzpatrick

List of Publications by Year
in descending order

172	20,287	13865 67	12597 132
papers	citations	h-index	g-index

201	201	201	28758
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Prevalence and architecture of de novo mutations in developmental disorders. Nature, 2017, 542, 433-438.	27.8	1,211
2	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
3	Large-scale discovery of novel genetic causes of developmental disorders. Nature, 2015, 519, 223-228.	27.8	998
4	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
5	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. Lancet, The, 2015, 385, 1305-1314.	13.7	651
6	Constitutional aneuploidy and cancer predisposition caused by biallelic mutations in BUB1B. Nature Genetics, 2004, 36, 1159-1161.	21.4	541
7	Mutations in SOX2 cause anophthalmia. Nature Genetics, 2003, 33, 462-463.	21.4	466
8	Mutations in the Small GTP-ase Late Endosomal Protein RAB7 Cause Charcot-Marie-Tooth Type 2B Neuropathy. American Journal of Human Genetics, 2003, 72, 722-727.	6.2	415
9	Disruption of an AP-2Î± binding site in an IRF6 enhancer is associated with cleft lip. Nature Genetics, 2008, 40, 1341-1347.	21.4	382
10	Paediatric genomics: diagnosing rare disease in children. Nature Reviews Genetics, 2018, 19, 253-268.	16.3	369
11	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	21.4	364
12	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065.	21.4	351
13	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
14	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
15	Human-Specific Gain of Function in a Developmental Enhancer. Science, 2008, 321, 1346-1350.	12.6	330
16	Mutations in the 3Î²-Hydroxysterol Î”24-Reductase Gene Cause Desmosterolosis, an Autosomal Recessive Disorder of Cholesterol Biosynthesis. American Journal of Human Genetics, 2001, 69, 685-694.	6.2	318
17	Mutations in STRA6 Cause a Broad Spectrum of Malformations Including Anophthalmia, Congenital Heart Defects, Diaphragmatic Hernia, Alveolar Capillary Dysplasia, Lung Hypoplasia, and Mental Retardation. American Journal of Human Genetics, 2007, 80, 550-560.	6.2	316
18	Anophthalmia and microphthalmia. Orphanet Journal of Rare Diseases, 2007, 2, 47.	2.7	310

#	Article	IF	Citations
19	Mutations that Cause Osteoglophonic Dysplasia Define Novel Roles for FGFR1 in Bone Elongation. American Journal of Human Genetics, 2005, 76, 361-367.	6.2	295
20	Heterozygous Mutations of OTX2 Cause Severe Ocular Malformations. American Journal of Human Genetics, 2005, 76, 1008-1022.	6.2	266
21	Making new genetic diagnoses with old data: iterative reanalysis and reporting from genome-wide data in 1,133 families with developmental disorders. Genetics in Medicine, 2018, 20, 1216-1223.	2.4	255
22	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	251
23	Identification of SATB2 as the cleft palate gene on 2q32-q33. Human Molecular Genetics, 2003, 12, 2491-2501.	2.9	248
24	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. Nature, 2018, 562, 268-271.	27.8	246
25	De novo mutations in regulatory elements in neurodevelopmental disorders. Nature, 2018, 555, 611-616.	27.8	232
26	3q29 Microdeletion Syndrome: Clinical and Molecular Characterization of a New Syndrome. American Journal of Human Genetics, 2005, 77, 154-160.	6.2	228
27	Cornelia de Lange syndrome: Clinical review, diagnostic and scoring systems, and anticipatory guidance. American Journal of Medical Genetics, Part A, 2007, 143A, 1287-1296.	1.2	223
28	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. Nature Reviews Genetics, 2018, 19, 649-666.	16.3	223
29	The genetic architecture of microphthalmia, anophthalmia and coloboma. European Journal of Medical Genetics, 2014, 57, 369-380.	1.3	213
30	Medical Sequencing of Candidate Genes for Nonsyndromic Cleft Lip and Palate. PLoS Genetics, 2005, 1, e64.	3.5	212
31	Fine Tuning of Craniofacial Morphology by Distant-Acting Enhancers. Science, 2013, 342, 1241006.	12.6	209
32	Loss-of-function mutations in SMARCE1 cause an inherited disorder of multiple spinal meningiomas. Nature Genetics, 2013, 45, 295-298.	21.4	208
33	Mutations in SOX2 cause anophthalmia-esophageal-genital (AEG) syndrome. Human Molecular Genetics, 2006, 15, 1413-1422.	2.9	202
34	Variant detection sensitivity and biases in whole genome and exome sequencing. BMC Bioinformatics, 2014, 15, 247.	2.6	197
35	SOX2 anophthalmia syndrome. American Journal of Medical Genetics, Part A, 2005, 135A, 1-7.	1.2	194
36	A Chromosomal Deletion Map of Human Malformations. American Journal of Human Genetics, 1998, 63, 1153-1159.	6.2	170

#	Article	IF	Citations
37	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	12.6	158
38	Clinical phenotype of desmosterolosis. , 1998, 75, 145-152.		157
39	Transcriptome analysis of human autosomal trisomy. Human Molecular Genetics, 2002, 11, 3249-3256.	2.9	150
40	Long-range regulation at the SOX9 locus in development and disease. Journal of Medical Genetics, 2009, 46, 649-656.	3.2	148
41	The Gene for Cherubism Maps to Chromosome 4p16.3. American Journal of Human Genetics, 1999, 65, 151-157.	6.2	145
42	Mapping of Deletion and Translocation Breakpoints in 1q44 Implicates the Serine/Threonine Kinase AKT3 in Postnatal Microcephaly and Agenesis of the Corpus Callosum. American Journal of Human Genetics, 2007, 81, 292-303.	6.2	144
43	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	3.2	141
44	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	21.4	133
45	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	21.4	131
46	A Chromosomal Duplication Map of Malformations: Regions of Suspected Haplo- and Triplolethalityâ€”and Tolerance of Segmental Aneuploidyâ€”in Humans. American Journal of Human Genetics, 1999, 64, 1702-1708.	6.2	130
47	Diagnostically relevant facial gestalt information from ordinary photos. ELife, 2014, 3, e02020.	6.0	129
48	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.	2.9	120
49	Clinical and molecular genetic features of ARC syndrome. Human Genetics, 2006, 120, 396-409.	3.8	118
50	Mutations in CEP57 cause mosaic variegated aneuploidy syndrome. Nature Genetics, 2011, 43, 527-529.	21.4	117
51	Mutations in autism susceptibility candidate 2 (AUTS2) in patients with mental retardation. Human Genetics, 2007, 121, 501-509.	3.8	116
52	Enhancer-adoption as a mechanism of human developmental disease. Human Mutation, 2011, 32, 1492-1499.	2.5	103
53	Gain-of-Function Mutations of ARHGAP31, a Cdc42/Rac1 GTPase Regulator, Cause Syndromic Cutis Aplasia and Limb Anomalies. American Journal of Human Genetics, 2011, 88, 574-585.	6.2	100
54	De novo, heterozygous, lossâ€ofâ€function mutations in SYNGAP1 cause a syndromic form of intellectual disability. American Journal of Medical Genetics, Part A, 2015, 167, 2231-2237.	1.2	96

#	Article	IF	Citations
55	BRD4 interacts with NIPBL and BRD4 is mutated in a Cornelia de Langeâ€“like syndrome. Nature Genetics, 2018, 50, 329-332.	21.4	96
56	Developmental eye disorders. Current Opinion in Genetics and Development, 2005, 15, 348-353.	3.3	95
57	Heterozygous Loss-of-Function Mutations in YAP1 Cause Both Isolated and Syndromic Optic Fissure Closure Defects. American Journal of Human Genetics, 2014, 94, 295-302.	6.2	93
58	CGG-Repeat Expansion in the DIP2B Gene Is Associated with the Fragile Site FRA12A on Chromosome 12q13.1. American Journal of Human Genetics, 2007, 80, 221-231.	6.2	92
59	B56Î´-related protein phosphatase 2A dysfunction identified in patients with intellectual disability. Journal of Clinical Investigation, 2015, 125, 3051-3062.	8.2	91
60	Xq28 duplication presenting with intestinal and bladder dysfunction and a distinctive facial appearance. European Journal of Human Genetics, 2009, 17, 434-443.	2.8	87
61	Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature Communications, 2019, 10, 2373.	12.8	86
62	Role of SOX2 Mutations in Human Hippocampal Malformations and Epilepsy. Epilepsia, 2006, 47, 534-542.	5.1	85
63	BCOR analysis in patients with OFCD and Lenz microphthalmia syndromes, mental retardation with ocular anomalies, and cardiac laterality defects. European Journal of Human Genetics, 2009, 17, 1325-1335.	2.8	85
64	Loss of the BMP Antagonist, SMOC-1, Causes Ophthalmo-Acromelic (Waardenburg Anophthalmia) Syndrome in Humans and Mice. PLoS Genetics, 2011, 7, e1002114.	3.5	81
65	A Restricted Repertoire of De Novo Mutations in ITPR1 Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. American Journal of Human Genetics, 2016, 98, 981-992.	6.2	81
66	Clinical and mutation analysis of 51 probands with anophthalmia and/or severe microphthalmia from a single center. Molecular Genetics & Genomic Medicine, 2013, 1, 15-31.	1.2	79
67	Monoallelic and Biallelic Mutations in MAB21L2 Cause a Spectrum of Major Eye Malformations. American Journal of Human Genetics, 2014, 94, 915-923.	6.2	79
68	Transcriptional consequences of autosomal trisomy: primary gene dosage with complex downstream effects. Trends in Genetics, 2005, 21, 249-253.	6.7	77
69	Clinical and mutation data in 12 patients with the clinical diagnosis of Nager syndrome. Human Genetics, 2013, 132, 885-898.	3.8	77
70	The clinical significance of small copy number variants in neurodevelopmental disorders. Journal of Medical Genetics, 2014, 51, 677-688.	3.2	72
71	De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462.	2.5	72
72	Miller (Genee-Wiedemann) syndrome represents a clinically and biochemically distinct subgroup of postaxial acrofacial dysostosis associated with partial deficiency of DHODH. Human Molecular Genetics, 2012, 21, 3969-3983.	2.9	70

#	Article	IF	Citations
73	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	5.5	70
74	Identification of Novel Craniofacial Regulatory Domains Located far Upstream ofSOX9and Disrupted in Pierre Robin Sequence. Human Mutation, 2014, 35, 1011-1020.	2.5	69
75	A rare de novo nonsense mutation in OTX2 causes early onset retinal dystrophy and pituitary dysfunction. Molecular Vision, 2009, 15, 2442-7.	1.1	68
76	Cardiac Genetic Predisposition in SuddenÂInfant Death Syndrome. Journal of the American College of Cardiology, 2018, 71, 1217-1227.	2.8	66
77	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	8.2	65
78	A Locus for Isolated Cleft Palate, Located on Human Chromosome 2q32. American Journal of Human Genetics, 1999, 65, 387-396.	6.2	63
79	Dysfunction of NaV1.4, a skeletal muscle voltage-gated sodium channel, in sudden infant death syndrome: a case-control study. Lancet, The, 2018, 391, 1483-1492.	13.7	63
80	The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. Orphanet Journal of Rare Diseases, 2013, 8, 63.	2.7	60
81	An excess of chromosome 1 breakpoints in male infertility. European Journal of Human Genetics, 2004, 12, 993-1000.	2.8	56
82	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. American Journal of Human Genetics, 2017, 100, 650-658.	6.2	56
83	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	2.5	54
84	Transforming growth factor betas in mammalian embryogenesis. Progress in Growth Factor Research, 1990, 2, 153-168.	1.6	51
85	The Molecular Basis of Malonyl-CoA Decarboxylase Deficiency. American Journal of Human Genetics, 1999, 65, 318-326.	6.2	51
86	Recurrence ofSOX2 anophthalmia syndrome with gonosomal mosaicism in a phenotypically normal mother. American Journal of Medical Genetics, Part A, 2006, 140A, 636-639.	1.2	51
87	Disruption of SATB2 or its long-range cis-regulation by SOX9 causes a syndromic form of Pierre Robin sequence. Human Molecular Genetics, 2014, 23, 2569-2579.	2.9	51
88	The genetic architecture of aniridia and Gillespie syndrome. Human Genetics, 2019, 138, 881-898.	3.8	51
89	Î”3,5-Î”2,4-Dienoyl-CoA Isomerase from Rat Liver. Journal of Biological Chemistry, 1998, 273, 349-355.	3.4	50
90	Clinical features associated with CTNNB1 de novo loss of function mutations in ten individuals. European Journal of Medical Genetics, 2017, 60, 130-135.	1.3	47

#	Article	IF	Citations
91	Warburg Micro syndrome is caused by RAB18 deficiency or dysregulation. Open Biology, 2015, 5, 150047.	3.6	46
92	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
93	Novel de novo <scp>EEF</scp>1A2 missense mutations causing epilepsy and intellectual disability. Molecular Genetics & Genomic Medicine, 2016, 4, 465-474.	1.2	44
94	Molecular and clinical characterization of de novo and familial cases with microduplication 3q29: guidelines for copy number variation case reporting. Cytogenetic and Genome Research, 2008, 123, 65-78.	1.1	43
95	Contribution of retrotransposition to developmental disorders. Nature Communications, 2019, 10, 4630.	12.8	43
96	Recurrent heterozygous PAX6 missense variants cause severe bilateral microphthalmia via predictable effects on DNAâ€“protein interaction. Genetics in Medicine, 2020, 22, 598-609.	2.4	43
97	PUF60 variants cause a syndrome of ID, short stature, microcephaly, coloboma, craniofacial, cardiac, renal and spinal features. European Journal of Human Genetics, 2017, 25, 552-559.	2.8	42
98	FISH Mapping of De Novo Apparently Balanced Chromosome Rearrangements Identifies Characteristics Associated with Phenotypic Abnormality. American Journal of Human Genetics, 2008, 82, 916-926.	6.2	41
99	FRA2A Is a CGG Repeat Expansion Associated with Silencing of AFF3. PLoS Genetics, 2014, 10, e1004242.	3.5	41
100	The RNA-binding landscape of RBM10 and its role in alternative splicing regulation in models of mouse early development. RNA Biology, 2017, 14, 45-57.	3.1	41
101	Tandem Mass Spectrometric Determination of Malonylcarnitine: Diagnosis and Neonatal Screening of Malonyl-CoA Decarboxylase Deficiency. Clinical Chemistry, 2003, 49, 660-662.	3.2	40
102	A secreted WNT-ligand-binding domain of FZD5 generated by a frameshift mutation causes autosomal dominant coloboma. Human Molecular Genetics, 2016, 25, 1382-1391.	2.9	40
103	The 12q14 microdeletion syndrome: six new cases confirming the role of HMGA2 in growth. European Journal of Human Genetics, 2011, 19, 534-539.	2.8	39
104	Managing clinically significant findings in research: the UK10K example. European Journal of Human Genetics, 2014, 22, 1100-1104.	2.8	38
105	MLYCD mutation analysis: Evidence for protein mistargeting as a cause of MLYCD deficiency. Human Mutation, 2003, 22, 288-300.	2.5	37
106	Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431.	2.8	37
107	MCS9.7 enhancer activity is highly, but not completely, associated with expression of Irf6 and p63. Developmental Dynamics, 2012, 241, 340-349.	1.8	35
108	Heterozygous truncation mutations of the <scp>SMC</scp>1A gene cause a severe early onset epilepsy with cluster seizures in females: Detailed phenotyping of 10 new cases. Epilepsia, 2017, 58, 565-575.	5.1	35

#	Article	IF	Citations
109	A Male with Unilateral Microphthalmia Reveals a Role for TMX3 in Eye Development. PLoS ONE, 2010, 5, e10565.	2.5	34
110	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. Genome Research, 2019, 29, 1047-1056.	5.5	34
111	Inherited PAX6, NF1 and OTX2 mutations in a child with microphthalmia and aniridia. European Journal of Human Genetics, 2007, 15, 898-901.	2.8	33
112	Folate and Clefts of the Lip and Palateâ€"A U.K.-Based Case-Control Study: Part II: Biochemical and Genetic Analysis. Cleft Palate-Craniofacial Journal, 2008, 45, 428-438.	0.9	33
113	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	12.8	33
114	The role of TGF-?s in mammalian development and neoplasia. Molecular Reproduction and Development, 1992, 32, 127-135.	2.0	31
115	Isolation and Characterization of Rat and Human cDNAs Encoding a Novel Putative Peroxisomal Enoyl-CoA Hydratase. Genomics, 1995, 27, 457-466.	2.9	31
116	Bilateral Renal Agenesis/Hypoplasia/Dysplasia (BRAHD): Postmortem Analysis of 45 Cases with Breakpoint Mapping of Two De Novo Translocations. PLoS ONE, 2010, 5, e12375.	2.5	31
117	Esrrg functions in early branch generation of the ureteric bud and is essential for normal development of the renal papilla. Human Molecular Genetics, 2011, 20, 917-926.	2.9	31
118	Long-range evolutionary constraints reveal cis-regulatory interactions on the human X chromosome. Nature Communications, 2015, 6, 6904.	12.8	31
119	Delineation of an estimated 6.7â€‰MB candidate interval for an anophthalmia gene at 3q26.33-q28 and description of the syndrome associated with visible chromosome deletions of this region. European Journal of Human Genetics, 2002, 10, 807-812.	2.8	29
120	Absence ofSIX6Mutations in Microphthalmia, Anophthalmia, and Coloboma. , 2004, 45, 3871.		28
121	Folate and Clefts of the Lip and Palateâ€"A U.K.-Based Case-Control Study: Part I: Dietary and Supplemental Folate. Cleft Palate-Craniofacial Journal, 2008, 45, 420-427.	0.9	28
122	A CGG-Repeat Expansion Mutation inZNF713Causes FRA7A: Association with Autistic Spectrum Disorder in two Families. Human Mutation, 2014, 35, n/a-n/a.	2.5	28
123	NAA10 polyadenylation signal variants cause syndromic microphthalmia. Journal of Medical Genetics, 2019, 56, 444-452.	3.2	28
124	Familial recurrence of SOX2 anophthalmia syndrome: Phenotypically normal mother with two affected daughters. American Journal of Medical Genetics, Part A, 2008, 146A, 2794-2798.	1.2	27
125	The molecular basis of the folate-sensitive fragile site FRA11A at 11q13. Cytogenetic and Genome Research, 2007, 119, 9-14.	1.1	25
126	A Novel Oculo-Skeletal syndrome with intellectual disability caused by a particular MAB21L2 mutation. European Journal of Medical Genetics, 2015, 58, 387-391.	1.3	25

#	Article	IF	Citations
127	Novel PEX11B Mutations Extend the Peroxisome Biogenesis Disorder 14B Phenotypic Spectrum and Underscore Congenital Cataract as an Early Feature. , 2017, 58, 594.		25
128	ITPase deficiency causes a Martsolf-like syndrome with a lethal infantile dilated cardiomyopathy. PLoS Genetics, 2019, 15, e1007605.	3.5	25
129	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
130	FRA18C: a new aphidicolin-inducible fragile site on chromosome 18q22, possibly associated with in vivo chromosome breakage. Journal of Medical Genetics, 2007, 44, 347-352.	3.2	24
131	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	3.8	24
132	Returning genome sequences to research participants: Policy and practice. Wellcome Open Research, 2017, 2, 15.	1.8	24
133	Interstitial deletion of the long arm of chromosome 5 in a boy with multiple congenital anomalies and mental retardation: Molecular characterization of the deleted region to 5q22.3q23.3. , 2005, 132A, 402-410.		22
134	GATAD2B-associatedneurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-relateddisorder. Genetics in Medicine, 2020, 22, 878-888.	2.4	22
135	A recurrent de novo mutation inACTG1causes isolated ocular coloboma. Human Mutation, 2017, 38, 942-946.	2.5	21
136	NALCN Dysfunction as a Cause of Disordered Respiratory Rhythm With Central Apnea. Pediatrics, 2018, 141, S485-S490.	2.1	21
137	Genotypeâ€“phenotype correlations in Cornelia de Lange syndrome: Behavioral characteristics and changes with age. American Journal of Medical Genetics, Part A, 2017, 173, 1566-1574.	1.2	20
138	Filling in the gaps in cranial suture biology. Nature Genetics, 2013, 45, 231-232.	21.4	19
139	A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX. European Journal of Medical Genetics, 2014, 57, 587-595.	1.3	18
140	Genetic Interactions in Nonsyndromic Orofacial Clefts in Europeâ€”EUROCRAN Study. Cleft Palate-Craniofacial Journal, 2017, 54, 623-630.	0.9	18
141	CorneliaÂde Lange syndrome-associated mutations cause a DNA damage signalling and repair defect. Nature Communications, 2021, 12, 3127.	12.8	18
142	Iris coloboma and a microdeletion of chromosome 22: del(22)(q11.22). British Journal of Ophthalmology, 2002, 86, 1316-1316.	3.9	17
143	Evaluating variants classified as pathogenic in ClinVar in the DDD Study. Genetics in Medicine, 2021, 23, 571-575.	2.4	16
144	Expansion of Ocular Phenotypic Features Associated With Mutations inADAMTS18. JAMA Ophthalmology, 2014, 132, 996.	2.5	15

#	Article	IF	Citations
145	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
146	Decreased cholesterol synthesis as a possible aetiological factor in malformations of Trisomy 18. European Journal of Medical Genetics, 2006, 49, 195-199.	1.3	13
147	Disruption of ST5 is associated with mental retardation and multiple congenital anomalies. Journal of Medical Genetics, 2010, 47, 91-98.	3.2	12
148	Detecting cryptic clinically relevant structural variation in exome-sequencing data increases diagnostic yield for developmental disorders. American Journal of Human Genetics, 2021, 108, 2186-2194.	6.2	12
149	Carey-Fineman-Ziter (CFZ) syndrome: Report on affected sibs. , 1999, 82, 110-113.		11
150	A Trans-Acting Protein Effect Causes Severe Eye Malformation in the Mp Mouse. PLoS Genetics, 2013, 9, e1003998.	3.5	11
151	Chromosome analysis: what and when to request. Archives of Disease in Childhood, 2005, 90, 1264-1269.	1.9	10
152	A3243G mitochondrial mutation associated with polymicrogyria. Developmental Medicine and Child Neurology, 2003, 45, 704-8.	2.1	9
153	Recurrences of trisomy 18 and trisomy 13 after trisomy 21. Human Genetics, 1989, 82, 301-301.	3.8	8
154	Beckwithâ€“Wiedemannâ€like macroglossia and 18q23 haploinsufficiency. American Journal of Medical Genetics, Part A, 2007, 143A, 2796-2803.	1.2	8
155	Definition of 5q11.2 microdeletion syndrome reveals overlap with CHARGE syndrome and 22q11 deletion syndrome phenotypes. American Journal of Medical Genetics, Part A, 2014, 164, 2843-2848.	1.2	8
156	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	6.2	8
157	Status dystonicus in two patients with SOX2â€anophthalmia syndrome and nonsense mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3048-3050.	1.2	7
158	An atypical facial appearance and growth pattern in a child with Cornelia de Lange Syndrome. Clinical Dysmorphology, 2012, 21, 22-23.	0.3	6
159	MS-PCR assay to detect 677Câ†'T mutation in the 5,10-methylenetetrahydrofolate reductase gene. Journal of Inherited Metabolic Disease, 1998, 21, 694-695.	3.6	5
160	â€˜Crommelin-typeâ€™ symmetrical tetramelic reduction deformity: a new case and breakpoint mapping of a reported case with de-novo t(2;12)(p25.1;q23.3). Clinical Dysmorphology, 2010, 19, 5-13.	0.3	4
161	Mutational bias in spermatogonia impacts the anatomy of regulatory sites in the human genome. Genome Research, 2021, 31, 1994-2007.	5.5	4
162	Cornelia de Lange syndrome: Clinical review, diagnostic and scoring systems, and anticipatory guidance Am J Med Genet Part A 143A:1287-1296. American Journal of Medical Genetics, Part A, 2008, 146A, 2713-2713.	1.2	3

#	Article	IF	Citations
163	Resequencing at scale in neurodevelopmental disorders. Nature Genetics, 2017, 49, 488-489.	21.4	3
164	Genomically Aided Diagnosis of Severe Developmental Disorders. Annual Review of Genomics and Human Genetics, 2020, 21, 327-349.	6.2	3
165	Identification and functional modelling of plausibly causative cis-regulatory variants in a highly-selected cohort with X-linked intellectual disability. PLoS ONE, 2021, 16, e0256181.	2.5	3
166	Genetic Metabolic Disease. , 2007, , 162-183.		3
167	A new complementation assay for peroxisome-deficient cell lines. Journal of Inherited Metabolic Disease, 1996, 19, 94-95.	3.6	2
168	A case of Menkes syndrome with pyloric stenosis: An effect of copper deficiency on NOS1?. Journal of Inherited Metabolic Disease, 1999, 22, 197-198.	3.6	2
169	De novo translocation (1; 2)(q32; p25) associated with bilateral renal dysplasia. Clinical Genetics, 2003, 63, 239-240.	2.0	2
170	Developmental biology of the eye. , 2013, , 16-22.		1
171	Characterization of a cDNA Library Enriched for a Novel Peroxisomal Gene. Annals of the New York Academy of Sciences, 1996, 804, 739-741.	3.8	0
172	Mutations in SOX2 cause anophthalmiaâ€“esophagealâ€“genital (AEG) syndrome. Human Molecular Genetics, 2006, 15, 2030-2030.	2.9	0