Andrew Oliver Mungo Wilkie

List of Publications by Year
in descending order

193 papers	18,072 citations	15504 65 h-index	15266 126 g-index
258 all docs	258 docs citations	258 times ranked	19331 citing authors

#	Article	IF	Citations
1	Integrating mapping-, assembly- and haplotype-based approaches for calling variants in clinical sequencing applications. Nature Genetics, 2014, 46, 912-918.	21.4	937
2	Apert syndrome results from localized mutations of FGFR2 and is allelic with Crouzon syndrome. Nature Genetics, 1995, 9, 165-172.	21.4	892
3	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. Nucleic Acids Research, 2014, 42, D966-D974.	14.5	698
4	The detection of subtelomeric chromosomal rearrangements in idiopathic mental retardation. Nature Genetics, 1995, 9, 132-140.	21.4	482
5	Identical mutations in the FGFR2 gene cause both Pfeiffer and Crouzon syndrome phenotypes. Nature Genetics, 1995, 9, 173-176.	21.4	450
6	Growth of the normal skull vault and its alteration in craniosynostosis: insights from human genetics and experimental studies. Journal of Anatomy, 2005, 207, 637-653.	1.5	382
7	Genetics of craniofacial development and malformation. Nature Reviews Genetics, 2001, 2, 458-468.	16.3	380
8	Localized mutations in the gene encoding the cytoskeletal protein filamin A cause diverse malformations in humans. Nature Genetics, 2003, 33, 487-491.	21.4	375
9	Craniosynostosis. European Journal of Human Genetics, 2011, 19, 369-376.	2.8	367
10	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
11	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
12	Mutations of ephrin-B1 (EFNB1), a marker of tissue boundary formation, cause craniofrontonasal syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 8652-8657.	7.1	320
13	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. Nature Genetics, 2015, 47, 717-726.	21.4	310
14	A truncated human chromosome 16 associated with Î± thalassaemia is stabilized by addition of telomeric repeat (TTAGGG)n. Nature, 1990, 346, 868-871.	27.8	300
15	Functional haploinsufficiency of the human homeobox gene MSX2 causes defects in skull ossification. Nature Genetics, 2000, 24, 387-390.	21.4	295
16	Paternal Age Effect Mutations and Selfish Spermatogonial Selection: Causes and Consequences for Human Disease. American Journal of Human Genetics, 2012, 90, 175-200.	6.2	294
17	Evidence for Selective Advantage of Pathogenic FGFR2 Mutations in the Male Germ Line. Science, 2003, 301, 643-646.	12.6	291
18	Exclusive paternal origin of new mutations in Apert syndrome. Nature Genetics, 1996, 13, 48-53.	21.4	285

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19	Recessive Robinow syndrome, allelic to dominant brachydactyly type B, is caused by mutation of ROR2. Nature Genetics, 2000, 25, 419-422.	21.4	277
20	Oculofaciocardiodental and Lenz microphthalmia syndromes result from distinct classes of mutations in BCOR. Nature Genetics, 2004, 36, 411-416.	21.4	272
21	Functions of fibroblast growth factors and their receptors. Current Biology, 1995, 5, 500-507.	3.9	253
22	Genomic Screening of Fibroblast Growth-Factor Receptor 2 Reveals a Wide Spectrum of Mutations in Patients with Syndromic Craniosynostosis. American Journal of Human Genetics, 2002, 70, 472-486.	6.2	238
23	Prevalence and Complications of Single-Gene and Chromosomal Disorders in Craniosynostosis. Pediatrics, 2010, 126, e391-e400.	2.1	236
24	RAB23 Mutations in Carpenter Syndrome Imply an Unexpected Role for Hedgehog Signaling in Cranial-Suture Development and Obesity. American Journal of Human Genetics, 2007, 80, 1162-1170.	6.2	229
25	De Novo Alu-Element Insertions in FGFR2 Identify a Distinct Pathological Basis for Apert Syndrome. American Journal of Human Genetics, 1999, 64, 446-461.	6.2	225
26	Bad bones, absent smell, selfish testes: The pleiotropic consequences of human FGF receptor mutations. Cytokine and Growth Factor Reviews, 2005, 16, 187-203.	7.2	223
27	A Genetic-Pathophysiological Framework for Craniosynostosis. American Journal of Human Genetics, 2015, 97, 359-377.	6.2	213
28	Dominant mutations in ROR2, encoding an orphan receptor tyrosine kinase, cause brachydactyly type B. Nature Genetics, 2000, 24, 275-278.	21.4	210
29	Paternal Origin of FGFR2 Mutations in Sporadic Cases of Crouzon Syndrome and Pfeiffer Syndrome. American Journal of Human Genetics, 2000, 66, 768-777.	6.2	191
30	A Comprehensive Screen for TWIST Mutations in Patients with Craniosynostosis Identifies a New Microdeletion Syndrome of Chromosome Band 7p21.1. American Journal of Human Genetics, 1998, 63, 1282-1293.	6.2	187
31	FGFs, their receptors, and human limb malformations: Clinical and molecular correlations. American Journal of Medical Genetics Part A, 2002, 112, 266-278.	2.4	186
32	Cell mixing at a neural crest-mesoderm boundary and deficient ephrin-Eph signaling in the pathogenesis of craniosynostosis. Human Molecular Genetics, 2006, 15, 1319-1328.	2.9	184
33	Activating mutations in FGFR3 and HRAS reveal a shared genetic origin for congenital disorders and testicular tumors. Nature Genetics, 2009, 41, 1247-1252.	21.4	184
34	Mutations in TCF12, encoding a basic helix-loop-helix partner of TWIST1, are a frequent cause of coronal craniosynostosis. Nature Genetics, 2013, 45, 304-307.	21.4	181
35	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	12.6	178
36	Stable length polymorphism of up to 260 kb at the tip of the short arm of human chromosome 16. Cell, 1991, 64, 595-606.	28.9	169

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37	Distinct Mutations in the Receptor Tyrosine Kinase Gene ROR2 Cause Brachydactyly Type B. American Journal of Human Genetics, 2000, 67, 822-831.	6.2	166
38	Inactivation of IL11 Signaling Causes Craniosynostosis, Delayed Tooth Eruption, and Supernumerary Teeth. American Journal of Human Genetics, 2011, 89, 67-81.	6.2	164
39	A biallelic mutation in IL6ST encoding the GP130 co-receptor causes immunodeficiency and craniosynostosis. Journal of Experimental Medicine, 2017, 214, 2547-2562.	8.5	158
40	Epidermal mosaicism producing localised acne: somatic mutation in FGFR2. Lancet, The, 1998, 352, 704-705.	13.7	151
41	Haploinsufficiency of the human homeobox gene ALX4 causes skull ossification defects. Nature Genetics, 2001, 27, 17-18.	21.4	142
42	Clinical genetics of craniosynostosis. Current Opinion in Pediatrics, 2017, 29, 622-628.	2.0	142
43	Reduced dosage of ERF causes complex craniosynostosis in humans and mice and links ERK1/2 signaling to regulation of osteogenesis. Nature Genetics, 2013, 45, 308-313.	21.4	141
44	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
45	Gain-of-function amino acid substitutions drive positive selection of FGFR2 mutations in human spermatogonia. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 6051-6056.	7.1	125
46	New insights into craniofacial malformations. Human Molecular Genetics, 2015, 24, R50-R59.	2.9	122
47	A genome-wide association study identifies susceptibility loci for nonsyndromic sagittal craniosynostosis near BMP2 and within BBS9. Nature Genetics, 2012, 44, 1360-1364.	21.4	120
48	Next-generation sequencing (NGS) as a diagnostic tool for retinal degeneration reveals a much higher detection rate in early-onset disease. European Journal of Human Genetics, 2013, 21, 274-280.	2.8	119
49	Frontorhiny, a Distinctive Presentation of Frontonasal Dysplasia Caused by Recessive Mutations in the ALX3 Homeobox Gene. American Journal of Human Genetics, 2009, 84, 698-705.	6.2	118
50	Potential Gene Conversion and Source Genes for Recently Integrated Alu Elements. Genome Research, 2000, 10, 1485-1495.	5.5	108
51	Diagnostic value of exome and whole genome sequencing in craniosynostosis. Journal of Medical Genetics, 2017, 54, 260-268.	3.2	107
52	Duplications of noncoding elements 5â€² of SOX9 are associated with brachydactyly-anonychia. Nature Genetics, 2009, 41, 862-863.	21.4	105
53	A Noncoding Expansion in EIF4A3 Causes Richieri-Costa-Pereira Syndrome, a Craniofacial Disorder Associated with Limb Defects. American Journal of Human Genetics, 2014, 94, 120-128.	6.2	99
54	Missense Mutations in the Homeodomain of HOXD13 Are Associated with Brachydactyly Types D and E. American Journal of Human Genetics, 2003, 72, 984-997.	6.2	96

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55	The Origin of EFNB1 Mutations in Craniofrontonasal Syndrome: Frequent Somatic Mosaicism and Explanation of the Paucity of Carrier Males. American Journal of Human Genetics, 2006, 78, 999-1010.	6.2	96
56	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
57	Mutations in CDC45 , Encoding an Essential Component of the Pre-initiation Complex, Cause Meier-Gorlin Syndrome and Craniosynostosis. American Journal of Human Genetics, 2016, 99, 125-138.	6.2	92
58	OCT2, SSX and SAGE1 reveal the phenotypic heterogeneity of spermatocytic seminoma reflecting distinct subpopulations of spermatogonia. Journal of Pathology, 2011, 224, 473-483.	4.5	79
59	â€œSelfish Spermatogonial Selectionâ€: A Novel Mechanism for the Association Between Advanced Paternal Age and Neurodevelopmental Disorders. American Journal of Psychiatry, 2013, 170, 599-608.	7.2	79
60	Clinical dividends from the molecular genetic diagnosis of craniosynostosisâ€. American Journal of Medical Genetics, Part A, 2007, 143A, 1941-1949.	1.2	75
61	A variant in the sonic hedgehog regulatory sequence (ZRS) is associated with triphalangeal thumb and deregulates expression in the developing limb. Human Molecular Genetics, 2008, 17, 2417-2423.	2.9	74
62	Selective loss of function variants in IL6ST cause Hyper-IgE syndrome with distinct impairments of T-cell phenotype and function. Haematologica, 2019, 104, 609-621.	3.5	74
63	Skeletal analysis of the Fgfr3^{P244R} mouse, a genetic model for the Muenke craniosynostosis syndrome. Developmental Dynamics, 2009, 238, 331-342.	1.8	73
64	Mutations in Multidomain Protein MEGF8 Identify a Carpenter Syndrome Subtype Associated with Defective Lateralization. American Journal of Human Genetics, 2012, 91, 897-905.	6.2	72
65	HUWE1 variants cause dominant X-linked intellectual disability: a clinical study of 21 patients. European Journal of Human Genetics, 2018, 26, 64-74.	2.8	72
66	Reoperation for Intracranial Hypertension in TWIST1-Confirmed Saethre-Chotzen Syndrome: A 15-Year Review. Plastic and Reconstructive Surgery, 2009, 123, 1801-1810.	1.4	70
67	Contributions of intrinsic mutation rate and selfish selection to levels of de novo HRAS mutations in the paternal germline. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 20152-20157.	7.1	70
68	A Recurrent Mosaic Mutation in SMO , Encoding the Hedgehog Signal Transducer Smoothened, Is the Major Cause of Curry-Jones Syndrome. American Journal of Human Genetics, 2016, 98, 1256-1265.	6.2	70
69	A novel mutation, Ala315Ser, in FGFR2: a geneâ€“environment interaction leading to craniosynostosis?. European Journal of Human Genetics, 2000, 8, 571-577.	2.8	68
70	Paternal origin of FGFR3 mutations in Muenke-type craniosynostosis. Human Genetics, 2004, 115, 200-207.	3.8	67
71	Enlarged parietal foramina caused by mutations in the homeobox genes ALX4 and MSX2: from genotype to phenotype. European Journal of Human Genetics, 2006, 14, 151-158.	2.8	67
72	Frontometaphyseal dysplasia: Mutations inFLNA and phenotypic diversity. American Journal of Medical Genetics, Part A, 2006, 140A, 1726-1736.	1.2	67

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73	Cellular interference in craniofrontonasal syndrome: males mosaic for mutations in the X-linked EFNB1 gene are more severely affected than true hemizygotes. Human Molecular Genetics, 2013, 22, 1654-1662.	2.9	66
74	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. American Journal of Human Genetics, 2018, 102, 468-479.	6.2	63
75	Clinical dividends from the molecular genetic diagnosis of craniosynostosis. American Journal of Medical Genetics, Part A, 2006, 140A, 2631-2639.	1.2	60
76	Skeletal development is regulated by fibroblast growth factor receptor 1 signalling dynamics. Development (Cambridge), 2004, 131, 325-335.	2.5	58
77	Hearing loss in a mouse model of Muenke syndrome. Human Molecular Genetics, 2009, 18, 43-50.	2.9	57
78	Gain-of-Function Mutations in ZIC1 Are Associated with Coronal Craniosynostosis and Learning Disability. American Journal of Human Genetics, 2015, 97, 378-388.	6.2	56
79	Selfish mutations dysregulating RAS-MAPK signaling are pervasive in aged human testes. Genome Research, 2018, 28, 1779-1790.	5.5	56
80	Rare mutations ofFGFR2causing apert syndrome: identification of the first partial gene deletion, and anAluelement insertion from a new subfamily. Human Mutation, 2009, 30, 204-211.	2.5	55
81	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. American Journal of Human Genetics, 2016, 99, 392-406.	6.2	52
82	FGFR3 P250R Mutation Increases the Risk of Reoperation in Apparent â€˜Nonsyndromicâ€™ Coronal Craniosynostosis. Journal of Craniofacial Surgery, 2005, 16, 347-352.	0.7	51
83	Craniosynostosis. Current Opinion in Neurology, 1996, 9, 146.	3.6	48
84	Expanding the phenotype of craniofrontonasal syndrome: two unrelated boys with EFNB1 mutations and congenital diaphragmatic hernia. European Journal of Human Genetics, 2006, 14, 884-887.	2.8	48
85	The developing mouse coronal suture at single-cell resolution. Nature Communications, 2021, 12, 4797.	12.8	48
86	The genetics of mental retardation. British Medical Bulletin, 1996, 52, 453-464.	6.9	46
87	Visualizing the origins of selfish de novo mutations in individual seminiferous tubules of human testes. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 2454-2459.	7.1	45
88	Alx4 and Msx2 play phenotypically similar and additive roles in skull vault differentiation. Journal of Anatomy, 2004, 204, 487-499.	1.5	44
89	Parietal foramina with cleidocranial dysplasia is caused by mutation in MSX2. European Journal of Human Genetics, 2003, 11, 892-895.	2.8	42
90	De Novo Missense Substitutions in the Gene Encoding CDK8, a Regulator of the Mediator Complex, Cause a Syndromic Developmental Disorder. American Journal of Human Genetics, 2019, 104, 709-720.	6.2	41

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91	Interstitial deletion of 2q associated with craniosynostosis, ocular coloboma, and limb abnormalities: Cytogenetic and molecular investigation. American Journal of Medical Genetics Part A, 1997, 70, 324-327.	2.4	40
92	A survey ofTWIST for mutations in craniosynostosis reveals a variable length polyglycine tract in asymptomatic individuals. Human Mutation, 2001, 18, 535-541.	2.5	39
93	Nonsenseâ€mediated decay and the molecular pathogenesis of mutations in SALL1 and GLI3. American Journal of Medical Genetics, Part A, 2007, 143A, 3150-3160.	1.2	39
94	Missing heritability: paternal age effect mutations and selfish spermatogonia. Nature Reviews Genetics, 2010, 11, 589-589.	16.3	39
95	Postzygotic mutation and germline mosaicism in the otopalatodigital syndrome spectrum disorders. European Journal of Human Genetics, 2006, 14, 549-554.	2.8	38
96	Why study human limb malformations?. Journal of Anatomy, 2003, 202, 27-35.	1.5	37
97	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	6.2	37
98	Heterozygous mutations affecting the protein kinase domain of CDK13 cause a syndromic form of developmental delay and intellectual disability. Journal of Medical Genetics, 2018, 55, 28-38.	3.2	36
99	Whole-genome sequencing of spermatocytic tumors provides insights into the mutational processes operating in the male germline. PLoS ONE, 2017, 12, e0178169.	2.5	36
100	Limited Proteolysis and Proton NMR Spectroscopy of Bacillus stearothermophilusPyruvate Dehydrogenase Multienzyme Complex. FEBS Journal, 1982, 124, 63-69.	0.2	35
101	Germline and somatic mosaicism for FGFR2 mutation in the mother of a child with Crouzon syndrome: Implications for genetic testing in â€œpaternal ageâ€effectâ€•syndromes. American Journal of Medical Genetics, Part A, 2010, 152A, 2067-2073.	1.2	35
102	Efficient use of a 'dead-end' GA 5' splice site in the human fibroblast growth factor receptor genes. EMBO Journal, 2003, 22, 1620-1631.	7.8	34
103	Implications of a Vertex Bulge following Modified Strip Craniectomy for Sagittal Synostosis. Plastic and Reconstructive Surgery, 2008, 122, 217-224.	1.4	34
104	Carpenter syndrome: extended RAB23 mutation spectrum and analysis of nonsenseâ€mediated mRNA decay. Human Mutation, 2011, 32, E2069-78.	2.5	34
105	Raised Intracranial Pressure Is Frequent in Untreated Nonsyndromic Unicoronal Synostosis and Does Not Correlate with Severity of Phenotypic Features. Plastic and Reconstructive Surgery, 2012, 130, 690e-697e.	1.4	34
106	Selfish Spermatogonial Selection: Evidence from an Immunohistochemical Screen in Testes of Elderly Men. PLoS ONE, 2012, 7, e42382.	2.5	32
107	De novoand rare inherited mutations implicate the transcriptional coregulator TCF20/SPBP in autism spectrum disorder. Journal of Medical Genetics, 2014, 51, 737-747.	3.2	31
108	SMAD6 variants in craniosynostosis: genotype and phenotype evaluation. Genetics in Medicine, 2020, 22, 1498-1506.	2.4	31

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109	Laband syndrome. Oral Surgery, Oral Medicine, and Oral Pathology, 1994, 78, 57-63.	0.6	30
110	Cancer drugs to treat birth defects. Nature Genetics, 2007, 39, 1057-1059.	21.4	30
111	Craniosynostosis and Related Limb Anomalies. Novartis Foundation Symposium, 2008, 232, 122-143.	1.1	30
112	Metopic and sagittal synostosis in Greig cephalopolysyndactyly syndrome: five cases with intragenic mutations or complete deletions of GLI3. European Journal of Human Genetics, 2011, 19, 757-762.	2.8	30
113	ACTH receptor mutation in a girl with familial glucocorticoid deficiency. Clinical Genetics, 1998, 53, 57-62.	2.0	30
114	An acceptor splice site mutation in HOXD13 results in variable hand, but consistent foot malformations. American Journal of Medical Genetics Part A, 2003, 121A, 69-74.	2.4	29
115	ERFâ€related craniosynostosis: The phenotypic and developmental profile of a new craniosynostosis syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 615-627.	1.2	29
116	The Gene for Spondyloepiphyseal Dysplasia (SEDL) Maps to Xp22 between DXS16 and DXS92. Genomics, 1993, 18, 100-104.	2.9	28
117	Epidemiology and genetics of craniosynostosis. , 2000, 90, 82-83.		28
118	Truncated SALL1 Impedes Primary Cilia Function in Townes-Brocks Syndrome. American Journal of Human Genetics, 2018, 102, 249-265.	6.2	27
119	Polydactyly in the mouse mutant Doublefoot involves altered Gli3 processing and is caused by a large deletion in cis to Indian hedgehog. Mechanisms of Development, 2008, 125, 517-526.	1.7	26
120	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	2.5	26
121	ImplementationÂof a genomic medicine multi-disciplinary team approach for rare diseaseÂin the clinical setting: a prospective exome sequencingÂcase series. Genome Medicine, 2019, 11, 46.	8.2	25
122	Brachydactyly Type B: Linkage to Chromosome 9q22 and Evidence for Genetic Heterogeneity. American Journal of Human Genetics, 1999, 64, 578-585.	6.2	24
123	Insights from early experience of a Rare Disease Genomic Medicine Multidisciplinary Team: a qualitative study. European Journal of Human Genetics, 2017, 25, 680-686.	2.8	24
124	A genome-wide association study implicates the BMP7 locus as a risk factor for nonsyndromic metopic craniosynostosis. Human Genetics, 2020, 139, 1077-1090.	3.8	24
125	Fibroblast growth factor receptor 2, gain-of-function mutations, and tumourigenesis: investigating a potential link. Journal of Pathology, 2005, 207, 27-31.	4.5	23
126	Frank-ter Haar syndrome associated with sagittal craniosynostosis and raised intracranial pressure. BMC Medical Genetics, 2012, 13, 104.	2.1	23

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127	Genetic mapping of Xp22.12â€“p22.31, with a refined localization for spondyloepiphyseal dysplasia (SEDL). Human Genetics, 1995, 96, 407-410.	3.8	22
128	Linkage of Otopalatodigital Syndrome Type 2 (OPD2) to Distal Xq28: Evidence for Allelism with OPD1. American Journal of Human Genetics, 2001, 69, 223-227.	6.2	22
129	A further mutation of the FGFR2 tyrosine kinase domain in mild Crouzon syndrome. European Journal of Human Genetics, 2005, 13, 503-505.	2.8	22
130	Etiological heterogeneity and clinical characteristics of metopic synostosis: Evidence from a tertiary craniofacial unit. American Journal of Medical Genetics, Part A, 2010, 152A, 1383-1389.	1.2	22
131	Dominant coloboma-microphthalmos syndrome associated with sensorineural hearing loss, hematuria, and cleft lip/palate. , 1997, 72, 227-236.		21
132	Localized TWIST1 and TWIST2 basic domain substitutions cause four distinct human diseases that can be modeled in Caenorhabditis elegans. Human Molecular Genetics, 2017, 26, 2118-2132.	2.9	21
133	A variant in IL6ST with a selective IL-11 signaling defect in human and mouse. Bone Research, 2020, 8, 24.	11.4	21
134	A new locus for split hand/foot malformation with long bone deficiency (SHFLD) at 2q14.2 identified from a chromosome translocation. Human Genetics, 2007, 122, 191-199.	3.8	20
135	Recessive omodysplasia: five new cases and review of the literature. Pediatric Radiology, 2004, 34, 75-82.	2.0	19
136	Monozygotic twins discordant for frontonasal malformation. American Journal of Medical Genetics Part A, 2004, 130A, 384-388.	2.4	19
137	Functional analysis of natural mutations in two TWIST protein motifs. Human Mutation, 2005, 25, 550-556.	2.5	19
138	Identification of Intragenic Exon Deletions and Duplication ofTCF12by Whole Genome or Targeted Sequencing as a Cause ofTCF12-Related Craniosynostosis. Human Mutation, 2016, 37, 732-736.	2.5	19
139	Pure de novo partial trisomy 6p in a girl with craniosynostosis. American Journal of Medical Genetics, Part A, 2013, 161, 343-351.	1.2	18
140	Gonadal mosaicism and nonâ€invasive prenatal diagnosis for â€˜reassuranceâ€™ in sporadic paternal age effect (PAE) disorders. Prenatal Diagnosis, 2017, 37, 946-948.	2.3	18
141	An unusually large (CA)n repeat in the region of divergence between subtelomeric alleles of human chromosome 16p. Genomics, 1992, 13, 81-88.	2.9	17
142	Homozygous SALL1 Mutation Causes a Novel Multiple Congenital Anomalyâ€”Mental Retardation Syndrome. Journal of Pediatrics, 2013, 162, 612-617.	1.8	17
143	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. American Journal of Human Genetics, 2020, 106, 830-845.	6.2	17
144	A deletion of FGFR2 creating a chimeric IIIb/IIIc exon in a child with Apert syndrome. BMC Medical Genetics, 2011, 12, 122.	2.1	16

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145	Atypical Crouzon Syndrome with a Novel Cys62Arg Mutation in FGFR2 Presenting with Sagittal Synostosis. Cleft Palate-Craniofacial Journal, 2012, 49, 373-377.	0.9	16
146	Mutational Screening of FGFR1, CER1, and CDON in a Large Cohort of Trigonocephalic Patients. Cleft Palate-Craniofacial Journal, 2006, 43, 148-151.	0.9	15
147	Scalp fibroblasts have a shared expression profile in monogenic craniosynostosis. Journal of Medical Genetics, 2010, 47, 803-808.	3.2	15
148	Duplication of theEFNB1gene in familial hypertelorism: imbalance in ephrinâ€B1 expression and abnormal phenotypes in humans and mice. Human Mutation, 2011, 32, 930-938.	2.5	15
149	Genetic aspects of birth defects: new understandings of old problems. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2007, 92, F308-F314.	2.8	14
150	Apparently synonymous substitutions in FGFR2affect splicing and result in mild Crouzon syndrome. BMC Medical Genetics, 2014, 15, 95.	2.1	14
151	Many faces of SMCHD1. Nature Genetics, 2017, 49, 176-178.	21.4	14
152	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
153	Isodisomy in BWS chromosomes. Nature, 1991, 353, 802-802.	27.8	13
154	Evaluating the performance of a clinical genome sequencing program for diagnosis of rare genetic disease, seen through the lens of craniosynostosis. Genetics in Medicine, 2021, 23, 2360-2368.	2.4	13
155	Clinical hypochondroplasia in a family caused by a heterozygous double mutation inFGFR3 encoding GLY380LYS. American Journal of Medical Genetics, Part A, 2007, 143A, 355-359.	1.2	12
156	TCF12 microdeletion in a 72â€yearâ€old woman with intellectual disability. American Journal of Medical Genetics, Part A, 2015, 167, 1897-1901.	1.2	12
157	Gregor Mendel and the concepts of dominance and recessiveness. Nature Reviews Genetics, 2022, 23, 387-388.	16.3	11
158	Burning down DEFECT11. American Journal of Medical Genetics Part A, 2001, 100, 331-332.	2.4	10
159	Association of mutations in FLNA with craniosynostosis. European Journal of Human Genetics, 2015, 23, 1684-1688.	2.8	10
160	Cellular correlates of selfish spermatogonial selection. Andrology, 2016, 4, 550-553.	3.5	10
161	Disruption ofTWIST1translation by 5â€² UTR variants in Saethre-Chotzen syndrome. Human Mutation, 2018, 39, 1360-1365.	2.5	10
162	Biallelic GINS2 variant p.(Arg114Leu) causes Meier-Gorlin syndrome with craniosynostosis. Journal of Medical Genetics, 2022, 59, 776-780.	3.2	10

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163	Identification of causative variants in TXNL4A in Burn-McKeown syndrome and isolated choanal atresia. European Journal of Human Genetics, 2017, 25, 1126-1133.	2.8	10
164	The Fibroblast Growth Factor Receptor 2 p.<scp>A</scp>la172<scp>P</scp>he Mutation in Pfeiffer Syndromeâ€"<scp>H</scp>istory Repeating Itself. American Journal of Medical Genetics, Part A, 2013, 161, 1158-1163.	1.2	9
165	amplimap: a versatile tool to process and analyze targeted NGS data. Bioinformatics, 2019, 35, 5349-5350.	4.1	9
166	Fibroblast growth factor receptor mutations and craniosynostosis: Three receptors, five syndromes. Indian Journal of Pediatrics, 1996, 63, 351-356.	0.8	8
167	Language Development, Hearing Loss, and Intracranial Hypertension in Children With TWIST1-Confirmed Saethre-Chotzen Syndrome. Journal of Craniofacial Surgery, 2019, 30, 1506-1511.	0.7	8
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