Andrew Oliver Mungo Wilkie

List of Publications by Citations

190 papers	14,115 citations	61 h-index	116 g-index
258 ext. papers	16,371 ext. citations	12 avg, IF	6.13 L-index

#	Paper	IF	Citations
190	Apert syndrome results from localized mutations of FGFR2 and is allelic with Crouzon syndrome. *Nature Genetics*, **1995**, 9, 165-72	36.3	757
189	Integrating mapping-, assembly- and haplotype-based approaches for calling variants in clinical sequencing applications. *Nature Genetics*, **2014**, 46, 912-918	36.3	671
188	The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. *Nucleic Acids Research*, **2014**, 42, D966-74	20.1	565
187	The detection of subtelomeric chromosomal rearrangements in idiopathic mental retardation. *Nature Genetics*, **1995**, 9, 132-40	36.3	423
186	Identical mutations in the FGFR2 gene cause both Pfeiffer and Crouzon syndrome phenotypes. *Nature Genetics*, **1995**, 9, 173-6	36.3	384
185	Localized mutations in the gene encoding the cytoskeletal protein filamin A cause diverse malformations in humans. *Nature Genetics*, **2003**, 33, 487-91	36.3	337
184	Growth of the normal skull vault and its alteration in craniosynostosis: insights from human genetics and experimental studies. *Journal of Anatomy*, **2005**, 207, 637-53	2.9	320
183	Genetics of craniofacial development and malformation. *Nature Reviews Genetics*, **2001**, 2, 458-68	30.1	317
182	Craniosynostosis. *European Journal of Human Genetics*, **2011**, 19, 369-76	5.3	287
181	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, 8652-7	11.5	272
180	A truncated human chromosome 16 associated with alpha thalassaemia is stabilized by addition of telomeric repeat (TTAGGG)n. *Nature*, **1990**, 346, 868-71	50.4	264
179	Functional haploinsufficiency of the human homeobox gene MSX2 causes defects in skull ossification. *Nature Genetics*, **2000**, 24, 387-90	36.3	260
178	Exclusive paternal origin of new mutations in Apert syndrome. *Nature Genetics*, **1996**, 13, 48-53	36.3	251
177	Evidence for selective advantage of pathogenic FGFR2 mutations in the male germ line. *Science*, **2003**, 301, 643-6	33.3	248
176	Recessive Robinow syndrome, allelic to dominant brachydactyly type B, is caused by mutation of ROR2. *Nature Genetics*, **2000**, 25, 419-22	36.3	247
175	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. *Nature Genetics*, **2015**, 47, 717-726	36.3	244
174	Oculofaciocardiodental and Lenz microphthalmia syndromes result from distinct classes of mutations in BCOR. *Nature Genetics*, **2004**, 36, 411-6	36.3	234

173	Functions of fibroblast growth factors and their receptors. *Current Biology*, **1995**, 5, 500-7	6.3	229
172	Paternal age effect mutations and selfish spermatogonial selection: causes and consequences for human disease. *American Journal of Human Genetics*, **2012**, 90, 175-200	11	217
171	Bad bones, absent smell, selfish testes: the pleiotropic consequences of human FGF receptor mutations. *Cytokine and Growth Factor Reviews*, **2005**, 16, 187-203	17.9	203
170	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, 472-86	11	203
169	De novo alu-element insertions in FGFR2 identify a distinct pathological basis for Apert syndrome. *American Journal of Human Genetics*, **1999**, 64, 446-61	11	198
168	Prevalence and complications of single-gene and chromosomal disorders in craniosynostosis. *Pediatrics*, **2010**, 126, e391-400	7.4	194
167	Dominant mutations in ROR2, encoding an orphan receptor tyrosine kinase, cause brachydactyly type B. *Nature Genetics*, **2000**, 24, 275-8	36.3	187
166	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, 1162-70	11	182
165	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, 1282-93	11	165
164	FGFs, their receptors, and human limb malformations: clinical and molecular correlations. *American Journal of Medical Genetics Part A*, **2002**, 112, 266-78		162
163	Cell mixing at a neural crest-mesoderm boundary and deficient ephrin-Eph signaling in the pathogenesis of craniosynostosis. *Human Molecular Genetics*, **2006**, 15, 1319-28	5.6	161
162	Paternal origin of FGFR2 mutations in sporadic cases of Crouzon syndrome and Pfeiffer syndrome. *American Journal of Human Genetics*, **2000**, 66, 768-77	11	161
161	Activating mutations in FGFR3 and HRAS reveal a shared genetic origin for congenital disorders and testicular tumors. *Nature Genetics*, **2009**, 41, 1247-52	36.3	154
160	Stable length polymorphism of up to 260 kb at the tip of the short arm of human chromosome 16. *Cell*, **1991**, 64, 595-606	56.2	151
159	Mutations in TCF12, encoding a basic helix-loop-helix partner of TWIST1, are a frequent cause of coronal craniosynostosis. *Nature Genetics*, **2013**, 45, 304-7	36.3	146
158	Distinct mutations in the receptor tyrosine kinase gene ROR2 cause brachydactyly type B. *American Journal of Human Genetics*, **2000**, 67, 822-31	11	145
157	A Genetic-Pathophysiological Framework for Craniosynostosis. *American Journal of Human Genetics* , **2015**, 97, 359-77	11	144
156	Epidermal mosaicism producing localised acne: somatic mutation in FGFR2. *Lancet, The*, **1998**, 352, 704-540		128

155	Haploinsufficiency of the human homeobox gene ALX4 causes skull ossification defects. *Nature Genetics*, **2001**, 27, 17-8	36.3	126
154	Inactivation of IL11 signaling causes craniosynostosis, delayed tooth eruption, and supernumerary teeth. *American Journal of Human Genetics*, **2011**, 89, 67-81	11	123
153	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, 659-68	5.8	111
152	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, 6051-6	11.5	108
151	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, 274-80	5.3	107
150	Reduced dosage of ERF causes complex craniosynostosis in humans and mice and links ERK1/2 signaling to regulation of osteogenesis. *Nature Genetics*, **2013**, 45, 308-13	36.3	107
149	Germline selection shapes human mitochondrial DNA diversity. *Science*, **2019**, 364,	33.3	105
148	Evidence for 28 genetic disorders discovered by combining healthcare and research data. *Nature*, **2020**, 586, 757-762	50.4	103
147	A biallelic mutation in encoding the GP130 co-receptor causes immunodeficiency and craniosynostosis. *Journal of Experimental Medicine*, **2017**, 214, 2547-2562	16.6	102
146	Duplications of noncoding elements 5Rof SOX9 are associated with brachydactyly-anonychia. *Nature Genetics*, **2009**, 41, 862-3	36.3	94
145	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	11	94
144	A genome-wide association study identifies susceptibility loci for nonsyndromic sagittal craniosynostosis near BMP2 and within BBS9. *Nature Genetics*, **2012**, 44, 1360-4	36.3	93
143	Clinical genetics of craniosynostosis. *Current Opinion in Pediatrics*, **2017**, 29, 622-628	3.2	86
142	Potential gene conversion and source genes for recently integrated Alu elements. *Genome Research*, **2000**, 10, 1485-95	9.7	84
141	New insights into craniofacial malformations. *Human Molecular Genetics*, **2015**, 24, R50-9	5.6	82
140	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	11	82
139	Missense mutations in the homeodomain of HOXD13 are associated with brachydactyly types D and E. *American Journal of Human Genetics*, **2003**, 72, 984-97	11	79
138	Diagnostic value of exome and whole genome sequencing in craniosynostosis. *Journal of Medical Genetics*, **2017**, 54, 260-268	5.8	77

137	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	11	74
136	A noncoding expansion in EIF4A3 causes Richieri-Costa-Pereira syndrome, a craniofacial disorder associated with limb defects. *American Journal of Human Genetics*, **2014**, 94, 120-8	11	67
135	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, 125-38	11	67
134	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, 2417-23	5.6	65
133	Skeletal analysis of the Fgfr3(P244R) mouse, a genetic model for the Muenke craniosynostosis syndrome. *Developmental Dynamics*, **2009**, 238, 331-42	2.9	64
132	Clinical dividends from the molecular genetic diagnosis of craniosynostosis. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1941-9	2.5	64
131	OCT2, SSX and SAGE1 reveal the phenotypic heterogeneity of spermatocytic seminoma reflecting distinct subpopulations of spermatogonia. *Journal of Pathology*, **2011**, 224, 473-83	9.4	62
130	Frontometaphyseal dysplasia: mutations in FLNA and phenotypic diversity. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1726-36	2.5	61
129	A novel mutation, Ala315Ser, in FGFR2: a gene-environment interaction leading to craniosynostosis?. *European Journal of Human Genetics*, **2000**, 8, 571-7	5.3	61
128	"Selfish spermatogonial selection": a novel mechanism for the association between advanced paternal age and neurodevelopmental disorders. *American Journal of Psychiatry*, **2013**, 170, 599-608	11.9	60
127	Reoperation for intracranial hypertension in TWIST1-confirmed Saethre-Chotzen syndrome: a 15-year review. *Plastic and Reconstructive Surgery*, **2009**, 123, 1801-1810	2.7	57
126	Enlarged parietal foramina caused by mutations in the homeobox genes ALX4 and MSX2: from genotype to phenotype. *European Journal of Human Genetics*, **2006**, 14, 151-8	5.3	57
125	Mutations in multidomain protein MEGF8 identify a Carpenter syndrome subtype associated with defective lateralization. *American Journal of Human Genetics*, **2012**, 91, 897-905	11	55
124	Paternal origin of FGFR3 mutations in Muenke-type craniosynostosis. *Human Genetics*, **2004**, 115, 200-7	6.3	55
123	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	11	53
122	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, 20152-7	11.5	51
121	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, 1654-62	5.6	50
120	Hearing loss in a mouse model of Muenke syndrome. *Human Molecular Genetics*, **2009**, 18, 43-50	5.6	48

119	Rare mutations of FGFR2 causing apert syndrome: identification of the first partial gene deletion, and an Alu element insertion from a new subfamily. *Human Mutation*, **2009**, 30, 204-11	4.7	46
118	Skeletal development is regulated by fibroblast growth factor receptor 1 signalling dynamics. *Development (Cambridge)*, **2004**, 131, 325-35	6.6	46
117	FGFR3 P250R mutation increases the risk of reoperation in apparent nonsyndromic coronal craniosynostosis. *Journal of Craniofacial Surgery*, **2005**, 16, 347-52; discussion 353-4	1.2	45
116	HUWE1 variants cause dominant X-linked intellectual disability: a clinical study of 21 patients. *European Journal of Human Genetics*, **2018**, 26, 64-74	5.3	43
115	Selective loss of function variants in cause Hyper-IgE syndrome with distinct impairments of T-cell phenotype and function. *Haematologica*, **2019**, 104, 609-621	6.6	43
114	Expanding the phenotype of craniofrontonasal syndrome: two unrelated boys with EFNB1 mutations and congenital diaphragmatic hernia. *European Journal of Human Genetics*, **2006**, 14, 884-7	5.3	41
113	Gain-of-Function Mutations in ZIC1 Are Associated with Coronal Craniosynostosis and Learning Disability. *American Journal of Human Genetics*, **2015**, 97, 378-88	11	38
112	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		38
111	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. *American Journal of Human Genetics*, **2018**, 102, 468-479	11	37
110	Craniosynostosis. *Current Opinion in Neurology*, **1996**, 9, 146	7.1	37
109	Alx4 and Msx2 play phenotypically similar and additive roles in skull vault differentiation. *Journal of Anatomy*, **2004**, 204, 487-99	2.9	36
108	A survey of TWIST for mutations in craniosynostosis reveals a variable length polyglycine tract in asymptomatic individuals. *Human Mutation*, **2001**, 18, 535-41	4.7	36
107	Selfish mutations dysregulating RAS-MAPK signaling are pervasive in aged human testes. *Genome Research*, **2018**, 28, 1779-1790	9.7	36
106	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. *American Journal of Human Genetics*, **2016**, 99, 392-406	11	34
105	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, 2454-9	11.5	34
104	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. *New England Journal of Medicine*, **2021**, 385, 1868-1880	59.2	34
103	Parietal foramina with cleidocranial dysplasia is caused by mutation in MSX2. *European Journal of Human Genetics*, **2003**, 11, 892-5	5.3	33
102	Limited proteolysis and proton NMR spectroscopy of Bacillus stearothermophilus pyruvate dehydrogenase multienzyme complex. *FEBS Journal*, **1982**, 124, 63-9		33

101	Efficient use of a R dead-end RGA 5R splice site in the human fibroblast growth factor receptor genes. *EMBO Journal*, **2003**, 22, 1620-31	13	32
100	The genetics of mental retardation. *British Medical Bulletin*, **1996**, 52, 453-64	5.4	31
99	Carpenter syndrome: extended RAB23 mutation spectrum and analysis of nonsense-mediated mRNA decay. *Human Mutation*, **2011**, 32, E2069-78	4.7	30
98	Germline and somatic mosaicism for FGFR2 mutation in the mother of a child with Crouzon syndrome: Implications for genetic testing in "paternal age-effect" syndromes. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2067-73	2.5	30
97	Implications of a vertex bulge following modified strip craniectomy for sagittal synostosis. *Plastic and Reconstructive Surgery*, **2008**, 122, 217-224	2.7	30
96	Postzygotic mutation and germline mosaicism in the otopalatodigital syndrome spectrum disorders. *European Journal of Human Genetics*, **2006**, 14, 549-54	5.3	30
95	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	11	29
94	Missing heritability: paternal age effect mutations and selfish spermatogonia. *Nature Reviews Genetics*, **2010**, 11, 589	30.1	29
93	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	2.7	28
92	Nonsense-mediated decay and the molecular pathogenesis of mutations in SALL1 and GLI3. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 3150-60	2.5	28
91	Why study human limb malformations?. *Journal of Anatomy*, **2003**, 202, 27-35	2.9	28
90	ACTH receptor mutation in a girl with familial glucocorticoid deficiency. *Clinical Genetics*, **1998**, 53, 57-62	4	28
89	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, 757-62	5.3	27
88	Epidemiology and genetics of craniosynostosis. *American Journal of Medical Genetics Part A*, **2000**, 90, 82-4		27
87	Selfish spermatogonial selection: evidence from an immunohistochemical screen in testes of elderly men. *PLoS ONE*, **2012**, 7, e42382	3.7	26
86	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, 517-26	1.7	26
85	Craniosynostosis and related limb anomalies. *Novartis Foundation Symposium*, **2001**, 232, 122-33; discussion 133-43		25
84	Heterozygous mutations affecting the protein kinase domain of cause a syndromic form of developmental delay and intellectual disability. *Journal of Medical Genetics*, **2018**, 55, 28-38	5.8	24

83	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	11	24
82	The gene for spondyloepiphyseal dysplasia (SEDL) maps to Xp22 between DXS16 and DXS92. *Genomics*, **1993**, 18, 100-4	4.3	24
81	Laband syndrome. Report of two cases, review of the literature, and identification of additional manifestations. *Oral Surgery, Oral Medicine, and Oral Pathology*, **1994**, 78, 57-63		24
80	Whole-genome sequencing of spermatocytic tumors provides insights into the mutational processes operating in the male germline. *PLoS ONE*, **2017**, 12, e0178169	3.7	24
79	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		23
78	Frank-ter Haar syndrome associated with sagittal craniosynostosis and raised intracranial pressure. *BMC Medical Genetics*, **2012**, 13, 104	2.1	21
77	Clinical dividends from the molecular genetic diagnosis of craniosynostosis. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2631-9	2.5	21
76	Fibroblast growth factor receptor 2, gain-of-function mutations, and tumourigenesis: investigating a potential link. *Journal of Pathology*, **2005**, 207, 27-31	9.4	21
75	Brachydactyly type B: linkage to chromosome 9q22 and evidence for genetic heterogeneity. *American Journal of Human Genetics*, **1999**, 64, 578-85	11	21
74	Dominant coloboma-microphthalmos syndrome associated with sensorineural hearing loss, hematuria, and cleft lip/palate. *American Journal of Medical Genetics Part A*, **1997**, 72, 227-36		20
73	A further mutation of the FGFR2 tyrosine kinase domain in mild Crouzon syndrome. *European Journal of Human Genetics*, **2005**, 13, 503-5	5.3	20
72	Genetic mapping of Xp22.12-p22.31, with a refined localization for spondyloepiphyseal dysplasia (SEDL). *Human Genetics*, **1995**, 96, 407-10	6.3	20
71	De novo and rare inherited mutations implicate the transcriptional coregulator TCF20/SPBP in autism spectrum disorder. *Journal of Medical Genetics*, **2014**, 51, 737-47	5.8	19
70	Linkage of otopalatodigital syndrome type 2 (OPD2) to distal Xq28: evidence for allelism with OPD1. *American Journal of Human Genetics*, **2001**, 69, 223-7	11	19
69	Etiological heterogeneity and clinical characteristics of metopic synostosis: Evidence from a tertiary craniofacial unit. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1383-9	2.5	18
68	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, 191-9	6.3	18
67	Insights from early experience of a Rare Disease Genomic Medicine Multidisciplinary Team: a qualitative study. *European Journal of Human Genetics*, **2017**, 25, 680-686	5.3	17
66	ERF-related craniosynostosis: The phenotypic and developmental profile of a new craniosynostosis syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 615-627	2.5	17

65	Monozygotic twins discordant for frontonasal malformation. *American Journal of Medical Genetics Part A*, **2004**, 130A, 384-8		17
64	An unusually large (CA)n repeat in the region of divergence between subtelomeric alleles of human chromosome 16p. *Genomics*, **1992**, 13, 81-8	4.3	17
63	Truncated SALL1 Impedes Primary Cilia Function in Townes-Brocks Syndrome. *American Journal of Human Genetics*, **2018**, 102, 249-265	11	16
62	Pure de novo partial trisomy 6p in a girl with craniosynostosis. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 343-51	2.5	16
61	Functional analysis of natural mutations in two TWIST protein motifs. *Human Mutation*, **2005**, 25, 550-6	4.7	16
60	Identification of Intragenic Exon Deletions and Duplication of TCF12 by Whole Genome or Targeted Sequencing as a Cause of TCF12-Related Craniosynostosis. *Human Mutation*, **2016**, 37, 732-6	4.7	16
59	Mutational screening of FGFR1, CER1, and CDON in a large cohort of trigonocephalic patients. *Cleft Palate-Craniofacial Journal*, **2006**, 43, 148-51	1.9	14
58	Implementation of a genomic medicine multi-disciplinary team approach for rare disease in the clinical setting: a prospective exome sequencing case series. *Genome Medicine*, **2019**, 11, 46	14.4	13
57	Homozygous SALL1 mutation causes a novel multiple congenital anomaly-mental retardation syndrome. *Journal of Pediatrics*, **2013**, 162, 612-7	3.6	13
56	Gonadal mosaicism and non-invasive prenatal diagnosis for reassurance in sporadic paternal age effect (PAE) disorders. *Prenatal Diagnosis*, **2017**, 37, 946-948	3.2	13
55	A deletion of FGFR2 creating a chimeric IIIb/IIIc exon in a child with Apert syndrome. *BMC Medical Genetics*, **2011**, 12, 122	2.1	13
54	Duplication of the EFNB1 gene in familial hypertelorism: imbalance in ephrin-B1 expression and abnormal phenotypes in humans and mice. *Human Mutation*, **2011**, 32, 930-8	4.7	13
53	Scalp fibroblasts have a shared expression profile in monogenic craniosynostosis. *Journal of Medical Genetics*, **2010**, 47, 803-8	5.8	13
52	Atypical Crouzon syndrome with a novel Cys62Arg mutation in FGFR2 presenting with sagittal synostosis. *Cleft Palate-Craniofacial Journal*, **2012**, 49, 373-7	1.9	13
51	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	5.6	12
50	Clinical hypochondroplasia in a family caused by a heterozygous double mutation in FGFR3 encoding GLY380LYS. *American Journal of Medical Genetics, Part A*, **2007**, 143, 355-9	2.5	12
49	Genetic aspects of birth defects: new understandings of old problems. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **2007**, 92, F308-14	4.7	12
48	Recessive omodysplasia: five new cases and review of the literature. *Pediatric Radiology*, **2004**, 34, 75-82	2.8	12

47	A variant in with a selective IL-11 signaling defect in human and mouse. *Bone Research*, **2020**, 8, 24	13.3	11
46	Isodisomy in BWS chromosomes. *Nature*, **1991**, 353, 802	50.4	11
45	Many faces of SMCHD1. *Nature Genetics*, **2017**, 49, 176-178	36.3	10
44	Association of mutations in FLNA with craniosynostosis. *European Journal of Human Genetics*, **2015**, 23, 1684-8	5.3	10
43	TCF12 microdeletion in a 72-year-old woman with intellectual disability. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1897-901	2.5	10
42	A genome-wide association study implicates the BMP7 locus as a risk factor for nonsyndromic metopic craniosynostosis. *Human Genetics*, **2020**, 139, 1077-1090	6.3	9
41	The fibroblast growth factor receptor 2 p.Ala172Phe mutation in Pfeiffer syndrome--history repeating itself. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1158-63	2.5	9
40	SMAD6 variants in craniosynostosis: genotype and phenotype evaluation. *Genetics in Medicine*, **2020**, 22, 1498-1506	8.1	8
39	Burning down DEFECT11. *American Journal of Medical Genetics Part A*, **2001**, 100, 331-5		8
38	The developing mouse coronal suture at single-cell resolution. *Nature Communications*, **2021**, 12, 4797	17.4	8
37	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. *Frontiers in Genetics*, **2019**, 10, 611	4.5	7
36	Apparently synonymous substitutions in FGFR2 affect splicing and result in mild Crouzon syndrome. *BMC Medical Genetics*, **2014**, 15, 95	2.1	7
35	Identification of causative variants in TXNL4A in Burn-McKeown syndrome and isolated choanal atresia. *European Journal of Human Genetics*, **2017**, 25, 1126-1133	5.3	7
34	Disruption of TWIST1 translation by 5RUTR variants in Saethre-Chotzen syndrome. *Human Mutation*, **2018**, 39, 1360-1365	4.7	6
33	Patterson-Stevenson-Fontaine syndrome: 30-year follow-up and clinical details of a further affected case. *American Journal of Medical Genetics Part A*, **1997**, 69, 433-4		6
32	Dominant inheritance of optic pits. *American Journal of Ophthalmology*, **1998**, 125, 124-5	4.9	6
31	Fibroblast growth factor receptor mutations and craniosynostosis: three receptors, five syndromes. *Indian Journal of Pediatrics*, **1996**, 63, 351-6	3	6
30	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. *American Journal of Human Genetics*, **2020**, 106, 830-845	11	6

29	Cellular correlates of selfish spermatogonial selection. *Andrology*, **2016**, 4, 550-3	4.2	6
28	Language Development, Hearing Loss, and Intracranial Hypertension in Children With TWIST1-Confirmed Saethre-Chotzen Syndrome. *Journal of Craniofacial Surgery*, **2019**, 30, 1506-1511	1.2	5
27	Gastrointestinal disorders in Curry-Jones syndrome: Clinical and molecular insights from an affected newborn. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1586-1592	2.5	4
26	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. *Human Mutation*, **2019**, 40, 1993-2000	4.7	4
25	amplimap: a versatile tool to process and analyze targeted NGS data. *Bioinformatics*, **2019**, 35, 5349-5350	7.2	4
24	Sifting the significance from the data—the impact of high-throughput genomic technologies on human genetics and health care. *Human Genomics*, **2012**, 6, 11	6.8	4
23	Genetics of disease. *Current Opinion in Genetics and Development*, **1996**, 6, 271-4	4.9	4
22	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. *Human Mutation*, **2021**, 42, 445-459	4.7	4
21	The Drosophila homologue of MEGF8 is essential for early development. *Scientific Reports*, **2018**, 8, 8790	4.9	3
20	Pitfalls in the phylogenomic evaluation of human disease-causing mutations. *Journal of Biology*, **2009**, 8, 26		3
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