David Chitayat

List of Publications by Citations

169 papers	4,569 citations	35 h-index	61 g-index
173 ext. papers	5,564 ext. citations	6.4 avg, IF	4.75 L-index

#	Paper	IF	Citations
169	Evidence for multi-site closure of the neural tube in humans. *American Journal of Medical Genetics Part A*, **1993**, 47, 723-43		317
168	Heterozygous mutations in the gene encoding noggin affect human joint morphogenesis. *Nature Genetics*, **1999**, 21, 302-4	36.3	285
167	Discordant KCNQ1OT1 imprinting in sets of monozygotic twins discordant for Beckwith-Wiedemann syndrome. *Human Molecular Genetics*, **2002**, 11, 1317-25	5.6	258
166	Mutations in CDMP1 cause autosomal dominant brachydactyly type C. *Nature Genetics*, **1997**, 17, 18-9	36.3	235
165	Whole Genome Sequencing Expands Diagnostic Utility and Improves Clinical Management in Pediatric Medicine. *Npj Genomic Medicine*, **2016**, 1,	6.2	208
164	PDE3A mutations cause autosomal dominant hypertension with brachydactyly. *Nature Genetics*, **2015**, 47, 647-53	36.3	118
163	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. *Nature Genetics*, **2017**, 49, 1529-1538	36.3	105
162	CHARGE and Kabuki Syndromes: Gene-Specific DNA Methylation Signatures Identify Epigenetic Mechanisms Linking These Clinically Overlapping Conditions. *American Journal of Human Genetics*, **2017**, 100, 773-788	11	99
161	A new mutation in the type II hair cortex keratin hHb1 involved in the inherited hair disorder monilethrix. *Human Genetics*, **1997**, 101, 165-9	6.3	89
160	FGFR2 mutation associated with clinical manifestations consistent with Antley-Bixler syndrome. *American Journal of Medical Genetics Part A*, **1998**, 77, 219-24		83
159	Prenatally diagnosed neural tube defects: ultrasound, chromosome, and autopsy or postnatal findings in 212 cases. *American Journal of Medical Genetics Part A*, **1998**, 77, 317-21		83
158	Findings in children exposed in utero to phenytoin and carbamazepine monotherapy: Independent effects of epilepsy and medications **1997**, 68, 18-24		74
157	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. *American Journal of Human Genetics*, **2017**, 101, 466-477	11	73
156	The functional O-mannose glycan on □dystroglycan contains a phospho-ribitol primed for matriglycan addition. *ELife*, **2016**, 5,	8.9	73
155	Lymphatic abnormalities in fetuses with posterior cervical cystic hygroma. *American Journal of Medical Genetics Part A*, **1989**, 33, 352-6		70
154	Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S, which Encode Subcomponents C1r and C1s of Complement. *American Journal of Human Genetics*, **2016**, 99, 1005-1014	11	70
153	Alagille syndrome: clinical perspectives. *The Application of Clinical Genetics*, **2016**, 9, 75-82	3.1	62

152	Clustering of FBN2 mutations in patients with congenital contractural arachnodactyly indicates an important role of the domains encoded by exons 24 through 34 during human development **1998**, 78, 350-355		59
151	No. 348-Joint SOGC-CCMG Guideline: Update on Prenatal Screening for Fetal Aneuploidy, Fetal Anomalies, and Adverse Pregnancy Outcomes. *Journal of Obstetrics and Gynaecology Canada*, **2017**, 39, 805-817	1.3	55
150	Prenatal screening, diagnosis, and pregnancy management of fetal neural tube defects. *Journal of Obstetrics and Gynaecology Canada*, **2014**, 36, 927-939	1.3	52
149	Congenital diaphragmatic hernia, coarse facies, and acral hypoplasia: Fryns syndrome. *American Journal of Medical Genetics Part A*, **1989**, 32, 93-9		51
148	CpG Methylation, a Parent-of-Origin Effect for Maternal-Biased Transmission of Congenital Myotonic Dystrophy. *American Journal of Human Genetics*, **2017**, 100, 488-505	11	48
147	Familial growth hormone deficiency associated with MRI abnormalities. *American Journal of Medical Genetics Part A*, **1998**, 80, 128-132		47
146	Folic acid supplementation for pregnant women and those planning pregnancy: 2015 update. *Journal of Clinical Pharmacology*, **2016**, 56, 170-5	2.9	46
145	Hypomelanosis of Ito--a nonspecific marker of somatic mosaicism: report of case with trisomy 18 mosaicism. *American Journal of Medical Genetics Part A*, **1990**, 35, 422-4		44
144	Biallelic mutations in the 3' exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. *Nature Genetics*, **2017**, 49, 457-464	36.3	43
143	Anesthesia for Freeman-Sheldon syndrome using a laryngeal mask airway. *Canadian Journal of Anaesthesia*, **1999**, 46, 783-7	3	41
142	Early-infantile galactosialidosis: Prenatal presentation and postnatal follow-up. *American Journal of Medical Genetics Part A*, **1999**, 85, 38-47		41
141	Hydrops-ectopic calcification-moth-eaten skeletal dysplasia (Greenberg dysplasia): prenatal diagnosis and further delineation of a rare genetic disorder. *American Journal of Medical Genetics Part A*, **1993**, 47, 272-7		40
140	Genomic study of severe fetal anomalies and discovery of GREB1L mutations in renal agenesis. *Genetics in Medicine*, **2018**, 20, 745-753	8.1	38
139	The molecular and phenotypic spectrum of IQSEC2-related epilepsy. *Epilepsia*, **2016**, 57, 1858-1869	6.4	38
138	Fetal reprogramming and senescence in hypoplastic left heart syndrome and in human pluripotent stem cells during cardiac differentiation. *American Journal of Pathology*, **2013**, 183, 720-34	5.8	36
137	Novel mutations of the tyrosinase (TYR) gene in type I oculocutaneous albinism (OCA1). *Human Mutation*, **1997**, 10, 171-4	4.7	35
136	The PDAC syndrome (pulmonary hypoplasia/agenesis, diaphragmatic hernia/eventration, anophthalmia/microphthalmia, and cardiac defect) (Spear syndrome, Matthew-Wood syndrome): report of eight cases including a living child and further evidence for autosomal recessive inheritance. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1268-81	2.5	35
135	Chondrodysplasia punctata associated with maternal autoimmune diseases: expanding the spectrum from systemic lupus erythematosus (SLE) to mixed connective tissue disease (MCTD) and scleroderma report of eight cases. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 3038-53	2.5	35

134	Novel mutations of the P gene in type II oculocutaneous albinism (OCA2). *Human Mutation*, **1997**, 10, 175-7	4.7	34
133	Familial Dandy-Walker malformation associated with macrocephaly, facial anomalies, developmental delay, and brain stem dysgenesis: prenatal diagnosis and postnatal outcome in brothers. A new syndrome?. *American Journal of Medical Genetics Part A*, **1994**, 52, 406-15		34
132	Intrinsic Endocardial Defects Contribute to Hypoplastic Left Heart Syndrome. *Cell Stem Cell*, **2020**, 27, 574-589.e8	18	34
131	Mutations in Plasmalemma Vesicle Associated Protein Result in Sieving Protein-Losing Enteropathy Characterized by Hypoproteinemia, Hypoalbuminemia, and Hypertriglyceridemia. *Cellular and Molecular Gastroenterology and Hepatology*, **2015**, 1, 381-394.e7	7.9	32
130	Double-positive maternal serum screening results for down syndrome and open neural tube defects: An indicator for fetal structural or chromosomal abnormalities and adverse obstetric outcomes. *American Journal of Obstetrics and Gynecology*, **2002**, 187, 758-63	6.4	32
129	Apparent postnatal onset of some manifestations of the Wiedemann-Beckwith syndrome. *American Journal of Medical Genetics Part A*, **1990**, 36, 434-9		32
128	TRPV6 Variants Interfere with Maternal-Fetal Calcium Transport through the Placenta and Cause Transient Neonatal Hyperparathyroidism. *American Journal of Human Genetics*, **2018**, 102, 1104-1114	11	32
127	Deletion of 15q11.2(BP1-BP2) region: further evidence for lack of phenotypic specificity in a pediatric population. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2098-102	2.5	31
126	Partial tetrasomy with triplication of chromosome (5) (p14-p15.33) in a patient with severe multiple congenital anomalies. *American Journal of Medical Genetics Part A*, **1998**, 79, 103-107		31
125	Brain abnormalities in patients with Beckwith-Wiedemann syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1388-94	2.5	29
124	Penoscrotal transposition: a case report and review. *American Journal of Medical Genetics Part A*, **1994**, 49, 103-7		29
123	Clinical delineation of the PACS1-related syndrome--Report on 19 patients. *American Journal of Medical Genetics, Part A*, **2016**, 170, 670-5	2.5	28
122	Syndrome of proximal interstitial deletion 4p15: report of three cases and review of the literature. *American Journal of Medical Genetics Part A*, **1995**, 55, 147-54		28
121	Spectrum and outcome of primary cardiomyopathies diagnosed during fetal life. *JACC: Heart Failure*, **2014**, 2, 403-11	7.9	27
120	Limb defects in homozygous alpha-thalassemia: report of three cases. *American Journal of Medical Genetics Part A*, **1997**, 68, 162-7		27
119	Terminal deletion of the long arm of chromosome 3 [46,XX,del(3)(q27-->qter)]. *American Journal of Medical Genetics Part A*, **1996**, 61, 45-8		27
118	Prenatal diagnosis of retinal nonattachment in the Walker-Warburg syndrome. *American Journal of Medical Genetics Part A*, **1995**, 56, 351-8		27
117	Tissue-specific methylation differences and cognitive function in fragile X premutation females. *American Journal of Medical Genetics Part A*, **1996**, 64, 329-33		26

116	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. *American Journal of Human Genetics*, **2020**, 106, 596-610	11	26
115	Omphalocele in Miller-Dieker syndrome: expanding the phenotype. *American Journal of Medical Genetics Part A*, **1997**, 69, 293-8		25
114	Prenatal diagnosis and fetopathological findings in five fetuses with trisomy 9. *American Journal of Medical Genetics Part A*, **1995**, 56, 247-51		25
113	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. *American Journal of Human Genetics*, **2018**, 103, 154-162	11	25
112	Compound heterozygosity for the achondroplasia-hypochondroplasia FGFR3 mutations: Prenatal diagnosis and postnatal outcome. *American Journal of Medical Genetics Part A*, **1999**, 84, 401-405		24
111	Chitayat-Hall and Schaaf-Yang syndromes:a common aetiology: expanding the phenotype of -related disorders. *Journal of Medical Genetics*, **2018**, 55, 316-321	5.8	23
110	De novo 46,XX,t(6;7)(q27;q11;23) associated with severe cardiovascular manifestations characteristic of supravalvular aortic stenosis and Williams syndrome. *American Journal of Medical Genetics Part A*, **2000**, 90, 270-5		23
109	Fetal arthrogryposis multiplex congenita/fetal akinesia deformation sequence (FADS)-Aetiology, diagnosis, and management. *Prenatal Diagnosis*, **2019**, 39, 720-731	3.2	22
108	Dual loss of p110PI3-kinase and SKAP (KNSTRN) expression leads to combined immunodeficiency and multisystem syndromic features. *Journal of Allergy and Clinical Immunology*, **2018**, 142, 618-629	11.5	22
107	The Relationship Between Burnout and Occupational Stress in Genetic Counselors. *Journal of Genetic Counseling*, **2016**, 25, 731-41	2.5	21
106	Role of amniotic fluid interphase fluorescence in situ hybridization (FISH) analysis in patient management. *Prenatal Diagnosis*, **2001**, 21, 327-32	3.2	21
105	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. *American Journal of Human Genetics*, **2019**, 105, 1005-1015	11	20
104	Inverted duplication of the distal short arm of chromosome 3 associated with lobar holoprosencephaly and lumbosacral meningomyelocele **2000**, 91, 167-170		20
103	Congenital heart disease and Robinow syndrome: coincidence or an additional component of the syndrome?. *American Journal of Medical Genetics Part A*, **1990**, 37, 519-21		20
102	Raine syndrome: a rare lethal osteosclerotic bone dysplasia. Prenatal diagnosis, autopsy, and neuropathological findings. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 3280-5	2.5	19
101	Congenital toxoplasmosis: prenatal diagnosis, treatment and postnatal outcome. *Prenatal Diagnosis*, **1999**, 19, 330-3	3.2	18
100	Genetic homogeneity of cartilage-hair hypoplasia. *Human Genetics*, **1995**, 95, 157-60	6.3	18
99	The expanding clinical phenotype of the tRNA(Leu(UUR)) A-->G mutation at np 3243 of mitochondrial DNA: diabetic embryopathy associated with mitochondrial cytopathy. *American Journal of Medical Genetics Part A*, **1996**, 62, 404-9		18

98	ATP6AP2 variant impairs CNS development and neuronal survival to cause fulminant neurodegeneration. *Journal of Clinical Investigation*, **2019**, 129, 2145-2162	15.9	18
97	De novo missense variants in RAC3 cause a novel neurodevelopmental syndrome. *Genetics in Medicine*, **2019**, 21, 1021-1026	8.1	17
96	Juvenile galactosialidosis in a white male: a new variant. *American Journal of Medical Genetics Part A* , **1988**, 31, 887-901		16
95	Hepatocellular carcinoma in a child with familial Russell-Silver syndrome. *American Journal of Medical Genetics Part A*, **1988**, 31, 909-14		16
94	Syndrome of mental retardation, facial anomalies, hypopituitarism, and distal arthrogryposis in sibs. *American Journal of Medical Genetics Part A*, **1990**, 37, 65-70		14
93	Loss-of-Function Variants in PPP1R12A: From Isolated Sex Reversal to Holoprosencephaly Spectrum and Urogenital Malformations. *American Journal of Human Genetics*, **2020**, 106, 121-128	11	14
92	New insights into DNA methylation signatures: SMARCA2 variants in Nicolaides-Baraitser syndrome. *BMC Medical Genomics*, **2019**, 12, 105	3.7	13
91	Congenital abnormalities in two sibs exposed to valproic acid in utero. *American Journal of Medical Genetics Part A*, **1988**, 31, 369-73		13
90	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. *JAMA Network Open*, **2020**, 3, e2018109	10.4	13
89	Warsaw breakage syndrome: Further clinical and genetic delineation. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2404-2418	2.5	13
88	Evidence for somatic and germline mosaicism in CRASH syndrome. *Human Mutation*, **1998**, Suppl 1, S284-7	4.7	12
87	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6,	14.3	12
86	Mutations in the NEB gene cause fetal akinesia/arthrogryposis multiplex congenita. *Prenatal Diagnosis*, **2017**, 37, 144-150	3.2	11
85	Experience with genetic counseling: the adolescent perspective. *Journal of Genetic Counseling*, **2016**, 25, 583-95	2.5	11
84	An Additional Individual with a De Novo Variant in Myelin Regulatory Factor (MYRF) with Cardiac and Urogenital Anomalies: Further Proof of Causality: Comments on the article by Pinz et al. (). *American Journal of Medical Genetics, Part A*, **2018**, 176, 2041-2043	2.5	11
83	The pathology of incipient polymicrogyria. *Brain and Development*, **2017**, 39, 23-39	2.2	11
82	Brachydactyly-short stature-hypertension (Bilginturan) syndrome: report on two families. *American Journal of Medical Genetics Part A*, **1997**, 73, 279-85		11
81	Perinatal and first year follow-up of patients with Prader-Willi syndrome: normal size of hands and feet. *Clinical Genetics*, **1989**, 35, 161-6	4	11

80	Meconium peritonitis: the role of postnatal radiographic and sonographic findings in predicting the need for surgery. *Pediatric Radiology*, **2018**, 48, 1755-1762	2.8	10
79	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. *American Journal of Human Genetics*, **2019**, 104, 422-438	11	10
78	Homozygous/compound heterozygote RYR1 gene variants: Expanding the clinical spectrum. *American Journal of Medical Genetics, Part A*, **2019**, 179, 386-396	2.5	10
77	Prenatal detection of isolated bilateral hyperechogenic kidneys: Etiologies and outcomes. *Prenatal Diagnosis*, **2019**, 39, 693-700	3.2	9
76	SON haploinsufficiency causes impaired pre-mRNA splicing of CAKUT genes and heterogeneous renal phenotypes. *Kidney International*, **2019**, 95, 1494-1504	9.9	9
75	GATAD2B-associated neurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-related disorder. *Genetics in Medicine*, **2020**, 22, 878-888	8.1	9
74	Risk estimates for complex disorders: comparing personal genome testing and family history. *Genetics in Medicine*, **2014**, 16, 231-7	8.1	9
73	Fetal myelomeningocele surgery: Only treating the tip of the iceberg. *Prenatal Diagnosis*, **2019**, 39, 10-15	3.2	9
72	Characteristic MR Imaging Findings of the Neonatal Brain in RASopathies. *American Journal of Neuroradiology*, **2018**, 39, 1146-1152	4.4	8
71	De novo pathogenic variant in TUBB2A presenting with arthrogryposis multiplex congenita, brain abnormalities, and severe developmental delay. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2725-2730	2.5	8
70	Severe classical congenital muscular dystrophy and merosin expression. *Clinical Genetics*, **1998**, 54, 193-8	4	8
69	Interstitial 7q deletion [46,XX,del(7)(pter----q21.1::q22----qter)] and the location of genes for beta-glucuronidase and cystic fibrosis. *American Journal of Medical Genetics Part A*, **1988**, 31, 655-61		8
68	Mapping the cellular origin and early evolution of leukemia in Down syndrome. *Science*, **2021**, 373,	33.3	8
67	"A change in perspective": Exploring the experiences of adolescents with hereditary tumor predisposition. *Pediatric Blood and Cancer*, **2019**, 66, e27445	3	8
66	Abnormal fetal cerebral and vascular development in hypoplastic left heart syndrome. *Prenatal Diagnosis*, **2019**, 39, 38-44	3.2	8
65	Human IFT52 mutations uncover a novel role for the protein in microtubule dynamics and centrosome cohesion. *Human Molecular Genetics*, **2019**, 28, 2720-2737	5.6	7
64	Hb FM-Fort Ripley: confirmation of autosomal dominant inheritance and diagnosis by PCR and direct nucleotide sequencing. *Human Mutation*, **1994**, 3, 239-42	4.7	7
63	Lethal congenital muscular dystrophy with cataracts and a minor brain anomaly: new entity or variant of Walker-Warburg syndrome?. *American Journal of Medical Genetics Part A*, **1991**, 39, 19-24		7

62	A homozygous pathogenic variant in SHROOM3 associated with anencephaly and cleft lip and palate. *Clinical Genetics*, **2020**, 98, 299-302	4	6
61	Mitochondrial POLG related disorder presenting prenatally with fetal cerebellar growth arrest. *Metabolic Brain Disease*, **2018**, 33, 1369-1373	3.9	6
60	Maternal uniparental disomy for chromosome 6 in a patient with IUGR, ambiguous genitalia, and persistent mullerian structures. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3227-3230	2.5	6
59	Arthrogryposis multiplex congenita, craniofacial, and ophthalmological abnormalities and normal intelligence: a new syndrome?. *American Journal of Medical Genetics Part A*, **1997**, 71, 401-5		6
58	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. *Journal of Human Genetics*, **2021**, 66, 995-1008	4.3	6
57	Fetal chondrodysplasia punctata associated with maternal autoimmune diseases: a review. *The Application of Clinical Genetics*, **2018**, 11, 31-44	3.1	6
56	Williams syndrome presenting with findings consistent with Alagille syndrome. *Clinical Case Reports (discontinued)*, **2015**, 3, 24-8	0.7	5
55	Syndrome of mental retardation and distal arthrogryposis in sibs. *American Journal of Medical Genetics Part A*, **1991**, 41, 49-51		5
54	Prenatally diagnosed omphaloceles: Report of 92 cases and association with Beckwith-Wiedemann syndrome. *Prenatal Diagnosis*, **2021**, 41, 798-816	3.2	5
53	Brain and Placental Pathology in Fetal COL4A1 Related Disease. *Pediatric and Developmental Pathology*, **2021**, 24, 175-186	2.2	5
52	The point-of-care use of a facial phenotyping tool in the genetics clinic: Enhancing diagnosis and education with machine learning. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1151-1158	2.5	5
51	Perplexed by PGx? Exploring the impact of pharmacogenomic results on medical management, disclosures and patient behavior. *Pharmacogenomics*, **2019**, 20, 319-329	2.6	4
50	Informed Decision-Making in the Context of Prenatal Chromosomal Microarray. *Journal of Genetic Counseling*, **2018**, 27, 1130-1147	2.5	4
49	Rationale for dopa-responsive CTNNB1/Ecatenin deficient dystonia. *Movement Disorders*, **2018**, 33, 656-657		4
48	Congenital limb deficiencies with vascular etiology: Possible association with maternal thrombophilia. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3083-3089	2.5	4
47	Prenatal genomic microarray and sequencing in Canadian medical practice: towards consensus. *Journal of Medical Genetics*, **2015**, 52, 585-6	5.8	4
46	Detection and enumeration of colonic mucosal cells in amniotic fluid using a colon epithelial-specific monoclonal antibody. *Prenatal Diagnosis*, **1990**, 10, 725-32	3.2	4
45	Familial renal hypophosphatemia, minor facial anomalies, intracerebral calcifications, and non-rachitic bone changes: apparently new syndrome?. *American Journal of Medical Genetics Part A*, **1990**, 35, 406-14		4

44	Pancreatic Cell-Specific Deletion of VPS41 Causes Diabetes Due to Defects in Insulin Secretion. *Diabetes*, **2021**, 70, 436-448	0.9	4
43	Haploinsufficiency of SF3B2 causes craniofacial microsomia. *Nature Communications*, **2021**, 12, 4680	17.4	4
42	Agenesis of the corpus callosum, developmental delay, autism spectrum disorder, facial dysmorphism, and posterior polymorphous corneal dystrophy associated with ZEB1 gene deletion. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2467-2471	2.5	4
41	Alveolar capillary dysplasia with misalignment of the pulmonary veins and hypoplastic left heart sequence caused by an in frame deletion within FOXF1. *American Journal of Medical Genetics, Part A* , **2019**, 179, 1325-1329	2.5	3
40	Impact of introduction of noninvasive prenatal testing on uptake of genetic testing in fetuses with central nervous system anomalies. *Prenatal Diagnosis*, **2019**, 39, 544-548	3.2	3
39	Expanding the genotypic and phenotypic spectrum of severe serine biosynthesis disorders. *Human Mutation*, **2020**, 41, 1615-1628	4.7	3
38	Prenatal presentation of hereditary hemorrhagic telangiectasia - a report of two sibs. *Prenatal Diagnosis*, **2016**, 36, 891-3	3.2	3
37	Challenges in Diagnosing Rare Genetic Causes of Common In Utero Presentations: Report of Two Patients with Mucolipidosis Type II (I-Cell Disease). *Journal of Pediatric Genetics*, **2018**, 7, 134-137	0.7	3
36	Prenatal and preconception genetic counseling for consanguinity: Consanguineous couples' expectations, experiences, and perspectives. *Journal of Genetic Counseling*, **2019**, 28, 982-992	2.5	3
35	Homozygous GLUL deletion is embryonically viable and leads to glutamine synthetase deficiency. *Clinical Genetics*, **2020**, 98, 613-619	4	3
34	The role of digital tools in the delivery of genomic medicine: enhancing patient-centered care. *Genetics in Medicine*, **2021**, 23, 1086-1094	8.1	3
33	Neurodegenerative VPS41 variants inhibit HOPS function and mTORC1-dependent TFEB/TFE3 regulation. *EMBO Molecular Medicine*, **2021**, 13, e13258	12	3
32	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. *American Journal of Human Genetics*, **2021**, 108, 1053-1068	11	3
31	Characteristic Cochlear Hypoplasia in Patients with Walker-Warburg Syndrome: A Radiologic Study of the Inner Ear in Dystroglycan-Related Muscular Disorders. *American Journal of Neuroradiology*, **2021**, 42, 167-172	4.4	3
30	Fetal Renal Echogenicity Associated with Maternal Focal Segmental Glomerulosclerosis: The Effect of Transplacental Transmission of Permeability Factor suPAR. *Journal of Clinical Medicine*, **2018**, 7,	5.1	3
29	Congenital hypothyroidism, cardiac defects, and pancreatic agenesis in an infant with GATA6 mutation. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1496-1499	2.5	2
28	Reply to the letter to the editor by Gripp et al.Not Antley-Bixler syndrome*American Journal of Medical Genetics Part A*, **1999**, 83, 67-68		2
27	Diagnostic yield of genome sequencing for fetal structural anomalies.. *Prenatal Diagnosis*, **2022**,	3.2	2

26	Exome and genome sequencing in adults with undiagnosed disease: a prospective cohort study. *Journal of Medical Genetics*, **2021**, 58, 275-283	5.8	2
25	Gene therapy: perspectives from young adults with Leber's congenital amaurosis. *Eye*, **2021**,	4.4	2
24	Clinical application of fetal genome-wide sequencing during pregnancy: position statement of the Canadian College of Medical Geneticists. *Journal of Medical Genetics*, **2021**,	5.8	2
23	Findings in children exposed in utero to phenytoin and carbamazepine monotherapy: Independent effects of epilepsy and medications **1997**, 68, 18		2
22	Partial tetrasomy with triplication of chromosome (5) (p14-p15.33) in a patient with severe multiple congenital anomalies **1998**, 79, 103		2
21	Pregnancy in 3M syndrome. *Journal of Obstetrics and Gynaecology*, **2018**, 38, 421-422	1.3	1
20	Mosaicism for a small marker chromosome resulting from a familial Robertsonian translocation (21;22). *Clinical Genetics*, **1999**, 56, 362-6	4	1
19	Genetic counselling for infertile men of known and unknown etiology. *Translational Andrology and Urology*, **2021**, 10, 1479-1485	2.3	1
18	Heterozygous NOTCH1 deletion associated with variable congenital heart defects. *Clinical Genetics*, **2021**, 99, 836-841	4	1
17	Mild Idiopathic Infantile Hypercalcemia-Part 1: Biochemical and Genetic Findings. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, 2915-2937	5.6	1
16	Newborn with bilateral congenital cataracts: Never forget congenital rubella syndrome. *Paediatrics and Child Health*, **2020**, 25, 72-76	0.7	1
15	Tumor surveillance for children and adolescents with cancer predisposition syndromes: The psychosocial impact reported by adolescents and caregivers. *Pediatric Blood and Cancer*, **2021**, 68, e29021	3	1
14	The phenotypic spectrum of AMER1-related osteopathia striata with cranial sclerosis: The first Canadian cohort. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3793-3803	2.5	1
13	Tissue-specific methylation differences and cognitive function in fragile X premutation females **1996**, 64, 329		1
12	Novel mutations of the P gene in type II oculocutaneous albinism (OCA2) **1997**, 10, 175		1
11	Autopsy findings in pontine tegmental cap dysplasia. *Canadian Journal of Neurological Sciences*, **2015**, 42, S6-S6	1	0
10	Adapting obstetric and neonatal services during the COVID-19 pandemic: a scoping review.. *BMC Pregnancy and Childbirth*, **2022**, 22, 119	3.2	0
9	Variants in cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy.. *Brain Communications*, **2021**, 3, fcab245	4.5	0

List of Publications

8	Maternal SLE and brachytelephalangic chondrodysplasia punctata in a patient with unrelated de novo RAF1 and SIX2 variants. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1807-1811	2.5
7	CHROMOSOMAL MICROARRAYS: THE BENEFITS AND CHALLENGES OF INTRODUCTION INTO PRENATAL DIAGNOSIS. *Fetal and Maternal Medicine Review*, **2010**, 21, 307-322	
6	Prenatal and Neonatal Screening **2006**, 163-218	
5	Dandy-Walker malformation syndromes: Reply to Fiumara et al.. *American Journal of Medical Genetics Part A*, **1996**, 63, 413-413	
4	Incomplete Platelet Dense Granule Formation in Normal Neonates.. *Blood*, **2007**, 110, 3210-3210	2.2
3	Fetal Macrocephaly: A Novel Sonographic Finding in Congenital Myotonic Dystrophy. *AJP Reports*, **2020**, 10, e294-e299	1.2
2	Helping nephrologists find answers: hyperinsulinism and tubular dysfunction: Questions. *Pediatric Nephrology*, **2020**, 35, 253-255	3.2
1	Helping nephrologists find answers: hyperinsulinism and tubular dysfunction: Answers. *Pediatric Nephrology*, **2020**, 35, 257-260	3.2