

David Chitayat

List of Publications by Year in descending order

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Version: 2024-02-01

169
papers

6,420
citations

87723

38
h-index

95083

68
g-index

173
all docs

173
docs citations

173
times ranked

9260
citing authors

#	ARTICLE	IF	CITATIONS
1	Evidence for multi-site closure of the neural tube in humans. American Journal of Medical Genetics Part A, 1993, 47, 723-743.	2.4	364
2	Heterozygous mutations in the gene encoding noggin affect human joint morphogenesis. Nature Genetics, 1999, 21, 302-304.	9.4	329
3	Discordant KCNQ1OT1 imprinting in sets of monozygotic twins discordant for Beckwith-Wiedemann syndrome. Human Molecular Genetics, 2002, 11, 1317-1325.	1.4	322
4	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	1.7	295
5	Mutations in CDMP1 cause autosomal dominant brachydactyly type C. Nature Genetics, 1997, 17, 18-19.	9.4	255
6	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.	2.6	166
7	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	9.4	164
8	PDE3A mutations cause autosomal dominant hypertension with brachydactyly. Nature Genetics, 2015, 47, 647-653.	9.4	146
9	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. American Journal of Human Genetics, 2017, 101, 466-477.	2.6	119
10	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.	2.6	100
11	A new mutation in the type II hair cortex keratin hHb1 involved in the inherited hair disorder monilethrix. Human Genetics, 1997, 101, 165-169.	1.8	99
12	Findings in children exposed in utero to phenytoin and carbamazepine monotherapy: Independent effects of epilepsy and medications. , 1997, 68, 18-24.		98
13	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.	0.3	98
14	The functional O-mannose glycan on Î±-dystroglycan contains a phospho-ribitol primed for matriglycan addition. ELife, 2016, 5, .	2.8	98
15	FGFR2 mutation associated with clinical manifestations consistent with Antley-Bixler syndrome. , 1998, 77, 219-224.		97
16	Prenatally diagnosed neural tube defects: Ultrasound, chromosome, and autopsy or postnatal findings in 212 cases. , 1998, 77, 317-321.		95
17	Alagille syndrome: clinical perspectives. The Application of Clinical Genetics, 2016, Volume 9, 75-82.	1.4	94
18	Intrinsic Endocardial Defects Contribute to Hypoplastic Left Heart Syndrome. Cell Stem Cell, 2020, 27, 574-589.e8.	5.2	89

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19	Lymphatic abnormalities in fetuses with posterior cervical cystic hygroma. American Journal of Medical Genetics Part A, 1989, 33, 352-356.	2.4	75
20	CpG Methylation, a Parent-of-Origin Effect for Maternal-Biased Transmission of Congenital Myotonic Dystrophy. American Journal of Human Genetics, 2017, 100, 488-505.	2.6	74
21	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.		72
22	Folic acid supplementation for pregnant women and those planning pregnancy: 2015 update. Journal of Clinical Pharmacology, 2016, 56, 170-175.	1.0	67
23	RETIRED: Prenatal Screening, Diagnosis, and Pregnancy Management of Fetal Neural Tube Defects. Journal of Obstetrics and Gynaecology Canada, 2014, 36, 927-939.	0.3	66
24	Biallelic mutations in the 3' exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. Nature Genetics, 2017, 49, 457-464.	9.4	66
25	Fetal Reprogramming and Senescence in Hypoplastic Left Heart Syndrome and in Human Pluripotent Stem Cells during Cardiac Differentiation. American Journal of Pathology, 2013, 183, 720-734.	1.9	65
26	Genomic study of severe fetal anomalies and discovery of GREB1L mutations in renal agenesis. Genetics in Medicine, 2018, 20, 745-753.	1.1	60
27	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	2.6	59
28	Congenital diaphragmatic hernia, coarse facies, and acral hypoplasia: Fryns syndrome. American Journal of Medical Genetics Part A, 1989, 32, 93-99.	2.4	56
29	Familial growth hormone deficiency associated with MRI abnormalities. American Journal of Medical Genetics Part A, 1998, 80, 128-132.	2.4	56
30	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	2.6	56
31	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, 272-277.	2.4	51
32	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, 422-424.	2.4	49
33	Anesthesia for Freeman-Sheldon syndrome using a laryngeal mask airway. Canadian Journal of Anaesthesia, 1999, 46, 783-787.	0.7	47
34	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.	2.6	47
35	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. JAMA Network Open, 2020, 3, e2018109.	2.8	47
36	The molecular and phenotypic spectrum of IQSEC2-related epilepsy. Epilepsia, 2016, 57, 1858-1869.	2.6	46

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37	Novel mutations of the tyrosinase (TYR) gene in type I oculocutaneous albinism (OCA1). , 1997, 10, 171-174.		45
38	Early-infantile galactosialidosis: Prenatal presentation and postnatal follow-up. American Journal of Medical Genetics Part A, 1999, 85, 38-47.	2.4	44
39	Clinical delineation of the <i>PACS1</i>-related syndromeâ€”Report on 19 patients. American Journal of Medical Genetics, Part A, 2016, 170, 670-675.	0.7	44
40	The PDAC syndrome (pulmonary hypoplasia/agenesis, diaphragmatic hernia/eventration,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 632 Td (Report of eight cases including a living child and further evidence for autosomal recessive inheritance. American Journal of Medical Genetics, Part A, 2007, 143A, 1268-1281.	0.7	43
41	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.	2.3	43
42	Histone H3.3 beyond cancer: Germline mutations in <i>Histone 3 Family 3A and 3B</i> cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
43	Haploinsufficiency of SF3B2 causes craniofacial microsomia. Nature Communications, 2021, 12, 4680.	5.8	43
44	Mapping the cellular origin and early evolution of leukemia in Down syndrome. Science, 2021, 373, .	6.0	42
45	Apparent postnatal onset of some manifestations of the Wiedemann-Beckwith syndrome. American Journal of Medical Genetics Part A, 1990, 36, 434-439.	2.4	41
46	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, 3038-3053.	0.7	38
47	Novel mutations of theP gene in type II oculocutaneous albinism (OCA2). Human Mutation, 1997, 10, 175-177.	1.1	37
48	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, 758-763.	0.7	37
49	ATP6AP2 variant impairs CNS development and neuronal survival to cause fulminant neurodegeneration. Journal of Clinical Investigation, 2019, 129, 2145-2162.	3.9	37
50	Spectrum and Outcome of Primary Cardiomyopathies Diagnosed During Fetal Life. JACC: Heart Failure, 2014, 2, 403-411.	1.9	36
51	Fetal arthrogryposis multiplex congenita/fetal akinesia deformation sequence (FADS)â€”Aetiology, diagnosis, and management. Prenatal Diagnosis, 2019, 39, 720-731.	1.1	35
52	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, 406-415.	2.4	34
53	Brain abnormalities in patients with Beckwithâ€”Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 1388-1394.	0.7	34
54	Penoscrotal transposition: A case report and review. American Journal of Medical Genetics Part A, 1994, 49, 103-107.	2.4	33

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55	Omphalocele in Miller-Dieker syndrome: Expanding the phenotype. , 1997, 69, 293-298.		33
56	Compound heterozygosity for the achondroplasia-hypochondroplasia FGFR3 mutations: Prenatal diagnosis and postnatal outcome. American Journal of Medical Genetics Part A, 1999, 84, 401-405.	2.4	33
57	Deletion of 15q11.2(BP1â€BP2) region; Further evidence for lack of phenotypic specificity in a pediatric population. American Journal of Medical Genetics, Part A, 2015, 167, 2098-2102.	0.7	33
58	Dual loss of p110Î PI3-kinase and SKAP (KNSTRN) expression leads to combined immunodeficiency and multisystem syndromic features. Journal of Allergy and Clinical Immunology, 2018, 142, 618-629.	1.5	33
59	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.	2.4	32
60	De novo missense variants in RAC3 cause a novel neurodevelopmental syndrome. Genetics in Medicine, 2019, 21, 1021-1026.	1.1	32
61	Syndrome of proximal interstitial deletion 4p15: Report of three cases and review of the literature. American Journal of Medical Genetics Part A, 1995, 55, 147-154.	2.4	31
62	Prenatal diagnosis of retinal nonattachment in the Walker-Warburg syndrome. American Journal of Medical Genetics Part A, 1995, 56, 351-358.	2.4	31
63	Chitayat-Hall and Schaaf-Yang syndromes:a common aetiology: expanding the phenotype of <i>MAGEL2</i>-related disorders. Journal of Medical Genetics, 2018, 55, 316-321.	1.5	31
64	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.	2.6	31
65	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.	2.6	30
66	Prenatal diagnosis and fetopathological findings in five fetuses with trisomy 9. American Journal of Medical Genetics Part A, 1995, 56, 247-251.	2.4	29
67	Tissue-specific methylation differences and cognitive function in fragile X premutation females. , 1996, 64, 329-333.		29
68	The Relationship Between Burnout and Occupational Stress in Genetic Counselors. Journal of Genetic Counseling, 2016, 25, 731-741.	0.9	29
69	Terminal deletion of the long arm of chromosome 3 [46,XX,del(3)(q27â†qter)]. , 1996, 61, 45-48.		28
70	Limb defects in homozygous Î±-thalassemia: Report of three cases. , 1997, 68, 162-167.		28
71	Congenital toxoplasmosis: prenatal diagnosis, treatment and postnatal outcome. , 1999, 19, 330-333.		28
72	De novo 46,XX,t(6;7)(q27;q11;23) associated with severe cardiovascular manifestations characteristic of supraaortic stenosis and Williams syndrome. , 2000, 90, 270-275.		28

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73	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	2.6	27
74	Congenital heart disease and Robinow syndrome: Coincidence or an additional component of the syndrome?. American Journal of Medical Genetics Part A, 1990, 37, 519-521.	2.4	26
75	Raine syndrome: A rare lethal osteosclerotic bone dysplasia. Prenatal diagnosis, autopsy, and neuropathological findings. American Journal of Medical Genetics, Part A, 2007, 143A, 3280-3285.	0.7	26
76	Neurodegenerative <i>VPS41</i> variants inhibit HOPS function and mTORC1-dependent TFEB/TFE3 regulation. EMBO Molecular Medicine, 2021, 13, e13258.	3.3	26
77	The expanding clinical phenotype of the tRNA ^{Leu} (UUR) A→G mutation at np 3243 of mitochondrial DNA: Diabetic embryopathy associated with mitochondrial cytopathy. , 1996, 62, 404-409.		25
78	New insights into DNA methylation signatures: SMARCA2 variants in Nicolaides-Baraitser syndrome. BMC Medical Genomics, 2019, 12, 105.	0.7	25
79	Inverted duplication of the distal short arm of chromosome 3 associated with lobar holoprosencephaly and lumbosacral meningomyelocele. , 2000, 91, 167-170.		24
80	Role of amniotic fluid interphase fluorescence in situ hybridization (FISH) analysis in patient management. Prenatal Diagnosis, 2001, 21, 327-332.	1.1	24
81	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	2.6	24
82	Human IFT52 mutations uncover a novel role for the protein in microtubule dynamics and centrosome cohesion. Human Molecular Genetics, 2019, 28, 2720-2737.	1.4	23
83	GATAD2B-associated neurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-related disorder. Genetics in Medicine, 2020, 22, 878-888.	1.1	22
84	Prenatal detection of isolated bilateral hyperechogenic kidneys: Etiologies and outcomes. Prenatal Diagnosis, 2019, 39, 693-700.	1.1	21
85	Hepatocellular carcinoma in a child with familial Russell-Silver syndrome. American Journal of Medical Genetics Part A, 1988, 31, 909-914.	2.4	19
86	Genetic homogeneity of cartilage-hair hypoplasia. Human Genetics, 1995, 95, 157-160.	1.8	19
87	Homozygous/compound heterozygote <i>RYR1</i> gene variants: Expanding the clinical spectrum. American Journal of Medical Genetics, Part A, 2019, 179, 386-396.	0.7	19
88	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.	1.1	19
89	Experience with genetic counseling: the adolescent perspective. Journal of Genetic Counseling, 2016, 25, 583-595.	0.9	18
90	Mutations in the <i>NEB</i> gene cause fetal akinesia/arthrogryposis multiplex congenita. Prenatal Diagnosis, 2017, 37, 144-150.	1.1	18

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91	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.	0.7	18
92	<scp>The pointâ€ofâ€care</scp> 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.	0.7	18
93	The role of digital tools in the delivery of genomic medicine: enhancing patient-centered care. Genetics in Medicine, 2021, 23, 1086-1094.	1.1	18
94	Juvenile galactosialidosis in a white male: A new variant. American Journal of Medical Genetics Part A, 1988, 31, 887-901.	2.4	17
95	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.	0.7	17
96	Fetal myelomeningocele surgery: Only treating the tip of the iceberg. Prenatal Diagnosis, 2019, 39, 10-15.	1.1	17
97	SON haploinsufficiency causes impaired pre-mRNA splicing of CAKUT genes and heterogeneous renal phenotypes. Kidney International, 2019, 95, 1494-1504.	2.6	17
98	Syndrome of mental retardation, facial anomalies, hypopituitarism, and distal arthrogryposis in sibs. American Journal of Medical Genetics Part A, 1990, 37, 65-70.	2.4	16
99	Perinatal and first year followâ€up of patients with Praderâ€Willi syndrome: normal size of hands and feet. Clinical Genetics, 1989, 35, 161-166.	1.0	16
100	Warsaw breakage syndrome: Further clinical and genetic delineation. American Journal of Medical Genetics, Part A, 2018, 176, 2404-2418.	0.7	16
101	Congenital abnormalities in two sibs exposed to valproic acid in utero. American Journal of Medical Genetics Part A, 1988, 31, 369-373.	2.4	15
102	De novo pathogenic variant in TUBB2A presenting with arthrogryposis multiplex congenita, brain abnormalities, and severe developmental delay. , 2017, 173, 2725-2730.		15
103	The pathology of incipient polymicrogyria. Brain and Development, 2017, 39, 23-39.	0.6	15
104	Abnormal fetal cerebral and vascular development in hypoplastic left heart syndrome. Prenatal Diagnosis, 2019, 39, 38-44.	1.1	15
105	Brachydactyly-short stature-hypertension (Bilginturan) syndrome: Report on two families. , 1997, 73, 279-285.		14
106	Evidence for somatic and germline mosaicism in CRASH syndrome. Human Mutation, 1998, 11, S284-S287.	1.1	14
107	Meconium peritonitis: the role of postnatal radiographic and sonographic findings in predicting the need for surgery. Pediatric Radiology, 2018, 48, 1755-1762.	1.1	14
108	Expanding the genotypic and phenotypic spectrum of severe serine biosynthesis disorders. Human Mutation, 2020, 41, 1615-1628.	1.1	14

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109	Exome and genome sequencing in adults with undiagnosed disease: a prospective cohort study. <i>Journal of Medical Genetics</i> , 2021, 58, 275-283.	1.5	14
110	â€œA change in perspectiveâ€ Exploring the experiences of adolescents with hereditary tumor predisposition. <i>Pediatric Blood and Cancer</i> , 2019, 66, e27445.	0.8	13
111	Clinical application of fetal genome-wide sequencing during pregnancy: position statement of the Canadian College of Medical Geneticists. <i>Journal of Medical Genetics</i> , 2022, 59, 931-937.	1.5	13
112	Agenesis of the corpus callosum, developmental delay, autism spectrum disorder, facial dysmorphism, and posterior polymorphous corneal dystrophy associated with ZEB1 gene deletion. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2467-2471.	0.7	13
113	Hb FM-Fort Ripley: Confirmation of autosomal dominant inheritance and diagnosis by PCR and direct nucleotide sequencing. <i>Human Mutation</i> , 1994, 3, 239-242.	1.1	12
114	Characteristic MR Imaging Findings of the Neonatal Brain in RASopathies. <i>American Journal of Neuroradiology</i> , 2018, 39, 1146-1152.	1.2	12
115	Diagnostic yield of genome sequencing for prenatal diagnosis of fetal structural anomalies. <i>Prenatal Diagnosis</i> , 2022, 42, 822-830.	1.1	12
116	Prenatally diagnosed omphaloceles: Report of 92 cases and association with Beckwithâ€Wiedemann syndrome. <i>Prenatal Diagnosis</i> , 2021, 41, 798-816.	1.1	11
117	Risk estimates for complex disorders: comparing personal genome testing and family history. <i>Genetics in Medicine</i> , 2014, 16, 231-237.	1.1	10
118	Prenatal and preconception genetic counseling for consanguinity: Consanguineous couples' expectations, experiences, and perspectives. <i>Journal of Genetic Counseling</i> , 2019, 28, 982-992.	0.9	10
119	Alveolar capillary dysplasia with misalignment of the pulmonary veins and hypoplastic left heart sequence caused by an in frame deletion within <i>FOXF1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1325-1329.	0.7	10
120	Pancreatic Î²-Cellâ€Specific Deletion of VPS41 Causes Diabetes Due to Defects in Insulin Secretion. <i>Diabetes</i> , 2021, 70, 436-448.	0.3	10
121	Variants in <i>ATP6VOA1</i> cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy. <i>Brain Communications</i> , 2021, 3, fcab245.	1.5	10
122	Adapting obstetric and neonatal services during the COVID-19 pandemic: a scoping review. <i>BMC Pregnancy and Childbirth</i> , 2022, 22, 119.	0.9	10
123	Arthrogyrosis multiplex congenita, craniofacial, and ophthalmological abnormalities and normal intelligence: A new syndrome?. , 1997, 71, 401-405.		9
124	Severe classical congenital muscular dystrophy and merosin expression. <i>Clinical Genetics</i> , 1998, 54, 193-198.	1.0	9
125	Perplexed by PGx? Exploring the impact of pharmacogenomic results on medical management, disclosures and patient behavior. <i>Pharmacogenomics</i> , 2019, 20, 319-329.	0.6	9
126	Characteristic Cochlear Hypoplasia in Patients with Walker-Warburg Syndrome: A Radiologic Study of the Inner Ear in Î±-Dystroglycanâ€Related Muscular Disorders. <i>American Journal of Neuroradiology</i> , 2021, 42, 167-172.	1.2	9

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127	Tumor surveillance for children and adolescents with cancer predisposition syndromes: The psychosocial impact reported by adolescents and caregivers. <i>Pediatric Blood and Cancer</i> , 2021, 68, e29021.	0.8	9
128	Interstitial 7q deletion [46,XX,del(7)(pter â†’ q21.1::q22 â†’ qter)] and the location of genes for Î²-glucuronidase and cystic fibrosis. <i>American Journal of Medical Genetics Part A</i> , 1988, 31, 655-661.	2.4	8
129	Lethal congenital muscular dystrophy with cataracts and a minor brain anomaly: New entity or variant of Walker-Warburg syndrome?. <i>American Journal of Medical Genetics Part A</i> , 1991, 39, 19-24.	2.4	8
130	Congenital limb deficiencies with vascular etiology: Possible association with maternal thrombophilia. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3083-3089.	0.7	8
131	Rationale for dopaâ€responsive <i>CTNNB1/ÃŸ</i>â€catenin deficient dystonia. <i>Movement Disorders</i> , 2018, 33, 656-657.	2.2	8
132	Fetal chondrodysplasia punctata associated with maternal autoimmune diseases: a review. <i>The Application of Clinical Genetics</i> , 2018, Volume 11, 31-44.	1.4	8
133	A homozygous pathogenic variant in <scp><i>SHROOM3</i></scp> associated with anencephaly and cleft lip and palate. <i>Clinical Genetics</i> , 2020, 98, 299-302.	1.0	8
134	Congenital hypothyroidism, cardiac defects, and pancreatic agenesis in an infant with <scp>GATA6</scp> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1496-1499.	0.7	8
135	Brain and Placental Pathology in Fetal COL4A1 Related Disease. <i>Pediatric and Developmental Pathology</i> , 2021, 24, 175-186.	0.5	8
136	Mild Idiopathic Infantile Hypercalcemiaâ€”Part 1: Biochemical and Genetic Findings. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, 2915-2937.	1.8	8
137	Williams syndrome presenting with findings consistent with Alagille syndrome. <i>Clinical Case Reports (discontinued)</i> , 2015, 3, 24-28.	0.2	7
138	Informed Decisionâ€Making in the Context of Prenatal Chromosomal Microarray. <i>Journal of Genetic Counseling</i> , 2018, 27, 1130-1147.	0.9	7
139	Mitochondrial POLG related disorder presenting prenatally with fetal cerebellar growth arrest. <i>Metabolic Brain Disease</i> , 2018, 33, 1369-1373.	1.4	7
140	Heterozygous <scp><i>NOTCH1</i></scp> deletion associated with variable congenital heart defects. <i>Clinical Genetics</i> , 2021, 99, 836-841.	1.0	7
141	Detection and enumeration of colonic mucosal cells in amniotic fluid using a colon epithelial-specific monoclonal antibody. <i>Prenatal Diagnosis</i> , 1990, 10, 725-732.	1.1	6
142	Familial renal hypophosphatemia, minor facial anomalies, intracerebral calcifications, and non-rachitic bone changes: Apparently new syndrome?. <i>American Journal of Medical Genetics Part A</i> , 1990, 35, 406-414.	2.4	6
143	Prenatal genomic microarray and sequencing in Canadian medical practice: towards consensus. <i>Journal of Medical Genetics</i> , 2015, 52, 585-586.	1.5	6
144	Syndrome of mental retardation and distal arthrogryposis in sibs. <i>American Journal of Medical Genetics Part A</i> , 1991, 41, 49-51.	2.4	5

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145	Fetal Renal Echogenicity Associated with Maternal Focal Segmental Glomerulosclerosis: The Effect of Transplacental Transmission of Permeability Factor suPAR. <i>Journal of Clinical Medicine</i> , 2018, 7, 324.	1.0	5
146	Impact of introduction of noninvasive prenatal testing on uptake of genetic testing in fetuses with central nervous system anomalies. <i>Prenatal Diagnosis</i> , 2019, 39, 544-548.	1.1	5
147	Homozygous GLUL deletion is embryonically viable and leads to glutamine synthetase deficiency. <i>Clinical Genetics</i> , 2020, 98, 613-619.	1.0	5
148	Genetic counselling for infertile men of known and unknown etiology. <i>Translational Andrology and Urology</i> , 2021, 10, 1479-1485.	0.6	5
149	Challenges in Diagnosing Rare Genetic Causes of Common In Utero Presentations: Report of Two Patients with Mucopolipidosis Type II (I-Cell Disease). <i>Journal of Pediatric Genetics</i> , 2018, 07, 134-137.	0.3	4
150	Gene therapy: perspectives from young adults with Leber's congenital amaurosis. <i>Eye</i> , 2022, 36, 2088-2093.	1.1	4
151	Prenatal presentation of hereditary hemorrhagic telangiectasia – a report of two sibs. <i>Prenatal Diagnosis</i> , 2016, 36, 891-893.	1.1	3
152	Novel mutations of the P gene in type II oculocutaneous albinism (OCA2). <i>Human Mutation</i> , 1997, 10, 175-177.	1.1	3
153	KMT2D-NOTCH Mediates Coronary Abnormalities in Hypoplastic Left Heart Syndrome. <i>Circulation Research</i> , 2022, 131, 280-282.	2.0	3
154	Reply to the letter to the editor by Gripp et al.??Not Antley-Bixler syndrome?. <i>American Journal of Medical Genetics Part A</i> , 1999, 83, 67-68.	2.4	2
155	Pregnancy in 3M syndrome. <i>Journal of Obstetrics and Gynaecology</i> , 2018, 38, 421-422.	0.4	2
156	Newborn with bilateral congenital cataracts: Never forget congenital rubella syndrome. <i>Paediatrics and Child Health</i> , 2020, 25, 72-76.	0.3	2
157	The phenotypic spectrum of AMER1 related osteopathia striata with cranial sclerosis: The first Canadian cohort. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3793-3803.	0.7	2
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