David Chitayat

List of Publications by Year
in descending order

169 papers	6,420 citations	87888 38 h-index	88630 70 g-index
173 all docs	173 docs citations	173 times ranked	9260 citing authors

#	Article	IF	Citations
1	Gene therapy: perspectives from young adults with Leberâ€™s congenital amaurosis. Eye, 2022, 36, 2088-2093.	2.1	4
2	Clinical application of fetal genome-wide sequencing during pregnancy: position statement of the Canadian College of Medical Geneticists. Journal of Medical Genetics, 2022, 59, 931-937.	3.2	13
3	Diagnostic yield of genome sequencing for prenatal diagnosis of fetal structural anomalies. Prenatal Diagnosis, 2022, 42, 822-830.	2.3	12
4	Adapting obstetric and neonatal services during the COVID-19 pandemic: a scoping review. BMC Pregnancy and Childbirth, 2022, 22, 119.	2.4	10
5	KMT2D-NOTCH Mediates Coronary Abnormalities in Hypoplastic Left Heart Syndrome. Circulation Research, 2022, 131, 280-282.	4.5	3
6	Exome and genome sequencing in adults with undiagnosed disease: a prospective cohort study. Journal of Medical Genetics, 2021, 58, 275-283.	3.2	14
7	Pancreatic Î²-Cellâ€“Specific Deletion of VPS41 Causes Diabetes Due to Defects in Insulin Secretion. Diabetes, 2021, 70, 436-448.	0.6	10
8	Characteristic Cochlear Hypoplasia in Patients with Walker-Warburg Syndrome: A Radiologic Study of the Inner Ear in Î±-Dystroglycanâ€“Related Muscular Disorders. American Journal of Neuroradiology, 2021, 42, 167-172.	2.4	9
9	Brain and Placental Pathology in Fetal COL4A1 Related Disease. Pediatric and Developmental Pathology, 2021, 24, 175-186.	1.0	8
10	<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.	1.2	18
11	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.	1.5	9
12	Genetic counselling for infertile men of known and unknown etiology. Translational Andrology and Urology, 2021, 10, 1479-1485.	1.4	5
13	The role of digital tools in the delivery of genomic medicine: enhancing patient-centered care. Genetics in Medicine, 2021, 23, 1086-1094.	2.4	18
14	Prenatally diagnosed omphaloceles: Report of 92 cases and association with Beckwithâ€Wiedemann syndrome. Prenatal Diagnosis, 2021, 41, 798-816.	2.3	11
15	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.	2.3	19
16	Neurodegenerative VPS41 variants inhibit HOPS function and mTORC1â€dependent TFEB/TFE3 regulation. EMBO Molecular Medicine, 2021, 13, e13258.	6.9	26
17	Heterozygous <scp>NOTCH1</scp> deletion associated with variable congenital heart defects. Clinical Genetics, 2021, 99, 836-841.	2.0	7
18	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.	6.2	31

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19	Mild Idiopathic Infantile Hypercalcemiaâ€"Part 1: Biochemical and Genetic Findings. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 2915-2937.	3.6	8
20	Mapping the cellular origin and early evolution of leukemia in Down syndrome. Science, 2021, 373, .	12.6	42
21	Haploinsufficiency of SF3B2 causes craniofacial microsomia. Nature Communications, 2021, 12, 4680.	12.8	43
22	The phenotypic spectrum of AMER1 â€related osteopathia striata with cranial sclerosis: The first Canadian cohort. American Journal of Medical Genetics, Part A, 2021, 185, 3793-3803.	1.2	2
23	Variants in ATP6V0A1 cause progressive myoclonus epilepsy and developmental and epileptic encephalopathy. Brain Communications, 2021, 3, fcab245.	3.3	10
24	Newborn with bilateral congenital cataracts: Never forget congenital rubella syndrome. Paediatrics and Child Health, 2020, 25, 72-76.	0.6	2
25	Helping nephrologists find answers: hyperinsulinism and tubular dysfunction: Questions. Pediatric Nephrology, 2020, 35, 253-255.	1.7	0
26	Helping nephrologists find answers: hyperinsulinism and tubular dysfunction: Answers. Pediatric Nephrology, 2020, 35, 257-260.	1.7	0
27	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.	6.2	30
28	Intrinsic Endocardial Defects Contribute to Hypoplastic Left Heart Syndrome. Cell Stem Cell, 2020, 27, 574-589.e8.	11.1	89
29	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. JAMA Network Open, 2020, 3, e2018109.	5.9	47
30	Fetal Macrocephaly: A Novel Sonographic Finding in Congenital Myotonic Dystrophy. AJP Reports, 2020, 10, e294-e299.	0.7	1
31	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
32	Homozygous GLUL deletion is embryonically viable and leads to glutamine synthetase deficiency. Clinical Genetics, 2020, 98, 613-619.	2.0	5
33	Expanding the genotypic and phenotypic spectrum of severe serine biosynthesis disorders. Human Mutation, 2020, 41, 1615-1628.	2.5	14
34	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.	1.2	0
35	A homozygous pathogenic variant in <scp>SHROOM3</scp> associated with anencephaly and cleft lip and palate. Clinical Genetics, 2020, 98, 299-302.	2.0	8
36	GATAD2B-associatedneurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-relateddisorder. Genetics in Medicine, 2020, 22, 878-888.	2.4	22

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37	Congenital hypothyroidism, cardiac defects, and pancreatic agenesis in an infant with <scp>GATA6</scp> mutation. American Journal of Medical Genetics, Part A, 2020, 182, 1496-1499.	1.2	8
38	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	6.2	59
39	Fetal myelomeningocele surgery: Only treating the tip of the iceberg. Prenatal Diagnosis, 2019, 39, 10-15.	2.3	17
40	Prenatal and preconception genetic counseling for consanguinity: Consanguineous couples' expectations, experiences, and perspectives. Journal of Genetic Counseling, 2019, 28, 982-992.	1.6	10
41	New insights into DNA methylation signatures: SMARCA2 variants in Nicolaides-Baraitser syndrome. BMC Medical Genomics, 2019, 12, 105.	1.5	25
42	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	6.2	24
43	Prenatal detection of isolated bilateral hyperechogenic kidneys: Etiologies and outcomes. Prenatal Diagnosis, 2019, 39, 693-700.	2.3	21
44	Fetal arthrogryposis multiplex congenita/fetal akinesia deformation sequence (FADS)â€"Aetiology, diagnosis, and management. Prenatal Diagnosis, 2019, 39, 720-731.	2.3	35
45	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.	1.2	10
46	Perplexed by PGx? Exploring the impact of pharmacogenomic results on medical management, disclosures and patient behavior. Pharmacogenomics, 2019, 20, 319-329.	1.3	9
47	Human IFT52 mutations uncover a novel role for the protein in microtubule dynamics and centrosome cohesion. Human Molecular Genetics, 2019, 28, 2720-2737.	2.9	23
48	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.	2.3	5
49	SON haploinsufficiency causes impaired pre-mRNA splicing of CAKUT genes and heterogeneous renalÂphenotypes. Kidney International, 2019, 95, 1494-1504.	5.2	17
50	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
51	â€œA change in perspectiveâ€: Exploring the experiences of adolescents with hereditary tumor predisposition. Pediatric Blood and Cancer, 2019, 66, e27445.	1.5	13
52	Abnormal fetal cerebral and vascular development in hypoplastic left heart syndrome. Prenatal Diagnosis, 2019, 39, 38-44.	2.3	15
53	Homozygous/compound heterozygote RYR1 gene variants: Expanding the clinical spectrum. American Journal of Medical Genetics, Part A, 2019, 179, 386-396.	1.2	19
54	De novo missense variants in RAC3 cause a novel neurodevelopmental syndrome. Genetics in Medicine, 2019, 21, 1021-1026.	2.4	32

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55	ATP6AP2 variant impairs CNS development and neuronal survival to cause fulminant neurodegeneration. Journal of Clinical Investigation, 2019, 129, 2145-2162.	8.2	37
56	Informed Decisionâ€Making in the Context of Prenatal Chromosomal Microarray. Journal of Genetic Counseling, 2018, 27, 1130-1147.	1.6	7
57	Characteristic MR Imaging Findings of the Neonatal Brain in RASopathies. American Journal of Neuroradiology, 2018, 39, 1146-1152.	2.4	12
58	Rationale for dopaâ€responsive CTNNB1/ÃŸâ€catenin deficient dystonia. Movement Disorders, 2018, 33, 656-657.	3.9	8
59	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.	2.9	33
60	Chitayat-Hall and Schaaf-Yang syndromes:a common aetiology: expanding the phenotype of MAGEL2-related disorders. Journal of Medical Genetics, 2018, 55, 316-321.	3.2	31
61	Mitochondrial POLG related disorder presenting prenatally with fetal cerebellar growth arrest. Metabolic Brain Disease, 2018, 33, 1369-1373.	2.9	7
62	Pregnancy in 3M syndrome. Journal of Obstetrics and Gynaecology, 2018, 38, 421-422.	0.9	2
63	Genomic study of severe fetal anomalies and discovery of GREB1L mutations in renal agenesis. Genetics in Medicine, 2018, 20, 745-753.	2.4	60
64	Fetal Renal Echogenicity Associated with Maternal Focal Segmental Glomerulosclerosis: The Effect of Transplacental Transmission of Permeability Factor suPAR. Journal of Clinical Medicine, 2018, 7, 324.	2.4	5
65	Warsaw breakage syndrome: Further clinical and genetic delineation. American Journal of Medical Genetics, Part A, 2018, 176, 2404-2418.	1.2	16
66	Fetal chondrodysplasia punctata associated with maternal autoimmune diseases: a review. The Application of Clinical Genetics, 2018, Volume 11, 31-44.	3.0	8
67	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.	6.2	47
68	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	6.2	56
69	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.	1.2	18
70	Meconium peritonitis: the role of postnatal radiographic and sonographic findings in predicting the need for surgery. Pediatric Radiology, 2018, 48, 1755-1762.	2.0	14
71	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, 07, 134-137.	0.7	4
72	Biallelic mutations in the 3â€² exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. Nature Genetics, 2017, 49, 457-464.	21.4	66

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73	CpG Methylation, a Parent-of-Origin Effect for Maternal-Biased Transmission of Congenital Myotonic Dystrophy. American Journal of Human Genetics, 2017, 100, 488-505.	6.2	74
74	Mutations in the NEB gene cause fetal akinesia/arthrogryposis multiplex congenita. Prenatal Diagnosis, 2017, 37, 144-150.	2.3	18
75	De novo pathogenic variant in TUBB2A presenting with arthrogryposis multiplex congenita, brain abnormalities, and severe developmental delay. , 2017, 173, 2725-2730.		15
76	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.	0.7	98
77	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. American Journal of Human Genetics, 2017, 101, 466-477.	6.2	119
78	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	21.4	164
79	The pathology of incipient polymicrogyria. Brain and Development, 2017, 39, 23-39.	1.1	15
80	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.	1.2	13
81	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.	6.2	166
82	Alagille syndrome: clinical perspectives. The Application of Clinical Genetics, 2016, Volume 9, 75-82.	3.0	94
83	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.	6.2	100
84	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
85	The molecular and phenotypic spectrum of <scp>IQSEC</scp>2â€related epilepsy. Epilepsia, 2016, 57, 1858-1869.	5.1	46
86	Congenital limb deficiencies with vascular etiology: Possible association with maternal thrombophilia. American Journal of Medical Genetics, Part A, 2016, 170, 3083-3089.	1.2	8
87	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.	1.2	17
88	Prenatal presentation of hereditary hemorrhagic telangiectasia â€“ a report of two sibs. Prenatal Diagnosis, 2016, 36, 891-893.	2.3	3
89	The Relationship Between Burnout and Occupational Stress in Genetic Counselors. Journal of Genetic Counseling, 2016, 25, 731-741.	1.6	29
90	Experience with genetic counseling: the adolescent perspective. Journal of Genetic Counseling, 2016, 25, 583-595.	1.6	18

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91	Clinical delineation of the PACS1â€related syndromeâ€"Report on 19 patients. American Journal of Medical Genetics, Part A, 2016, 170, 670-675.	1.2	44
92	Folic acid supplementation for pregnant women and those planning pregnancy: 2015 update. Journal of Clinical Pharmacology, 2016, 56, 170-175.	2.0	67
93	The functional O-mannose glycan on Î±-dystroglycan contains a phospho-ribitol primed for matriglycan addition. ELife, 2016, 5, .	6.0	98
94	Autopsy findings in pontine tegmental cap dysplasia. Canadian Journal of Neurological Sciences, 2015, 42, S6-S6.	0.5	1
95	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.	1.2	33
96	Prenatal genomic microarray and sequencing in Canadian medical practice: towards consensus. Journal of Medical Genetics, 2015, 52, 585-586.	3.2	6
97	PDE3A mutations cause autosomal dominant hypertension with brachydactyly. Nature Genetics, 2015, 47, 647-653.	21.4	146
98	Williams syndrome presenting with findings consistent with Alagille syndrome. Clinical Case Reports (discontinued), 2015, 3, 24-28.	0.5	7
99	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.	4.5	43
100	RETIRED: Prenatal Screening, Diagnosis, and Pregnancy Management of Fetal Neural Tube Defects. Journal of Obstetrics and Gynaecology Canada, 2014, 36, 927-939.	0.7	66
101	Risk estimates for complex disorders: comparing personal genome testing and family history. Genetics in Medicine, 2014, 16, 231-237.	2.4	10
102	Spectrum and Outcome of Primary Cardiomyopathies Diagnosed During Fetal Life. JACC: Heart Failure, 2014, 2, 403-411.	4.1	36
103	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.	3.8	65
104	Brain abnormalities in patients with Beckwithâ€"Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 1388-1394.	1.2	34
105	CHROMOSOMAL MICROARRAYS: THE BENEFITS AND CHALLENGES OF INTRODUCTION INTO PRENATAL DIAGNOSIS. Fetal and Maternal Medicine Review, 2010, 21, 307-322.	0.3	0
106	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.	1.2	38
107	The PDAC syndrome (pulmonary hypoplasia/agenesis, diaphragmatic hernia/eventration,) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf 5 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.	1.2	43
108	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.	1.2	26

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109	Incomplete Platelet Dense Granule Formation in Normal Neonates.. Blood, 2007, 110, 3210-3210.	1.4	0
110	Discordant KCNQ1OT1 imprinting in sets of monozygotic twins discordant for Beckwith-Wiedemann syndrome. Human Molecular Genetics, 2002, 11, 1317-1325.	2.9	322
111	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.	1.3	37
112	Role of amniotic fluid interphase fluorescencein situ hybridization (FISH) analysis in patient management. Prenatal Diagnosis, 2001, 21, 327-332.	2.3	24
113	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, 270-275.	2.4	28
114	Inverted duplication of the distal short arm of chromosome 3 associated with lobar holoprosencephaly and lumbosacral meningomyelocele. , 2000, 91, 167-170.		24
115	Mosaicism for a small marker chromosome resulting from a familial Robertsonian translocation (21;22). Clinical Genetics, 1999, 56, 363-367.	2.0	1
116	Heterozygous mutations in the gene encoding noggin affect human joint morphogenesis. Nature Genetics, 1999, 21, 302-304.	21.4	329
117	Anesthesia for Freeman-Sheldon syndrome using a laryngeal mask airway. Canadian Journal of Anaesthesia, 1999, 46, 783-787.	1.6	47
118	Congenital toxoplasmosis: prenatal diagnosis, treatment and postnatal outcome. , 1999, 19, 330-333.		28
119	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.	2.4	2
120	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
121	Early-infantile galactosialidosis: Prenatal presentation and postnatal follow-up. American Journal of Medical Genetics Part A, 1999, 85, 38-47.	2.4	44
122	Evidence for somatic and germline mosaicism in CRASH syndrome. Human Mutation, 1998, 11, S284-S287.	2.5	14
123	FGFR2 mutation associated with clinical manifestations consistent with Antley-Bixler syndrome. American Journal of Medical Genetics Part A, 1998, 77, 219-224.	2.4	97
124	Prenatally diagnosed neural tube defects: Ultrasound, chromosome, and autopsy or postnatal findings in 212 cases. American Journal of Medical Genetics Part A, 1998, 77, 317-321.	2.4	95
125	Clustering ofFBN2 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
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.	2.4	32

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127	Familial growth hormone deficiency associated with MRI abnormalities. American Journal of Medical Genetics Part A, 1998, 80, 128-132.	2.4	56
128	Severe classical congenital muscular dystrophy and merosin expression. Clinical Genetics, 1998, 54, 193-198.	2.0	9
129	Partial tetrasomy with triplication of chromosome (5) (p14â€p15.33) in a patient with severe multiple congenital anomalies. American Journal of Medical Genetics Part A, 1998, 79, 103-107.	2.4	2
130	Mutations in CDMP1 cause autosomal dominant brachydactyly type C. Nature Genetics, 1997, 17, 18-19.	21.4	255
131	A new mutation in the type II hair cortex keratin hHb1 involved in the inherited hair disorder monilethrix. Human Genetics, 1997, 101, 165-169.	3.8	99
132	Findings in children exposed in utero to phenytoin and carbamazepine monotherapy: Independent effects of epilepsy and medications. , 1997, 68, 18-24.		98
133	Limb defects in homozygous Î±-thalassemia: Report of three cases. American Journal of Medical Genetics Part A, 1997, 68, 162-167.	2.4	28
134	Omphalocele in Miller-Dieker syndrome: Expanding the phenotype. , 1997, 69, 293-298.		33
135	Arthrogryposis multiplex congenita, craniofacial, and ophthalmological abnormalities and normal intelligence: A new syndrome?. , 1997, 71, 401-405.		9
136	Brachydactyly-short stature-hypertension (Bilginturan) syndrome: Report on two families. , 1997, 73, 279-285.		14
137	Novel mutations of the tyrosinase (TYR) gene in type I oculocutaneous albinism (OCA1). Human Mutation, 1997, 10, 171-174.	2.5	45
138	Novel mutations of theP gene in type II oculocutaneous albinism (OCA2). Human Mutation, 1997, 10, 175-177.	2.5	37
139	Findings in children exposed in utero to phenytoin and carbamazepine monotherapy: Independent effects of epilepsy and medications. American Journal of Medical Genetics Part A, 1997, 68, 18-24.	2.4	2
140	Novel mutations of the P gene in type II oculocutaneous albinism (OCA2). Human Mutation, 1997, 10, 175-177.	2.5	3
141	Dandy-Walker malformation syndromes: Reply to Fiumara et al.. American Journal of Medical Genetics Part A, 1996, 63, 413-413.	2.4	0
142	Terminal deletion of the long arm of chromosome 3 [46,XX,del(3)(q27â†'qter)]. American Journal of Medical Genetics Part A, 1996, 61, 45-48.	2.4	28
143	The expanding clinical phenotype of the tRNALeu(UUR) Aâ†'G mutation at np 3243 of mitochondrial DNA: Diabetic embryopathy associated with mitochondrial cytopathy. , 1996, 62, 404-409.		25
144	Tissue-specific methylation differences and cognitive function in fragile X premutation females. , 1996, 64, 329-333.		29

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145	Tissueâ€specific methylation differences and cognitive function in fragile X premutation females. American Journal of Medical Genetics Part A, 1996, 64, 329-333.	2.4	1
146	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
147	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
148	Prenatal diagnosis of retinal nonattachment in the Walker-Warburg syndrome. American Journal of Medical Genetics Part A, 1995, 56, 351-358.	2.4	31
149	Genetic homogeneity of cartilage-hair hypoplasia. Human Genetics, 1995, 95, 157-160.	3.8	19
150	Hb FM-Fort Ripley: Confirmation of autosormal dominant inheritance and diagnosis by PCR and direct nucleotide sequencing. Human Mutation, 1994, 3, 239-242.	2.5	12
151	Penoscrotal transposition: A case report and review. American Journal of Medical Genetics Part A, 1994, 49, 103-107.	2.4	33
152	Familial Dandyâ€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
153	Hydrops-ectopic calcificationâ€"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
154	Evidence for multiâ€site closure of the neural tube in humans. American Journal of Medical Genetics Part A, 1993, 47, 723-743.	2.4	364
155	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.	2.4	8
156	Syndrome of mental retardation and distal arthrogryposis in sibs. American Journal of Medical Genetics Part A, 1991, 41, 49-51.	2.4	5
157	Detection and enumeration of colonic mucosal cells in amniotic fluid using a colon epithelial-specific monoclonal antibody. Prenatal Diagnosis, 1990, 10, 725-732.	2.3	6
158	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, 406-414.	2.4	6
159	Hypomelanosis of Itoâ€"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
160	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
161	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
162	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

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163	Congenital diaphragmatic hernia, coarse facies, and acral hypoplasia: Fryns syndrome. American Journal of Medical Genetics Part A, 1989, 32, 93-99.	2.4	56
164	Lymphatic abnormalities in fetuses with posterior cervical cystic hygroma. American Journal of Medical Genetics Part A, 1989, 33, 352-356.	2.4	75
165	Perinatal and first year followâ€up of patients with Praderâ€Willi syndrome: normal size of hands and feet. Clinical Genetics, 1989, 35, 161-166.	2.0	16
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