Lionel Val Maldergem

List of Publications by Year
in descending order

204 papers	15,506 citations	18482 62 h-index	20358 116 g-index
211 all docs	211 docs citations	211 times ranked	19706 citing authors

#	Article	IF	Citations
1	Identification of the gene altered in Berardinelliâ€“Seip congenital lipodystrophy on chromosome 11q13. Nature Genetics, 2001, 28, 365-370.	21.4	665
2	Human TUBB3 Mutations Perturb Microtubule Dynamics, Kinesin Interactions, and Axon Guidance. Cell, 2010, 140, 74-87.	28.9	515
3	GJB2 Mutations and Degree of Hearing Loss: A Multicenter Study. American Journal of Human Genetics, 2005, 77, 945-957.	6.2	455
4	Genome-Wide Scan for Autism Susceptibility Genes. Human Molecular Genetics, 1999, 8, 805-812.	2.9	453
5	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	21.4	431
6	Mutations in the gene encoding the human matrix Gla protein cause Keutel syndrome. Nature Genetics, 1999, 21, 142-144.	21.4	362
7	Heterozygous missense mutations in BSCL2 are associated with distal hereditary motor neuropathy and Silver syndrome. Nature Genetics, 2004, 36, 271-276.	21.4	349
8	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. Nature Genetics, 2008, 40, 32-34.	21.4	330
9	Mutations in the gene encoding immunoglobulin Î¼-binding protein 2 cause spinal muscular atrophy with respiratory distress type 1. Nature Genetics, 2001, 29, 75-77.	21.4	317
10	Periventricular heterotopia: phenotypic heterogeneity and correlation with Filamin A mutations. Brain, 2006, 129, 1892-1906.	7.6	315
11	Homozygosity for a missense mutation in fibulin-5 (FBLN5) results in a severe form of cutis laxa. Human Molecular Genetics, 2002, 11, 2113-2118.	2.9	283
12	Mutations in the DNA methyltransferase gene DNMT3A cause an overgrowth syndrome with intellectual disability. Nature Genetics, 2014, 46, 385-388.	21.4	280
13	Hepatocyte transplantation in a 4-year-old girl with peroxisomal biogenesis disease: technique, safety, and metabolic follow-up1. Transplantation, 2003, 76, 735-738.	1.0	254
14	Mutations in genes encoding the cadherin receptor-ligand pair DCHS1 and FAT4 disrupt cerebral cortical development. Nature Genetics, 2013, 45, 1300-1308.	21.4	247
15	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. Molecular Psychiatry, 2016, 21, 133-148.	7.9	243
16	Positional cloning of a novel gene on chromosome 16q causing Bardet-Biedl syndrome (BBS2). Human Molecular Genetics, 2001, 10, 865-874.	2.9	240
17	GJA1mutations, variants, and connexin 43 dysfunction as it relates to the oculodentodigital dysplasia phenotype. Human Mutation, 2009, 30, 724-733.	2.5	240
18	Spectrum of FOXL2 gene mutations in blepharophimosis-ptosis-epicanthus inversus (BPES) families demonstrates a genotype-phenotype correlation. Human Molecular Genetics, 2001, 10, 1591-1600.	2.9	238

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19	RAD21 Mutations Cause a Human Cohesinopathy. American Journal of Human Genetics, 2012, 90, 1014-1027.	6.2	238
20	Genotype-phenotype relationships in Berardinelli-Seip congenital lipodystrophy. Journal of Medical Genetics, 2002, 39, 722-733.	3.2	233
21	FOXL2 and BPES: Mutational Hotspots, Phenotypic Variability, and Revision of the Genotype-Phenotype Correlation. American Journal of Human Genetics, 2003, 72, 478-487.	6.2	219
22	The Thr124Met mutation in the peripheral myelin protein zero (MPZ) gene is associated with a clinically distinct Charcotâ€“Marieâ€“Tooth phenotype. Brain, 1999, 122, 281-290.	7.6	215
23	Camurati-Engelmann disease: review of the clinical, radiological, and molecular data of 24 families and implications for diagnosis and treatment. Journal of Medical Genetics, 2005, 43, 1-11.	3.2	211
24	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	21.4	211
25	Paternal Origin of FGFR2 Mutations in Sporadic Cases of Crouzon Syndrome and Pfeiffer Syndrome. American Journal of Human Genetics, 2000, 66, 768-777.	6.2	191
26	3-Phosphoglycerate dehydrogenase deficiency: an inborn error of serine biosynthesis.. Archives of Disease in Childhood, 1996, 74, 542-545.	1.9	179
27	Revisiting the craniosynostosis-radial ray hypoplasia association: Baller-Gerold syndrome caused by mutations in the RECQL4 gene. Journal of Medical Genetics, 2005, 43, 148-152.	3.2	179
28	Mutational analysis of the SOX9 gene in campomelic dysplasia and autosomal sex reversal: lack of genotype/phenotype correlations. Human Molecular Genetics, 1997, 6, 91-98.	2.9	175
29	The Spectrum of Mutations in TBX3: Genotype/Phenotype Relationship in Ulnar-Mammary Syndrome. American Journal of Human Genetics, 1999, 64, 1550-1562.	6.2	158
30	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	2.8	149
31	The phenotype of motor neuropathies associated with BSCL2 mutations is broader than Silver syndrome and distal HMN type V. Brain, 2004, 127, 2124-2130.	7.6	146
32	The Spectrum of Pathogenic Mutations in SPINK5 in 19 Families with Netherton Syndrome: Implications for Mutation Detection and First Case of Prenatal Diagnosis. Journal of Investigative Dermatology, 2001, 117, 179-187.	0.7	145
33	Primary ciliary dyskinesia: a genome-wide linkage analysis reveals extensive locus heterogeneity. European Journal of Human Genetics, 2000, 8, 109-118.	2.8	143
34	Sporadic Imprinting Defects in Prader-Willi Syndrome and Angelman Syndrome: Implications for Imprint-Switch Models, Genetic Counseling, and Prenatal Diagnosis. American Journal of Human Genetics, 1998, 63, 170-180.	6.2	142
35	Infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). Annals of Neurology, 2003, 54, 719-724.	5.3	141
36	Clinical, molecular, and genotype-phenotype correlation studies from 25 cases of oral-facial-digital syndrome type 1: a French and Belgian collaborative study. Journal of Medical Genetics, 2005, 43, 54-61.	3.2	137

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37	Coenzyme Q- responsive Leigh's encephalopathy in two sisters. Annals of Neurology, 2002, 52, 750-754.	5.3	136
38	Mechanisms for Nonrecurrent Genomic Rearrangements Associated with CMT1A or HNPP: Rare CNVs as a Cause for Missing Heritability. American Journal of Human Genetics, 2010, 86, 892-903.	6.2	125
39	SOX10 mutations in chronic intestinal pseudo-obstruction suggest a complex physiopathological mechanism. Human Genetics, 2002, 111, 198-206.	3.8	123
40	Hereditary vitamin D resistant rickets caused by a novel mutation in the vitamin D receptor that results in decreased affinity for hormone and cellular hyporesponsiveness.. Journal of Clinical Investigation, 1997, 99, 297-304.	8.2	121
41	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 2972-2980.	1.2	119
42	TBX5 Genetic Testing Validates Strict Clinical Criteria for Holt-Oram Syndrome. Pediatric Research, 2005, 58, 981-986.	2.3	118
43	Mutations in the RP2 Gene Cause Disease in 10% of Families with Familial X-Linked Retinitis Pigmentosa Assessed in This Study. American Journal of Human Genetics, 1999, 64, 1210-1215.	6.2	115
44	Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. Human Molecular Genetics, 2009, 18, 2149-2165.	2.9	115
45	Disruption of the ATXN1â€“CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. Nature Genetics, 2017, 49, 527-536.	21.4	113
46	The Phenotype of Leber Congenital Amaurosis in Patients With AIPL1 Mutations. JAMA Ophthalmology, 2004, 122, 1029.	2.4	105
47	Molecular Characterization of 3-Phosphoglycerate Dehydrogenase Deficiencyâ€”a Neurometabolic Disorder Associated with Reduced L-Serine Biosynthesis. American Journal of Human Genetics, 2000, 67, 1389-1399.	6.2	104
48	An update on serine deficiency disorders. Journal of Inherited Metabolic Disease, 2013, 36, 613-619.	3.6	103
49	Novel comprehensive diagnostic strategy in Pitt-Hopkins syndrome: Clinical score and further delineation of the TCF4 mutational spectrum. Human Mutation, 2012, 33, 64-72.	2.5	102
50	New insights into genotypeâ€“phenotype correlation for GLI3 mutations. European Journal of Human Genetics, 2015, 23, 92-102.	2.8	97
51	A panel study on patients with dominant cerebellar ataxia highlights the frequency of channelopathies. Brain, 2017, 140, 1579-1594.	7.6	89
52	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	6.2	88
53	Prevalence of Mutations in AGPAT2 Among Human Lipodystrophies. Diabetes, 2003, 52, 1573-1578.	0.6	87
54	Pontocerebellar hypoplasia type 1. Neurology, 2013, 80, 438-446.	1.1	84

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55	Increased risk for developmental delay in Saethre-Chotzen syndrome is associated with TWIST deletions: an improved strategy for TWIST mutation screening. Human Genetics, 2003, 114, 68-76.	3.8	83
56	Phenotypic Spectrum of Simpsonâ€“<scp>G</scp>olabiâ€“<scp>B</scp>ehmel Syndrome in a Series of 42 Cases With a Mutation in <scp>GPC</scp>3 and Review of the Literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 92-105.	1.6	78
57	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46.	1.8	75
58	Gain-of-function mutations in the phosphatidylserine synthase 1 (PTDSS1) gene cause Lenz-Majewski syndrome. Nature Genetics, 2014, 46, 70-76.	21.4	74
59	Further characterization of ATP6V0A2-related autosomal recessive cutis laxa. Human Genetics, 2012, 131, 1761-1773.	3.8	73
60	Congenital microcephaly and seizures due to 3-phosphoglycerate dehydrogenase deficiency: Outcome of treatment with amino acids. Journal of Inherited Metabolic Disease, 2002, 25, 119-125.	3.6	72
61	Hypomyelination and Reversible White Matter Attenuation in 3-Phosphoglycerate Dehydrogenase Deficiency. Neuropediatrics, 2000, 31, 287-292.	0.6	68
62	Phenotypic and molecular insights into CASK-related disorders in males. Orphanet Journal of Rare Diseases, 2015, 10, 44.	2.7	68
63	SIMPLE mutation in demyelinating neuropathy and distribution in sciatic nerve. Annals of Neurology, 2004, 55, 713-720.	5.3	67
64	Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. Human Mutation, 2013, 34, 111-121.	2.5	67
65	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	2.4	66
66	Phenotypic expansion in <scp>DDX</scp>3X â€“ a common cause of intellectual disability in females. Annals of Clinical and Translational Neurology, 2018, 5, 1277-1285.	3.7	66
67	Human liver pathology in peroxisomal diseases: A review including novel data. Biochimie, 1993, 75, 281-292.	2.6	64
68	A position effect on TRPS1 is associated with Ambras syndrome in humans and the Koala phenotype in mice. Human Molecular Genetics, 2008, 17, 3539-3551.	2.9	63
69	Autosomal recessive POLR1D mutation with decrease of TCOF1 mRNA is responsible for Treacher Collins syndrome. Genetics in Medicine, 2014, 16, 720-724.	2.4	63
70	FLNC pathogenic variants in patients with cardiomyopathies: Prevalence and genotypeâ€phenotype correlations. Clinical Genetics, 2019, 96, 317-329.	2.0	63
71	Loss of function of KIAA2022 causes mild to severe intellectual disability with an autism spectrum disorder and impairs neurite outgrowth. Human Molecular Genetics, 2013, 22, 3306-3314.	2.9	62
72	Clinical and molecular findings in 39 patients with KBG syndrome caused by deletion or mutation of ANKRD11. American Journal of Medical Genetics, Part A, 2016, 170, 2847-2859.	1.2	62

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73	Disruption of a new X linked gene highly expressed in brain in a family with two mentally retarded males. Journal of Medical Genetics, 2004, 41, 736-742.	3.2	60
74	The Natural Course of Infantile Spinal Muscular Atrophy With Respiratory Distress Type 1 (SMARD1). Pediatrics, 2012, 129, e148-e156.	2.1	59
75	Autism spectrum disorder associated with low serotonin in CSF and mutations in the SLC29A4 plasma membrane monoamine transporter (PMAT) gene. Molecular Autism, 2014, 5, 43.	4.9	59
76	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	1.1	57
77	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. Biological Psychiatry, 2018, 84, 253-264.	1.3	56
78	Relief of gastrointestinal symptoms under enzyme replacement therapy in patients with Fabry disease. Journal of Inherited Metabolic Disease, 2004, 27, 499-505.	3.6	55
79	Exome Sequencing in Brown-Vialetto-Van Laere Syndrome. American Journal of Human Genetics, 2010, 87, 567-569.	6.2	54
80	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	2.5	54
81	Variable phenotypes associated with 10q23 microdeletions involving the PTEN and BMPR1A genes. Clinical Genetics, 2008, 74, 145-154.	2.0	52
82	A homozygous ATAD1 mutation impairs postsynaptic AMPA receptor trafficking and causes a lethal encephalopathy. Brain, 2018, 141, 651-661.	7.6	52
83	Townes-Brocks syndrome: twenty novelSALL1 mutations in sporadic and familial cases and refinement of theSALL1 hot spot region. Human Mutation, 2007, 28, 204-205.	2.5	51
84	De novo mutations ofKIAA2022in females cause intellectual disability and intractable epilepsy. Journal of Medical Genetics, 2016, 53, 850-858.	3.2	47
85	An Application of NGS for Molecular Investigations in Perrault Syndrome: Study of 14 Families and Review of the Literature. Human Mutation, 2016, 37, 1354-1362.	2.5	46
86	A common ancestor for COCH related cochleovestibular (DFNA9) patients in Belgium and The Netherlands bearing the P51S mutation. Journal of Medical Genetics, 2001, 38, 61-65.	3.2	43
87	3-Phosphoglycerate dehydrogenase deficiency and 3-phosphoserine phosphatase deficiency: Inborn errors of serine biosynthesis. Journal of Inherited Metabolic Disease, 1996, 19, 223-226.	3.6	42
88	Multiple exostoses, mental retardation, hypertrichosis, and brain abnormalities in a boy with a de novo 8q24 submicroscopic interstitial deletion. American Journal of Medical Genetics Part A, 2002, 113, 326-332.	2.4	42
89	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
90	Multiple founder effects in spinal and bulbar muscular atrophy (SBMA, Kennedy disease) around the world. European Journal of Human Genetics, 2001, 9, 431-436.	2.8	41

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91	Neonatal progeroid variant of Marfan syndrome with congenital lipodystrophy results from mutations at the 3â€² end of FBN1 gene. European Journal of Medical Genetics, 2014, 57, 230-234.	1.3	41
92	Localisation of the gene causing diaphyseal dysplasia Camurati-Engelmann to chromosome 19q13. Journal of Medical Genetics, 2000, 37, 245-249.	3.2	40
93	Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. Genetics in Medicine, 2019, 21, 2025-2035.	2.4	40
94	Identification of three novel SEDL mutations, including mutation in the rare, non-canonical splice site of exon 4. Clinical Genetics, 2003, 64, 235-242.	2.0	39
95	Cobblestone-like brain dysgenesis and altered glycosylation in congenital cutis laxa, Debrè•type. Neurology, 2008, 71, 1602-1608.	1.1	39
96	Van Maldergem syndrome: further characterisation and evidence for neuronal migration abnormalities and autosomal recessive inheritance. European Journal of Human Genetics, 2012, 20, 1024-1031.	2.8	39
97	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. Genome Research, 2019, 29, 1057-1066.	5.5	38
98	Systematic Profiling of DNMT3A Variants Reveals Protein Instability Mediated by the DCAF8 E3 Ubiquitin Ligase Adaptor. Cancer Discovery, 2022, 12, 220-235.	9.4	38
99	Chromosomal rearrangements in the 11p15 imprinted region: 17 new 11p15.5 duplications with associated phenotypes and putative functional consequences. Journal of Medical Genetics, 2018, 55, 205-213.	3.2	36
100	CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays. American Journal of Human Genetics, 2017, 101, 391-403.	6.2	35
101	Syndrome of lipoatrophic diabetes, vitamin D resistant rickets, and persistent MÃ¼llerian ducts in a Turkish boy born to consanguineous parents. , 1996, 64, 506-513.		34
102	Pathogenic variants in the DEAH-box RNA helicase DHX37 are a frequent cause of 46,XY gonadal dysgenesis and 46,XY testicular regression syndrome. Genetics in Medicine, 2020, 22, 150-159.	2.4	34
103	The CHD8 overgrowth syndrome: A detailed evaluation of an emerging overgrowth phenotype in 27 patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 557-564.	1.6	33
104	Peroxisomal localization of the immunoreactiveÎ²-oxidation enzymes in a neonate with aÎ²-oxidation defect. Virchows Archiv A, Pathological Anatomy and Histopathology, 1991, 419, 301-308.	1.4	32
105	Orthotopic liver transplantation from a living-related donor in an infant with a peroxisome biogenesis defect of the infantile Refsum disease type. Journal of Inherited Metabolic Disease, 2005, 28, 593-600.	3.6	32
106	OFD1mutations in males: phenotypic spectrum and ciliary basal body docking impairment. Clinical Genetics, 2013, 84, 86-90.	2.0	32
107	Treatment outcome of creatine transporter deficiency: international retrospective cohort study. Metabolic Brain Disease, 2018, 33, 875-884.	2.9	32
108	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	2.5	31

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109	Neonatal seizures and severe hypotonia in a male infant suffering from a defect in peroxisomal Î²-oxidation. Neuromuscular Disorders, 1992, 2, 217-224.	0.6	30
110	Oral-facial-digital syndrome type I in a newborn male. American Journal of Medical Genetics Part A, 1993, 46, 335-338.	2.4	30
111	Severe congenital cutis laxa with pulmonary emphysema: A family with three affected sibs. American Journal of Medical Genetics Part A, 1988, 31, 455-464.	2.4	29
112	Major decrease in the incidence of trisomyÂ21 at birth in south Belgium: mass impact of triple test?. European Journal of Human Genetics, 2001, 9, 1-4.	2.8	29
113	Severe cardiac phenotype of Berardinelli-Seip congenital lipodystrophy in an infant with homozygous E189X BSCL2 mutation. European Journal of Medical Genetics, 2009, 52, 14-16.	1.3	29
114	Renal insufficiency, a frequent complication with age in oralâ€facialâ€digital syndrome type I. Clinical Genetics, 2010, 77, 258-265.	2.0	29
115	Unraveling the genetic landscape of autosomal recessive Charcot-Marie-Tooth neuropathies using a homozygosity mapping approach. Neurogenetics, 2015, 16, 33-42.	1.4	29
116	IL11RAâ€related Crouzonâ€like autosomal recessive craniosynostosis in 10 new patients: Resemblances and differences. Clinical Genetics, 2018, 94, 373-380.	2.0	29
117	<i>IRF6</i> Screening of Syndromic and a priori Non-Syndromic Cleft Lip and Palate Patients: Identification of a New Type of Minor VWS Sign. Molecular Syndromology, 2010, 1, 67-74.	0.8	28
118	Acromelic frontonasal â€œdysplasiaâ€: Further delineation of a subtype with brain malformation and polydactyly (Toriello syndrome). American Journal of Medical Genetics Part A, 1992, 42, 180-183.	2.4	27
119	Substitution of aspartic acid for glycine at position 310 in type II collagen produces achondrogenesis II, and substitution of serine at position 805 produces hypochondrogenesis: analysis of genotype-phenotype relationships. Biochemical Journal, 1995, 307, 823-830.	3.7	27
120	Genomic deletions ofOFD1account for 23% of oral-facial-digital type 1 syndrome after negative DNA sequencing. Human Mutation, 2009, 30, E320-E329.	2.5	27
121	Type I hyperprolinemia: genotype/phenotype correlations. Human Mutation, 2010, 31, 961-965.	2.5	26
122	Severe sex differentiation disorder in a boy with a 3.8â€‰Mb 10q25.3â€“q26.12 microdeletion encompassing EMX2. American Journal of Medical Genetics, Part A, 2014, 164, 2618-2622.	1.2	25
123	A Postural Tremor Highly Responsive to Transcranial Cerebello-Cerebral DCS in ARCA3. Frontiers in Neurology, 2017, 8, 71.	2.4	25
124	Heterogeneity versus variability in megalocornea-mental retardation (MMR) syndromes: Report of new cases and delineation of 4 probable types. American Journal of Medical Genetics Part A, 1993, 46, 132-137.	2.4	24
125	Patients with lipodystrophic diabetes mellitus of the Seip-Berardinelli type, express normal insulin receptors. Diabetologia, 1993, 36, 172-174.	6.3	24
126	Extension of the phenotype of biallelic lossâ€ofâ€function mutations in <scp>SLC25A46</scp> to the severe form of pontocerebellar hypoplasia type I. Clinical Genetics, 2018, 93, 255-265.	2.0	24

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127	Mapping the RP2 locus for X-linked retinitis pigmentosa on proximal Xp: a genetically defined 5-cM critical region and exclusion of candidate genes by physical mapping.. Genome Research, 1996, 6, 1093-1102.	5.5	23
128	Thiamine pyrophosphate: An essential cofactor for the Î±-oxidation in mammals â€“ implications for thiamine deficiencies?. Cellular and Molecular Life Sciences, 2006, 63, 1553-1563.	5.4	23
129	Hearing loss and deafness in the pediatric population. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1527-1538.	1.8	23
130	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	8.2	23
131	The Baller-Gerold syndrome.. Journal of Medical Genetics, 1992, 29, 266-268.	3.2	22
132	Congenital generalized lipodystrophy: identification of novel variants and expansion of clinical spectrum. Clinical Genetics, 2016, 89, 434-441.	2.0	22
133	Maternal vitamin K deficient embryopathy: Association with hyperemesis gravidarum and Crohn disease. American Journal of Medical Genetics, Part A, 2013, 161, 417-429.	1.2	21
134	Nextâ€generation sequencing in a series of 80 fetuses with complex cardiac malformations and/or heterotaxy. Human Mutation, 2020, 41, 2167-2178.	2.5	21
135	Brachymorphism-onychodysplasia-dysphalangism syndrome.. Journal of Medical Genetics, 1993, 30, 158-161.	3.2	20
136	Cerebral Venous Thrombosis and Procoagulant Factors. Angiology, 1998, 49, 563-571.	1.8	20
137	Diagnostic Value of Immunostaining in Cultured Skin Fibroblasts from Patients with Oxidative Phosphorylation Defects. Pediatric Research, 2006, 59, 2-6.	2.3	20
138	Copy number variation analysis in adults with catatonia confirms haploinsufficiency of SHANK3 as a predisposing factor. European Journal of Medical Genetics, 2016, 59, 436-443.	1.3	20
139	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. Human Molecular Genetics, 2018, 27, 589-600.	2.9	20
140	Molecular and neurological characterizations of three Saudi families with lipoid proteinosis. BMC Medical Genetics, 2011, 12, 31.	2.1	19
141	The Marshall-smith syndrome. European Journal of Pediatrics, 1990, 150, 54-55.	2.7	17
142	Lethal femoral-facial syndrome: a case with unusual manifestations.. Journal of Medical Genetics, 1997, 34, 518-519.	3.2	17
143	TCOF1 mutations excluded from a role in other first and second branchial arch-related disorders. American Journal of Medical Genetics Part A, 2002, 111, 324-327.	2.4	17
144	Spatiotemporal expression in mouse brain of Kiaa2022, a gene disrupted in two patients with severe mental retardation. Gene Expression Patterns, 2009, 9, 423-429.	0.8	17

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145	Facial anomalies in congenital cutis laxa with retarded growth and skeletal dysplasia. American Journal of Medical Genetics Part A, 1989, 32, 265-265.	2.4	16
146	Morphological spectrum and clinical features of myopathies with tubular aggregates. Histology and Histopathology, 2013, 28, 1041-54.	0.7	16
147	Prenatal ultrasound detection of congenital cataract in trisomy 21. Prenatal Diagnosis, 1999, 19, 780-782.	2.3	15
148	Early infantile cardiomyopathy and liver disease: A multisystemic disorder caused by congenital lipodystrophy. Molecular Genetics and Metabolism, 2013, 109, 227-229.	1.1	15
149	Clinical features of SMARCA2 duplication overlap with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2662-2670.	1.2	15
150	Validation of a clinical practice-based algorithm for the diagnosis of autosomal recessive cerebellar ataxias based on NGS identified cases. Journal of Neurology, 2016, 263, 1314-1322.	3.6	15
151	Genetic assessment and folate receptor autoantibodies in infantile-onset cerebral folate deficiency (CFD) syndrome. Molecular Genetics and Metabolism, 2018, 124, 87-93.	1.1	15
152	Perturbed hematopoiesis in individuals with germline DNMT3A overgrowth Tatton-Brown-Rahman syndrome. Haematologica, 2022, 107, 887-898.	3.5	15
153	X-linked recessive chondrodysplasia punctata with XY translocation in a stillborn fetus. Human Genetics, 1991, 87, 661-4.	3.8	14
154	Hepatic Ultrastructure in Congenital Total Lipodystrophy with Special Reference to Peroxisomes. Ultrastructural Pathology, 1992, 16, 307-316.	0.9	14
155	Comprehensive methylation analysis in typical and atypical PWS and AS patients with normal biparental chromosomes 15. European Journal of Human Genetics, 2001, 9, 519-526.	2.8	13
156	Autosomal recessive cutis laxa type 2A (ARCL2A) mimicking Ehlersâ€Danlos syndrome by its dermatological manifestations: Report of three affected patients. American Journal of Medical Genetics, Part A, 2014, 164, 1245-1253.	1.2	13
157	POLG2 deficiency causes adultâ€onset syndromic sensory neuropathy, ataxia and parkinsonism. Annals of Clinical and Translational Neurology, 2017, 4, 4-14.	3.7	13
158	Livingâ€donor liver transplantation for mild Zellweger spectrum disorder: Up to 17Âyears followâ€up. Pediatric Transplantation, 2018, 22, e13112.	1.0	13
159	The <scp>GRIA3</scp> c.<scp>2477G</scp> >â€‰A Variant Causes an Exaggerated Startle Reflex, Chorea, and Multifocal Myoclonus. Movement Disorders, 2020, 35, 1224-1232.	3.9	13
160	Implementation of fetal clinical exome sequencing: Comparing prospective and retrospective cohorts. Genetics in Medicine, 2022, 24, 344-363.	2.4	13
161	Congenital generalized lipodystrophy in an Indian patient with a novel mutation in BSCL2 gene. Journal of Inherited Metabolic Disease, 2008, 31, 317-322.	3.6	12
162	Congenital cutis laxa with ligamentous laxity and delayed development, Dandyâ€Walker malformation and minor heart and osseous defects. Clinical Genetics, 1994, 45, 318-322.	2.0	12

#	Article	IF	Citations
163	Templeâ€“Baraitser syndrome: A rare and possibly unrecognized condition. American Journal of Medical Genetics, Part A, 2010, 152A, 2322-2326.	1.2	12
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