Lionel Val Maldergem

List of Publications by Year
in descending order

204 papers	15,506 citations	18436 62 h-index	20307 116 g-index
211 all docs	211 docs citations	211 times ranked	19706 citing authors

#	Article	IF	Citations
1	Perturbed hematopoiesis in individuals with germline DNMT3A overgrowth Tatton-Brown-Rahman syndrome. Haematologica, 2022, 107, 887-898.	1.7	15
2	Systematic Profiling of DNMT3A Variants Reveals Protein Instability Mediated by the DCAF8 E3 Ubiquitin Ligase Adaptor. Cancer Discovery, 2022, 12, 220-235.	7.7	38
3	Implementation of fetal clinical exome sequencing: Comparing prospective and retrospective cohorts. Genetics in Medicine, 2022, 24, 344-363.	1.1	13
4	CYLDâ€related cutaneous syndrome: variable p.Pro482fs*6 phenotype in five individuals from two unrelated families. Journal of the European Academy of Dermatology and Venereology, 2021, 35, e81-e83.	1.3	1
5	<scp>IQSEC2</scp> disorder: A new disease entity or a Rett spectrum continuum?. Clinical Genetics, 2021, 99, 462-474.	1.0	11
6	<scp>Skrabanâ€Deardorff</scp> syndrome: Six new cases of <scp>WDR26</scp>â€related disease and expansion of the clinical phenotype. Clinical Genetics, 2021, 99, 732-739.	1.0	4
7	Touch and olfaction/taste differentiate children carrying a 16p11.2 deletion from children with ASD. Molecular Autism, 2021, 12, 8.	2.6	6
8	Biallelic KDSR mutations mitigate the formation of novel ketoâ€type ceramides in human stratum corneum. FASEB Journal, 2021, 35, .	0.2	0
9	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.	0.7	42
10	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.	1.1	34
11	Split hand/foot malformation associated with 20p12.1 deletion: A case report. European Journal of Medical Genetics, 2020, 63, 103805.	0.7	1
12	A new case of KIAA0753-related variant of Jeune asphyxiating thoracic dystrophy. European Journal of Medical Genetics, 2020, 63, 103823.	0.7	4
13	Bifid nose as the sole manifestation of <scp>BNAR</scp> syndrome, a <scp>FREM1</scp>â€related condition. Clinical Genetics, 2020, 98, 515-516.	1.0	4
14	Nextâ€generation sequencing in a series of 80 fetuses with complex cardiac malformations and/or heterotaxy. Human Mutation, 2020, 41, 2167-2178.	1.1	21
15	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.	2.2	13
16	Congenital posterior cervical spine malformation due to biallelic c.240â€4T>G RIPPLY2 variant: A discrete entity. American Journal of Medical Genetics, Part A, 2020, 182, 1466-1472.	0.7	5
17	FLNC pathogenic variants in patients with cardiomyopathies: Prevalence and genotypeâ€phenotype correlations. Clinical Genetics, 2019, 96, 317-329.	1.0	63
18	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.	0.7	33

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19	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.	1.1	40
20	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. Genome Research, 2019, 29, 1057-1066.	2.4	38
21	Phenotypic Overlap of Roberts and Baller-Gerold Syndromes in Two Patients With Craniosynostosis, Limb Reductions, and ESCO2 Mutations. Frontiers in Pediatrics, 2019, 7, 210.	0.9	0
22	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.	3.6	23
23	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	2.6	88
24	Autopsy findings of ectodermal dysplasia and sex development disorder in a fetus with 19q12q13 microdeletion. European Journal of Medical Genetics, 2019, 62, 103539.	0.7	1
25	Livingâ€donor liver transplantation for mild Zellweger spectrum disorder: Up to 17Âyears followâ€up. Pediatric Transplantation, 2018, 22, e13112.	0.5	13
26	Genetic assessment and folate receptor autoantibodies in infantile-onset cerebral folate deficiency (CFD) syndrome. Molecular Genetics and Metabolism, 2018, 124, 87-93.	0.5	15
27	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. Biological Psychiatry, 2018, 84, 253-264.	0.7	56
28	A homozygous ATAD1 mutation impairs postsynaptic AMPA receptor trafficking and causes a lethal encephalopathy. Brain, 2018, 141, 651-661.	3.7	52
29	Cutis laxa and excessive bone growth due to de novo mutations in PTDSS1. American Journal of Medical Genetics, Part A, 2018, 176, 668-675.	0.7	11
30	Treatment outcome of creatine transporter deficiency: international retrospective cohort study. Metabolic Brain Disease, 2018, 33, 875-884.	1.4	32
31	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	1.1	66
32	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. Human Molecular Genetics, 2018, 27, 589-600.	1.4	20
33	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.	1.0	24
34	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.	1.5	36
35	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.	1.7	66
36	Autosomal-dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function. Human Mutation, 2018, 39, 1076-1080.	1.1	8

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37	Reply: ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. Brain, 2018, 141, e50-e50.	3.7	1
38	IL11RAâ€related Crouzonâ€like autosomal recessive craniosynostosis in 10 new patients: Resemblances and differences. Clinical Genetics, 2018, 94, 373-380.	1.0	29
39	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	1.1	31
40	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46.	0.9	75
41	POLG2 deficiency causes adultâ€onset syndromic sensory neuropathy, ataxia and parkinsonism. Annals of Clinical and Translational Neurology, 2017, 4, 4-14.	1.7	13
42	A panel study on patients with dominant cerebellar ataxia highlights the frequency of channelopathies. Brain, 2017, 140, 1579-1594.	3.7	89
43	Disruption of the ATXN1â€“CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. Nature Genetics, 2017, 49, 527-536.	9.4	113
44	CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays. American Journal of Human Genetics, 2017, 101, 391-403.	2.6	35
45	A Postural Tremor Highly Responsive to Transcranial Cerebello-Cerebral DCS in ARCA3. Frontiers in Neurology, 2017, 8, 71.	1.1	25
46	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	1.1	54
47	Clinical features of SMARCA2 duplication overlap with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2662-2670.	0.7	15
48	Congenital generalized lipodystrophy: identification of novel variants and expansion of clinical spectrum. Clinical Genetics, 2016, 89, 434-441.	1.0	22
49	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.	1.8	15
50	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.	0.7	62
51	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.	1.1	46
52	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.	0.7	20
53	De novo mutations ofKIAA2022in females cause intellectual disability and intractable epilepsy. Journal of Medical Genetics, 2016, 53, 850-858.	1.5	47
54	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. Molecular Psychiatry, 2016, 21, 133-148.	4.1	243

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55	Phenotypic and molecular insights into CASK-related disorders in males. Orphanet Journal of Rare Diseases, 2015, 10, 44.	1.2	68
56	TCF12 microdeletion in a 72â€yearâ€old woman with intellectual disability. American Journal of Medical Genetics, Part A, 2015, 167, 1897-1901.	0.7	12
57	Berardinelli-Seip syndrome and achalasia: a shared pathomechanism?. European Journal of Pediatrics, 2015, 174, 975-980.	1.3	10
58	Unraveling the genetic landscape of autosomal recessive Charcot-Marie-Tooth neuropathies using a homozygosity mapping approach. Neurogenetics, 2015, 16, 33-42.	0.7	29
59	New insights into genotypeâ€“phenotype correlation for GLI3 mutations. European Journal of Human Genetics, 2015, 23, 92-102.	1.4	97
60	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.	2.6	59
61	Li-Fraumeni syndrome: Multiple distinct brain tumours in two brothers. Neurochirurgie, 2014, 60, 51-54.	0.6	0
62	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.	0.7	41
63	Autosomal recessive POLR1D mutation with decrease of TCOF1 mRNA is responsible for Treacher Collins syndrome. Genetics in Medicine, 2014, 16, 720-724.	1.1	63
64	Mutations in the DNA methyltransferase gene DNMT3A cause an overgrowth syndrome with intellectual disability. Nature Genetics, 2014, 46, 385-388.	9.4	280
65	Gain-of-function mutations in the phosphatidylserine synthase 1 (PTDSS1) gene cause Lenz-Majewski syndrome. Nature Genetics, 2014, 46, 70-76.	9.4	74
66	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.	0.7	25
67	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.	0.7	13
68	Aquagenic Palmoplantar Keratoderma as a CFTR-related Disorder. Acta Dermato-Venereologica, 2014, 96, 848-9.	0.6	7
69	Early infantile cardiomyopathy and liver disease: A multisystemic disorder caused by congenital lipodystrophy. Molecular Genetics and Metabolism, 2013, 109, 227-229.	0.5	15
70	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	0.5	57
71	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.	1.0	23
72	Mutations in genes encoding the cadherin receptor-ligand pair DCHS1 and FAT4 disrupt cerebral cortical development. Nature Genetics, 2013, 45, 1300-1308.	9.4	247

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73	OFD1mutations in males: phenotypic spectrum and ciliary basal body docking impairment. Clinical Genetics, 2013, 84, 86-90.	1.0	32
74	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.	0.7	78
75	An update on serine deficiency disorders. Journal of Inherited Metabolic Disease, 2013, 36, 613-619.	1.7	103
76	Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. Human Mutation, 2013, 34, 111-121.	1.1	67
77	Pontocerebellar hypoplasia type 1. Neurology, 2013, 80, 438-446.	1.5	84
78	Association of iniencephaly, anencephaly, and fusion of cervical vertebral bodies. Clinical Dysmorphology, 2013, 22, 29-32.	0.1	2
79	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.	1.4	62
80	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 2972-2980.	0.7	119
81	Maternal vitamin K deficient embryopathy: Association with hyperemesis gravidarum and Crohn disease. American Journal of Medical Genetics, Part A, 2013, 161, 417-429.	0.7	21
82	Morphological spectrum and clinical features of myopathies with tubular aggregates. Histology and Histopathology, 2013, 28, 1041-54.	0.5	16
83	Van Maldergem syndrome: further characterisation and evidence for neuronal migration abnormalities and autosomal recessive inheritance. European Journal of Human Genetics, 2012, 20, 1024-1031.	1.4	39
84	Further characterization of ATP6V0A2-related autosomal recessive cutis laxa. Human Genetics, 2012, 131, 1761-1773.	1.8	73
85	Coronal craniosynostosis and radial ray hypoplasia: A third report of Twist mutation in a 33 weeks fetus with diaphragmatic hernia. European Journal of Medical Genetics, 2012, 55, 719-722.	0.7	5
86	The Natural Course of Infantile Spinal Muscular Atrophy With Respiratory Distress Type 1 (SMARD1). Pediatrics, 2012, 129, e148-e156.	1.0	59
87	RAD21 Mutations Cause a Human Cohesinopathy. American Journal of Human Genetics, 2012, 90, 1014-1027.	2.6	238
88	Novel comprehensive diagnostic strategy in Pitt-Hopkins syndrome: Clinical score and further delineation of the TCF4 mutational spectrum. Human Mutation, 2012, 33, 64-72.	1.1	102
89	Genetic Update on Auditory Neuropathy. Audiology and Neurotology Extra, 2011, 1, 20-29.	2.0	2
90	Molecular and neurological characterizations of three Saudi families with lipoid proteinosis. BMC Medical Genetics, 2011, 12, 31.	2.1	19

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91	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	3.1	149
92	828 A Novel De Novo Mutation of Chromosome 7 [46, XX, DEL(7)(P14.2 P15.1)] in a Child with Feeding Problems. Pediatric Research, 2010, 68, 416-416.	1.1	0
93	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.	2.6	125
94	Exome Sequencing in Brown-Vialetto-Van Laere Syndrome. American Journal of Human Genetics, 2010, 87, 567-569.	2.6	54
95	Type I hyperprolinemia: genotype/phenotype correlations. Human Mutation, 2010, 31, 961-965.	1.1	26
96	Templeâ€“Baraitser syndrome: A rare and possibly unrecognized condition. American Journal of Medical Genetics, Part A, 2010, 152A, 2322-2326.	0.7	12
97	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	9.4	431
98	Renal insufficiency, a frequent complication with age in oralâ€facialâ€digital syndrome type I. Clinical Genetics, 2010, 77, 258-265.	1.0	29
99	<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.	0.3	28
100	Human TUBB3 Mutations Perturb Microtubule Dynamics, Kinesin Interactions, and Axon Guidance. Cell, 2010, 140, 74-87.	13.5	515
101	Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. Human Molecular Genetics, 2009, 18, 2149-2165.	1.4	115
102	Spatiotemporal expression in mouse brain of Kiaa2022, a gene disrupted in two patients with severe mental retardation. Gene Expression Patterns, 2009, 9, 423-429.	0.3	17
103	Genomic deletions ofOFD1account for 23% of oral-facial-digital type 1 syndrome after negative DNA sequencing. Human Mutation, 2009, 30, E320-E329.	1.1	27
104	GJA1mutations, variants, and connexin 43 dysfunction as it relates to the oculodentodigital dysplasia phenotype. Human Mutation, 2009, 30, 724-733.	1.1	240
105	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	9.4	211
106	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.	0.7	29
107	Congenital generalized lipodystrophy in an Indian patient with a novel mutation in BSCL2 gene. Journal of Inherited Metabolic Disease, 2008, 31, 317-322.	1.7	12
108	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. Nature Genetics, 2008, 40, 32-34.	9.4	330

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109	Variable phenotypes associated with 10q23 microdeletions involving the PTEN and BMPR1A genes. Clinical Genetics, 2008, 74, 145-154.	1.0	52
110	Cobblestone-like brain dysgenesis and altered glycosylation in congenital cutis laxa, Debrel•type. Neurology, 2008, 71, 1602-1608.	1.5	39
111	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.	1.4	63
112	Clinical findings and PDS mutations in 15 patients with hearing loss and dilatation of the vestibular aqueduct. Journal of Laryngology and Otology, 2007, 121, 312-317.	0.4	8
113	Funtional characterization of four novel MAN2B1 mutations causing juvenile onset alpha-mannosidosis. Clinica Chimica Acta, 2007, 375, 136-139.	0.5	11
114	Townes-Brocks syndrome: twenty novelSALL1 mutations in sporadic and familial cases and refinement of theSALL1 hot spot region. Human Mutation, 2007, 28, 204-205.	1.1	51
115	Novel human pathological mutations. Gene symbol: KRIT1. Disease: cerebral cavernous malformation. Human Genetics, 2007, 122, 552.	1.8	3
116	Periventricular heterotopia: phenotypic heterogeneity and correlation with Filamin A mutations. Brain, 2006, 129, 1892-1906.	3.7	315
117	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.	2.4	23
118	Diagnostic Value of Immunostaining in Cultured Skin Fibroblasts from Patients with Oxidative Phosphorylation Defects. Pediatric Research, 2006, 59, 2-6.	1.1	20
119	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.	1.7	32
120	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.	1.5	211
121	TBX5 Genetic Testing Validates Strict Clinical Criteria for Holt-Oram Syndrome. Pediatric Research, 2005, 58, 981-986.	1.1	118
122	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.	1.5	137
123	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.	1.5	179
124	GJB2 Mutations and Degree of Hearing Loss: A Multicenter Study. American Journal of Human Genetics, 2005, 77, 945-957.	2.6	455
125	The phenotype of motor neuropathies associated with BSCL2 mutations is broader than Silver syndrome and distal HMN type V. Brain, 2004, 127, 2124-2130.	3.7	146
126	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.	1.5	60

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127	Heterozygous missense mutations in BSCL2 are associated with distal hereditary motor neuropathy and Silver syndrome. Nature Genetics, 2004, 36, 271-276.	9.4	349
128	Relief of gastrointestinal symptoms under enzyme replacement therapy in patients with Fabry disease. Journal of Inherited Metabolic Disease, 2004, 27, 499-505.	1.7	55
129	SIMPLE mutation in demyelinating neuropathy and distribution in sciatic nerve. Annals of Neurology, 2004, 55, 713-720.	2.8	67
130	The Phenotype of Leber Congenital Amaurosis in Patients With AIPL1 Mutations. JAMA Ophthalmology, 2004, 122, 1029.	2.6	105
131	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.	1.8	83
132	Infantile spinal muscular atrophy with respiratory distress type 1 (SMARD1). Annals of Neurology, 2003, 54, 719-724.	2.8	141
133	Identification of three novel SEDL mutations, including mutation in the rare, non-canonical splice site of exon 4. Clinical Genetics, 2003, 64, 235-242.	1.0	39
134	FOXL2 and BPES: Mutational Hotspots, Phenotypic Variability, and Revision of the Genotype-Phenotype Correlation. American Journal of Human Genetics, 2003, 72, 478-487.	2.6	219
135	Prevalence of Mutations in AGPAT2 Among Human Lipodystrophies. Diabetes, 2003, 52, 1573-1578.	0.3	87
136	Hepatocyte transplantation in a 4-year-old girl with peroxisomal biogenesis disease: technique, safety, and metabolic follow-up1. Transplantation, 2003, 76, 735-738.	0.5	254
137	Genotype-phenotype relationships in Berardinelli-Seip congenital lipodystrophy. Journal of Medical Genetics, 2002, 39, 722-733.	1.5	233
138	Pure terminal duplication of the short arm of chromosome 19 in a boy with mild microcephaly. Journal of Medical Genetics, 2002, 39, 60e-60.	1.5	8
139	Homozygosity for a missense mutation in fibulin-5 (FBLN5) results in a severe form of cutis laxa. Human Molecular Genetics, 2002, 11, 2113-2118.	1.4	283
140	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
141	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
142	Coenzyme Q- responsive Leigh's encephalopathy in two sisters. Annals of Neurology, 2002, 52, 750-754.	2.8	136
143	SOX10 mutations in chronic intestinal pseudo-obstruction suggest a complex physiopathological mechanism. Human Genetics, 2002, 111, 198-206.	1.8	123
144	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.	1.7	72

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145	Spectrum of FOXL2 gene mutations in blepharophimosis-ptosis-epicanthus inversus (BPES) families demonstrates a genotype-phenotype correlation. Human Molecular Genetics, 2001, 10, 1591-1600.	1.4	238
146	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.	0.3	145
147	Identification of the gene altered in Berardinelliâ€“Seip congenital lipodystrophy on chromosome 11q13. Nature Genetics, 2001, 28, 365-370.	9.4	665
148	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.	9.4	317
149	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.	1.4	29
150	Multiple founder effects in spinal and bulbar muscular atrophy (SBMA, Kennedy disease) around the world. European Journal of Human Genetics, 2001, 9, 431-436.	1.4	41
151	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.	1.4	13
152	Positional cloning of a novel gene on chromosome 16q causing Bardet-Biedl syndrome (BBS2). Human Molecular Genetics, 2001, 10, 865-874.	1.4	240
153	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.	1.5	43
154	Association of cleft palate with Sprengel anomaly: Confirmation of the Hodgson-Chiu syndrome. American Journal of Medical Genetics Part A, 2000, 95, 401-403.	2.4	4
155	Primary ciliary dyskinesia: a genome-wide linkage analysis reveals extensive locus heterogeneity. European Journal of Human Genetics, 2000, 8, 109-118.	1.4	143
156	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency and early-onset liver cirrhosis in two siblings. European Journal of Pediatrics, 2000, 159, 108-112.	1.3	7
157	Localisation of the gene causing diaphyseal dysplasia Camurati-Engelmann to chromosome 19q13. Journal of Medical Genetics, 2000, 37, 245-249.	1.5	40
158	Hypomyelination and Reversible White Matter Attenuation in 3-Phosphoglycerate Dehydrogenase Deficiency. Neuropediatrics, 2000, 31, 287-292.	0.3	68
159	Atypical Features in Angelman Syndrome Due to Imprinting Defect or Uniparental Disomy of Chromosome 15. Neuropediatrics, 2000, 31, 109-110.	0.3	9
160	Paternal Origin of FGFR2 Mutations in Sporadic Cases of Crouzon Syndrome and Pfeiffer Syndrome. American Journal of Human Genetics, 2000, 66, 768-777.	2.6	191
161	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.	2.6	104
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