Didier Lacombe

List of Publications by Year
in descending order

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322	20,527	9784	14208
papers	citations	73 h-index	128 g-index

351	351	351	24649
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	28.9	2,055
2	Identification of the gene altered in Berardinelliâ€“Seip congenital lipodystrophy on chromosome 11q13. Nature Genetics, 2001, 28, 365-370.	21.4	665
3	A human homologue of the Drosophila eyes absent gene underlies Branchio-Oto-Renal (BOR) syndrome and identifies a novel gene family. Nature Genetics, 1997, 15, 157-164.	21.4	628
4	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-GoutiÃ¨res syndrome and mimic congenital viral brain infection. Nature Genetics, 2006, 38, 910-916.	21.4	592
5	Mutation spectrum and genotype-phenotype analyses in Cowden disease and Bannayan-Zonana syndrome, two hamartoma syndromes with germline PTEN mutation. Human Molecular Genetics, 1998, 7, 507-515.	2.9	578
6	Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. Nature Genetics, 2013, 45, 639-647.	21.4	399
7	Localized mutations in the gene encoding the cytoskeletal protein filamin A cause diverse malformations in humans. Nature Genetics, 2003, 33, 487-491.	21.4	375
8	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	6.2	375
9	Mutations in LRP2, which encodes the multiligand receptor megalin, cause Donnai-Barrow and facio-oculo-acoustico-renal syndromes. Nature Genetics, 2007, 39, 957-959.	21.4	284
10	BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. Nature Genetics, 2006, 38, 521-524.	21.4	259
11	Genotype-phenotype relationships in Berardinelli-Seip congenital lipodystrophy. Journal of Medical Genetics, 2002, 39, 722-733.	3.2	233
12	Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing. Journal of Medical Genetics, 2014, 51, 724-736.	3.2	229
13	CHARGE syndrome: Report of 47 cases and review. American Journal of Medical Genetics Part A, 1998, 76, 402-409.	2.4	228
14	Cardio-facio-cutaneous and Noonan syndromes due to mutations in the RAS/MAPK signalling pathway: genotype phenotype relationships and overlap with Costello syndrome. Journal of Medical Genetics, 2007, 44, 763-771.	3.2	221
15	NF1 microdeletions in neurofibromatosis type 1: from genotype to phenotype. Human Mutation, 2010, 31, E1506-E1518.	2.5	208
16	Genotype-phenotype correlation in Costello syndrome: HRAS mutation analysis in 43 cases. Journal of Medical Genetics, 2005, 43, 401-405.	3.2	207
17	SLC26A4 gene is frequently involved in nonsyndromic hearing impairment with enlarged vestibular aqueduct in Caucasian populations. European Journal of Human Genetics, 2006, 14, 773-779.	2.8	204
18	Comparison of Clinical Presentations and Outcomes Between Patients With TGFBR2 and FBN1 Mutations in Marfan Syndrome and Related Disorders. Circulation, 2009, 120, 2541-2549.	1.6	203

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19	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	11.0	195
20	Mutations in PHF8 are associated with X linked mental retardation and cleft lip/cleft palate. Journal of Medical Genetics, 2005, 42, 780-786.	3.2	194
21	Five new TTF1/NKX2.1 mutations in brain-lung-thyroid syndrome: rescue by PAX8 synergism in one case. Human Molecular Genetics, 2009, 18, 2266-2276.	2.9	187
22	Alteration of Fatty-Acid-Metabolizing Enzymes Affects Mitochondrial Form and Function in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1051-1064.	6.2	179
23	Germline mutations in WTX cause a sclerosing skeletal dysplasia but do not predispose to tumorigenesis. Nature Genetics, 2009, 41, 95-100.	21.4	178
24	Heterozygous endothelin receptor B (EDNRB) mutations in isolated Hirschsprung disease. Human Molecular Genetics, 1996, 5, 355-357.	2.9	174
25	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	6.2	171
26	Mutations in SRCAP, Encoding SNF2-Related CREBBP Activator Protein, Cause Floating-Harbor Syndrome. American Journal of Human Genetics, 2012, 90, 308-313.	6.2	157
27	Thrombocytopenia resulting from mutations in filamin A can be expressed as an isolated syndrome. Blood, 2011, 118, 5928-5937.	1.4	148
28	Involvement of hyperprolinemia in cognitive and psychiatric features of the 22q11 deletion syndrome. Human Molecular Genetics, 2007, 16, 83-91.	2.9	147
29	Identification of a hot spot for microdeletions in patients with X- linked deafness type 3 (DFN3) 900 kb proximal to the DFN3 gene POU3F4. Human Molecular Genetics, 1996, 5, 1229-1235.	2.9	138
30	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	2.5	134
31	Structural variation in Xq28: MECP2 duplications in 1% of patients with unexplained XLMR and in 2% of male patients with severe encephalopathy. European Journal of Human Genetics, 2009, 17, 444-453.	2.8	130
32	Schizencephaly: clinical and imaging features in 30 infantile cases. Brain and Development, 2000, 22, 475-483.	1.1	128
33	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. Genetics in Medicine, 2016, 18, 49-56.	2.4	125
34	COL4A1 mutation in Axenfeldâ€“Rieger anomaly with leukoencephalopathy and stroke. Annals of Neurology, 2007, 62, 177-184.	5.3	124
35	A proposed new contiguous gene syndrome on 8q consists of Branchio-Oto-Renal (BOR) syndrome, Duane syndrome, a dominant form of hydrocephalus and trapeze aplasia; implications for the mapping of the BOR gene. Human Molecular Genetics, 1994, 3, 1859-1866.	2.9	121
36	Spectrum of NSD1 mutations in Sotos and Weaver syndromes. Journal of Medical Genetics, 2003, 40, 436-440.	3.2	116

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37	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	2.8	115
38	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. Journal of Clinical Investigation, 2017, 127, 1991-2006.	8.2	115
39	Molecular analysis of the CBP gene in 60 patients with Rubinstein-Taybi syndrome. Journal of Medical Genetics, 2002, 39, 415-421.	3.2	109
40	Phenotype and Outcome in Hereditary Tubulointerstitial Nephritis Secondary to UMOD Mutations. Clinical Journal of the American Society of Nephrology: CJASN, 2011, 6, 2429-2438.	4.5	109
41	Large-Scale Deletions and SMADIP1 Truncating Mutations in Syndromic Hirschsprung Disease with Involvement of Midline Structures. American Journal of Human Genetics, 2001, 69, 1370-1377.	6.2	105
42	Targeted high-throughput sequencing for diagnosis of genetically heterogeneous diseases: efficient mutation detection in Bardet-Biedl and AlstrÃ¶m Syndromes. Journal of Medical Genetics, 2012, 49, 502-512.	3.2	104
43	Mutations in RNF135, a gene within the NF1 microdeletion region, cause phenotypic abnormalities including overgrowth. Nature Genetics, 2007, 39, 963-965.	21.4	103
44	Definition of a Critical Region on Chromosome 18 for Congenital Aural Atresia by ArrayCGH. American Journal of Human Genetics, 2003, 72, 1578-1584.	6.2	102
45	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
46	New insights into genotypeâ€“phenotype correlation for GLI3 mutations. European Journal of Human Genetics, 2015, 23, 92-102.	2.8	97
47	Mutations inSETD2cause a novel overgrowth condition. Journal of Medical Genetics, 2014, 51, 512-517.	3.2	96
48	Screening of SLC26A4 (PDS) gene in Pendred's syndrome: a large spectrum of mutations in France and phenotypic heterogeneity. Clinical Genetics, 2004, 66, 333-340.	2.0	93
49	Complete exon sequencing of all known Usher syndrome genes greatly improves molecular diagnosis. Orphanet Journal of Rare Diseases, 2011, 6, 21.	2.7	93
50	Spectrum of PTCH1 Mutations in French Patients with Gorlin Syndrome. Journal of Investigative Dermatology, 2003, 121, 478-481.	0.7	92
51	Molecular characterization of a series of 990 index patients with albinism. Pigment Cell and Melanoma Research, 2018, 31, 466-474.	3.3	92
52	Lymphedema-lymphangiectasia-mental retardation (Hennekam) syndrome: A review. American Journal of Medical Genetics Part A, 2002, 112, 412-421.	2.4	91
53	Phenotype and genotype in 52 patients with Rubinsteinâ€“Taybi syndrome caused by EP300 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3069-3082.	1.2	91
54	STXBP1-related encephalopathy presenting as infantile spasms and generalized tremor in three patients. Epilepsia, 2011, 52, 1820-1827.	5.1	90

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55	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	2.4	90
56	EFTUD2haploinsufficiency leads to syndromic oesophageal atresia. Journal of Medical Genetics, 2012, 49, 737-746.	3.2	89
57	Molecular findings and clinical data in a cohort of 150 patients with anophthalmia/microphthalmia. Clinical Genetics, 2014, 86, 326-334.	2.0	88
58	Prevalence of Mutations in AGPAT2 Among Human Lipodystrophies. Diabetes, 2003, 52, 1573-1578.	0.6	87
59	Consensus recommendations for diagnosis, management and treatment of Fabry disease in paediatric patients. Clinical Genetics, 2019, 96, 107-117.	2.0	87
60	Identification of 23TGFBR2and 6TGFBR1gene mutations and genotype-phenotype investigations in 457 patients with Marfan syndrome type I and II, Loeys-Dietz syndrome and related disorders. Human Mutation, 2008, 29, E284-E295.	2.5	86
61	Whole mitochondrial genome screening in maternally inherited non-syndromic hearing impairment using a microarray resequencing mitochondrial DNA chip. European Journal of Human Genetics, 2007, 15, 1145-1155.	2.8	85
62	Further delineation of Kabuki syndrome in 48 wellâ€defined new individuals. American Journal of Medical Genetics, Part A, 2005, 132A, 265-272.	1.2	84
63	Molecular diagnosis of oculocutaneous albinism: new mutations in the OCA1â€“4 genes and practical aspects. Pigment Cell and Melanoma Research, 2008, 21, 583-587.	3.3	84
64	The Gene for Bazex-DuprÃ©-Christol Syndrome Maps to Chromosome Xq. Journal of Investigative Dermatology, 1995, 105, 87-91.	0.7	83
65	REEP1 mutations in SPG31: Frequency, mutational spectrum, and potential association with mitochondrial morpho-functional dysfunction. Human Mutation, 2011, 32, 1118-1127.	2.5	83
66	Spectrum of pontocerebellar hypoplasia in 13 girls and boys with CASK mutations: confirmation of a recognizable phenotype and first description of a male mosaic patient. Orphanet Journal of Rare Diseases, 2012, 7, 18.	2.7	83
67	Prenatal diagnosis of female monozygotic twins discordant for Turner syndrome: implications for prenatal genetic counselling. Prenatal Diagnosis, 2002, 22, 697-702.	2.3	81
68	The Prevalence ofCHD7Missense Versus Truncating Mutations Is Higher in Patients With Kallmann Syndrome Than in Typical CHARGE Patients. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E2138-E2143.	3.6	81
69	LRP5 mutations in osteoporosis-pseudoglioma syndrome and high-bone-mass disorders. Joint Bone Spine, 2005, 72, 207-214.	1.6	80
70	Twelve new patients with 13q deletion syndrome: Genotypeâ€“phenotype analyses in progress. European Journal of Medical Genetics, 2009, 52, 41-46.	1.3	80
71	A mutation in the 3â€²-UTR of the HDAC6 gene abolishing the post-transcriptional regulation mediated by hsa-miR-433 is linked to a new form of dominant X-linked chondrodysplasia. Human Molecular Genetics, 2010, 19, 2015-2027.	2.9	80
72	Factor V Leiden, prothrombin 20210A, methylenetetrahydrofolate reductase 677T, and population genetics. Molecular Genetics and Metabolism, 2005, 86, 91-99.	1.1	78

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73	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
74	The 2q37-deletion syndrome: an update of the clinical spectrum including overweight, brachydactyly and behavioural features in 14 new patients. European Journal of Human Genetics, 2013, 21, 602-612.	2.8	78
75	Hydrometrocolpos and polydactyly: a common neonatal presentation of Bardet-Biedl and McKusick-Kaufman syndromes. Journal of Medical Genetics, 1999, 36, 599-603.	3.2	78
76	Metabolic Correction Induced by Leptin Replacement Treatment in Young Children With Berardinelli-Seip Congenital Lipoatrophy. Pediatrics, 2007, 120, e291-e296.	2.1	76
77	Blepharophimosis-mental retardation (BMR) syndromes: A proposed clinical classification of the so-called Ohdo syndrome, and delineation of two new BMR syndromes, one X-linked and one autosomal recessive. American Journal of Medical Genetics, Part A, 2006, 140A, 1285-1296.	1.2	73
78	Mutations in Î³-secretase subunitâ€“encoding PSENEN underlie Dowling-Degos disease associated with acne inversa. Journal of Clinical Investigation, 2017, 127, 1485-1490.	8.2	73
79	AGC1/2, the mitochondrial aspartate-glutamate carriers. Biochimica Et Biophysica Acta - Molecular Cell Research, 2016, 1863, 2394-2412.	4.1	72
80	High glucose repatterns human podocyte energy metabolism during differentiation and diabetic nephropathy. FASEB Journal, 2017, 31, 294-307.	0.5	72
81	C5orf42 is the major gene responsible for OFD syndrome type VI. Human Genetics, 2014, 133, 367-377.	3.8	71
82	Further delineation of the congenital form of X-linked dyskeratosis congenita (Hoyeraal-Hreidarsson) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	2.7	70
83	Testing for triallelism: analysis of six BBS genes in a Bardetâ€“Biedl syndrome family cohort. European Journal of Human Genetics, 2005, 13, 607-616.	2.8	69
84	Metabolic Reprogramming in Amyotrophic Lateral Sclerosis. Scientific Reports, 2018, 8, 3953.	3.3	69
85	Progressive Osseous Heteroplasia: A Model for the Imprinting Effects of GNAS Inactivating Mutations in Humans. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3028-3038.	3.6	68
86	Progressive bulbospinal amyotrophy in Triple A syndrome with AAAS gene mutation. Neurology, 2002, 58, 962-965.	1.1	67
87	Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	2.4	67
88	Confirmation of assigment of a locus for rubinstein-taybi syndrome gene to 16p13.3. American Journal of Medical Genetics Part A, 1992, 44, 126-128.	2.4	65
89	Phenotype and genotype analysis of a French cohort of 119 patients with CHARGE syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 417-430.	1.6	65
90	Identification of a novel mutation confirms the implication of IFT172 (BBS20) in Bardetâ€“Biedl syndrome. Journal of Human Genetics, 2016, 61, 447-450.	2.3	64

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91	Molecular characterization of 1q44 microdeletion in 11 patients reveals three candidate genes for intellectual disability and seizures. American Journal of Medical Genetics, Part A, 2012, 158A, 1633-1640.	1.2	63
92	Heterogeneity ofNSD1alterations in 116 patients with Sotos syndrome. Human Mutation, 2007, 28, 1098-1107.	2.5	62
93	New practical definitions for the diagnosis of autosomal recessive spastic ataxia of <scp>C</scp>harlevoixâ€“<scp>S</scp>aguenay. Annals of Neurology, 2015, 78, 871-886.	5.3	62
94	Mutations of the ImprintedCDKN1CGene as a Cause of the Overgrowth Beckwith-Wiedemann Syndrome: Clinical Spectrum and Functional Characterization. Human Mutation, 2015, 36, 894-902.	2.5	62
95	Townes-Brocks syndrome: Detection of aSALL1 mutation hot spot and evidence for a position effect in one patient. Human Mutation, 1999, 14, 377-386.	2.5	61
96	The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. Orphanet Journal of Rare Diseases, 2013, 8, 63.	2.7	60
97	Spectrum of epilepsy in terminal 1p36 deletion syndrome. Epilepsia, 2008, 49, 509-515.	5.1	59
98	Case with autistic syndrome and chromosome 22q13.3 deletion detected by FISH. American Journal of Medical Genetics Part A, 2000, 96, 839-844.	2.4	58
99	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	3.8	58
100	Wiedemannâ€Steiner syndrome as a major cause of syndromic intellectual disability: A study of 33 French cases. Clinical Genetics, 2018, 94, 141-152.	2.0	57
101	Ondine-Hirschsprung syndrome (Haddad syndrome). European Journal of Pediatrics, 1993, 152, 75-77.	2.7	56
102	Mutational spectrum of COH1 and clinical heterogeneity in Cohen syndrome. Journal of Medical Genetics, 2005, 43, e22-e22.	3.2	56
103	New clinicoâ€genetic classification of trichothiodystrophy. American Journal of Medical Genetics, Part A, 2009, 149A, 2020-2030.	1.2	56
104	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
105	SETD2andDNMT3Ascreen in the Sotos-like syndrome French cohort. Journal of Medical Genetics, 2016, 53, 743-751.	3.2	54
106	Mutations in WNT10A are frequently involved in oligodontia associated with minor signs of ectodermal dysplasia. American Journal of Medical Genetics, Part A, 2013, 161, 671-678.	1.2	52
107	Neurodevelopmental outcome following prenatal diagnosis of an isolated anomaly of the corpus callosum. Ultrasound in Obstetrics and Gynecology, 2011, 37, 290-295.	1.7	51
108	Mutations inMYT1, encoding the myelin transcription factor 1, are a rare cause of OAVS. Journal of Medical Genetics, 2016, 53, 752-760.	3.2	51

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109	Resistance to leptin-replacement therapy in Berardinelliâ€“Seip congenital lipodystrophy: an immunological origin. European Journal of Endocrinology, 2010, 162, 1083-1091.	3.7	50
110	Delineation ofEFTUD2Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. Human Mutation, 2014, 35, 478-485.	2.5	50
111	Costello syndrome and neurological abnormalities. American Journal of Medical Genetics Part A, 2003, 123A, 301-305.	2.4	49
112	Identification of Novel Mutations ConfirmsPde4das a Major Gene Causing Acrodysostosis. Human Mutation, 2013, 34, 97-102.	2.5	49
113	New candidate loci identified by arrayâ€CGH in a cohort of 100 children presenting with syndromic obesity. American Journal of Medical Genetics, Part A, 2014, 164, 1965-1975.	1.2	49
114	Four-Year Follow-up of Diagnostic Service in USH1 Patients. , 2011, 52, 4063.		47
115	Mutations in the Endothelin Receptor Type A Cause Mandibulofacial Dysostosis with Alopecia. American Journal of Human Genetics, 2015, 96, 519-531.	6.2	47
116	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
117	Neuroimaging findings in Mowatâ€“Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	2.4	45
118	Further delineation of theMECP2duplication syndrome phenotype in 59 French male patients, with a particular focus on morphological and neurological features. Journal of Medical Genetics, 2018, 55, 359-371.	3.2	45
119	BLOC1S5 pathogenic variants cause a new type of Hermanskyâ€“Pudlak syndrome. Genetics in Medicine, 2020, 22, 1613-1622.	2.4	44
120	Analysis of CBP (CREBBP) gene deletions in Rubinstein-Taybi syndrome patients using real-time quantitative PCR. Human Mutation, 2004, 23, 278-284.	2.5	43
121	The phenotype of Floatingâ€“Harbor syndrome in 10 patients. American Journal of Medical Genetics, Part A, 2010, 152A, 821-829.	1.2	43
122	CREBBP mutations in individuals without Rubinsteinâ€“Taybi syndrome phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2681-2693.	1.2	43
123	Molecular characterisation of patients with subtelomeric 22q abnormalities using chromosome specific array-based comparative genomic hybridisation. European Journal of Human Genetics, 2005, 13, 1019-1024.	2.8	42
124	Spectrum of CREBBP gene dosage anomalies in Rubinsteinâ€“Taybi Syndrome patients. European Journal of Human Genetics, 2007, 15, 843-847.	2.8	41
125	Reduced Placental Telomere Length during Pregnancies Complicated by Intrauterine Growth Restriction. PLoS ONE, 2013, 8, e54013.	2.5	41
126	Exome sequencing in congenital ataxia identifies two new candidate genes and highlights a pathophysiological link between some congenital ataxias and early infantile epileptic encephalopathies. Genetics in Medicine, 2019, 21, 553-563.	2.4	41

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127	Growth patterns of patients with Noonan syndrome: correlation with age and genotype. European Journal of Endocrinology, 2016, 174, 641-650.	3.7	40
128	Socio-Behavioral Characteristics of Children with Rubinstein-Taybi Syndrome. Journal of Autism and Developmental Disorders, 2009, 39, 1252-1260.	2.7	39
129	Leberâ€™s optic neuropathy associated with disseminated white matter disease: A case report and review. Clinical Neurology and Neurosurgery, 2009, 111, 83-86.	1.4	39
130	Neurologic Aspects of MECP2 Gene Duplication in Male Patients. Pediatric Neurology, 2009, 41, 187-191.	2.1	39
131	Mutations inSNRPB, Encoding Components of the Core Splicing Machinery, Cause Cerebro-Costo-Mandibular Syndrome. Human Mutation, 2015, 36, 187-190.	2.5	39
132	The expanding spectrum of COL2A1 gene variants IN 136 patients with a skeletal dysplasia phenotype. European Journal of Human Genetics, 2016, 24, 992-1000.	2.8	39
133	Mitochondrial functions and rare diseases. Molecular Aspects of Medicine, 2020, 71, 100842.	6.4	39
134	Submicroscopic deletion of chromosome 16p13.3 in patients with Rubinstein-Taybi syndrome. American Journal of Medical Genetics Part A, 1998, 78, 267-270.	2.4	38
135	Osteopathia striata cranial sclerosis: Non-random X-inactivation suggestive of X-linked dominant inheritance. American Journal of Medical Genetics Part A, 2002, 107, 1-4.	2.4	38
136	Poikiloderma with neutropenia, Clericuzio type, in a family from Morocco. American Journal of Medical Genetics, Part A, 2008, 146A, 2762-2769.	1.2	38
137	Bilateral periventricular nodular heterotopia in France: frequency of mutations in FLNA, phenotypic heterogeneity and spectrum of mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 1394-1398.	1.9	38
138	Targeted resequencing identifies PTCH1 as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. Genome Research, 2016, 26, 474-485.	5.5	37
139	Screening of ARX in mental retardation families: consequences for the strategy of molecular diagnosis. Neurogenetics, 2006, 7, 39-46.	1.4	36
140	Bardet-Biedl Syndrome and Brain Abnormalities. Neuropediatrics, 2007, 38, 5-9.	0.6	36
141	SLC24A5 Mutations Are Associated with Non-Syndromic Oculocutaneous Albinism. Journal of Investigative Dermatology, 2014, 134, 568-571.	0.7	36
142	Combination of WAGR and Potockiâ€“Shaffer contiguous deletion syndromes in a patient with an 11p11.2â€“p14 deletion. European Journal of Human Genetics, 2005, 13, 409-413.	2.8	35
143	A novel de novo mutation in MYT1, the unique OAVS gene identified so far. European Journal of Human Genetics, 2017, 25, 1083-1086.	2.8	35
144	Floating-Harbor syndrome: description of a further patient, review of the literature, and suggestion of autosomal dominant inheritance. European Journal of Pediatrics, 1995, 154, 658-661.	2.7	34

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145	Towards a new point of view on the phenotype of patients with a 17q12 microdeletion syndrome. Archives of Disease in Childhood, 2015, 100, 259-264.	1.9	34
146	Clinical and genetic heterogeneity in familial steroid-sensitive nephrotic syndrome. Pediatric Nephrology, 2018, 33, 473-483.	1.7	34
147	Rubinstein-Taybi Syndrome: A Model of Epigenetic Disorder. Genes, 2021, 12, 968.	2.4	34
148	A second locus for Aicardi-Goutieres syndrome at chromosome 13q14-21. Journal of Medical Genetics, 2005, 43, 444-450.	3.2	33
149	Further delineation of the phenotype caused by biallelic variants in the <scp>WDR4</scp> gene. Clinical Genetics, 2018, 93, 374-377.	2.0	33
150	Dopachrome tautomerase variants in patients with oculocutaneous albinism. Genetics in Medicine, 2021, 23, 479-487.	2.4	33
151	Osteoporosis in late-diagnosed adult homocystinuric patients. Journal of Inherited Metabolic Disease, 2000, 23, 338-340.	3.6	32
152	Costello syndrome: report of six patients including one with an embryonal rhabdomyosarcoma. European Journal of Pediatrics, 2000, 159, 139-142.	2.7	32
153	2.3ÂMb terminal deletion in 12p13.33 associated with oculoauriculovertebral spectrum and evaluation of WNT5B as a candidate gene. European Journal of Medical Genetics, 2009, 52, 446-449.	1.3	32
154	Molecular, clinical and neuropsychological study in 31 patients with Kabuki syndrome and <scp>KMT2D</scp> mutations. Clinical Genetics, 2017, 92, 298-305.	2.0	32
155	Search for the best indicators for the presence of a VPS13B gene mutation and confirmation of diagnostic criteria in a series of 34 patients genotyped for suspected Cohen syndrome. Journal of Medical Genetics, 2010, 47, 549-553.	3.2	31
156	The male phenotype in osteopathia striata congenita with cranial sclerosis. American Journal of Medical Genetics, Part A, 2011, 155, 2397-2408.	1.2	31
157	Prenatal diagnosis using array-CGH: A French experience. European Journal of Medical Genetics, 2013, 56, 341-345.	1.3	31
158	Functional validation of ABHD12 mutations in the neurodegenerative disease PHARC. Neurobiology of Disease, 2017, 98, 36-51.	4.4	31
159	Congenital hypotrichosis and milia: Report of a large family suggesting X-linked dominant inheritance. American Journal of Medical Genetics Part A, 1994, 52, 487-490.	2.4	30
160	Cutaneous Manifestations in Costello and Cardiofaciocutaneous Syndrome: Report of 18 Cases and Literature Review. Pediatric Dermatology, 2013, 30, 665-673.	0.9	30
161	Homozygous and compound heterozygous mutations in theFBN1gene: unexpected findings in molecular diagnosis of Marfan syndrome. Journal of Medical Genetics, 2017, 54, 100-103.	3.2	30
162	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. Genetics in Medicine, 2020, 22, 181-188.	2.4	30

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