Didier Lacombe

List of Publications by Year in Descending Order

319 papers	16,361 citations	64 h-index	116 g-index
351 ext. papers	18,626 ext. citations	5.4 avg, IF	5.39 L-index

#	Paper	IF	Citations
319	HRAS germline mutations impair LKB1/AMPK signaling and mitochondrial homeostasis in Costello syndrome models.. *Journal of Clinical Investigation*, **2022**,	15.9	2
318	CHN1 and duane retraction syndrome: Expanding the phenotype to cranial nerves development disease. *European Journal of Medical Genetics*, **2021**, 64, 104188	2.6	2
317	Rubinstein-Taybi Syndrome: A Model of Epigenetic Disorder. *Genes*, **2021**, 12,	4.2	4
316	CHEDDA syndrome is an underrecognized neurodevelopmental disorder with a highly restricted ATN1 mutation spectrum. *Clinical Genetics*, **2021**, 100, 468-477	4	2
315	Effects of eight neuropsychiatric copy number variants on human brain structure. *Translational Psychiatry*, **2021**, 11, 399	8.6	3
314	Psychomotor, cognitive, and socio-emotional developmental profiles of children with Rubinstein-Taybi Syndrome and a severe intellectual disability. *Journal of Intellectual and Developmental Disability*, **2021**, 46, 80-89	1.9	4
313	Dopachrome tautomerase variants in patients with oculocutaneous albinism. *Genetics in Medicine*, **2021**, 23, 479-487	8.1	8
312	Clinical and neuroimaging findings in 33 patients with MCAP syndrome: A survey to evaluate relevant endpoints for future clinical trials. *Clinical Genetics*, **2021**, 99, 650-661	4	4
311	A recurrent missense variant in EYA3 gene is associated with oculo-auriculo-vertebral spectrum. *Human Genetics*, **2021**, 140, 933-944	6.3	2
310	Clinical description and mutational profile of a Moroccan series of patients with Rubinstein Taybi syndrome. *African Health Sciences*, **2021**, 21, 960-967	1.1	0
309	Proteomic Study of Low-Birth-Weight Nephropathy in Rats. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
308	A comparative study of cognitive and socio-emotional development in children with Rubinstein-Taybi syndrome and children with Autism Spectrum Disorder associated with a severe intellectual disability, and in young typically developing children with matched developmental ages. *Research in Developmental Disabilities*, **2021**, 116, 104029	2.7	1
307	BLOC1S5 pathogenic variants cause a new type of Hermansky-Pudlak syndrome. *Genetics in Medicine*, **2020**, 22, 1613-1622	8.1	25
306	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. *American Journal of Human Genetics*, **2020**, 106, 356-370	11	51
305	Primrose syndrome: a phenotypic comparison of patients with a ZBTB20 missense variant versus a 3q13.31 microdeletion including ZBTB20. *European Journal of Human Genetics*, **2020**, 28, 1044-1055	5.3	2
304	Mitochondrial functions and rare diseases. *Molecular Aspects of Medicine*, **2020**, 71, 100842	16.7	18
303	Excess of de novo variants in genes involved in chromatin remodelling in patients with marfanoid habitus and intellectual disability. *Journal of Medical Genetics*, **2020**, 57, 466-474	5.8	2

302	Growth charts in Kabuki syndrome 1. *American Journal of Medical Genetics, Part A*, **2020**, 182, 446-453	2.5	4
301	Description of a family with X-linked oculo-auriculo-vertebral spectrum associated with polyalanine tract expansion in ZIC3. *Clinical Genetics*, **2020**, 98, 384-389	4	6
300	Functional and genetic analyses of ZYG11B provide evidences for its involvement in OAVS. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1375	2.3	8
299	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. *Genetics in Medicine*, **2020**, 22, 181-188	8.1	12
298	Cornea verticillata and acroparesthesia efficiently discriminate clusters of severity in Fabry disease **2020**, 15, e0233460		
297	Cornea verticillata and acroparesthesia efficiently discriminate clusters of severity in Fabry disease **2020**, 15, e0233460		
296	Cornea verticillata and acroparesthesia efficiently discriminate clusters of severity in Fabry disease **2020**, 15, e0233460		
295	Cornea verticillata and acroparesthesia efficiently discriminate clusters of severity in Fabry disease **2020**, 15, e0233460		
294	Semen and male genital tract characteristics of patients with Fabry disease: the FERTIFABRY multicentre observational study. *Basic and Clinical Andrology*, **2019**, 29, 7	2.8	2
293	Fetal phenotype of Rubinstein-Taybi syndrome caused by CREBBP mutations. *Clinical Genetics*, **2019**, 95, 420-426	4	4
292	Consensus recommendations for diagnosis, management and treatment of Fabry disease in paediatric patients. *Clinical Genetics*, **2019**, 96, 107-117	4	39
291	Dermatological manifestations in cardiofaciocutaneous syndrome: a prospective multicentric study of 45 mutation-positive patients. *British Journal of Dermatology*, **2019**, 180, 172-180	4	8
290	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	8.1	25
289	Developmental trajectories of neuroanatomical alterations associated with the 16p11.2 Copy Number Variations. *NeuroImage*, **2019**, 203, 116155	7.9	6
288	Treatment needs and expectations for Fabry disease in France: development of a new Patient Needs Questionnaire. *Orphanet Journal of Rare Diseases*, **2019**, 14, 284	4.2	5
287	Dermatological manifestations in Noonan syndrome: a´prospective multicentric study of 129 patients positive for mutation. *British Journal of Dermatology*, **2019**, 180, 1438-1448	4	12
286	Metabolic Reprogramming in Amyotrophic Lateral Sclerosis. *Scientific Reports*, **2018**, 8, 3953	4.9	45
285	Wiedemann-Steiner syndrome as a major cause of syndromic intellectual disability: A study of 33 French cases. *Clinical Genetics*, **2018**, 94, 141-152	4	33

284	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. *Biological Psychiatry*, **2018**, 84, 253-264	7.9	33
283	Further delineation of the duplication syndrome phenotype in 59 French male patients, with a particular focus on morphological and neurological features. *Journal of Medical Genetics*, **2018**, 55, 359-371	5.8	25
282	A neuropathological study of novel RTTN gene mutations causing a familial microcephaly with simplified gyral pattern. *Birth Defects Research*, **2018**, 110, 598-602	2.9	6
281	Molecular characterization of a series of 990 index patients with albinism. *Pigment Cell and Melanoma Research*, **2018**, 31, 466-474	4.5	55
280	Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. *Genetics in Medicine*, **2018**, 20, 965-975	8.1	37
279	Non-specific gastrointestinal features: Could it be Fabry disease?. *Digestive and Liver Disease*, **2018**, 50, 429-437	3.3	21
278	Further delineation of the phenotype caused by biallelic variants in the WDR4 gene. *Clinical Genetics*, **2018**, 93, 374-377	4	17
277	Clinical and genetic heterogeneity in familial steroid-sensitive nephrotic syndrome. *Pediatric Nephrology*, **2018**, 33, 473-483	3.2	22
276	CYP2U1 activity is altered by missense mutations in hereditary spastic paraplegia 56. *Human Mutation*, **2018**, 39, 140-151	4.7	12
275	Deep characterization of the anti-drug antibodies developed in Fabry disease patients, a prospective analysis from the French multicenter cohort FFABRY. *Orphanet Journal of Rare Diseases* , **2018**, 13, 127	4.2	15
274	Apport des puces ˆADN et nouveaux syndromes microdltionnels. *Bulletin De LhAcademie Nationale De Medecine*, **2018**, 202, 693-705	0.1	
273	19p13 microduplications encompassing NFIX are responsible for intellectual disability, short stature and small head circumference. *European Journal of Human Genetics*, **2018**, 26, 85-93	5.3	4
272	Prenatal retinoic acid exposure reveals candidate genes for craniofacial disorders. *Scientific Reports* , **2018**, 8, 17492	4.9	7
271	Interest of chromosomal microarray analysis in the prenatal diagnosis of fetal intrauterine growth restriction. *Prenatal Diagnosis*, **2018**, 38, 1111-1119	3.2	7
270	Novel KDM5B splice variants identified in patients with developmental disorders: Functional consequences. *Gene*, **2018**, 679, 305-313	3.8	10
269	IL11RA-related Crouzon-like autosomal recessive craniosynostosis in 10 new patients: Resemblances and differences. *Clinical Genetics*, **2018**, 94, 373-380	4	21
268	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. *Genetics in Medicine*, **2017**, 19, 989-997	8.1	62
267	Homozygous and compound heterozygous mutations in the FBN1 gene: unexpected findings in molecular diagnosis of Marfan syndrome. *Journal of Medical Genetics*, **2017**, 54, 100-103	5.8	21

266	A novel de novo mutation in MYT1, the unique OAVS gene identified so far. *European Journal of Human Genetics*, **2017**, 25, 1083-1086	5.3	21
265	Molecular, clinical and neuropsychological study in 31 patients with Kabuki syndrome and KMT2D mutations. *Clinical Genetics*, **2017**, 92, 298-305	4	20
264	Deletion of the transcription factor SOX4 is implicated in syndromic nephroblastoma. *Clinical Genetics*, **2017**, 92, 449-450	4	1
263	Copy Number Variations Found in Patients with a Corpus Callosum Abnormality and Intellectual Disability. *Journal of Pediatrics*, **2017**, 185, 160-166.e1	3.6	16
262	First female prenatal case of osteopathia striata with cranial sclerosis in a fetus carrying a de-novo 1.9 Mbp interstitial deletion at Xq11.1q11.2. *Clinical Dysmorphology*, **2017**, 26, 231-234	0.9	1
261	Recommendations for the inclusion of Fabry disease as a rare febrile condition in existing algorithms for fever of unknown origin. *Internal and Emergency Medicine*, **2017**, 12, 1059-1067	3.7	2
260	Clinico-molecular analysis of eleven patients with Hermansky-Pudlak type 5 syndrome, a mild form of HPS. *Pigment Cell and Melanoma Research*, **2017**, 30, 563-570	4.5	11
259	Evaluation of Motor Skills in Children with Rubinstein-Taybi Syndrome. *Journal of Autism and Developmental Disorders*, **2017**, 47, 3321-3332	4.6	7
258	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	3.1	41
257	High glucose repatterns human podocyte energy metabolism during differentiation and diabetic nephropathy. *FASEB Journal*, **2017**, 31, 294-307	0.9	47
256	Neuroimaging findings in Mowat-Wilson syndrome: a study of 54 patients. *Genetics in Medicine*, **2017**, 19, 691-700	8.1	28
255	Functional validation of ABHD12 mutations in the neurodegenerative disease PHARC. *Neurobiology of Disease*, **2017**, 98, 36-51	7.5	23
254	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. *Npj Genomic Medicine*, **2017**, 2, 32	6.2	32
253	Mutations in γ-secretase subunit-encoding PSENEN underlie Dowling-Degos disease associated with acne inversa. *Journal of Clinical Investigation*, **2017**, 127, 1485-1490	15.9	53
252	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. *Journal of Clinical Investigation*, **2017**, 127, 1991-2006	15.9	73
251	Mitochondrial morphology and cellular distribution are altered in SPG31 patients and are linked to DRP1 hyperphosphorylation. *Human Molecular Genetics*, **2017**, 26, 674-685	5.6	19
250	Albinism in a patient with mutations at both the OA1 and OCA3 loci. *Pigment Cell and Melanoma Research*, **2016**, 29, 107-9	4.5	3
249	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. *Genetics in Medicine*, **2016**, 18, 49-56	8.1	78

248	Phenotype and genotype in 52 patients with Rubinstein-Taybi syndrome caused by EP300 mutations. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3069-3082	2.5	60
247	Consultations d'oncogénétique pédiatrique : Quelles indications et quelles pratiques dans un service spécialisé de cancérologie pédiatrique ? Enquête dans le centre spécialisé du CHU de Bordeaux en 2011-2012. *Revue D'Oncologie Hématologie Pédiatrique*, **2016**, 4, 35-45		
246	Duplication of PTHLH causes osteochondroplasia with a combined brachydactyly type E/A1 phenotype with disturbed bone maturation and rhizomelia. *European Journal of Human Genetics*, **2016**, 24, 1132-6	5.3	5
245	Identification of a novel mutation confirms the implication of IFT172 (BBS20) in Bardet-Biedl syndrome. *Journal of Human Genetics*, **2016**, 61, 447-50	4.3	45
244	Mutations in RIT1 cause Noonan syndrome with possible juvenile myelomonocytic leukemia but are not involved in acute lymphoblastic leukemia. *European Journal of Human Genetics*, **2016**, 24, 1124-31	5.3	20
243	The expanding spectrum of COL2A1 gene variants IN 136 patients with a skeletal dysplasia phenotype. *European Journal of Human Genetics*, **2016**, 24, 992-1000	5.3	29
242	Targeted resequencing identifies PTCH1 as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. *Genome Research*, **2016**, 26, 474-85	9.7	27
241	Growth patterns of patients with Noonan syndrome: correlation with age and genotype. *European Journal of Endocrinology*, **2016**, 174, 641-50	6.5	25
240	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. *JAMA Psychiatry*, **2016**, 73, 20-30	14.5	120
239	Complete loss of function of the ubiquitin ligase HERC2 causes a severe neurodevelopmental phenotype. *European Journal of Human Genetics*, **2016**, 25, 52-58	5.3	17
238	Fetal phenotypes in otopalatodigital spectrum disorders. *Clinical Genetics*, **2016**, 89, 371-7	4	15
237	Mutation Update for Kabuki Syndrome Genes KMT2D and KDM6A and Further Delineation of X-Linked Kabuki Syndrome Subtype 2. *Human Mutation*, **2016**, 37, 847-64	4.7	89
236	CREBBP mutations in individuals without Rubinstein-Taybi syndrome phenotype. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2681-93	2.5	28
235	and screen in the Sotos-like syndrome French cohort. *Journal of Medical Genetics*, **2016**, 53, 743-751	5.8	36
234	Mutations in , encoding the myelin transcription factor 1, are a rare cause of OAVS. *Journal of Medical Genetics*, **2016**, 53, 752-760	5.8	32
233	Otopalatodigital spectrum disorders: refinement of the phenotypic and mutational spectrum. *Journal of Human Genetics*, **2016**, 61, 693-9	4.3	14
232	AGC1/2, the mitochondrial aspartate-glutamate carriers. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2016**, 1863, 2394-412	4.9	49
231	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	4.7	34

230	Typical facial gestalt in X-linked Kabuki syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3363-3364	2.5	3
229	Large national series of patients with Xq28 duplication involving MECP2: Delineation of brain MRI abnormalities in 30 affected patients. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 116-29	2.5	15
228	Identification of a homozygous mutation of SLC24A5 (OCA6) in two patients with oculocutaneous albinism from French Guiana. *Pigment Cell and Melanoma Research*, **2016**, 29, 104-6	4.5	14
227	ARID1B mutations are the major genetic cause of corpus callosum anomalies in patients with intellectual disability. *Brain*, **2016**, 139, e64	11.2	16
226	Mutations in SNRPB, encoding components of the core splicing machinery, cause cerebro-costo-mandibular syndrome. *Human Mutation*, **2015**, 36, 187-90	4.7	30
225	Towards a new point of view on the phenotype of patients with a 17q12 microdeletion syndrome. *Archives of Disease in Childhood*, **2015**, 100, 259-64	2.2	25
224	Mutations in the endothelin receptor type A cause mandibulofacial dysostosis with alopecia. *American Journal of Human Genetics*, **2015**, 96, 519-31	11	28
223	Mutations of the Imprinted CDKN1C Gene as a Cause of the Overgrowth Beckwith-Wiedemann Syndrome: Clinical Spectrum and Functional Characterization. *Human Mutation*, **2015**, 36, 894-902	4.7	47
222	Phenotype-genotype correlations in 17 new patients with an Xp11.23p11.22 microduplication and review of the literature. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 111-22	2.5	17
221	Baraitser-Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. *European Journal of Human Genetics*, **2015**, 23, 292-301	5.3	83
220	New insights into genotype-phenotype correlation for GLI3 mutations. *European Journal of Human Genetics*, **2015**, 23, 92-102	5.3	75
219	Homozygous 16p13.11 duplication associated with mild intellectual disability and urinary tract malformations in two siblings born from consanguineous parents. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2714-9	2.5	7
218	New practical definitions for the diagnosis of autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Annals of Neurology*, **2015**, 78, 871-86	9.4	45
217	SCA27 is a cause of early-onset ataxia and developmental delay. *European Journal of Paediatric Neurology*, **2015**, 19, 271-3	3.8	5
216	C5orf42 is the major gene responsible for OFD syndrome type VI. *Human Genetics*, **2014**, 133, 367-77	6.3	59
215	Mutations in SETD2 cause a novel overgrowth condition. *Journal of Medical Genetics*, **2014**, 51, 512-7	5.8	64
214	High-resolution array-CGH in patients with oculocutaneous albinism identifies new deletions of the TYR, OCA2, and SLC45A2 genes and a complex rearrangement of the OCA2 gene. *Pigment Cell and Melanoma Research*, **2014**, 27, 59-71	4.5	19
213	Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing. *Journal of Medical Genetics*, **2014**, 51, 724-36	5.8	177

212	A novel FTL mutation responsible for neuroferritinopathy with asymmetric clinical features and brain anomalies. *Parkinsonism and Related Disorders*, **2014**, 20, 935-7	3.6	11
211	New candidate loci identified by array-CGH in a cohort of 100 children presenting with syndromic obesity. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1965-75	2.5	37
210	The c.429_452 duplication of the ARX gene: a unique developmental-model of limb kinetic apraxia. *Orphanet Journal of Rare Diseases*, **2014**, 9, 25	4.2	9
209	The prevalence of CHD7 missense versus truncating mutations is higher in patients with Kallmann syndrome than in typical CHARGE patients. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E2138-43	5.6	56
208	Delineation of EFTUD2 haploinsufficiency-related phenotypes through a series of 36 patients. *Human Mutation*, **2014**, 35, 478-85	4.7	38
207	Atypical hematologic and renal manifestations in neurofibromatosis type I: coincidence or pathophysiological link?. *European Journal of Medical Genetics*, **2014**, 57, 639-42	2.6	4
206	Molecular findings and clinical data in a cohort of 150 patients with anophthalmia/microphthalmia. *Clinical Genetics*, **2014**, 86, 326-34	4	74
205	Rare genetic diseases, signalling pathways, and keloid scar formation. *British Journal of Dermatology*, **2014**, 171, 452-3	4	2
204	SLC24A5 mutations are associated with non-syndromic oculocutaneous albinism. *Journal of Investigative Dermatology*, **2014**, 134, 568-571	4.3	29
203	Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. *Nature Genetics*, **2013**, 45, 639-47	36.3	309
202	Expanding the clinical phenotype at the 3q13.31 locus with a new case of microdeletion and first characterization of the reciprocal duplication. *Molecular Genetics and Metabolism*, **2013**, 110, 90-7	3.7	12
201	Identification of novel mutations confirms PDE4D as a major gene causing acrodysostosis. *Human Mutation*, **2013**, 34, 97-102	4.7	46
200	Germline mosaicism in Rubinstein-Taybi syndrome. *Gene*, **2013**, 518, 476-8	3.8	12
199	Detailed clinical, genetic and neuroimaging characterization of OFD VI syndrome. *European Journal of Medical Genetics*, **2013**, 56, 301-8	2.6	14
198	Clinical and molecular spectrum of renal malformations in Kabuki syndrome. *Journal of Pediatrics*, **2013**, 163, 742-6	3.6	15
197	Mutations in WNT10A are frequently involved in oligodontia associated with minor signs of ectodermal dysplasia. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 671-8	2.5	39
196	Systematic molecular and cytogenetic screening of 100 patients with marfanoid syndromes and intellectual disability. *Clinical Genetics*, **2013**, 84, 507-21	4	20
195	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	4.2	47

194	Prenatal diagnosis using array-CGH: a French experience. *European Journal of Medical Genetics*, **2013**, 56, 341-5	2.6	27
193	Phenotypic spectrum of Simpson-Golabi-Behmel syndrome in a series of 42 cases with a mutation in GPC3 and review of the literature. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2013**, 163C, 92-105	3.1	65
192	Cutaneous manifestations in Costello and cardiofaciocutaneous syndrome: report of 18 cases and literature review. *Pediatric Dermatology*, **2013**, 30, 665-73	1.9	22
191	Finger creases lend a hand in Kabuki syndrome. *European Journal of Medical Genetics*, **2013**, 56, 556-60	2.6	9
190	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, 602-12	5.3	64
189	Adaptative capacity of mitochondrial biogenesis and of mitochondrial dynamics in response to pathogenic respiratory chain dysfunction. *Antioxidants and Redox Signaling*, **2013**, 19, 350-65	8.4	15
188	Reduced placental telomere length during pregnancies complicated by intrauterine growth restriction. *PLoS ONE*, **2013**, 8, e54013	3.7	31
187	Novel comprehensive diagnostic strategy in Pitt-Hopkins syndrome: clinical score and further delineation of the TCF4 mutational spectrum. *Human Mutation*, **2012**, 33, 64-72	4.7	84
186	Search for a gene responsible for Floating-Harbor syndrome on chromosome 12q15q21.1. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 333-9	2.5	6
185	A homozygous balanced reciprocal translocation suggests LINC00237 as a candidate gene for MOMO (macrosomia, obesity, macrocephaly, and ocular abnormalities) syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2849-56	2.5	14
184	Oral manifestations of patients with Kenny-Caffey Syndrome. *European Journal of Medical Genetics*, **2012**, 55, 441-5	2.6	18
183	1.5 Mb microdeletion in 15q24 in a patient with mild OAVS phenotype. *European Journal of Medical Genetics*, **2012**, 55, 135-9	2.6	18
182	Dysmorphic features in subtelomeric 20p13 deletion excluding JAG1: a recognizable microdeletion phenotype?. *European Journal of Medical Genetics*, **2012**, 55, 151-5	2.6	6
181	Atypical male and female presentations of FLNA-related periventricular nodular heterotopia. *European Journal of Medical Genetics*, **2012**, 55, 313-8	2.6	17
180	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	4.2	61
179	Alteration of fatty-acid-metabolizing enzymes affects mitochondrial form and function in hereditary spastic paraplegia. *American Journal of Human Genetics*, **2012**, 91, 1051-64	11	150
178	Blepharophimosis, ptosis, epicanthus inversus syndrome with translocation and deletion at chromosome 3q23 in a black African female. *European Journal of Medical Genetics*, **2012**, 55, 630-4	2.6	8
177	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, 1633-40	2.5	49

176	Mutations in SRCAP, encoding SNF2-related CREBBP activator protein, cause Floating-Harbor syndrome. *American Journal of Human Genetics*, **2012**, 90, 308-13	11	130
175	EFTUD2 haploinsufficiency leads to syndromic oesophageal atresia. *Journal of Medical Genetics*, **2012**, 49, 737-46	5.8	75
174	Targeted high-throughput sequencing for diagnosis of genetically heterogeneous diseases: efficient mutation detection in Bardet-Biedl and Alstrm syndromes. *Journal of Medical Genetics*, **2012**, 49, 502-12	5.8	96
173	Mutations of TSEN and CASK genes are prevalent in pontocerebellar hypoplasias type 2 and 4. *Brain*, **2012**, 135, e199; author reply e200	11.2	12
172	An unusual chromosome 22q11 deletion associated with an apparent complementary ring chromosome in a phenotypically normal woman. *European Journal of Medical Genetics*, **2011**, 54, 292-4	2.6	2
171	Rheumatologic and neurological events in an elderly patient with tricho-rhino-phalangeal syndrome type I. *European Journal of Medical Genetics*, **2011**, 54, e405-8	2.6	6
170	Thrombocytopenia resulting from mutations in filamin A can be expressed as an isolated syndrome. *Blood*, **2011**, 118, 5928-37	2.2	128
169	STXBP1-related encephalopathy presenting as infantile spasms and generalized tremor in three patients. *Epilepsia*, **2011**, 52, 1820-7	6.4	75
168	Rubinstein-Taybi syndrome (CREBBP, EP300). *European Journal of Human Genetics*, **2011**, 19, preceeding 118-20	5.3	20
167	Neurodevelopmental outcome following prenatal diagnosis of an isolated anomaly of the corpus callosum. *Ultrasound in Obstetrics and Gynecology*, **2011**, 37, 290-5	5.8	43
166	High resolution mapping of OCA2 intragenic rearrangements and identification of a founder effect associated with a deletion in Polish albino patients. *Human Genetics*, **2011**, 129, 199-208	6.3	14
165	Mutation screening of ASMT, the last enzyme of the melatonin pathway, in a large sample of patients with intellectual disability. *BMC Medical Genetics*, **2011**, 12, 17	2.1	21
164	Complete exon sequencing of all known Usher syndrome genes greatly improves molecular diagnosis. *Orphanet Journal of Rare Diseases*, **2011**, 6, 21	4.2	78
163	The male phenotype in osteopathia striata congenita with cranial sclerosis. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2397-408	2.5	27
162	REEP1 mutations in SPG31: frequency, mutational spectrum, and potential association with mitochondrial morpho-functional dysfunction. *Human Mutation*, **2011**, 32, 1118-27	4.7	68
161	Phenotype and outcome in hereditary tubulointerstitial nephritis secondary to UMOD mutations. *Clinical Journal of the American Society of Nephrology: CJASN*, **2011**, 6, 2429-38	6.9	85
160	Four-year follow-up of diagnostic service in USH1 patients **2011**, 52, 4063-71		41
159	Evidence of postzygotic mosaicism in a transmitted form of Conradi-Hunermann-Happle syndrome associated with a novel EBP mutation. *Archives of Dermatology*, **2011**, 147, 1073-6		14

158	Array-based comparative genomic hybridization identifies a high frequency of copy number variations in patients with syndromic overgrowth. *European Journal of Human Genetics*, **2010**, 18, 227-32	5.3	18
157	Resistance to leptin-replacement therapy in Berardinelli-Seip congenital lipodystrophy: an immunological origin. *European Journal of Endocrinology*, **2010**, 162, 1083-91	6.5	36
156	A mutation in the 3'-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, 2015-27	5.6	71
155	Progressive osseous heteroplasia: a model for the imprinting effects of GNAS inactivating mutations in humans. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, 3028-38	5.6	60
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