Didier Lacombe

List of Publications by Citations

Source: https://exaly.com/author-pdf/6681198/didier-lacombe-publications-by-citations.pdf
Version: 2024-04-28

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.
The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	LDL receptor-related protein 5 (LRP5) affects bone accrual and eye development. *Cell*, **2001**, 107, 513-23	36.2	1827
318	Identification of the gene altered in Berardinelli-Seip congenital lipodystrophy on chromosome 11q13. *Nature Genetics*, **2001**, 28, 365-70	36.3	572
317	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, 157-64	36.3	566
316	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-Goutières syndrome and mimic congenital viral brain infection. *Nature Genetics*, **2006**, 38, 910-6	36.3	505
315	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, 507-15	5.6	458
314	Localized mutations in the gene encoding the cytoskeletal protein filamin A cause diverse malformations in humans. *Nature Genetics*, **2003**, 33, 487-91	36.3	337
313	Clinical and molecular phenotype of Aicardi-Goutieres syndrome. *American Journal of Human Genetics*, **2007**, 81, 713-25	11	310
312	Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. *Nature Genetics*, **2013**, 45, 639-47	36.3	309
311	Mutations in LRP2, which encodes the multiligand receptor megalin, cause Donnai-Barrow and facio-oculo-acoustico-renal syndromes. *Nature Genetics*, **2007**, 39, 957-9	36.3	230
310	BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. *Nature Genetics*, **2006**, 38, 521-4	36.3	214
309	CHARGE syndrome: report of 47 cases and review. *American Journal of Medical Genetics Part A*, **1998**, 76, 402-9		191
308	Genotype-phenotype relationships in Berardinelli-Seip congenital lipodystrophy. *Journal of Medical Genetics*, **2002**, 39, 722-33	5.8	190
307	SLC26A4 gene is frequently involved in nonsyndromic hearing impairment with enlarged vestibular aqueduct in Caucasian populations. *European Journal of Human Genetics*, **2006**, 14, 773-9	5.3	183
306	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, 763-71	5.8	178
305	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
304	NF1 microdeletions in neurofibromatosis type 1: from genotype to phenotype. *Human Mutation*, **2010**, 31, E1506-18	4.7	176
303	Genotype-phenotype correlation in Costello syndrome: HRAS mutation analysis in 43 cases. *Journal of Medical Genetics*, **2006**, 43, 401-5	5.8	174

302	Mutations in PHF8 are associated with X linked mental retardation and cleft lip/cleft palate. *Journal of Medical Genetics*, **2005**, 42, 780-6	5.8	165
301	Five new TTF1/NKX2.1 mutations in brain-lung-thyroid syndrome: rescue by PAX8 synergism in one case. *Human Molecular Genetics*, **2009**, 18, 2266-76	5.6	162
300	Germline mutations in WTX cause a sclerosing skeletal dysplasia but do not predispose to tumorigenesis. *Nature Genetics*, **2009**, 41, 95-100	36.3	161
299	Comparison of clinical presentations and outcomes between patients with TGFBR2 and FBN1 mutations in Marfan syndrome and related disorders. *Circulation*, **2009**, 120, 2541-9	16.7	160
298	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
297	Heterozygous endothelin receptor B (EDNRB) mutations in isolated Hirschsprung disease. *Human Molecular Genetics*, **1996**, 5, 355-7	5.6	138
296	Mutations in SRCAP, encoding SNF2-related CREBBP activator protein, cause Floating-Harbor syndrome. *American Journal of Human Genetics*, **2012**, 90, 308-13	11	130
295	Involvement of hyperprolinemia in cognitive and psychiatric features of the 22q11 deletion syndrome. *Human Molecular Genetics*, **2007**, 16, 83-91	5.6	129
294	Thrombocytopenia resulting from mutations in filamin A can be expressed as an isolated syndrome. *Blood*, **2011**, 118, 5928-37	2.2	128
293	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. *JAMA Psychiatry*, **2016**, 73, 20-30	14.5	120
292	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, 1229-35	5.6	116
291	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, 444-53	5.3	111
290	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, 1859-66	5.6	105
289	COL4A1 mutation in Axenfeld-Rieger anomaly with leukoencephalopathy and stroke. *Annals of Neurology*, **2007**, 62, 177-84	9.4	104
288	Schizencephaly: clinical and imaging features in 30 infantile cases. *Brain and Development*, **2000**, 22, 475-83	2.1	101
287	Large-scale deletions and SMADIP1 truncating mutations in syndromic Hirschsprung disease with involvement of midline structures. *American Journal of Human Genetics*, **2001**, 69, 1370-7	11	98
286	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
285	Spectrum of NSD1 mutations in Sotos and Weaver syndromes. *Journal of Medical Genetics*, **2003**, 40, 436-40	5.8	92

284	Definition of a critical region on chromosome 18 for congenital aural atresia by arrayCGH. *American Journal of Human Genetics*, **2003**, 72, 1578-84	11	92
283	Mutations in RNF135, a gene within the NF1 microdeletion region, cause phenotypic abnormalities including overgrowth. *Nature Genetics*, **2007**, 39, 963-5	36.3	90
282	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
281	Molecular analysis of the CBP gene in 60 patients with Rubinstein-Taybi syndrome. *Journal of Medical Genetics*, **2002**, 39, 415-21	5.8	87
280	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
279	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
278	Baraitser-Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. *European Journal of Human Genetics*, **2015**, 23, 292-301	5.3	83
277	Spectrum of PTCH1 mutations in French patients with Gorlin syndrome. *Journal of Investigative Dermatology*, **2003**, 121, 478-81	4.3	79
276	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
275	Complete exon sequencing of all known Usher syndrome genes greatly improves molecular diagnosis. *Orphanet Journal of Rare Diseases*, **2011**, 6, 21	4.2	78
274	Screening of SLC26A4 (PDS) gene in Pendred's syndrome: a large spectrum of mutations in France and phenotypic heterogeneity. *Clinical Genetics*, **2004**, 66, 333-40	4	78
273	New insights into genotype-phenotype correlation for GLI3 mutations. *European Journal of Human Genetics*, **2015**, 23, 92-102	5.3	75
272	STXBP1-related encephalopathy presenting as infantile spasms and generalized tremor in three patients. *Epilepsia*, **2011**, 52, 1820-7	6.4	75
271	EFTUD2 haploinsufficiency leads to syndromic oesophageal atresia. *Journal of Medical Genetics*, **2012**, 49, 737-46	5.8	75
270	Molecular findings and clinical data in a cohort of 150 patients with anophthalmia/microphthalmia. *Clinical Genetics*, **2014**, 86, 326-34	4	74
269	Lymphedema-lymphangiectasia-mental retardation (Hennekam) syndrome: a review. *American Journal of Medical Genetics Part A*, **2002**, 112, 412-21		74
268	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. *Journal of Clinical Investigation*, **2017**, 127, 1991-2006	15.9	73
267	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

266	Prevalence of mutations in AGPAT2 among human lipodystrophies. *Diabetes*, **2003**, 52, 1573-8	0.9	71
265	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, 1145-55	5.3	70
264	Molecular diagnosis of oculocutaneous albinism: new mutations in the OCA1-4 genes and practical aspects. *Pigment Cell and Melanoma Research*, **2008**, 21, 583-7	4.5	69
263	Hydrometrocolpos and polydactyly: a common neonatal presentation of Bardet-Biedl and McKusick-Kaufman syndromes. *Journal of Medical Genetics*, **1999**, 36, 599-603	5.8	69
262	REEP1 mutations in SPG31: frequency, mutational spectrum, and potential association with mitochondrial morpho-functional dysfunction. *Human Mutation*, **2011**, 32, 1118-27	4.7	68
261	Identification of 23 TGFBR2 and 6 TGFBR1 gene 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, E284-95	4.7	68
260	LRP5 mutations in osteoporosis-pseudoglioma syndrome and high-bone-mass disorders. *Joint Bone Spine*, **2005**, 72, 207-14	2.9	68
259	Further delineation of Kabuki syndrome in 48 well-defined new individuals **2005**, 132A, 265-72		68
258	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
257	Twelve new patients with 13q deletion syndrome: genotype-phenotype analyses in progress. *European Journal of Medical Genetics*, **2009**, 52, 41-6	2.6	65
256	Mutations in SETD2 cause a novel overgrowth condition. *Journal of Medical Genetics*, **2014**, 51, 512-7	5.8	64
255	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
254	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**, 140, 1285-96	2.5	64
253	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
252	Factor V Leiden, prothrombin 20210A, methylenetetrahydrofolate reductase 677T, and population genetics. *Molecular Genetics and Metabolism*, **2005**, 86, 91-9	3.7	62
251	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
250	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
249	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

248	Testing for triallelism: analysis of six BBS genes in a Bardet-Biedl syndrome family cohort. *European Journal of Human Genetics*, **2005**, 13, 607-16	5.3	60
247	C5orf42 is the major gene responsible for OFD syndrome type VI. *Human Genetics*, **2014**, 133, 367-77	6.3	59
246	Further delineation of the congenital form of X-linked dyskeratosis congenita (Hoyeraal-Hreidarsson syndrome). *European Journal of Pediatrics*, **2003**, 162, 863-7	4.1	57
245	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
244	Molecular characterization of a series of 990 index patients with albinism. *Pigment Cell and Melanoma Research*, **2018**, 31, 466-474	4.5	55
243	Metabolic correction induced by leptin replacement treatment in young children with Berardinelli-Seip congenital lipoatrophy. *Pediatrics*, **2007**, 120, e291-6	7.4	55
242	Progressive bulbospinal amyotrophy in triple A syndrome with AAAS gene mutation. *Neurology*, **2002**, 58, 962-5	6.5	55
241	The gene for Bazex-Dupr□Christol syndrome maps to chromosome Xq. *Journal of Investigative Dermatology*, **1995**, 105, 87-91	4.3	54
240	Mutations in □ecretase subunit-encoding PSENEN underlie Dowling-Degos disease associated with acne inversa. *Journal of Clinical Investigation*, **2017**, 127, 1485-1490	15.9	53
239	Townes-Brocks syndrome: detection of a SALL1 mutation hot spot and evidence for a position effect in one patient. *Human Mutation*, **1999**, 14, 377-86	4.7	53
238	Confirmation of assignment of a locus for Rubinstein-Taybi syndrome gene to 16p13.3. *American Journal of Medical Genetics Part A*, **1992**, 44, 126-8		52
237	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
236	Heterogeneity of NSD1 alterations in 116 patients with Sotos syndrome. *Human Mutation*, **2007**, 28, 1098-107	4.7	50
235	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
234	Spectrum of epilepsy in terminal 1p36 deletion syndrome. *Epilepsia*, **2008**, 49, 509-15	6.4	49
233	AGC1/2, the mitochondrial aspartate-glutamate carriers. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2016**, 1863, 2394-412	4.9	49
232	Case with autistic syndrome and chromosome 22q13.3 deletion detected by FISH. *American Journal of Medical Genetics Part A*, **2000**, 96, 839-844		48
231	Ondine-Hirschsprung syndrome (Haddad syndrome). Further delineation in two cases and review of the literature. *European Journal of Pediatrics*, **1993**, 152, 75-7	4.1	48

230	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
229	High glucose repatterns human podocyte energy metabolism during differentiation and diabetic nephropathy. *FASEB Journal*, **2017**, 31, 294-307	0.9	47
228	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
227	Prenatal diagnosis of female monozygotic twins discordant for Turner syndrome: implications for prenatal genetic counselling. *Prenatal Diagnosis*, **2002**, 22, 697-702	3.2	47
226	Identification of novel mutations confirms PDE4D as a major gene causing acrodysostosis. *Human Mutation*, **2013**, 34, 97-102	4.7	46
225	Metabolic Reprogramming in Amyotrophic Lateral Sclerosis. *Scientific Reports*, **2018**, 8, 3953	4.9	45
224	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
223	New practical definitions for the diagnosis of autosomal recessive spastic ataxia of Charlevoix-Saguenay. *Annals of Neurology*, **2015**, 78, 871-86	9.4	45
222	New clinico-genetic classification of trichothiodystrophy. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2020-30	2.5	44
221	Mutational spectrum of COH1 and clinical heterogeneity in Cohen syndrome. *Journal of Medical Genetics*, **2006**, 43, e22	5.8	44
220	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
219	Costello syndrome and neurological abnormalities. *American Journal of Medical Genetics Part A*, **2003**, 123A, 301-5		42
218	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
217	Four-year follow-up of diagnostic service in USH1 patients **2011**, 52, 4063-71		41
216	Consensus recommendations for diagnosis, management and treatment of Fabry disease in paediatric patients. *Clinical Genetics*, **2019**, 96, 107-117	4	39
215	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
214	Molecular characterisation of patients with subtelomeric 22q abnormalities using chromosome specific array-based comparative genomic hybridisation. *European Journal of Human Genetics*, **2005**, 13, 1019-24	5.3	39
213	Delineation of EFTUD2 haploinsufficiency-related phenotypes through a series of 36 patients. *Human Mutation*, **2014**, 35, 478-85	4.7	38

212	Neurologic aspects of MECP2 gene duplication in male patients. *Pediatric Neurology*, **2009**, 41, 187-91	2.9	38
211	The phenotype of Floating-Harbor syndrome in 10 patients. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 821-9	2.5	38
210	Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. *Genetics in Medicine*, **2018**, 20, 965-975	8.1	37
209	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
208	Resistance to leptin-replacement therapy in Berardinelli-Seip congenital lipodystrophy: an immunological origin. *European Journal of Endocrinology*, **2010**, 162, 1083-91	6.5	36
207	Analysis of CBP (CREBBP) gene deletions in Rubinstein-Taybi syndrome patients using real-time quantitative PCR. *Human Mutation*, **2004**, 23, 278-84	4.7	36
206	and screen in the Sotos-like syndrome French cohort. *Journal of Medical Genetics*, **2016**, 53, 743-751	5.8	36
205	Spectrum of CREBBP gene dosage anomalies in Rubinstein-Taybi syndrome patients. *European Journal of Human Genetics*, **2007**, 15, 843-7	5.3	35
204	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, 1394-8	5.5	34
203	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
202	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
201	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
200	Socio-behavioral characteristics of children with Rubinstein-Taybi syndrome. *Journal of Autism and Developmental Disorders*, **2009**, 39, 1252-60	4.6	33
199	Submicroscopic deletion of chromosome 16p13.3 in patients with Rubinstein-Taybi syndrome. *American Journal of Medical Genetics Part A*, **1998**, 78, 267-270		33
198	Poikiloderma with neutropenia, Clericuzio type, in a family from Morocco. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2762-9	2.5	33
197	Screening of ARX in mental retardation families: Consequences for the strategy of molecular diagnosis. *Neurogenetics*, **2006**, 7, 39-46	3	33
196	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. *Npj Genomic Medicine*, **2017**, 2, 32	6.2	32
195	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		32

194	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
193	Reduced placental telomere length during pregnancies complicated by intrauterine growth restriction. *PLoS ONE*, **2013**, 8, e54013	3.7	31
192	Leber's optic neuropathy associated with disseminated white matter disease: a case report and review. *Clinical Neurology and Neurosurgery*, **2009**, 111, 83-6	2	31
191	Mutations in SNRPB, encoding components of the core splicing machinery, cause cerebro-costo-mandibular syndrome. *Human Mutation*, **2015**, 36, 187-90	4.7	30
190	Bardet-biedl syndrome and brain abnormalities. *Neuropediatrics*, **2007**, 38, 5-9	1.6	30
189	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
188	SLC24A5 mutations are associated with non-syndromic oculocutaneous albinism. *Journal of Investigative Dermatology*, **2014**, 134, 568-571	4.3	29
187	2.3 Mb terminal deletion in 12p13.33 associated with oculoauriculovertebral spectrum and evaluation of WNT5B as a candidate gene. *European Journal of Medical Genetics*, **2009**, 52, 446-9	2.6	29
186	Mutations in the endothelin receptor type A cause mandibulofacial dysostosis with alopecia. *American Journal of Human Genetics*, **2015**, 96, 519-31	11	28
185	Neuroimaging findings in Mowat-Wilson syndrome: a study of 54 patients. *Genetics in Medicine*, **2017**, 19, 691-700	8.1	28
184	Osteoporosis in late-diagnosed adult homocystinuric patients. *Journal of Inherited Metabolic Disease*, **2000**, 23, 338-40	5.4	28
183	Congenital hypotrichosis and milia: report of a large family suggesting X-linked dominant inheritance. *American Journal of Medical Genetics Part A*, **1994**, 52, 487-90		28
182	CREBBP mutations in individuals without Rubinstein-Taybi syndrome phenotype. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2681-93	2.5	28
181	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
180	Prenatal diagnosis using array-CGH: a French experience. *European Journal of Medical Genetics*, **2013**, 56, 341-5	2.6	27
179	The male phenotype in osteopathia striata congenita with cranial sclerosis. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2397-408	2.5	27
178	A second locus for Aicardi-Goutieres syndrome at chromosome 13q14-21. *Journal of Medical Genetics*, **2006**, 43, 444-50	5.8	27
177	Costello syndrome: report of six patients including one with an embryonal rhabdomyosarcoma. *European Journal of Pediatrics*, **2000**, 159, 139-42	4.1	27

176	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, 549-53	5.8	26
175	A point mutation in the XNP gene, associated with an ATR-X phenotype without alpha-thalassemia. *European Journal of Human Genetics*, **1996**, 4, 316-20	5.3	26
174	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
173	BLOC1S5 pathogenic variants cause a new type of Hermansky-Pudlak syndrome. *Genetics in Medicine*, **2020**, 22, 1613-1622	8.1	25
172	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
171	Growth patterns of patients with Noonan syndrome: correlation with age and genotype. *European Journal of Endocrinology*, **2016**, 174, 641-50	6.5	25
170	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
169	Combination of WAGR and Potocki-Shaffer contiguous deletion syndromes in a patient with an 11p11.2-p14 deletion. *European Journal of Human Genetics*, **2005**, 13, 409-13	5.3	25
168	Floating-Harbor syndrome: description of a further patient, review of the literature, and suggestion of autosomal dominant inheritance. *European Journal of Pediatrics*, **1995**, 154, 658-61	4.1	25
167	Toriello-Carey syndrome: evidence for X-linked inheritance. *American Journal of Medical Genetics Part A*, **1996**, 65, 291-4		25
166	Perrault syndrome: report of four new cases, review and exclusion of candidate genes. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 661-4	2.5	24
165	Prenatal diagnosis of hypochondroplasia: three-dimensional multislice computed tomography findings and molecular analysis. *Fetal Diagnosis and Therapy*, **2006**, 21, 18-21	2.4	24
164	Donnai-Barrow syndrome: four additional patients. *American Journal of Medical Genetics Part A*, **2003**, 121A, 258-62		24
163	Familial occurrence of hereditary renal adysplasia with m□lerian anomalies. *Clinical Genetics*, **1993**, 43, 23-4	4	24
162	Functional validation of ABHD12 mutations in the neurodegenerative disease PHARC. *Neurobiology of Disease*, **2017**, 98, 36-51	7.5	23
161	Clinical and genetic heterogeneity in familial steroid-sensitive nephrotic syndrome. *Pediatric Nephrology*, **2018**, 33, 473-483	3.2	22
160	Cutaneous manifestations in Costello and cardiofaciocutaneous syndrome: report of 18 cases and literature review. *Pediatric Dermatology*, **2013**, 30, 665-73	1.9	22
159	Oro-dental features as useful diagnostic tool in Rubinstein-Taybi syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 570-3	2.5	22

158	The diagnosis of Costello syndrome: nomenclature in Ras/MAPK pathway disorders. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1218-20	2.5	22
157	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
156	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
155	Non-specific gastrointestinal features: Could it be Fabry disease?. *Digestive and Liver Disease*, **2018**, 50, 429-437	3.3	21
154	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
153	Behavioral and temperamental features of children with Costello syndrome. *American Journal of Medical Genetics, Part A*, **2006**, 140, 968-74	2.5	21
152	Medulloblastoma in the nevoid basal-cell carcinoma syndrome: case reports and review of the literature. *Genetic Counseling*, **1990**, 1, 273-7		21
151	IL11RA-related Crouzon-like autosomal recessive craniosynostosis in 10 new patients: Resemblances and differences. *Clinical Genetics*, **2018**, 94, 373-380	4	21
150	Molecular, clinical and neuropsychological study in 31 patients with Kabuki syndrome and KMT2D mutations. *Clinical Genetics*, **2017**, 92, 298-305	4	20
149	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
148	Systematic molecular and cytogenetic screening of 100 patients with marfanoid syndromes and intellectual disability. *Clinical Genetics*, **2013**, 84, 507-21	4	20
147	Rubinstein-Taybi syndrome (CREBBP, EP300). *European Journal of Human Genetics*, **2011**, 19, preceeding 118-20	5.3	20
146	Limb body wall complex and amniotic band sequence in sibs. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2682-7	2.5	20
145	Craniofacial dysmorphology and three-dimensional ultrasound: a prospective study on practicability for prenatal diagnosis. *Prenatal Diagnosis*, **2003**, 23, 810-8	3.2	20
144	No evidence for linkage to the type 1 or type 2 neurofibromatosis loci in Noonan syndrome families. *American Journal of Medical Genetics Part A*, **1993**, 46, 700-5		20
143	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
142	Oculo-dento-digital dysplasia: lack of genotype-phenotype correlation for GJA1 mutations and usefulness of neuro-imaging. *European Journal of Medical Genetics*, **2010**, 53, 19-22	2.6	19
141	Severe linear form of granuloma annulare along Blaschko's lines preceding the onset of a classical form of granuloma annulare in a child. *British Journal of Dermatology*, **2007**, 157, 1056-8	4	19

140	New case of Toriello-Carey syndrome. *American Journal of Medical Genetics Part A*, **1992**, 42, 374-6		19
139	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
138	Mitochondrial functions and rare diseases. *Molecular Aspects of Medicine*, **2020**, 71, 100842	16.7	18
137	Oral manifestations of patients with Kenny-Caffey Syndrome. *European Journal of Medical Genetics*, **2012**, 55, 441-5	2.6	18
136	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
135	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
134	Axenfeld-Rieger anomaly: a novel mutation in the forkhead box C1 (FOXC1) gene in a 4-generation family. *JAMA Ophthalmology*, **2004**, 122, 1527-33		18
133	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
132	Further delineation of the phenotype caused by biallelic variants in the WDR4 gene. *Clinical Genetics*, **2018**, 93, 374-377	4	17
131	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
130	Atypical male and female presentations of FLNA-related periventricular nodular heterotopia. *European Journal of Medical Genetics*, **2012**, 55, 313-8	2.6	17
129	Mosaic maternal uniparental isodisomy for chromosome 7q21-qter. *Clinical Genetics*, **2006**, 70, 207-13	4	17
128	A 22-year French experience with solid tumors in children with Down syndrome. *Pediatric Hematology and Oncology*, **2003**, 20, 517-29	1.7	17
127	Menkes disease: study of the mitochondrial respiratory chain in three cases. *European Journal of Paediatric Neurology*, **1999**, 3, 167-70	3.8	17
126	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
125	Oculo-ectodermal syndrome: a new tumour predisposition syndrome. *Clinical Dysmorphology*, **2004**, 13, 81-83	0.9	16
124	GDNF as a candidate modifier in a type 1 neurofibromatosis (NF1) enteric phenotype. *Journal of Medical Genetics*, **2001**, 38, 638-43	5.8	16
123	Clinical identification of a human equivalent to the short ear (se) murine phenotype. *Annales De Génétique*, **1994**, 37, 184-91		16

122	ARID1B mutations are the major genetic cause of corpus callosum anomalies in patients with intellectual disability. *Brain*, **2016**, 139, e64	11.2	16
121	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
120	Clinical and molecular spectrum of renal malformations in Kabuki syndrome. *Journal of Pediatrics*, **2013**, 163, 742-6	3.6	15
119	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
118	Association of external auditory canal atresia, vertical talus, and hypertelorism: confirmation of Rasmussen syndrome. *American Journal of Medical Genetics Part A*, **2002**, 110, 179-81		15
117	Fetal phenotypes in otopalatodigital spectrum disorders. *Clinical Genetics*, **2016**, 89, 371-7	4	15
116	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
115	Detailed clinical, genetic and neuroimaging characterization of OFD VI syndrome. *European Journal of Medical Genetics*, **2013**, 56, 301-8	2.6	14
114	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
113	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
112	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
111	Contribution of PTEN large rearrangements in Cowden disease: a multiplex amplifiable probe hybridisation (MAPH) screening approach. *Journal of Medical Genetics*, **2008**, 45, 657-65	5.8	14
110	The Basques: review of population genetics and Mendelian disorders. *Human Biology*, **2005**, 77, 619-37	1.2	14
109	Leucodystrophy and oculocutaneous albinism in a child with an 11q14 deletion. *Journal of Medical Genetics*, **2001**, 38, 35-8	5.8	14
108	Isolated macrodactyly and Proteus syndrome. *Clinical Dysmorphology*, **1996**, 5, 255???258	0.9	14
107	Split hand/split foot deformity and LADD syndrome in a family: overlap between the EEC and LADD syndromes. *Journal of Medical Genetics*, **1993**, 30, 700-3	5.8	14
106	Otopalatodigital spectrum disorders: refinement of the phenotypic and mutational spectrum. *Journal of Human Genetics*, **2016**, 61, 693-9	4.3	14
105	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

104	Identification of a homozygous deletion mutation in C16orf57 in a family with Clericuzio-type poikiloderma with neutropenia. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1347-8	2.5	13
103	Overlap between the Bazex syndrome and congenital hypotrichosis and milia. *American Journal of Medical Genetics Part A*, **1995**, 56, 423-4		13
102	Phenotypic variability in van der Woude syndrome. *Genetic Counseling*, **1995**, 6, 221-6		13
101	CYP2U1 activity is altered by missense mutations in hereditary spastic paraplegia 56. *Human Mutation*, **2018**, 39, 140-151	4.7	12
100	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
99	Germline mosaicism in Rubinstein-Taybi syndrome. *Gene*, **2013**, 518, 476-8	3.8	12
98	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
97	Molecular characterization of an 11q14.3 microdeletion associated with leukodystrophy. *European Journal of Human Genetics*, **2004**, 12, 245-50	5.3	12
96	Statistical analysis of mitochondrial pathologies in childhood: identification of deficiencies using principal component analysis. *Laboratory Investigation*, **2000**, 80, 1019-30	5.9	12
95	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
94	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. *Genetics in Medicine*, **2020**, 22, 181-188	8.1	12
93	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
92	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
91	Keratoconus associated with Williams-Beuren syndrome: first case reports. *Ophthalmic Genetics*, **2010**, 31, 252-6	1.2	11
90	Eight previously unidentified mutations found in the OA1 ocular albinism gene. *BMC Medical Genetics*, **2006**, 7, 41	2.1	11
89	Azoospermia as a new feature of Fabry disease. *Fertility and Sterility*, **2007**, 88, 212.e15-8	4.8	10
88	Novel KDM5B splice variants identified in patients with developmental disorders: Functional consequences. *Gene*, **2018**, 679, 305-313	3.8	10
87	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

86	Finger creases lend a hand in Kabuki syndrome. *European Journal of Medical Genetics*, **2013**, 56, 556-60	2.6	9
85	Genetic basis of oculocutaneous albinism. *Expert Review of Dermatology*, **2009**, 4, 611-622		9
84	Evaluation of a mutation screening strategy for sporadic cases of ATR-X syndrome. *Journal of Medical Genetics*, **1999**, 36, 183-6	5.8	9
83	Dermatological manifestations in cardiofaciocutaneous syndrome: a prospective multicentric study of 45 mutation-positive patients. *British Journal of Dermatology*, **2019**, 180, 172-180	4	8
82	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
81	A novel mutation in the fatty acid transport protein 4 gene in a patient initially described as affected by self-healing congenital verruciform hyperkeratosis. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2664-5	2.5	8
80	Fetal presentation of PHACES syndrome. *American Journal of Medical Genetics, Part A*, **2005**, 132A, 110	2.5	8
79	Functional and genetic analyses of ZYG11B provide evidences for its involvement in OAVS. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1375	2.3	8
78	Dopachrome tautomerase variants in patients with oculocutaneous albinism. *Genetics in Medicine*, **2021**, 23, 479-487	8.1	8
77	Evaluation of Motor Skills in Children with Rubinstein-Taybi Syndrome. *Journal of Autism and Developmental Disorders*, **2017**, 47, 3321-3332	4.6	7
76	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
75	Detection of an intragenic deletion expands the spectrum of CTSC mutations in Papillon-Lef□re syndrome. *Journal of Investigative Dermatology*, **2008**, 128, 322-5	4.3	7
74	A patient with hydranencephaly and PEHO-like dysmorphic features. *Annales De G□□ique*, **2003**, 46, 25-8		7
73	Prenatal retinoic acid exposure reveals candidate genes for craniofacial disorders. *Scientific Reports* , **2018**, 8, 17492	4.9	7
72	Interest of chromosomal microarray analysis in the prenatal diagnosis of fetal intrauterine growth restriction. *Prenatal Diagnosis*, **2018**, 38, 1111-1119	3.2	7
71	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
70	Developmental trajectories of neuroanatomical alterations associated with the 16p11.2 Copy Number Variations. *NeuroImage*, **2019**, 203, 116155	7.9	6
69	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

68	Dysmorphic features in subtelomeric 20p13 deletion excluding JAG1: a recognizable microdeletion phenotype?. *European Journal of Medical Genetics*, **2012**, 55, 151-5	2.6	6
67	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
66	Characterization of a de novo balanced translocation t(9;18)(p23;q12.2) in a patient with oculoauriculovertebral spectrum. *European Journal of Medical Genetics*, **2010**, 53, 104-7	2.6	6
65	X-linked dominant chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia. *American Journal of Medical Genetics, Part A*, **2005**, 136A, 307-12	2.5	6
64	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
63	Abdominal lymphatic dysplasia and 22q11 microdeletion. *Genetic Counseling*, **1999**, 10, 67-70		6
62	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
61	SCA27 is a cause of early-onset ataxia and developmental delay. *European Journal of Paediatric Neurology*, **2015**, 19, 271-3	3.8	5
60	Rubinstein-Taybi syndrome and Hirschsprung disease in a patient harboring an intragenic deletion of the CREBBP gene. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1847-8	2.5	5
59	Albinisme oculo-cutan. *Annales De Dermatologie Et De Venereologie*, **2007**, 134, 55-64	0.3	5
58	Fabry disease prenatal diagnosis. *Prenatal Diagnosis*, **2008**, 28, 268	3.2	5
57	Neonatal Proteus syndrome?. *American Journal of Medical Genetics Part A*, **2002**, 112, 228-30		5
56	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
55	Fetal phenotype of Rubinstein-Taybi syndrome caused by CREBBP mutations. *Clinical Genetics*, **2019**, 95, 420-426	4	4
54	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
53	Hallerman-Streiff-like syndrome presenting with laterality and cardiac defects. *Clinical Dysmorphology*, **2009**, 18, 116-119	0.9	4
52	A 580 kb microdeletion in 17q21.32 associated with mental retardation, microcephaly, cleft palate, and cardiac malformation. *European Journal of Medical Genetics*, **2008**, 51, 74-80	2.6	4
51	Testing and improving experimental parameters for the use of low molecular weight targets in array-CGH experiments. *Human Mutation*, **2006**, 27, 1143-50	4.7	4

50	SMAD4 germinal mosaicism in a family with juvenile polyposis and hypertrophic osteoarthropathy. *Journal of Pediatric Gastroenterology and Nutrition*, **2005**, 41, 117-20	2.8	4
49	W syndrome: Report of three cases and review. *American Journal of Medical Genetics Part A*, **1999**, 87, 446-449		4
48	Gonadal function in Smith-Lemli-Opitz syndrome. *American Journal of Medical Genetics Part A*, **1993**, 45, 119		4
47	Growth charts in Kabuki syndrome 1. *American Journal of Medical Genetics, Part A*, **2020**, 182, 446-453	2.5	4
46	Rubinstein-Taybi Syndrome: A Model of Epigenetic Disorder. *Genes*, **2021**, 12,	4.2	4
45	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
44	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
43	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
42	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
41	Cutaneous epidermal cysts as a presentation of gorlin syndrome. *Archives of Dermatology*, **2009**, 145, 1341-3		3
40	Brachydactyly type A1 with short humerus and associated skeletal features. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 3016-21	2.5	3
39	A review of malignancies in fragile X syndrome and report of an Ewing sarcoma. *International Journal on Disability and Human Development*, **2008**, 7,		3
38	Self-healing congenital verruciform hyperkeratosis **2004**, 130A, 303-6		3
37	Progressive bulbospinal amyotrophy in triple A syndrome with AAAS gene mutation. *Neurology*, **2002**, 59, 1823; author reply 1823	6.5	3
36	Germinal mosaicism in oculo-auriculo-vertebral dysplasia?. *American Journal of Medical Genetics Part A*, **1993**, 46, 609-10		3
35	Effects of eight neuropsychiatric copy number variants on human brain structure. *Translational Psychiatry*, **2021**, 11, 399	8.6	3
34	Typical facial gestalt in X-linked Kabuki syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3363-3364	2.5	3
33	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

32	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
31	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
30	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
29	Rare genetic diseases, signalling pathways, and keloid scar formation. *British Journal of Dermatology*, **2014**, 171, 452-3	4	2
28	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
27	A survey of ocular tumors in Down syndrome alone or associated with another genetic affection. *International Journal on Disability and Human Development*, **2006**, 5,		2
26	LRP5 : le gène mutédans le syndrome d'ostéoporose avec pseudogliome et le phénotype de masse osseuse élevé. *Revue Du Rhumatisme (Edition Francaise)*, **2005**, 72, 388-396	0.1	2
25	Transcription factors in dysmorphology. *Clinical Genetics*, **1999**, 55, 137-43	4	2
24	Maladies des gènes du développement.. *Medecine/Sciences*, **2000**, 16, 354		2
23	CHN1 and duane retraction syndrome: Expanding the phenotype to cranial nerves development disease. *European Journal of Medical Genetics*, **2021**, 64, 104188	2.6	2
22	CHEDDA syndrome is an underrecognized neurodevelopmental disorder with a highly restricted ATN1 mutation spectrum. *Clinical Genetics*, **2021**, 100, 468-477	4	2
21	A recurrent missense variant in EYA3 gene is associated with oculo-auriculo-vertebral spectrum. *Human Genetics*, **2021**, 140, 933-944	6.3	2
20	HRAS germline mutations impair LKB1/AMPK signaling and mitochondrial homeostasis in Costello syndrome models.. *Journal of Clinical Investigation*, **2022**,	15.9	2
19	Deletion of the transcription factor SOX4 is implicated in syndromic nephroblastoma. *Clinical Genetics*, **2017**, 92, 449-450	4	1
18	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
17	Refined localisation of the voltage-gated chloride channel, CLCN3, to 4q33. *Human Genetics*, **1998**, 102, 178-81	6.3	1
16	A new case of VACTERL association with unilateral amelia of upper limb. *Clinical Dysmorphology*, **2007**, 16, 185-187	0.9	1
15	Proteomic Study of Low-Birth-Weight Nephropathy in Rats. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1

14	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
13	Clinical dysmorphology beyond developmental genetics: recent advances in some human developmental genes. *Annales De Génétique*, **1995**, 38, 137-44		0
12	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
11	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		
10	Neurofibromatosis type 1 with undescribed osseous abnormalities: new features?. *Clinical Dysmorphology*, **2009**, 18, 92-94	0.9	
9	Genetic osteoarticular diseases under the molecular biology spotlight. *Joint Bone Spine*, **2004**, 71, 486-92	2.9	
8	Les pathologies génétiques ostéoarticulaires à l'ère de la biologie moléculaire. *Revue Du Rhumatisme (Edition Francaise)*, **2004**, 71, 865-871	0.1	
7	Phénotypes psycho-comportementaux de l'enfant et de l'adolescent dans les syndromes microdélétionnels. *Annales Medico-Psychologiques*, **2003**, 161, 54-58	0.2	
6	Apport des puces à ADN et nouveaux syndromes microdélétionnels. *Bulletin De L'Academie Nationale De Medecine*, **2018**, 202, 693-705	0.1	
5	Maladies des gènes du développement codant pour des facteurs de transcription. *Bulletin De L'Academie Nationale De Medecine*, **2009**, 193, 931-945	0.1	
4	Cornea verticillata and acroparesthesia efficiently discriminate clusters of severity in Fabry disease **2020**, 15, e0233460		
3	Cornea verticillata and acroparesthesia efficiently discriminate clusters of severity in Fabry disease **2020**, 15, e0233460		
2	Cornea verticillata and acroparesthesia efficiently discriminate clusters of severity in Fabry disease **2020**, 15, e0233460		
1	Cornea verticillata and acroparesthesia efficiently discriminate clusters of severity in Fabry disease **2020**, 15, e0233460		