Damien Sanlaville

List of Publications by Year
in descending order

171	7,995	57758	60623
papers	citations	44 h-index	81 g-index

180	180	180	13465
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	3.5	501
2	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	27.8	394
3	GRIN2A mutations in acquired epileptic aphasia and related childhood focal epilepsies and encephalopathies with speech and language dysfunction. Nature Genetics, 2013, 45, 1061-1066.	21.4	380
4	Selective predisposition to bacterial infections in IRAK-4â€“deficient children: IRAK-4â€“dependent TLRs are otherwise redundant in protective immunity. Journal of Experimental Medicine, 2007, 204, 2407-2422.	8.5	374
5	CHARGE syndrome: an update. European Journal of Human Genetics, 2007, 15, 389-399.	2.8	280
6	Association of TALS Developmental Disorder with Defect in Minor Splicing Component U4atac snRNA. Science, 2011, 332, 240-243.	12.6	195
7	Disruption of ROBO2 Is Associated with Urinary Tract Anomalies and Confers Risk of Vesicoureteral Reflux. American Journal of Human Genetics, 2007, 80, 616-632.	6.2	189
8	Submicroscopic Duplications of the Hydroxysteroid Dehydrogenase HSD17B10 and the E3 Ubiquitin Ligase HUWE1 Are Associated with Mental Retardation. American Journal of Human Genetics, 2008, 82, 432-443.	6.2	187
9	Benign hereditary chorea: phenotype, prognosis, therapeutic outcome and long term follow-up in a large series with new mutations in theTITF1/NKX2-1gene. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 956-962.	1.9	172
10	Disruption of a long distance regulatory region upstream of SOX9 in isolated disorders of sex development. Journal of Medical Genetics, 2011, 48, 825-830.	3.2	162
11	Epileptic encephalopathies of the Landauâ€Kleffner and continuous spike and waves during slowâ€wave sleep types: Genomic dissection makes the link with autism. Epilepsia, 2012, 53, 1526-1538.	5.1	148
12	Guidelines for molecular karyotyping in constitutional genetic diagnosis. European Journal of Human Genetics, 2007, 15, 1105-1114.	2.8	144
13	GRIN2A-related disorders: genotype and functional consequence predict phenotype. Brain, 2019, 142, 80-92.	7.6	143
14	Reinforcement of STAT3 activity reprogrammes human embryonic stem cells to naive-like pluripotency. Nature Communications, 2015, 6, 7095.	12.8	137
15	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
16	Genetic and neurodevelopmental spectrum ofSYNGAP1-associated intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 511-522.	3.2	135
17	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	2.5	134
18	Contiguous Gene Deletion within Chromosome Arm 10q Is Associated with Juvenile Polyposis of Infancy, Reflecting Cooperation between the BMPR1A and PTEN Tumor-Suppressor Genes. American Journal of Human Genetics, 2006, 78, 1066-1074.	6.2	127

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19	Mutations in SLC13A5 Cause Autosomal-Recessive Epileptic Encephalopathy with Seizure Onset in the First Days of Life. American Journal of Human Genetics, 2014, 95, 113-120.	6.2	112
20	Paradoxical NSD1 Mutations in Beckwith-Wiedemann Syndrome and 11p15 Anomalies in Sotos Syndrome. American Journal of Human Genetics, 2004, 74, 715-720.	6.2	110
21	Optical genome mapping enables constitutional chromosomal aberration detection. American Journal of Human Genetics, 2021, 108, 1409-1422.	6.2	108
22	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
23	Breakpoint mapping by next generation sequencing reveals causative gene disruption in patients carrying apparently balanced chromosome rearrangements with intellectual deficiency and/or congenital malformations. Journal of Medical Genetics, 2013, 50, 144-150.	3.2	99
24	Molecular and clinical characterization of 25 individuals with exonic deletions of NRXN1 and comprehensive review of the literature. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 388-403.	1.7	93
25	PRRT2 links infantile convulsions and paroxysmal dyskinesia with migraine. Neurology, 2012, 79, 2097-2103.	1.1	90
26	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	6.2	90
27	Cryptic genomic imbalances in de novo and inherited apparently balanced chromosomal rearrangements: Array CGH study of 47 unrelated cases. European Journal of Medical Genetics, 2009, 52, 291-296.	1.3	89
28	Functional variants of POC5 identified in patients with idiopathic scoliosis. Journal of Clinical Investigation, 2015, 125, 1124-1128.	8.2	87
29	Sex gap in aging and longevity: can sex chromosomes play a role?. Biology of Sex Differences, 2018, 9, 33.	4.1	82
30	Paternal deletion of the GNAS imprinted locus (including Gnasxl) in two girls presenting with severe pre- and post-natal growth retardation and intractable feeding difficulties. European Journal of Human Genetics, 2005, 13, 1033-1039.	2.8	80
31	Relapsing encephalopathy with cerebellar ataxia related to an <scp>ATP</scp>1A3 mutation. Developmental Medicine and Child Neurology, 2015, 57, 1183-1186.	2.1	78
32	Interstitial 9q22.3 microdeletion: clinical and molecular characterisation of a newly recognised overgrowth syndrome. European Journal of Human Genetics, 2006, 14, 759-767.	2.8	71
33	12p13.33 microdeletion including ELKS/ERC1, a new locus associated with childhood apraxia of speech. European Journal of Human Genetics, 2013, 21, 82-88.	2.8	70
34	Genome-wide arrays: Quality criteria and platforms to be used in routine diagnostics. Human Mutation, 2012, 33, 906-915.	2.5	69
35	A subset of genomic alterations detected in rolandic epilepsies contains candidate or known epilepsy genes including <scp>GRIN</scp>2A and <scp>PRRT</scp>2. Epilepsia, 2014, 55, 370-378.	5.1	69
36	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

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37	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	2.9	61
38	Distal Xq duplication and functional Xq disomy. Orphanet Journal of Rare Diseases, 2009, 4, 4.	2.7	58
39	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	3.8	58
40	Functional disomy of the Xq28 chromosome region. European Journal of Human Genetics, 2005, 13, 579-585.	2.8	57
41	The C20orf133 gene is disrupted in a patient with Kabuki syndrome. Journal of Medical Genetics, 2007, 44, 562-569.	3.2	56
42	Molecular characterization of a cohort of 73 patients with infantile spasms syndrome. European Journal of Medical Genetics, 2015, 58, 51-58.	1.3	56
43	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
44	A novel X-linked recessive form of Mendelian susceptibility to mycobaterial disease. Journal of Medical Genetics, 2006, 44, e65-e65.	3.2	52
45	Prenatal diagnosis of â€˜isolatedâ€™ Dandyâ€“Walker malformation: imaging findings and prenatal counselling. Prenatal Diagnosis, 2012, 32, 185-193.	2.3	52
46	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
47	A new intellectual disability syndrome caused by CTNNB1 haploinsufficiency. American Journal of Medical Genetics, Part A, 2014, 164, 1571-1575.	1.2	50
48	Clinical and molecular characterization of 17q21.31 microdeletion syndrome in 14 French patients with mental retardation. European Journal of Medical Genetics, 2011, 54, 144-151.	1.3	48
49	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogryposis. American Journal of Human Genetics, 2018, 102, 116-132.	6.2	46
50	Whole genome paired-end sequencing elucidates functional and phenotypic consequences of balanced chromosomal rearrangement in patients with developmental disorders. Journal of Medical Genetics, 2019, 56, 526-535.	3.2	46
51	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
52	Further delineation of the 17p13.3 microdeletion involving YWHAE but distal to PAFAH1B1: Four additional patients. European Journal of Medical Genetics, 2010, 53, 303-308.	1.3	44
53	Behavioral disturbance and treatment strategies in Smith-Magenis syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 111.	2.7	44
54	Guidelines for reporting secondary findings of genome sequencing in cancer genes: the SFMPP recommendations. European Journal of Human Genetics, 2018, 26, 1732-1742.	2.8	44

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55	16p13.11 microduplication in 45 new patients: refined clinical significance and genotypeâ€“phenotype correlations. Journal of Medical Genetics, 2020, 57, 301-307.	3.2	44
56	Heterozygous Bile Salt Export Pump Deficiency: A Possible Genetic Predisposition to Transient Neonatal Cholestasis. Journal of Pediatric Gastroenterology and Nutrition, 2006, 42, 114-116.	1.8	40
57	De novo Xq11.11 microdeletion including ARHGEF9 in a boy with mental retardation, epilepsy, macrosomia, and dysmorphic features. American Journal of Medical Genetics, Part A, 2011, 155, 1706-1711.	1.2	40
58	Clinical and molecular overlap in overgrowth syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 137C, 4-11.	1.6	39
59	Clinical and neurocognitive characterization of a family with a novel MED12 gene frameshift mutation. American Journal of Medical Genetics, Part A, 2013, 161, 3063-3071.	1.2	37
60	A new syndrome of intellectual disability with dysmorphism due to TBL1XR1 deletion. American Journal of Medical Genetics, Part A, 2015, 167, 164-168.	1.2	37
61	Xp22.3 interstitial deletion: A recognizable chromosomal abnormality encompassing VCX3A and STS genes in a patient with X-linked ichthyosis and mental retardation. Gene, 2013, 527, 578-583.	2.2	36
62	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. European Journal of Human Genetics, 2016, 24, 1761-1770.	2.8	36
63	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	7.6	35
64	A Distinct Class of Chromoanagenesis Events Characterized by Focal Copy Number Gains. Human Mutation, 2016, 37, 661-668.	2.5	30
65	Novel homozygous missense variant of GRIN1 in two sibs with intellectual disability and autistic features without epilepsy. European Journal of Human Genetics, 2017, 25, 376-380.	2.8	30
66	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. Genetics in Medicine, 2020, 22, 181-188.	2.4	30
67	Failure to detect an 8p22â€“8p23.1 duplication in patients with Kabuki (Niikawaâ€“Kuroki) syndrome. European Journal of Human Genetics, 2005, 13, 690-693.	2.8	28
68	Functional disomy of Xp including duplication ofDAX1gene with sex reversal due to t(X;Y)(p21.2;p11.3). , 2004, 128A, 325-330.		27
69	Clinical and Molecular Spectrum of Renal Malformations in Kabuki Syndrome. Journal of Pediatrics, 2013, 163, 742-746.	1.8	27
70	The role of CNVs in the etiology of rare autosomal recessive disorders: the example of TRAPPC9-associated intellectual disability. European Journal of Human Genetics, 2018, 26, 143-148.	2.8	26
71	Unusual Clinical Severity of Complement Membrane Cofactor Proteinâ€“Associated Hemolytic-Uremic Syndrome and Uniparental Isodisomy. American Journal of Kidney Diseases, 2007, 49, 323-329.	1.9	25
72	Fine Characterisation of a Recombination Hotspot at the DPY19L2 Locus and Resolution of the Paradoxical Excess of Duplications over Deletions in the General Population. PLoS Genetics, 2013, 9, e1003363.	3.5	25

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73	Refinement of genotypeâ€“phenotype correlation in 18 patients carrying a 1q24q25 deletion. American Journal of Medical Genetics, Part A, 2015, 167, 1008-1017.	1.2	25
74	Copy Number Variations Found in Patients with a Corpus Callosum Abnormality and Intellectual Disability. Journal of Pediatrics, 2017, 185, 160-166.e1.	1.8	25
75	Molecular karyotyping in human constitutional cytogenetics. European Journal of Medical Genetics, 2005, 48, 214-231.	1.3	24
76	Additive Effect of Variably Penetrant 22q11.2 Duplication and Pathogenic Mutations in Autism Spectrum Disorder: To Which Extent Does the Tree Hide the Forest?. Journal of Autism and Developmental Disorders, 2018, 48, 2886-2889.	2.7	24
77	Interstitial 12p13.1 deletion involving GRIN2B in three patients with intellectual disability. American Journal of Medical Genetics, Part A, 2013, 161, 2564-2569.	1.2	23
78	A systematic variant screening in familial cases of congenital heart defects demonstrates the usefulness of molecular genetics in this field. European Journal of Human Genetics, 2016, 24, 228-236.	2.8	23
79	Chromosome 21 Scan in Down Syndrome Reveals DSCAM as a Predisposing Locus in Hirschsprung Disease. PLoS ONE, 2013, 8, e62519.	2.5	22
80	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. Genetics in Medicine, 2019, 21, 1058-1064.	2.4	22
81	TWIST microdeletion identified by array CGH in a patient presenting Saethreâ€“Chotzen phenotype and a complex rearrangement involving chromosomes 2 and 7. European Journal of Medical Genetics, 2008, 51, 156-164.	1.3	21
82	17p13.1 microdeletion involving the TP53 gene in a boy presenting with mental retardation but no tumor. American Journal of Medical Genetics, Part A, 2010, 152A, 1278-1282.	1.2	20
83	Milk kinship is not an obstacle to using donor human milk to feed preterm infants in Muslim countries. Acta Paediatrica, International Journal of Paediatrics, 2016, 105, 462-467.	1.5	20
84	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	5.3	20
85	Chromosomal microarray analysis in fetuses with an isolated congenital heart defect: A retrospective, nationwide, multicenter study in France. Prenatal Diagnosis, 2019, 39, 464-470.	2.3	20
86	Chromosomal instability in the prediction of pituitary neuroendocrine tumors prognosis. Acta Neuropathologica Communications, 2020, 8, 190.	5.2	20
87	Comparison of two next-generation sequencing kits for diagnosis of epileptic disorders with a user-friendly tool for displaying gene coverage, DeCovA. Applied & Translational Genomics, 2015, 7, 19-25.	2.1	19
88	Sex chromosome aneuploidies and copy-number variants: a further explanation for neurodevelopmental prognosis variability?. European Journal of Human Genetics, 2017, 25, 930-934.	2.8	19
89	A novel lethal recognizable polymicrogyric syndrome caused by ATP1A2 homozygous truncating variants. Brain, 2019, 142, 3367-3374.	7.6	19
90	A proposed diagnostic approach for infantile spasms based on a spectrum of variable aetiology. European Journal of Paediatric Neurology, 2014, 18, 176-182.	1.6	18

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91	Exome sequencing in clinical settings: preferences and experiences of parents of children with rare diseases (SEQUAPRE study). European Journal of Human Genetics, 2019, 27, 701-710.	2.8	18
92	Anatomical and functional abnormalities on MRI in kabuki syndrome. NeuroImage: Clinical, 2019, 21, 101610.	2.7	17
93	Exome sequencing in 57 patients with self-limited focal epilepsies of childhood with typical or atypical presentations suggests novel candidate genes. European Journal of Paediatric Neurology, 2020, 27, 104-110.	1.6	17
94	Infantile Convulsions with Paroxysmal Dyskinesia (ICCA Syndrome) and Copy Number Variation at Human Chromosome 16p11. PLoS ONE, 2010, 5, e13750.	2.5	16
95	Direct Visualization of the Highly Polymorphic RNU2 Locus in Proximity to the BRCA1 Gene. PLoS ONE, 2013, 8, e76054.	2.5	16
96	Prenatal overgrowth and mosaic trisomy 15q25-qter including the IGF1 receptor gene. Prenatal Diagnosis, 2004, 24, 393-395.	2.3	15
97	An 800â€‰kb deletion at 17q23.2 including the MED13 (THRAP1) gene, revealed by aCGH in a patient with a SMC 17p. American Journal of Medical Genetics, Part A, 2012, 158A, 400-405.	1.2	15
98	Cell complementation using Genebridge 4 human:rodent hybrids for physical mapping of novel mitochondrial respiratory chain deficiency genes. Human Molecular Genetics, 2002, 11, 3273-3281.	2.9	14
99	Xp21 deletion in female patients with intellectual disability: Two new cases and a review of the literature. European Journal of Medical Genetics, 2015, 58, 341-345.	1.3	14
100	Autism spectrum disorder associated with 49,XYYYY: case report and review of the literature. BMC Medical Genetics, 2017, 18, 9.	2.1	14
101	Molecular characterisation of a prenatally diagnosed 5q15q21.3 deletion and review of the literature. Prenatal Diagnosis, 2006, 26, 231-238.	2.3	13
102	Arrayâ€CGH study of partial trisomy 9p without mental retardation. American Journal of Medical Genetics, Part A, 2011, 155, 1735-1739.	1.2	13
103	The psychological impact of cryptic chromosomal abnormalities diagnosis announcement. European Journal of Medical Genetics, 2013, 56, 585-590.	1.3	13
104	Neonatal tremor episodes and hyperekplexia-like presentation at onset in a child with SCN8A developmental and epileptic encephalopathy. Epileptic Disorders, 2018, 20, 289-294.	1.3	13
105	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.	2.7	12
106	Expression patterns of ERVWE1/Syncytin-1 and other placentally expressed human endogenous retroviruses along the malignant transformation process of hydatidiform moles. Placenta, 2016, 39, 116-124.	1.5	12
107	Reply to Salviati et al.. American Journal of Human Genetics, 2006, 79, 596-597.	6.2	11
108	Finger creases lend a hand in Kabuki syndrome. European Journal of Medical Genetics, 2013, 56, 556-560.	1.3	11

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109	Electrical status epilepticus in sleep, a constitutive feature of Christianson syndrome?. European Journal of Paediatric Neurology, 2018, 22, 1124-1132.	1.6	11
110	Novel truncating and missense variants extending the spectrum of EMC1-related phenotypes, causing autism spectrum disorder, severe global development delay and visual impairment. European Journal of Medical Genetics, 2020, 63, 103897.	1.3	11
111	Mosaic 18q21.2 deletions including the TCF4 gene: A clinical report. American Journal of Medical Genetics, Part A, 2012, 158A, 3174-3181.	1.2	10
112	Neuropathological features in a female fetus with OPHN1 deletion and cerebellar hypoplasia. European Journal of Medical Genetics, 2013, 56, 270-273.	1.3	10
113	Phenotype and Micro-array characterization of duplication 11q22.1-q25 and review of the literature. Gene, 2013, 519, 135-141.	2.2	10
114	A Novel Analog Reasoning Paradigm: New Insights in Intellectually Disabled Patients. PLoS ONE, 2016, 11, e0149717.	2.5	10
115	West syndrome caused by homozygous variant in the evolutionary conserved gene encoding the mitochondrial elongation factor GUF1. European Journal of Human Genetics, 2016, 24, 1001-1008.	2.8	10
116	Severe hemophilia A caused by an unbalanced chromosomal rearrangement identified using nanopore sequencing. Journal of Thrombosis and Haemostasis, 2019, 17, 1097-1103.	3.8	10
117	Genome sequencing in cytogenetics: Comparison of shortâ€read and linkedâ€read approaches for germline structural variant detection and characterization. Molecular Genetics & Genomic Medicine, 2020, 8, e1114.	1.2	10
118	Exclusion of the dymeclin and PAPSS2 genes in a novel form of spondyloepimetaphyseal dysplasia and mental retardation. European Journal of Human Genetics, 2005, 13, 541-546.	2.8	9
119	Childhood apraxia of speech without intellectual deficit in a patient with cri du chat syndrome. European Journal of Medical Genetics, 2012, 55, 433-436.	1.3	9
120	Chromosomal microarray analysis of functional Xq27-qter disomy and deletion 3p26.3 in a boy with Praderâ€“Willi like features and hypotonia. European Journal of Medical Genetics, 2012, 55, 461-465.	1.3	9
121	Multiple congenital anomaliesâ€intellectual disability (MCAâ€ID) and neuroblastoma in a patient harboring a de novo 14q23.1q23.3 deletion. American Journal of Medical Genetics, Part A, 2014, 164, 1310-1317.	1.2	9
122	Risk estimation of uniparental disomy of chromosome 14 or 15 in a fetus with a parent carrying a nonâ€homologous Robertsonian translocation. Should we still perform prenatal diagnosis?. Prenatal Diagnosis, 2019, 39, 986-992.	2.3	9
123	Beckwithâ€“Wiedemannâ€like macroglossia and 18q23 haploinsufficiency. American Journal of Medical Genetics, Part A, 2007, 143A, 2796-2803.	1.2	8
124	Clinical and molecular findings in nine new cases of tetrasomy 18p syndrome: FISH and array CGH characterization. Molecular Cytogenetics, 2019, 12, 5.	0.9	8
125	Cytogenetic investigation of a child with a mosaic isochromosome 18q and ring 18q. European Journal of Medical Genetics, 2007, 50, 379-385.	1.3	7
126	A novel telomeric (Â285 kb) Â-thalassemia deletion leading to a phenotypically unusual HbH disease. Haematologica, 2010, 95, 850-851.	3.5	7

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127	Search for a gene responsible for Floatingâ€Harbor syndrome on chromosome 12q15q21.1. American Journal of Medical Genetics, Part A, 2012, 158A, 333-339.	1.2	7
128	A novel truncating variant p.(Arg297*) in the GRM1 gene causing autosomal-recessive cerebellar ataxia with juvenile-onset. European Journal of Medical Genetics, 2019, 62, 103726.	1.3	7
129	Growth charts in Kabuki syndrome 1. American Journal of Medical Genetics, Part A, 2020, 182, 446-453.	1.2	7
130	HBB loss of heterozygosity in the hemopoietic lineage gives rise to an unusual sickle-cell trait phenotype. Haematologica, 2013, 98, e7-e8.	3.5	6
131	Complex mosaic CDKL5 deletion with two distinct mutant alleles in a 4â€yearâ€old girl. American Journal of Medical Genetics, Part A, 2014, 164, 2025-2028.	1.2	6
132	Clinical and molecular characterization of the 20q11.2 microdeletion syndrome: Six new patients. American Journal of Medical Genetics, Part A, 2015, 167, 504-511.	1.2	6
133	Pure proximal deletion of chromosome 21 and kyphosis. European Journal of Medical Genetics, 2007, 50, 469-474.	1.3	5
134	Molecular investigation, using chromosomal microarray and whole exome sequencing, of six patients affected by Williams Beuren syndrome and Autism Spectrum Disorder. Orphanet Journal of Rare Diseases, 2019, 14, 121.	2.7	5
135	Terminal 6q deletions cause brain malformations, a phenotype mimicking heterozygous DLL1 pathogenic variants: A multicenter retrospective case series. Prenatal Diagnosis, 2022, 42, 118-135.	2.3	5
136	No evidence of unbalanced growth-related gene inheritance in a series of overgrowth syndrome patients. American Journal of Medical Genetics Part A, 2001, 99, 166-167.	2.4	4
137	Characterization of two familial cases presenting with a syndromic specific learning disorder and carrying (17q;21q) unbalanced translocations. Clinical Case Reports (discontinued), 2018, 6, 827-834.	0.5	4
138	Supravalvular Aortic Stenosis Caused by a Familial Chromosome 7 Inversion Disrupting the ELN Gene Uncovered by Whole-Genome Sequencing. Molecular Syndromology, 2019, 10, 209-213.	0.8	4
139	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. Human Mutation, 2019, 40, 1993-2000.	2.5	4
140	The contribution of genomics in the medicine of tomorrow, clinical applications and issues. Therapie, 2019, 74, 9-15.	1.0	4
141	Regressive Autism Spectrum Disorder Expands theÂPhenotype of BSCL2/Seipin-Associated Neurodegeneration. Biological Psychiatry, 2019, 85, e17-e19.	1.3	4
142	A 14q distal chromoanagenesis elucidated by whole genome sequencing. European Journal of Medical Genetics, 2020, 63, 103776.	1.3	4
143	Prenatal cerebral imaging features of a new syndromic entity related to KIAA1109 pathogenic variants mimicking tubulinopathy. Prenatal Diagnosis, 2020, 40, 276-281.	2.3	4
144	X-linked partial corpus callosum agenesis with mild intellectual disability: identification of a novel L1CAM pathogenic variant. Neurogenetics, 2021, 22, 43-51.	1.4	4

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145	The C20orf133 gene is disrupted in a patient with Kabuki syndrome. BMJ Case Reports, 2009, 2009, bcr0620091994-bcr0620091994.	0.5	4
146	Genetic Counselling Pitfall: Co-Occurrence of an 11.8-Mb Xp22 Duplication and an Xp21.2 Duplication Disrupting IL1RAPL1. Molecular Syndromology, 2017, 8, 325-330.	0.8	4
147	Using deep-neural-network-driven facial recognition to identify distinct Kabuki syndrome 1 and 2 gestalt. European Journal of Human Genetics, 2022, 30, 682-686.	2.8	4
148	CNTNAP1-encephalopathy: Six novel patients surviving the neonatal period. European Journal of Paediatric Neurology, 2022, 37, 98-104.	1.6	4
149	Neurodevelopmental phenotype in 36 new patients with 8p inverted duplicationâ€“deletion: Genotypeâ€“phenotype correlation for anomalies of the corpus callosum. Clinical Genetics, 2022, 101, 307-316.	2.0	4
150	Complex phenotype with social communication disorder caused by mosaic supernumerary ring chromosome 19p. BMC Medical Genetics, 2014, 15, 132.	2.1	3
151	Microarray Analysis of 8p23.1 Deletion in New Patients with Atypical Phenotypical Traits. Journal of Pediatric Genetics, 2015, 04, 187-193.	0.7	3
152	Characterization of a de novo Supernumerary Neocentric Ring Chromosome Derived from Chromosome 7. Cytogenetic and Genome Research, 2015, 147, 111-117.	1.1	3
153	Clinical and molecular cytogenetic characterization of four unrelated patients carrying 2p14 microdeletions. American Journal of Medical Genetics, Part A, 2017, 173, 2268-2274.	1.2	3
154	Disruption and deletion of the proximal part of TCF4 are associated with mild intellectual disability: About three new patients. European Journal of Medical Genetics, 2022, 65, 104458.	1.3	3
155	Fertility in men with Klinefelter's syndrome. Annales D'Endocrinologie, 2022, 83, 172-176.	1.4	3
156	Possible association of 16p11.2 copy number variation with altered lymphocyte and neutrophil counts. Npj Genomic Medicine, 2022, 7, .	3.8	3
157	Screening for subtelomeric rearrangements using automated fluorescent genotyping of microsatellite markers: a Lebanese study. European Journal of Medical Genetics, 2006, 49, 117-126.	1.3	2
158	Monosomy 19pter and trisomy 19q13-qter in two siblings arising from a maternal pericentric inversion: Clinical data and molecular characterization. European Journal of Medical Genetics, 2008, 51, 622-630.	1.3	2
159	Unexpected diagnosis of 45,X/47,XX,+18 mosaicism in a girl with mild phenotype. American Journal of Medical Genetics, Part A, 2009, 149A, 2584-2587.	1.2	2
160	Jacobsen and Beckwithâ€“Wiedemann syndromes in a child with mosaicism for partial 11pter trisomy and partial 11qter monosomy. American Journal of Medical Genetics, Part A, 2013, 161, 331-337.	1.2	2
161	Diversity of Hepatocellular Carcinoma Clones Bearing Hematopoietic Malignancies-Related Chromosomal Translocation. Journal of Cellular Biochemistry, 2014, 115, 666-677.	2.6	2
162	Genomic Microarray in Intellectual Disability: The Usefulness of Existing Systems in the Interpretation of Copy Number Variation. Journal of Pediatric Genetics, 2017, 06, 084-091.	0.7	2

#	Article	IF	Citations
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