Damien Sanlaville

List of Publications by Year
in descending order

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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	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
2	CNTNAP1-encephalopathy: Six novel patients surviving the neonatal period. European Journal of Paediatric Neurology, 2022, 37, 98-104.	1.6	4
3	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
4	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
5	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
6	Toward clinical and molecular dissection of frontonasal dysplasia with facial skin polyps: From Pai syndrome to differential diagnosis through a series of 27 patients. American Journal of Medical Genetics, Part A, 2022, 188, 2036-2047.	1.2	1
7	Fertility in men with Klinefelter's syndrome. Annales D'Endocrinologie, 2022, 83, 172-176.	1.4	3
8	Possible association of 16p11.2 copy number variation with altered lymphocyte and neutrophil counts. Npj Genomic Medicine, 2022, 7, .	3.8	3
9	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
10	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	7.6	35
11	Optical genome mapping enables constitutional chromosomal aberration detection. American Journal of Human Genetics, 2021, 108, 1409-1422.	6.2	108
12	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
13	A 14q distal chromoanagenesis elucidated by whole genome sequencing. European Journal of Medical Genetics, 2020, 63, 103776.	1.3	4
14	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. Genetics in Medicine, 2020, 22, 181-188.	2.4	30
15	Growth charts in Kabuki syndrome 1. American Journal of Medical Genetics, Part A, 2020, 182, 446-453.	1.2	7
16	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
17	Chromosomal instability in the prediction of pituitary neuroendocrine tumors prognosis. Acta Neuropathologica Communications, 2020, 8, 190.	5.2	20
18	Multisystem disorders, severe developmental delay and seizures in two affected siblings, expanding the phenotype of PIGC deficiency. European Journal of Medical Genetics, 2020, 63, 103994.	1.3	1

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19	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
20	A Case of Trisomy 13 Mosaicism Presenting with a Severe Aortic Root Dilatation and Marfanoid Habitus due to an Unpredictable Cytogenetic Mechanism. Cytogenetic and Genome Research, 2020, 160, 72-79.	1.1	1
21	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
22	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
23	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
24	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
25	A novel lethal recognizable polymicrogyric syndrome caused by ATP1A2 homozygous truncating variants. Brain, 2019, 142, 3367-3374.	7.6	19
26	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
27	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
28	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. Human Mutation, 2019, 40, 1993-2000.	2.5	4
29	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
30	Molecular Characterization of a Familial 13.6-Mb 20p11.1p12.1 Duplication without Clinical Consequence. Cytogenetic and Genome Research, 2019, 157, 141-147.	1.1	0
31	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
32	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
33	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
34	The contribution of genomics in the medicine of tomorrow, clinical applications and issues. Therapie, 2019, 74, 9-15.	1.0	4
35	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
36	Regressive Autism Spectrum Disorder Expands theÂPhenotype of BSCL2/Seipin-Associated Neurodegeneration. Biological Psychiatry, 2019, 85, e17-e19.	1.3	4

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37	GRIN2A-related disorders: genotype and functional consequence predict phenotype. Brain, 2019, 142, 80-92.	7.6	143
38	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
39	Anatomical and functional abnormalities on MRI in kabuki syndrome. NeuroImage: Clinical, 2019, 21, 101610.	2.7	17
40	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
41	Centralization errors in comparative genomic hybridization array analysis of pituitary tumor samples. Genes Chromosomes and Cancer, 2018, 57, 320-328.	2.8	2
42	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
43	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
44	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
45	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	5.3	20
46	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
47	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
48	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
49	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
50	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
51	Sex gap in aging and longevity: can sex chromosomes play a role?. Biology of Sex Differences, 2018, 9, 33.	4.1	82
52	Electrical status epilepticus in sleep, a constitutive feature of Christianson syndrome?. European Journal of Paediatric Neurology, 2018, 22, 1124-1132.	1.6	11
53	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
54	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

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55	A novel disorder of sex development, characterized by progressive regression of testicular function and cystic leukoencephalopathy. American Journal of Medical Genetics, Part A, 2017, 173, 654-660.	1.2	1
56	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
57	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
58	Autism spectrum disorder associated with 49,XYYYY: case report and review of the literature. BMC Medical Genetics, 2017, 18, 9.	2.1	14
59	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
60	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
61	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
62	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
63	Familial X/Y Translocation Encompassing ARSE in Two Moroccan Siblings with Sensorineural Deafness. Cytogenetic and Genome Research, 2017, 153, 66-72.	1.1	1
64	Prenatal Diagnosis of Trisomy 2p due to Terminal 2p Duplication including Interstitial Telomeric Sequences. Cytogenetic and Genome Research, 2017, 153, 117-124.	1.1	2
65	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	3.8	58
66	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
67	A Novel Analog Reasoning Paradigm: New Insights in Intellectually Disabled Patients. PLoS ONE, 2016, 11, e0149717.	2.5	10
68	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	2.5	134
69	A Distinct Class of Chromoanagenesis Events Characterized by Focal Copy Number Gains. Human Mutation, 2016, 37, 661-668.	2.5	30
70	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
71	Genetic and neurodevelopmental spectrum ofSYNGAP1-associated intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 511-522.	3.2	135
72	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

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73	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
74	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
75	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
76	Microarray Analysis of 8p23.1 Deletion in New Patients with Atypical Phenotypical Traits. Journal of Pediatric Genetics, 2015, 04, 187-193.	0.7	3
77	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
78	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
79	Behavioral disturbance and treatment strategies in Smith-Magenis syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 111.	2.7	44
80	Functional variants of POC5 identified in patients with idiopathic scoliosis. Journal of Clinical Investigation, 2015, 125, 1124-1128.	8.2	87
81	Characterization of a de novo Supernumerary Neocentric Ring Chromosome Derived from Chromosome 7. Cytogenetic and Genome Research, 2015, 147, 111-117.	1.1	3
82	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
83	Molecular characterization of a cohort of 73 patients with infantile spasms syndrome. European Journal of Medical Genetics, 2015, 58, 51-58.	1.3	56
84	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
85	Reinforcement of STAT3 activity reprogrammes human embryonic stem cells to naive-like pluripotency. Nature Communications, 2015, 6, 7095.	12.8	137
86	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
87	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
88	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	2.9	61
89	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	3.5	501
90	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

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91	Diversity of Hepatocellular Carcinoma Clones Bearing Hematopoietic Malignancies-Related Chromosomal Translocation. Journal of Cellular Biochemistry, 2014, 115, 666-677.	2.6	2
92	A new intellectual disability syndrome caused by CTNNB1 haploinsufficiency. American Journal of Medical Genetics, Part A, 2014, 164, 1571-1575.	1.2	50
93	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
94	Complex phenotype with social communication disorder caused by mosaic supernumerary ring chromosome 19p. BMC Medical Genetics, 2014, 15, 132.	2.1	3
95	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
96	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
97	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
98	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
99	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
100	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
101	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
102	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
103	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
104	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
105	The psychological impact of cryptic chromosomal abnormalities diagnosis announcement. European Journal of Medical Genetics, 2013, 56, 585-590.	1.3	13
106	Clinical and Molecular Spectrum of Renal Malformations in Kabuki Syndrome. Journal of Pediatrics, 2013, 163, 742-746.	1.8	27
107	Neuropathological features in a female fetus with OPHN1 deletion and cerebellar hypoplasia. European Journal of Medical Genetics, 2013, 56, 270-273.	1.3	10
108	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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109	Phenotype and Micro-array characterization of duplication 11q22.1-q25 and review of the literature. Gene, 2013, 519, 135-141.	2.2	10
110	Finger creases lend a hand in Kabuki syndrome. European Journal of Medical Genetics, 2013, 56, 556-560.	1.3	11
111	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
112	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
113	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
114	Chromosome 21 Scan in Down Syndrome Reveals DSCAM as a Predisposing Locus in Hirschsprung Disease. PLoS ONE, 2013, 8, e62519.	2.5	22
115	Direct Visualization of the Highly Polymorphic RNU2 Locus in Proximity to the BRCA1 Gene. PLoS ONE, 2013, 8, e76054.	2.5	16
116	Molecular cytogenetic and phenotypic characterization of ring chromosome 13 in three unrelated patients. Journal of Pediatric Genetics, 2013, 2, 147-55.	0.7	2
117	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
118	PRRT2 links infantile convulsions and paroxysmal dyskinesia with migraine. Neurology, 2012, 79, 2097-2103.	1.1	90
119	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
120	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
121	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
122	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
123	Prenatal diagnosis of â€˜isolatedâ€™ Dandyâ€“Walker malformation: imaging findings and prenatal counselling. Prenatal Diagnosis, 2012, 32, 185-193.	2.3	52
124	Genome-wide arrays: Quality criteria and platforms to be used in routine diagnostics. Human Mutation, 2012, 33, 906-915.	2.5	69
125	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
126	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

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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	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	27.8	394
129	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
130	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
131	Arrayâ€CGH study of partial trisomy 9p without mental retardation. American Journal of Medical Genetics, Part A, 2011, 155, 1735-1739.	1.2	13
132	Association of TALS Developmental Disorder with Defect in Minor Splicing Component U4atac snRNA. Science, 2011, 332, 240-243.	12.6	195
133	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
134	A novel telomeric (Â285 kb) Â-thalassemia deletion leading to a phenotypically unusual HbH disease. Haematologica, 2010, 95, 850-851.	3.5	7
135	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
136	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
137	Infantile Convulsions with Paroxysmal Dyskinesia (ICCA Syndrome) and Copy Number Variation at Human Chromosome 16p11. PLoS ONE, 2010, 5, e13750.	2.5	16
138	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
139	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
140	Distal Xq duplication and functional Xq disomy. Orphanet Journal of Rare Diseases, 2009, 4, 4.	2.7	58
141	The C20orf133 gene is disrupted in a patient with Kabuki syndrome. BMJ Case Reports, 2009, 2009, bcr0620091994-bcr0620091994.	0.5	4
142	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
143	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
144	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

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145	The C20orf133 gene is disrupted in a patient with Kabuki syndrome. Journal of Medical Genetics, 2007, 44, 562-569.	3.2	56
146	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
147	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
148	Pure proximal deletion of chromosome 21 and kyphosis. European Journal of Medical Genetics, 2007, 50, 469-474.	1.3	5
149	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
150	Beckwithâ€“Wiedemannâ€like macroglossia and 18q23 haploinsufficiency. American Journal of Medical Genetics, Part A, 2007, 143A, 2796-2803.	1.2	8
151	CHARGE syndrome: an update. European Journal of Human Genetics, 2007, 15, 389-399.	2.8	280
152	Guidelines for molecular karyotyping in constitutional genetic diagnosis. European Journal of Human Genetics, 2007, 15, 1105-1114.	2.8	144
153	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
154	Reply to Salviati et al.. American Journal of Human Genetics, 2006, 79, 596-597.	6.2	11
155	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
156	A novel X-linked recessive form of Mendelian susceptibility to mycobaterial disease. Journal of Medical Genetics, 2006, 44, e65-e65.	3.2	52
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	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
159	Molecular characterisation of a prenatally diagnosed 5q15q21.3 deletion and review of the literature. Prenatal Diagnosis, 2006, 26, 231-238.	2.3	13
160	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
161	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
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