Orsetta Zuffardi

List of Publications by Year
in descending order

332 papers	17,525 citations	15880 67 h-index	22488 117 g-index
336 all docs	336 docs citations	336 times ranked	22041 citing authors

#	Article	IF	Citations
1	Seamless Gene Correction in the Human Cystic Fibrosis Transmembrane Conductance Regulator Locus by Vector Replacement and Vector Insertion Events. Frontiers in Genome Editing, 2022, 4, 843885.	2.7	0
2	The embryo battle against adverse genomes: Are de novo terminal deletions the rescue of unfavorable zygotic imbalances?. European Journal of Medical Genetics, 2022, 65, 104532.	0.7	4
3	Low penetrance COL5A1 variants in a young patient with intracranial aneurysm and very mild signs of Ehlers-Danlos syndrome. European Journal of Medical Genetics, 2021, 64, 104099.	0.7	2
4	Chiari 1 malformation and exome sequencing in 51 trios: the emerging role of rare missense variants in chromatin-remodeling genes. Human Genetics, 2021, 140, 625-647.	1.8	10
5	Improving the phenotype description of Basel-Vanagaite-Smirin-Yosef syndrome, MED25-related: polymicrogyria as a distinctive neuroradiological finding. Neurogenetics, 2021, 22, 19-25.	0.7	1
6	Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. Seizure: the Journal of the British Epilepsy Association, 2021, 88, 60-72.	0.9	6
7	Adducted Thumb and Peripheral Polyneuropathy: Diagnostic Supports in Suspecting Whiteâ€“Sutton Syndrome: Case Report and Review of the Literature. Genes, 2021, 12, 950.	1.0	5
8	Whole Exome Sequencing Is the Minimal Technological Approach in Probands Born to Consanguineous Couples. Genes, 2021, 12, 962.	1.0	0
9	Clinical Manifestations in a Girl with NAA10-Related Syndrome and Genotypeâ€“Phenotype Correlation in Females. Genes, 2021, 12, 900.	1.0	11
10	Posterior Lissencephaly Associated with Subcortical Band Heterotopia Due to a Variation in the CEP85L Gene: A Case Report and Refining of the Phenotypic Spectrum. Genes, 2021, 12, 1208.	1.0	2
11	NRF1 association with AUTS2-Polycomb mediates specific gene activation in the brain. Molecular Cell, 2021, 81, 4663-4676.e8.	4.5	23
12	Prenatal Noninvasive Trio-WES in a Case of Pregnancy-Related Liver Disorder. Diagnostics, 2021, 11, 1904.	1.3	3
13	RB1CC1 duplication and aberrant overexpression in a patient with schizophrenia: further phenotype delineation and proposal of a pathogenetic mechanism. Molecular Genetics & Genomic Medicine, 2021, 9, e1561.	0.6	2
14	Transcutaneous electrical stimulation therapy and genetic analysis in Dercum's disease. Medicine (United States), 2021, 100, e28360.	0.4	2
15	Alazami syndrome: the first case of papillary thyroid carcinoma. Journal of Human Genetics, 2020, 65, 133-141.	1.1	10
16	An additional piece in the <scp>TBX6</scp> gene dosage model: A novel nonsense variant in a fetus with severe spondylocostal dysostosis. Clinical Genetics, 2020, 98, 628-629.	1.0	2
17	Expanding the phenotype of Wiedemannâ€Steiner syndrome: Craniovertebral junction anomalies. American Journal of Medical Genetics, Part A, 2020, 182, 2877-2886.	0.7	9
18	Characterization of a novel loss-of-function variant in TDP2 in two adult patients with spinocerebellar ataxia autosomal recessive 23 (SCAR23). Journal of Human Genetics, 2020, 65, 1135-1141.	1.1	7

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19	FANCA, TP53, and del(5q)/RPS14 alterations in a patient with T-cell non-Hodgkin lymphoma and concomitant Fanconi anemia and Li-Fraumeni syndrome. Cancer Genetics, 2020, 256-257, 179-183.	0.2	1
20	Disseminated Mycobacterium Avium Infection in a Child with Complete Interferon-Î³ Receptor 1 Deficiency due to Compound Heterozygosis of IFNGR1 for a Subpolymorphic Copy Number Variation and a Novel Splice-Site Variant. Journal of Pediatric Genetics, 2020, 09, 186-192.	0.3	6
21	Targeted next-generation sequencing identifies the disruption of the SHANK3 and RYR2 genes in a patient carrying a de novo t(1;22)(q43;q13.3) associated with signs of Phelan-McDermid syndrome. Molecular Cytogenetics, 2020, 13, 22.	0.4	4
22	A novel de novo partial xq duplication in a girl with short stature, nonverbal learning disability and diminished ovarian reserve - effect of growth hormone treatment and fertility preservation strategies: a case report and up-to-date review. International Journal of Pediatric Endocrinology (Springer), 2020, 2020, 1.	1.6	5
23	Noninvasive prenatal diagnosis in a family at risk for Fraser syndrome. Prenatal Diagnosis, 2020, 40, 905-908.	1.1	4
24	Phenotypic Expansion in Nasu-Hakola Disease: Immunological Findings in Three Patients and Proposal of a Unifying Pathogenic Hypothesis. Frontiers in Immunology, 2019, 10, 1685.	2.2	15
25	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
26	PIEZO1 Hypomorphic Variants in Congenital Lymphatic Dysplasia Cause Shape and Hydration Alterations of Red Blood Cells. Frontiers in Physiology, 2019, 10, 258.	1.3	26
27	Insertional translocation involving an additional nonchromothriptic chromosome in constitutional chromothripsis: Rule or exception?. Molecular Genetics & Genomic Medicine, 2019, 7, e00496.	0.6	13
28	Small supernumerary marker chromosomes: A legacy of trisomy rescue?. Human Mutation, 2019, 40, 193-200.	1.1	33
29	Early-onset movement disorder as diagnostic marker in genetic syndromes: Three cases of FOXG1 -related syndrome. European Journal of Paediatric Neurology, 2018, 22, 336-339.	0.7	12
30	Chromothripsis and ring chromosome 22: a paradigm of genomic complexity in the Phelan-McDermid syndrome (22q13 deletion syndrome). Journal of Medical Genetics, 2018, 55, 269-277.	1.5	22
31	Three Reportedly Unrelated Families With Liddle Syndrome Inherited From a Common Ancestor. Hypertension, 2018, 71, 273-279.	1.3	14
32	De novo unbalanced translocations have a complex history/aetiology. Human Genetics, 2018, 137, 817-829.	1.8	23
33	SOX2: Not always eye malformations. Severe genital but no major ocular anomalies in a female patient with the recurrent c.70del20 variant. European Journal of Medical Genetics, 2018, 61, 335-340.	0.7	15
34	Inherited human IRAK-1 deficiency selectively impairs TLR signaling in fibroblasts. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E514-E523.	3.3	49
35	MCM5: a new actor in the link between DNA replication and Meier-Gorlin syndrome. European Journal of Human Genetics, 2017, 25, 646-650.	1.4	60
36	Whole exome sequencing in the differential diagnosis of Diamond-Blackfan anemia: Clinical and molecular study of three patients with novel RPL5 and mosaic RPS19 mutations. Blood Cells, Molecules, and Diseases, 2017, 64, 38-44.	0.6	12

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37	Guideline recommendations for diagnosis and clinical management of Ring14 syndromeâ€"first report of an ad hoc task force. Orphanet Journal of Rare Diseases, 2017, 12, 69.	1.2	18
38	Haploinsufficiency for ANKRD11-flanking genes makes the difference between KBG and 16q24.3 microdeletion syndromes: 12 new cases. European Journal of Human Genetics, 2017, 25, 694-701.	1.4	33
39	Developmental trends of communicative skills in children with chromosome 14 aberrations. European Journal of Pediatrics, 2017, 176, 455-464.	1.3	2
40	SMARCA4 inactivating mutations cause concomitant Coffinâ€"Siris syndrome, microphthalmia and smallâ€cell carcinoma of the ovary hypercalcaemic type. Journal of Pathology, 2017, 243, 9-15.	2.1	47
41	A novel APC promoter 1B deletion shows a founder effect in Italian patients with classical familial adenomatous polyposis phenotype. Genes Chromosomes and Cancer, 2017, 56, 846-854.	1.5	7
42	Clinical and Molecular Characteristics of SLC16A2 (MCT8) Mutations in Three Families with the Allan-Herndon-Dudley Syndrome. Human Mutation, 2017, 38, 260-264.	1.1	31
43	A Novel Strategy Combining Array-CGH, Whole-exome Sequencing and In Utero Electroporation in Rodents to Identify Causative Genes for Brain Malformations. Journal of Visualized Experiments, 2017, , .	0.2	0
44	A donor splice site mutation in CISD2 generates multiple truncated, non-functional isoforms in Wolfram syndrome type 2 patients. BMC Medical Genetics, 2017, 18, 147.	2.1	12
45	Partial monosomy 8p and trisomy 16q in two children with developmental delay detected by array comparative genomic hybridization. Molecular Medicine Reports, 2017, 16, 8808-8818.	1.1	3
46	A Data Fusion Approach to Enhance Association Study in Epilepsy. PLoS ONE, 2016, 11, e0164940.	1.1	4
47	Formation of new chromatin domains determines pathogenicity of genomic duplications. Nature, 2016, 538, 265-269.	13.7	582
48	Dissection of partial 21q monosomy in different phenotypes: clinical and molecular characterization of five cases and review of the literature. Molecular Cytogenetics, 2016, 9, 21.	0.4	21
49	Chromosome 17q21.31 duplication syndrome: Description of a new familiar case and further delineation of the clinical spectrum. European Journal of Paediatric Neurology, 2016, 20, 183-187.	0.7	10
50	Reply to Sajantila and Budowle. European Journal of Human Genetics, 2016, 24, 330-330.	1.4	1
51	The Diagnostic Yield of Array Comparative Genomic Hybridization Is High Regardless of Severity of Intellectual Disability/Developmental Delay in Children. Journal of Child Neurology, 2016, 31, 691-699.	0.7	37
52	Comprehensive characterization of mesenchymal stromal cells from patients with Fanconi anaemia. British Journal of Haematology, 2015, 170, 826-836.	1.2	23
53	A novel mutation in COL4A1 gene: A possible cause of early postnatal cerebrovascular events. American Journal of Medical Genetics, Part A, 2015, 167, 810-815.	0.7	7
54	Next Generation Sequencing for Systematic Assessment of Genetics of Small-Vessel Disease and Lacunar Stroke. Journal of Stroke and Cerebrovascular Diseases, 2015, 24, 759-765.	0.7	8

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55	Primary coenzyme Q10 deficiency presenting as fatal neonatal multiorgan failure. European Journal of Human Genetics, 2015, 23, 1254-1258.	1.4	42
56	TREX1 C-terminal frameshift mutations in the systemic variant of retinal vasculopathy with cerebral leukodystrophy. Neurological Sciences, 2015, 36, 323-330.	0.9	45
57	A Revised Genome Assembly of the Region 5â€² to Canine <i>SOX9</i> Includes the <i>RevSex</i> Orthologous Region. Sexual Development, 2015, 9, 155-161.	1.1	10
58	Wolfram syndrome 2: a novel CISD2 mutation identified in Italian siblings. Acta Diabetologica, 2015, 52, 175-178.	1.2	34
59	Loss-of-FunctionFANCLMutations Associate with Severe Fanconi Anemia Overlapping the VACTERL Association. Human Mutation, 2015, 36, 562-568.	1.1	23
60	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	1.1	45
61	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	1.4	56
62	Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3. European Journal of Human Genetics, 2015, 23, 1025-1032.	1.4	59
63	APC rearrangements in familial adenomatous polyposis: heterogeneity of deletion lengths and breakpoint sequences underlies similar phenotypes. Familial Cancer, 2015, 14, 41-49.	0.9	13
64	Improving molecular diagnosis in epilepsy by a dedicated high-throughput sequencing platform. European Journal of Human Genetics, 2015, 23, 354-362.	1.4	64
65	Proximal 10q duplication in a child with severe central hypotonia characterized by array-comparative genomic hybridization: A case report and review of the literature. Experimental and Therapeutic Medicine, 2014, 7, 953-957.	0.8	5
66	Hyper IgE syndrome: anaphylaxis in a patient carrying the N567DSTAT3mutation. Pediatric Allergy and Immunology, 2014, 25, 503-505.	1.1	9
67	Heterozygous deletion of CHL1 gene: Detailed array-CGH and clinical characterization of a new case and review of the literature. European Journal of Medical Genetics, 2014, 57, 626-629.	0.7	23
68	A Therapeutic Challenge: Liddleâ€™s Syndrome Managed with Amiloride during Pregnancy. Case Reports in Obstetrics and Gynecology, 2014, 2014, 1-4.	0.2	15
69	Lower motor neuron disease with respiratory failure caused by a novel MAPT mutation. Neurology, 2014, 82, 1990-1998.	1.5	21
70	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	1.4	109
71	MECP2 duplication phenotype in symptomatic females: report of three further cases. Molecular Cytogenetics, 2014, 7, 10.	0.4	21
72	Mutations in MAP3K1 tilt the balance from SOX9/FGF9 to WNT/Î²-catenin signaling. Human Molecular Genetics, 2014, 23, 1073-1083.	1.4	72

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73	Defining the phenotype associated with microduplication reciprocal to Sotos syndrome microdeletion. American Journal of Medical Genetics, Part A, 2014, 164, 2084-2090.	0.7	20
74	Deletions of the PRKAR1A Locus at 17q24.2-q24.3 in Carney Complex: Genotype-Phenotype Correlations and Implications for Genetic Testing. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E183-E188.	1.8	57
75	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. Human Molecular Genetics, 2014, 23, 2752-2768.	1.4	140
76	Seizures and EEG features in 74 patients with geneticâ€dysmorphic syndromes. American Journal of Medical Genetics, Part A, 2014, 164, 3154-3161.	0.7	16
77	PRKACB and Carney Complex. New England Journal of Medicine, 2014, 370, 1065-1067.	13.9	121
78	Functional and genetic aberrations of in vitro-cultured marrow-derived mesenchymal stromal cells of patients with classical Philadelphia-negative myeloproliferative neoplasms. Leukemia, 2014, 28, 1742-1745.	3.3	30
79	Severe growth hormone deficiency and pituitary malformation in a patient with chromosome 2p25 duplication and 2q37 deletion. Molecular Cytogenetics, 2014, 7, 41.	0.4	11
80	In vitro biosafety profile evaluation of multipotent mesenchymal stem cells derived from the bone marrow of sarcoma patients. Journal of Translational Medicine, 2014, 12, 95.	1.8	10
81	Constitutive Activation of PKA Catalytic Subunit in Adrenal Cushing's Syndrome. New England Journal of Medicine, 2014, 370, 1019-1028.	13.9	355
82	A patient with partial trisomy 21 and 7q deletion expresses mild Down syndrome phenotype. Gene, 2014, 536, 441-443.	1.0	20
83	Periventricular nodular heterotopia in Smithâ€Magenis syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 3142-3147.	0.7	14
84	Sox9 Duplications Are a Relevant Cause of Sry-Negative XX Sex Reversal Dogs. PLoS ONE, 2014, 9, e101244.	1.1	39
85	Genome-wide copy number analysis in pediatric glioblastoma multiforme. American Journal of Cancer Research, 2014, 4, 293-303.	1.4	10
86	A newborn with ambiguous genitalia and a complex X;Y rearrangement. Iranian Journal of Reproductive Medicine, 2014, 12, 351-6.	0.8	1
87	Neonatal suppressionâ€burst without epileptic seizures: expanding the electroclinical phenotype of STXBP1â€related, earlyâ€onset encephalopathy. Epileptic Disorders, 2013, 15, 55-61.	0.7	26
88	Dravet phenotype in a subject with a der(4)t(4;8)(p16.3;p23.3) without the involvement of the LETM1 gene. European Journal of Medical Genetics, 2013, 56, 551-555.	0.7	11
89	MCT8 Deficiency. Journal of Child Neurology, 2013, 28, 795-800.	0.7	48
90	Genomic alterations in human umbilical cordâ€“derived mesenchymal stromal cells call for stringent quality control before any possible therapeutic approach. Cytotherapy, 2013, 15, 1362-1373.	0.3	21

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91	MEF2C deletions and mutations versus duplications: A clinical comparison. European Journal of Medical Genetics, 2013, 56, 260-265.	0.7	24
92	5p13 microduplication syndrome: A new case and better clinical definition of the syndrome. European Journal of Medical Genetics, 2013, 56, 54-58.	0.7	14
93	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	3.7	85
94	Ade novoX;8 translocation creates aPTK2-THOC2gene fusion withTHOC2expression knockdown in a patient with psychomotor retardation and congenital cerebellar hypoplasia. Journal of Medical Genetics, 2013, 50, 543-551.	1.5	42
95	Idiopathic Central Precocious Puberty Associated with 11â€‰MbDe NovoDistal Deletion of the Chromosome 9 Short Arm. Case Reports in Genetics, 2013, 2013, 1-6.	0.1	6
96	Partial Trisomy 2p and Partial Monosomy 2q Arising from a Paternal Intrachromosomal 2q-into-2p Between-Arm Insertion and Paracentric Inversion: Molecular Cytogenetic Characterization of a Four-Break Rearrangement. Cytogenetic and Genome Research, 2013, 140, 12-20.	0.6	1
97	De novo 15.5-Mb Interstitial Deletion in 5p in a Male Ascertained by Oligospermia. Molecular Syndromology, 2013, 4, 250-254.	0.3	0
98	Spleen endothelial cells from patients with myelofibrosis harbor the JAK2V617F mutation. Blood, 2013, 121, 360-368.	0.6	102
99	Multiple clinical forms of dehydrated hereditary stomatocytosis arise from mutations in PIEZO1. Blood, 2013, 121, 3925-3935.	0.6	266
100	Prenatal diagnosis of two de novo 4q35-qter deletions characterized by array-CGH. Molecular Cytogenetics, 2013, 6, 47.	0.4	12
101	Haploinsufficiency ofCOQ4causes coenzyme Q₁₀deficiency. Journal of Medical Genetics, 2012, 49, 187-191.	1.5	95
102	Cognitive and Behavioral Phenotype of a Young Man With a Chromosome 13 Deletion del(13)(q21.32q31.1). Cognitive and Behavioral Neurology, 2012, 25, 154-158.	0.5	2
103	Agenesis of Internal Carotid Artery and Hypopituitarism: Case Report and Review of Literature. Journal of Clinical Endocrinology and Metabolism, 2012, 97, 3414-3420.	1.8	9
104	19q13.11 cryptic deletion: description of two new cases and indication for a role of WTIP haploinsufficiency in hypospadias. European Journal of Human Genetics, 2012, 20, 852-856.	1.4	40
105	The Genetics of Small-Vessel Disease. Current Medicinal Chemistry, 2012, 19, 4124-4141.	1.2	14
106	Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322.	4.9	61
107	22q11.2 Microduplication syndrome and epilepsy with continuous spikes and waves during sleep (CSWS). A case report and review of the literature. Epilepsy and Behavior, 2012, 25, 567-572.	0.9	14
108	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449.	9.4	207

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109	Unexpected results in the constitution of small supernumerary marker chromosomes. European Journal of Medical Genetics, 2012, 55, 185-190.	0.7	22
110	Interstitial deletion of chromosome 2p15-16.1: Report of two patients and critical review of current genotypeâ€“phenotype correlation. European Journal of Medical Genetics, 2012, 55, 238-244.	0.7	30
111	Microarray application in prenatal diagnosis: a position statement from the cytogenetics working group of the Italian Society of Human Genetics (SIGU), November 2011. Ultrasound in Obstetrics and Gynecology, 2012, 39, 384-388.	0.9	50
112	The strange case of the lost NRAS mutation in a child with juvenile myelomonocytic leukemia. Pediatric Blood and Cancer, 2012, 59, 580-582.	0.8	2
113	The introduction of arrays in prenatal diagnosis: A special challenge. Human Mutation, 2012, 33, 923-929.	1.1	63
114	Periventricular heterotopia with white matter abnormalities associated with 6p25 deletion. American Journal of Medical Genetics, Part A, 2012, 158A, 1793-1797.	0.7	29
115	Identification of de novo mutations and rare variants in hypoplastic left heart syndrome. Clinical Genetics, 2012, 81, 542-554.	1.0	97
116	Prenatal diagnosis of Wolf-Hirschhorn syndrome confirmed by comparative genomic hybridization array: report of two cases and review of the literature. Molecular Cytogenetics, 2012, 5, 12.	0.4	17
117	De Novo Unbalanced Translocations in Prader-Willi and Angelman Syndrome Might Be the Reciprocal Product of inv dup(15)s. PLoS ONE, 2012, 7, e39180.	1.1	5
118	Molecular Analysis of Primary Cutaneous Aggressive T-Cell Lymphomas: the Epidermotropic CD8+, the Pleomorphic CD8+ and the Gamma Delta Subsets.. Blood, 2012, 120, 2713-2713.	0.6	0
119	A wide methodological approach to identify a large duplication in CFTR gene in a CF patient uncharacterised by sequencing analysis. Journal of Cystic Fibrosis, 2011, 10, 412-417.	0.3	7
120	Array technology in prenatal diagnosis. Seminars in Fetal and Neonatal Medicine, 2011, 16, 94-98.	1.1	20
121	CD5â˜ diffuse large B-cell lymphoma with peculiar cyclin D1+ phenotype. Pathologic and molecular characterization of a single case. Human Pathology, 2011, 42, 1204-1208.	1.1	15
122	Correlation between genomic alterations assessed by array comparative genomic hybridization, prognostically informative histologic subtype, stage, and patient survival in gastric cancer. Human Pathology, 2011, 42, 1937-1945.	1.1	22
123	Twenty-one cases of blastic plasmacytoid dendritic cell neoplasm: focus on biallelic locus 9p21.3 deletion. Blood, 2011, 118, 4591-4594.	0.6	140
124	Duplications of FOXG1 in 14q12 are associated with developmental epilepsy, mental retardation, and severe speech impairment. European Journal of Human Genetics, 2011, 19, 102-107.	1.4	104
125	The phenotype of recurrent 10q22q23 deletions and duplications. European Journal of Human Genetics, 2011, 19, 400-408.	1.4	63
126	Definition of the neurological phenotype associated with dup (X)(p11.22-p11.23). Epileptic Disorders, 2011, 13, 240-251.	0.7	8

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127	Gene copy number variation in male breast cancer by aCGH. Cellular Oncology (Dordrecht), 2011, 34, 467-473.	2.1	12
128	Current controversies in prenatal diagnosis 3: is conventional chromosome analysis necessary in the postâ€array CGH era?. Prenatal Diagnosis, 2011, 31, 235-243.	1.1	50
129	Deletion 2q31.2â€q31.3 in a 4â€yearâ€old girl with microcephaly and severe mental retardation. American Journal of Medical Genetics, Part A, 2011, 155, 1476-1482.	0.7	8
130	Common structural features characterize interstitial intrachromosomal Xp and 18q triplications. American Journal of Medical Genetics, Part A, 2011, 155, 2681-2687.	0.7	6
131	Cellâ€cycle phases and genetic profile of bone marrowâ€derived mesenchymal stromal cells expanded in vitro from healthy donors. Journal of Cellular Biochemistry, 2011, 112, 1817-1821.	1.2	19
132	XX males SRY negative: a confirmed cause of infertility. Journal of Medical Genetics, 2011, 48, 710-712.	1.5	86
133	Identification of Novel Prognostic Markers in Relapsing Localized Resectable Neuroblastoma. OMICS A Journal of Integrative Biology, 2011, 15, 113-121.	1.0	4
134	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173.	1.5	172
135	Blastic Plasmacytoid Dendritic Cell Neoplasm: Clinical, Immunohistochemical and Molecular Evaluation of 23 Cases with Primary Cutaneous Involvement,. Blood, 2011, 118, 3565-3565.	0.6	2
136	Severe X-Linked Mitochondrial Encephalomyopathy Associated with a Mutation in Apoptosis-Inducing Factor. American Journal of Human Genetics, 2010, 86, 639-649.	2.6	199
137	Breakpoint determination of 15 large deletions in Peutzâ€“Jeghers subjects. Human Genetics, 2010, 128, 373-382.	1.8	26
138	A de novo 11p12-p15.4 duplication in a patient with pharmacoresistant epilepsy, mental retardation, and dysmorphisms. Brain and Development, 2010, 32, 248-252.	0.6	3
139	Distinct transcriptional profiles characterize bone microenvironment mesenchymal cells rather than osteoblasts in relationship with multiple myeloma bone disease. Experimental Hematology, 2010, 38, 141-153.	0.2	57
140	Mutations in SOX17 are associated with congenital anomalies of the kidney and the urinary tract. Human Mutation, 2010, 31, 1352-1359.	1.1	54
141	A t(7;12) balanced translocation with breakpoints overlapping those of the Williamsâ€“Beuren and 12q14 microdeletion syndromes. American Journal of Medical Genetics, Part A, 2010, 152A, 1285-1294.	0.7	1
142	A fetus with ring chromosome 21 characterized by aCGH shows no clinical findings after birth. Prenatal Diagnosis, 2010, 30, 586-588.	1.1	7
143	Xp22.3 genomic deletions involving the CDKL5 gene in girls with early onset epileptic encephalopathy. Epilepsia, 2010, 51, 647-654.	2.6	60
144	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. European Journal of Human Genetics, 2010, 18, 163-170.	1.4	71

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145	High frequency of copy number imbalances in Rubinsteinâ€“Taybi patients negative to CREBBP mutational analysis. European Journal of Human Genetics, 2010, 18, 768-775.	1.4	13
146	Genotypeâ€“phenotype relationship in three cases with overlapping 19p13.12 microdeletions. European Journal of Human Genetics, 2010, 18, 1302-1309.	1.4	46
147	Bone osteoblastic and mesenchymal stromal cells lack primarily tumoral features in multiple myeloma patients. Leukemia, 2010, 24, 1368-1370.	3.3	8
148	Eyebrow anomalies as a diagnostic sign of genomic disorders. Clinical Genetics, 2010, 77, 28-31.	1.0	4
149	Refining the phenotype associated with MEF2C haploinsufficiency. Clinical Genetics, 2010, 78, 471-477.	1.0	85
150	Gene Copy Number Variation in Male Breast Cancer by aCGH. Analytical Cellular Pathology, 2010, 33, 113-119.	0.7	15
151	Sixteen New Cases Contributing to the Characterization of Patients with Distal 22q11.2 Microduplications. Molecular Syndromology, 2010, 1, 246-254.	0.3	31
152	In Vitro Expanded MSCs From Patients with Myeloprliferative Neoplasms at Late Passages Show Recurrent Cytogenetic Abnormalities. Blood, 2010, 116, 4101-4101.	0.6	1
153	Medullary sponge kidney associated with primary distal renal tubular acidosis and mutations of the H+-ATPase genes. Nephrology Dialysis Transplantation, 2009, 24, 2734-2738.	0.4	29
154	Presence of 1q gain and absence of 7p gain are new predictors of local or metastatic relapse in localized resectable neuroblastoma. Neuro-Oncology, 2009, 11, 192-200.	0.6	22
155	Epigenetic analysis of the critical region I for premature ovarian failure: demonstration of a highly heterochromatic domain on the long arm of the mammalian X chromosome. Journal of Medical Genetics, 2009, 46, 585-592.	1.5	33
156	Multiple joint dislocations: An additional skeletal finding in Lowryâ€“Wood syndrome?. American Journal of Medical Genetics, Part A, 2009, 149A, 737-741.	0.7	7
157	Search for genomic imbalances in a cohort of 20 patients with oralâ€“facialâ€“digital syndromes negative for mutations and large rearrangements in the OFD1 gene. American Journal of Medical Genetics, Part A, 2009, 149A, 1846-1849.	0.7	6
158	Different molecular mechanisms causing 9p21 deletions in acute lymphoblastic leukemia of childhood. Human Genetics, 2009, 126, 511-520.	1.8	39
159	Chromosomal microarray mapping suggests a role for BSX and Neurogranin in neurocognitive and behavioral defects in the 11q terminal deletion disorder (Jacobsen syndrome). Neurogenetics, 2009, 10, 89-95.	0.7	49
160	Subtelomeric FISH analysis in 76 patients with syndromic developmental delay/intellectual disability. Italian Journal of Pediatrics, 2009, 35, 9.	1.0	7
161	Xq28 duplication presenting with intestinal and bladder dysfunction and a distinctive facial appearance. European Journal of Human Genetics, 2009, 17, 434-443.	1.4	87
162	Mosaic 22q13 deletions: evidence for concurrent mosaic segmental isodisomy and gene conversion. European Journal of Human Genetics, 2009, 17, 426-433.	1.4	16

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