Orsetta Zuffardi

List of Publications by Year in descending order

333 papers	17,525 citations	13865 67 h-index	19747 117 g-index
336 all docs	336 docs citations	336 times ranked	20363 citing authors

#	Article	IF	Citations
1	Localization of factors controlling spermatogenesis in the nonfluorescent portion of the human y chromosome long arm. Human Genetics, 1976, 34, 119-124.	3.8	925
2	Human Bone Marrowâ€“Derived Mesenchymal Stem Cells Do Not Undergo Transformation after Long-termIn vitroCulture and Do Not Exhibit Telomere Maintenance Mechanisms. Cancer Research, 2007, 67, 9142-9149.	0.9	649
3	A dosage sensitive locus at chromosome Xp21 is involved in male to female sex reversal. Nature Genetics, 1994, 7, 497-501.	21.4	605
4	Formation of new chromatin domains determines pathogenicity of genomic duplications. Nature, 2016, 538, 265-269.	27.8	582
5	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	21.4	509
6	Constitutive Activation of PKA Catalytic Subunit in Adrenal Cushing's Syndrome. New England Journal of Medicine, 2014, 370, 1019-1028.	27.0	355
7	Olfactory Receptorâ€“Gene Clusters, Genomic-Inversion Polymorphisms, and Common Chromosome Rearrangements. American Journal of Human Genetics, 2001, 68, 874-883.	6.2	338
8	Disruption of the ProSAP2 Gene in a t(12;22)(q24.1;q13.3) Is Associated with the 22q13.3 Deletion Syndrome. American Journal of Human Genetics, 2001, 69, 261-268.	6.2	273
9	Multiple clinical forms of dehydrated hereditary stomatocytosis arise from mutations in PIEZO1. Blood, 2013, 121, 3925-3935.	1.4	266
10	Optimization of in vitro expansion of human multipotent mesenchymal stromal cells for cell-therapy approaches: Further insights in the search for a fetal calf serum substitute. Journal of Cellular Physiology, 2007, 211, 121-130.	4.1	258
11	A Human Homologue of the Drosophila melanogaster diaphanous Gene Is Disrupted in a Patient with Premature Ovarian Failure: Evidence for Conserved Function in Oogenesis and Implications for Human Sterility. American Journal of Human Genetics, 1998, 62, 533-541.	6.2	248
12	Cryptic deletions are a common finding in "balanced" reciprocal and complex chromosome rearrangements: a study of 59 patients. Journal of Medical Genetics, 2007, 44, 750-762.	3.2	244
13	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449.	21.4	207
14	Severe X-Linked Mitochondrial Encephalomyopathy Associated with a Mutation in Apoptosis-Inducing Factor. American Journal of Human Genetics, 2010, 86, 639-649.	6.2	199
15	The 11q;22q translocation: A European collaborative analysis of 43 cases. Human Genetics, 1980, 56, 21-51.	3.8	192
16	Clinical and molecular delineation of the 17q21.31 microdeletion syndrome. Journal of Medical Genetics, 2008, 45, 710-720.	3.2	191
17	The ?Cat Eye syndrome?: Dicentric small marker chromosome probably derived from a No. 22 (Tetrasomy 22pter?q11) associated with a characteristic phenotype. Human Genetics, 1981, 57, 148-58.	3.8	188
18	Heterozygous Submicroscopic Inversions Involving Olfactory Receptorâ€“Gene Clusters Mediate the Recurrent t(4;8)(p16;p23) Translocation. American Journal of Human Genetics, 2002, 71, 276-285.	6.2	185

#	Article	IF	Citations
19	Identification and mapping of human cDNAs homologous to Drosophila mutant genes through EST database searching. Nature Genetics, 1996, 13, 167-174.	21.4	177
20	Characterization of a recurrent 15q24 microdeletion syndrome. Human Molecular Genetics, 2007, 16, 567-572.	2.9	173
21	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173.	3.5	172
22	Assignment by deletion mapping of the steroid sulfatase X-linked ichthyosis locus to Xp223. Human Genetics, 1980, 54, 205-206.	3.8	155
23	Identification of a recurrent breakpoint within the SHANK3 gene in the 22q13.3 deletion syndrome. Journal of Medical Genetics, 2006, 43, 822-828.	3.2	155
24	Localization of DNA sequences required for human centromere function through an analysis of rearranged Y chromosomes. Nature Genetics, 1993, 5, 368-375.	21.4	149
25	Guidelines for molecular karyotyping in constitutional genetic diagnosis. European Journal of Human Genetics, 2007, 15, 1105-1114.	2.8	144
26	Twenty-one cases of blastic plasmacytoid dendritic cell neoplasm: focus on biallelic locus 9p21.3 deletion. Blood, 2011, 118, 4591-4594.	1.4	140
27	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.	2.9	140
28	Chromosome 20 Ring: A Chromosomal Disorder Associated with a Particular Electroclinical Pattern. Epilepsia, 1998, 39, 942-951.	5.1	137
29	Genomic inversions of human chromosome 15q11-q13 in mothers of Angelman syndrome patients with class II (BP2/3) deletions. Human Molecular Genetics, 2003, 12, 849-858.	2.9	131
30	Molecular Cloning, Expression Pattern, and Chromosomal Localization of the Human Naâ€“Cl Thiazide-Sensitive Cotransporter (SLC12A3). Genomics, 1996, 35, 486-493.	2.9	123
31	A deletion map of the human Yq11 region: Implications for the evolution of the Y chromosome and tentative mapping of a locus involved in spermatogenesis. Genomics, 1991, 11, 443-451.	2.9	121
32	PRKACB and Carney Complex. New England Journal of Medicine, 2014, 370, 1065-1067.	27.0	121
33	Overexpression of the C-type natriuretic peptide (CNP) is associated with overgrowth and bone anomalies in an individual with balanced t(2;7) translocation. Human Mutation, 2007, 28, 724-731.	2.5	118
34	The 11q;22q translocation: A collaborative study of 20 new cases and analysis of 110 families. Human Genetics, 1983, 64, 343-355.	3.8	115
35	Cryptic telomeric rearrangements in subjects with mental retardation associated with dysmorphism and congenital malformations. Journal of Medical Genetics, 2001, 38, 417-420.	3.2	114
36	Transmission of a Fully Functional Human Neocentromere through Three Generations. American Journal of Human Genetics, 1999, 64, 1440-1444.	6.2	113

#	Article	IF	Citations
37	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) Tj ETQq1 1 0.784314 rgBT /Over	2.8	109
38	Infantile Spasms Is Associated with Deletion of the MAGI2 Gene on Chromosome 7q11.23-q21.11. American Journal of Human Genetics, 2008, 83, 106-111.	6.2	108
39	Neocentromeres in 15q24-26 Map to Duplicons Which Flanked an Ancestral Centromere in 15q25. Genome Research, 2003, 13, 2059-2068.	5.5	107
40	Chromosomal rearrangements in Xq and premature ovarian failure: mapping of 25 new cases and review of the literature. Human Reproduction, 2006, 21, 1477-1483.	0.9	105
41	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.	2.8	104
42	Spleen endothelial cells from patients with myelofibrosis harbor the JAK2V617F mutation. Blood, 2013, 121, 360-368.	1.4	102
43	13q Deletion and central nervous system anomalies: further insights from karyotype-phenotype analyses of 14 patients. Journal of Medical Genetics, 2006, 44, e60-e60.	3.2	97
44	Identification of de novo mutations and rare variants in hypoplastic left heart syndrome. Clinical Genetics, 2012, 81, 542-554.	2.0	97
45	Turner syndrome patients are H-Y positive. Human Genetics, 1980, 54, 315-318.	3.8	96
46	Haploinsufficiency ofCOQ4causes coenzyme Q₁₀deficiency. Journal of Medical Genetics, 2012, 49, 187-191.	3.2	95
47	The cDNA sequence and chromosomal location of the human SOX2 gene. Mammalian Genome, 1994, 5, 640-642.	2.2	91
48	Agenesis of corpus callosum, ocular, and skeletal anomalies (X-linked dominant aicardi's syndrome) in a girl with balanced X/3 translocation. Human Genetics, 1982, 61, 364-8.	3.8	90
49	A New Submicroscopic Deletion That Refines the 9p Region for Sex Reversal. Genomics, 2000, 65, 203-212.	2.9	89
50	Clinical and molecular characteristics of 1qter microdeletion syndrome: delineating a critical region for corpus callosum agenesis/hypogenesis. Journal of Medical Genetics, 2008, 45, 346-354.	3.2	87
51	Xq28 duplication presenting with intestinal and bladder dysfunction and a distinctive facial appearance. European Journal of Human Genetics, 2009, 17, 434-443.	2.8	87
52	XX males SRY negative: a confirmed cause of infertility. Journal of Medical Genetics, 2011, 48, 710-712.	3.2	86
53	Colocalization of (TTAGGG)n telomeric sequences and ribosomal genes in Atlantic eels. Chromosome Research, 1995, 3, 54-58.	2.2	85
54	Duplications in addition to terminal deletions are present in a proportion of ring chromosomes: clues to the mechanisms of formation. Journal of Medical Genetics, 2007, 45, 147-154.	3.2	85

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55	Refining the phenotype associated with MEF2C haploinsufficiency. Clinical Genetics, 2010, 78, 471-477.	2.0	85
56	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	7.6	85
57	Chromosomal localization of mitochondrial transcription factor A (TCF6), single-stranded DNA-binding protein (SSBP), and Endonuclease G (ENDOG), three human housekeeping genes involved in mitochondrial biogenesis. Genomics, 1995, 25, 559-564.	2.9	83
58	Deletion of a 5-cM Region at Chromosome 8p23 Is Associated With a Spectrum of Congenital Heart Defects. Circulation, 2000, 102, 432-437.	1.6	83
59	Mutation analysis of two candidate genes for premature ovarian failure, DACH2 and POF1B. Human Reproduction, 2004, 19, 2759-2766.	0.9	82
60	Endothelial colony-forming cells from patients with chronic myeloproliferative disorders lack the disease-specific molecular clonality marker. Blood, 2009, 114, 3127-3130.	1.4	79
61	Evidence for interaction between human PRUNE and nm23-H1 NDPKinase. Oncogene, 1999, 18, 7244-7252.	5.9	77
62	Inter- and Intrachromosomal Rearrangements Are Both Involved in the Origin of 15q11-q13 Deletions in Prader-Willi Syndrome. American Journal of Human Genetics, 1997, 61, 228-231.	6.2	76
63	Preferential maternal derivation in inv dup(15). Human Genetics, 1981, 57, 345-350.	3.8	74
64	Detailed phenotypeâ€“genotype study in five patients with chromosome 6q16 deletion: narrowing the critical region for Praderâ€“Willi-like phenotype. European Journal of Human Genetics, 2008, 16, 1443-1449.	2.8	74
65	Presence of telomeric and subtelomeric sequences at the fusion points of ring chromosomes indicates that the ring syndrome is caused by ring instability. Human Genetics, 1993, 92, 23-27.	3.8	73
66	Mutations in MAP3K1 tilt the balance from SOX9/FGF9 to WNT/Î²-catenin signaling. Human Molecular Genetics, 2014, 23, 1073-1083.	2.9	72
67	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. European Journal of Human Genetics, 2010, 18, 163-170.	2.8	71
68	The mouse Mid1 gene: implications for the pathogenesis of Opitz syndrome and the evolution of the mammalian pseudoautosomal region. Human Molecular Genetics, 1998, 7, 489-499.	2.9	68
69	A 12Mb deletion at 7q33â€“q35 associated with autism spectrum disorders and primary amenorrhea. European Journal of Medical Genetics, 2008, 51, 631-638.	1.3	68
70	Inverted duplications are recurrent rearrangements always associated with a distal deletion: description of a new case involving 2q. European Journal of Human Genetics, 2000, 8, 597-603.	2.8	66
71	Assignment of the Human Carnitine Palmitoyltransferase II Gene (CPT1) to Chromosome 1p32. Genomics, 1994, 24, 195-197.	2.9	65
72	Inverted duplications deletions: underdiagnosed rearrangements??. Clinical Genetics, 2009, 75, 505-513.	2.0	64

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73	Improving molecular diagnosis in epilepsy by a dedicated high-throughput sequencing platform. European Journal of Human Genetics, 2015, 23, 354-362.	2.8	64
74	The phenotype of recurrent 10q22q23 deletions and duplications. European Journal of Human Genetics, 2011, 19, 400-408.	2.8	63
75	The introduction of arrays in prenatal diagnosis: A special challenge. Human Mutation, 2012, 33, 923-929.	2.5	63
76	Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322.	4.5	61
77	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
78	Complex Segmental Duplications Mediate a Recurrent dup(X)(p11.22-p11.23) Associated with Mental Retardation, Speech Delay, and EEG Anomalies in Males and Females. American Journal of Human Genetics, 2009, 85, 394-400.	6.2	60
79	Xp22.3 genomic deletions involving the CDKL5 gene in girls with early onset epileptic encephalopathy. Epilepsia, 2010, 51, 647-654.	5.1	60
80	MCM5: a new actor in the link between DNA replication and Meier-Gorlin syndrome. European Journal of Human Genetics, 2017, 25, 646-650.	2.8	60
81	Phenotypical/functional characterization of in vitro-expanded mesenchymal stromal cells from patients with Crohn's disease. Cytotherapy, 2009, 11, 825-836.	0.7	59
82	Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3. European Journal of Human Genetics, 2015, 23, 1025-1032.	2.8	59
83	Distinct transcriptional profiles characterize bone microenvironment mesenchymal cells rather than osteoblasts in relationship with multiple myeloma bone disease. Experimental Hematology, 2010, 38, 141-153.	0.4	57
84	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.	3.6	57
85	Duplication of the short arm of chromosome 9. Analysis of five cases. Human Genetics, 1982, 61, 3-7.	3.8	56
86	Characterization ofCxorf5(71-7A), a Novel Human cDNA Mapping to Xp22 and Encoding a Protein Containing Coiled-Coil Î±-Helical Domains. Genomics, 1998, 51, 243-250.	2.9	56
87	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	2.8	56
88	Reciprocal translocations: a trap for cytogenetists?. Human Genetics, 2005, 117, 571-582.	3.8	54
89	Mutations in SOX17 are associated with congenital anomalies of the kidney and the urinary tract. Human Mutation, 2010, 31, 1352-1359.	2.5	54
90	The Gene Encoding a Cationic Amino Acid Transporter (SLC7A4) Maps to the Region Deleted in the Velocardiofacial Syndrome. Genomics, 1998, 49, 230-236.	2.9	52

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91	Familial XX true hermaphroditism and the H-Y antigen. Human Genetics, 1979, 48, 45-52.	3.8	50
92	Current controversies in prenatal diagnosis 3: is conventional chromosome analysis necessary in the postâ€array CGH era?. Prenatal Diagnosis, 2011, 31, 235-243.	2.3	50
93	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.	1.7	50
94	Deficiency, transposition, and duplication of one 15q region may be alternatively associated with Prader-Willi (or a similar) syndrome. Analysis of seven cases after varying ascertainment. Human Genetics, 1983, 64, 388-394.	3.8	49
95	Order of Six Loci at 2q24-q31 and Orientation of the HOXD Locus. Genomics, 1994, 24, 34-40.	2.9	49
96	2q24â€“q31 Deletion: Report of a case and review of the literature. European Journal of Medical Genetics, 2007, 50, 21-32.	1.3	49
97	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.	1.4	49
98	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.	7.1	49
99	MCT8 Deficiency. Journal of Child Neurology, 2013, 28, 795-800.	1.4	48
100	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.	4.5	47
101	Gene dosage of the spermidine/spermine N1-acetyltransferase (SSAT) gene with putrescine accumulation in a patient with a Xp21.1p22.12 duplication and keratosis follicularis spinulosa decalvans (KFSD). Human Genetics, 2002, 111, 235-241.	3.8	46
102	Genotypeâ€“phenotype relationship in three cases with overlapping 19p13.12 microdeletions. European Journal of Human Genetics, 2010, 18, 1302-1309.	2.8	46
103	Evidence for an ancestral alphoid domain on the long arm of human chromosome 2. Human Genetics, 1992, 89, 247-9.	3.8	45
104	A 2.3â€‰Mb duplication of chromosome 8q24.3 associated with severe mental retardation and epilepsy detected by standard karyotype. European Journal of Human Genetics, 2005, 13, 586-591.	2.8	45
105	TREX1 C-terminal frameshift mutations in the systemic variant of retinal vasculopathy with cerebral leukodystrophy. Neurological Sciences, 2015, 36, 323-330.	1.9	45
106	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	2.4	45
107	A susceptibility gene for premature ovarian failure (POF) maps to proximal Xq28. European Journal of Human Genetics, 2004, 12, 829-834.	2.8	44
108	Ring chromosome 12 and latent centromeres. Cytogenetic and Genome Research, 1980, 28, 151-157.	1.1	43

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109	Indirect immunofluorescence of inactive centromeres as indicator of centromeric function. Human Genetics, 1986, 73, 12-16.	3.8	43
110	Nullisomy for the distal portion of Xp in a male child with a X/Y translocation. Human Genetics, 1977, 39, 277-281.	3.8	42
111	Functional disomy of Xp22-pter in three males carrying a portion of Xp translocated to Yq. Human Genetics, 1993, 91, 333-8.	3.8	42
112	D8S7 is consistently deleted in inverted duplications of the short arm of chromosome 8 (inv dup 8p). Human Genetics, 1993, 92, 391-396.	3.8	42
113	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.	3.2	42
114	Primary coenzyme Q10 deficiency presenting as fatal neonatal multiorgan failure. European Journal of Human Genetics, 2015, 23, 1254-1258.	2.8	42
115	Two classes of low-copy repeats comediate a new recurrent rearrangement consisting of duplication at 8p23.1 and triplication at 8p23.2. Human Mutation, 2007, 28, 459-468.	2.5	41
116	Dysmorphic features, simplified gyral pattern and 7q11.23 duplication reciprocal to the Williams-Beuren deletion. European Journal of Human Genetics, 2008, 16, 880-887.	2.8	41
117	A new chromosome instability disorder. Clinical Genetics, 1986, 30, 353-365.	2.0	41
118	The role of Yp in sex determination: New evidence from X/Y translocations. American Journal of Medical Genetics Part A, 1982, 12, 175-184.	2.4	40
119	Human NRD Convertase: A Highly Conserved Metalloendopeptidase Expressed at Specific Sites during Development and in Adult Tissues. Genomics, 1998, 47, 238-245.	2.9	40
120	Identification and Characterization of CDS2, a Mammalian Homolog of theDrosophilaCDP-diacylglycerol Synthase Gene. Genomics, 1999, 55, 68-77.	2.9	40
121	Deletion of PTEN and BMPR1A on Chromosome 10q23 Is Not Always Associated with Juvenile Polyposis of Infancy. American Journal of Human Genetics, 2006, 79, 593-596.	6.2	40
122	Contiguous gene deletions involving EFNB1, OPHN1, PJA1 and EDA in patients with craniofrontonasal syndrome. Clinical Genetics, 2007, 72, 506-516.	2.0	40
123	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.	2.8	40
124	Mapping of a human centromere onto the DNA by topoisomerase II cleavage. EMBO Reports, 2000, 1, 489-493.	4.5	39
125	Cortical dysplasia of the left temporal lobe might explain severe expressive-language delay in patients with duplication of the Williamsâ€“Beuren locus. European Journal of Human Genetics, 2007, 15, 62-67.	2.8	39
126	Different molecular mechanisms causing 9p21 deletions in acute lymphoblastic leukemia of childhood. Human Genetics, 2009, 126, 511-520.	3.8	39

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127	Sox9 Duplications Are a Relevant Cause of Sry-Negative XX Sex Reversal Dogs. PLoS ONE, 2014, 9, e101244.	2.5	39
128	A familial inverted duplication/deletion of 2p25.1â€“25.3 provides new clues on the genesis of inverted duplications. European Journal of Human Genetics, 2009, 17, 179-186.	2.8	37
129	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.	1.4	37
130	Gene mapping and serendipity. The locus for torticollis, keloids, cryptorchidism and renal dysplasia (31430, McKusick) is at Xq28, distal to the G6PD locus. Human Genetics, 1982, 62, 280-281.	3.8	35
131	Epigenetic control of the critical region for premature ovarian failure on autosomal genes translocated to the X chromosome: a hypothesis. Human Genetics, 2007, 121, 441-450.	3.8	35
132	Contiguous gene syndrome due to an interstitial deletion in Xp22.3 in a boy with ichthyosis, chondrodysplasia punctata, mental retardation and ADHD. European Journal of Medical Genetics, 2007, 50, 301-308.	1.3	34
133	Wolfram syndrome 2: a novel CISD2 mutation identified in Italian siblings. Acta Diabetologica, 2015, 52, 175-178.	2.5	34
134	Inverted duplications: how many of them are mosaic?. European Journal of Human Genetics, 2004, 12, 713-717.	2.8	33
135	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.	3.2	33
136	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.	2.8	33
137	Small supernumerary marker chromosomes: A legacy of trisomy rescue?. Human Mutation, 2019, 40, 193-200.	2.5	33
138	Microphthalmia with linear skin defects (MLS) syndrome: Clinical, cytogenetic, and molecular characterization of 11 cases. American Journal of Medical Genetics, Part A, 2005, 137A, 190-198.	1.2	32
139	Presence of H-Y antigen in female patients with sex-chromosome mosaics and absence of testicular tissue. American Journal of Medical Genetics Part A, 1983, 15, 315-321.	2.4	31
140	Sixteen New Cases Contributing to the Characterization of Patients with Distal 22q11.2 Microduplications. Molecular Syndromology, 2010, 1, 246-254.	0.8	31
141	Clinical and Molecular Characteristics of SLC16A2 (MCT8) Mutations in Three Families with the Allan-Herndon-Dudley Syndrome. Human Mutation, 2017, 38, 260-264.	2.5	31
142	Genomic organization and chromosomal localization of the mouse Connexin36 (mCx36) gene. Gene, 2000, 251, 123-130.	2.2	30
143	ZPLD1 gene is disrupted in a patient with balanced translocation that exhibits cerebral cavernous malformations. Neuroscience, 2008, 155, 345-349.	2.3	30
144	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.	1.3	30

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145	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.	7.2	30
146	Medullary sponge kidney associated with primary distal renal tubular acidosis and mutations of the H+-ATPase genes. Nephrology Dialysis Transplantation, 2009, 24, 2734-2738.	0.7	29
147	Periventricular heterotopia with white matter abnormalities associated with 6p25 deletion. American Journal of Medical Genetics, Part A, 2012, 158A, 1793-1797.	1.2	29
148	Trisomy 16q21ï¿½qter. Human Genetics, 1980, 53, 165-7.	3.8	28
149	Identification and molecular modelling of a novel familial mutation in the SRY gene implicated in the pure gonadal dysgenesis. European Journal of Human Genetics, 2007, 15, 76-80.	2.8	28
150	Loss-of-function mutation of the AF9/MLLT3 gene in a girl with neuromotor development delay, cerebellar ataxia, and epilepsy. Human Genetics, 2005, 118, 76-81.	3.8	27
151	Inversion polymorphisms and non-contiguous terminal deletions: the cause and the (unpredicted) effect of our genome architecture. Journal of Medical Genetics, 2005, 43, e19-e19.	3.2	27
152	Dup(3)(p2â†'pter) in two families, including one infant with cyclopia. American Journal of Medical Genetics Part A, 1985, 20, 341-348.	2.4	26
153	CENP-G in neocentromeres and inactive centromeres. Chromosoma, 2000, 109, 328-333.	2.2	26
154	Breakpoint determination of 15 large deletions in Peutzâ€"Jeghers subjects. Human Genetics, 2010, 128, 373-382.	3.8	26
155	Neonatal suppressionâ€burst without epileptic seizures: expanding the electroclinical phenotype of STXBP1â€related, earlyâ€onset encephalopathy. Epileptic Disorders, 2013, 15, 55-61.	1.3	26
156	PIEZO1 Hypomorphic Variants in Congenital Lymphatic Dysplasia Cause Shape and Hydration Alterations of Red Blood Cells. Frontiers in Physiology, 2019, 10, 258.	2.8	26
157	?Jumping? satellites in three generations: A warning for paternity tests and prenatal diagnosis. Human Genetics, 1976, 34, 315-318.	3.8	25
158	Ataxic gait and mental retardation with absence of the paternal chromosome 8 and an idic(8)(p23.3): imprinting effect or nullisomy for distal 8p genes?. Human Genetics, 1997, 99, 766-771.	3.8	25
159	Multiple congenital anomalies, brain hypomyelination, and ocular albinism in a female with dup(X)(pterâ†'q24::q21.32â†'qter) and random X inactivation. American Journal of Medical Genetics Part A, 1997, 72, 329-334.	2.4	25
160	HumanFIGF:Cloning, Gene Structure, and Mapping to Chromosome Xp22.1 between thePIGAand theGRPRGenes. Genomics, 1998, 47, 207-216.	2.9	25
161	Cryptic t(1;12)(q44;p13.3) translocation in a previously described syndrome with polymicrogyria, segregating as an apparently X-linked trait. American Journal of Medical Genetics Part A, 2003, 117A, 65-71.	2.4	25
162	A locus for familial skewed X chromosome inactivation maps to chromosome Xq25 in a family with a female manifesting Lowe syndrome. Journal of Human Genetics, 2006, 51, 1030-1036.	2.3	25

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326	Familial translocation t(3;10) (p26.3;p12.31) leading to trisomy 10p12.31â€‰â†’â€‰pter and monosomy 3p26.3â€‰â†’â€‰pter in seven members. American Journal of Medical Genetics, Part A, 2008, 146A, 3242-3245.	1.2	0
327	De novo 15.5-Mb Interstitial Deletion in 5p in a Male Ascertained by Oligospermia. Molecular Syndromology, 2013, 4, 250-254.	0.8	0
328	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, , .	0.3	0
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330	Platelet-Lysate for In Vitro Expansion of Human Multipotent Mesenchymal Stromal Cells in Approaches of Cell-Therapy.. Blood, 2006, 108, 2577-2577.	1.4	0
331	Are the Myeloma Bone Microevironment Cells Tumoral or Not?.. Blood, 2009, 114, 1816-1816.	1.4	0
332	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.	1.4	0
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