Pawel Stankiewicz

List of Publications by Year
in descending order

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121 papers	10,639 citations	36303 51 h-index	33894 99 g-index
145 all docs	145 docs citations	145 times ranked	13817 citing authors

#	Article	IF	Citations
1	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
2	Phenotypic expansion of the <scp>BPTF</scp>â€related neurodevelopmental disorder with dysmorphic facies and distal limb anomalies. American Journal of Medical Genetics, Part A, 2021, 185, 1366-1378.	1.2	8
3	Variants in FLRT3 and SLC35E2B identified using exome sequencing in seven high myopia families from Central Europe. Advances in Medical Sciences, 2021, 66, 192-198.	2.1	5
4	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	6.2	15
5	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. Expert Review of Molecular Diagnostics, 2020, 20, 995-1002.	3.1	14
6	Parental somatic mosaicism for CNV deletions â€“ A need for more sensitive and precise detection methods in clinical diagnostics settings. Genomics, 2020, 112, 2937-2941.	2.9	14
7	Highly Sensitive Blocker Displacement Amplification and Droplet Digital PCR Reveal Low-Level Parental FOXF1 Somatic Mosaicism in Families with Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins. Journal of Molecular Diagnostics, 2020, 22, 447-456.	2.8	13
8	Disruption of normal patterns of FOXF1 expression in a lethal disorder of lung development. Journal of Medical Genetics, 2020, 57, 296-300.	3.2	7
9	The S52F FOXF1 Mutation Inhibits STAT3 Signaling and Causes Alveolar Capillary Dysplasia. American Journal of Respiratory and Critical Care Medicine, 2019, 200, 1045-1056.	5.6	51
10	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	8.2	55
11	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
12	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	8.2	42
13	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25.	8.2	22
14	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
15	Rare copy number variants contribute pathogenic alleles in patients with intestinal malrotation. Molecular Genetics & Genomic Medicine, 2019, 7, e549.	1.2	12
16	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. Cell, 2018, 172, 924-936.e11.	28.9	103
17	An estimation of the prevalence of genomic disorders using chromosomal microarray data. Journal of Human Genetics, 2018, 63, 795-801.	2.3	49
18	Infants with Atypical Presentations of Alveolar Capillary Dysplasia with Misalignment of the Pulmonary Veins Who Underwent Bilateral Lung Transplantation. Journal of Pediatrics, 2018, 194, 158-164.e1.	1.8	48

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19	Predicting human genes susceptible to genomic instability associated with Alu/Alu-mediated rearrangements. Genome Research, 2018, 28, 1228-1242.	5.5	74
20	Characterization of chromosomal abnormalities in pregnancy losses reveals critical genes and loci for human early development. Human Mutation, 2017, 38, 669-677.	2.5	28
21	Variants in SKP1, PROB1, and IL17B genes at keratoconus 5q31.1â€“q35.3 susceptibility locus identified by whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 73-78.	2.8	19
22	Evidence against <scp>ZNF</scp>469 being causative for keratoconus in Polish patients. Acta Ophthalmologica, 2016, 94, 289-294.	1.1	20
23	Complex translocation disrupting TCF4 and altering TCF4 isoform expression segregates as mild autosomal dominant intellectual disability. Orphanet Journal of Rare Diseases, 2016, 11, 62.	2.7	35
24	CAV3 mutation in a patient with transient hyperCKemia and myalgia. Neurologia I Neurochirurgia Polska, 2016, 50, 468-473.	1.2	8
25	Prenatal Diagnosis of Alveolar Capillary Dysplasia with Misalignment ofÂPulmonary Veins. Journal of Pediatrics, 2016, 170, 317-318.	1.8	17
26	The complex behavioral phenotype of 15q13.3 microdeletion syndrome. Genetics in Medicine, 2016, 18, 1111-1118.	2.4	45
27	Coâ€segregation of Freiberg's infraction with a familial translocation t(5;7)(p13.3;p22.2) ascertained by a child with cri du chat syndrome and brachydactyly type A1B. American Journal of Medical Genetics, Part A, 2015, 167, 445-449.	1.2	1
28	IncreasedSTAG2dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Human Molecular Genetics, 2015, 24, 7171-7181.	2.9	28
29	Somatic mosaicism: implications for disease and transmission genetics. Trends in Genetics, 2015, 31, 382-392.	6.7	234
30	Absence of Heterozygosity Due to Template Switching during Replicative Rearrangements. American Journal of Human Genetics, 2015, 96, 555-564.	6.2	45
31	Assessing structural variation in a personal genomeâ€”towards a human reference diploid genome. BMC Genomics, 2015, 16, 286.	2.8	153
32	Copy number variants in patients with intellectual disability affect the regulation of ARX transcription factor gene. Human Genetics, 2015, 134, 1163-1182.	3.8	14
33	Delineation of candidate genes responsible for structural brain abnormalities in patients with terminal deletions of chromosome 6q27. European Journal of Human Genetics, 2015, 23, 54-60.	2.8	45
34	Genomic and Epigenetic Complexity of the FOXF1 Locus in 16q24.1: Implications for Development and Disease. Current Genomics, 2015, 16, 107-116.	1.6	51
35	Molecular and clinical analyses of 16q24.1 duplications involving FOXF1 identify an evolutionarily unstable large minisatellite. BMC Medical Genetics, 2014, 15, 128.	2.1	11
36	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87.	2.8	112

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37	CHRNA7 triplication associated with cognitive impairment and neuropsychiatric phenotypes in a three-generation pedigree. European Journal of Human Genetics, 2014, 22, 1071-1076.	2.8	37
38	The Alu-Rich Genomic Architecture of SPAST Predisposes to Diverse and Functionally Distinct Disease-Associated CNV Alleles. American Journal of Human Genetics, 2014, 95, 143-161.	6.2	87
39	Somatic mosaicism detected by exon-targeted, high-resolution aCGH in 10â€‰362 consecutive cases. European Journal of Human Genetics, 2014, 22, 969-978.	2.8	51
40	Comparative Analyses of Lung Transcriptomes in Patients with Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins and in Foxf1 Heterozygous Knockout Mice. PLoS ONE, 2014, 9, e94390.	2.5	31
41	SOX12 and NRSN2 are candidate genes for 20p13 subtelomeric deletions associated with developmental delay. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 832-840.	1.7	15
42	Expanding the genotypeâ€“phenotype correlation in subtelomeric 19p13.3 microdeletions using high resolution clinical chromosomal microarray analysis. American Journal of Medical Genetics, Part A, 2013, 161, 2953-2963.	1.2	25
43	Rare DNA copy number variants in cardiovascular malformations with extracardiac abnormalities. European Journal of Human Genetics, 2013, 21, 173-181.	2.8	49
44	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	6.2	43
45	Comparison of chromosome analysis and chromosomal microarray analysis: what is the value of chromosome analysis in todayâ€™s genomic array era?. Genetics in Medicine, 2013, 15, 450-457.	2.4	63
46	Detection of copy-number variation in AUTS2 gene by targeted exonic array CGH in patients with developmental delay and autistic spectrum disorders. European Journal of Human Genetics, 2013, 21, 343-346.	2.8	56
47	Intragenic deletions of the IGF1 receptor gene in five individuals with psychiatric phenotypes and developmental delay. European Journal of Human Genetics, 2013, 21, 1304-1307.	2.8	8
48	A familial case of alveolar capillary dysplasia with misalignment of pulmonary veins supports paternal imprinting of FOXF1 in human. European Journal of Human Genetics, 2013, 21, 474-477.	2.8	42
49	NovelFOXF1Mutations in Sporadic and Familial Cases of Alveolar Capillary Dysplasia with Misaligned Pulmonary Veins Imply a Role for its DNA Binding Domain. Human Mutation, 2013, 34, 801-811.	2.5	97
50	Incidental copy-number variants identified by routine genome testing in a clinical population. Genetics in Medicine, 2013, 15, 45-54.	2.4	37
51	NAHR-mediated copy-number variants in a clinical population: Mechanistic insights into both genomic disorders and Mendelizing traits. Genome Research, 2013, 23, 1395-1409.	5.5	120
52	Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. Genome Research, 2013, 23, 1383-1394.	5.5	62
53	Early recurrence in standard-risk medulloblastoma patients with the common idic(17)(p11.2) rearrangement. Neuro-Oncology, 2012, 14, 831-840.	1.2	13
54	Small rare recurrent deletions and reciprocal duplications in 2q21.1, including brain-specific ARHGEF4 and GPR148. Human Molecular Genetics, 2012, 21, 3345-3355.	2.9	22

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55	Novel 9q34.11 gene deletions encompassing combinations of four Mendelian disease genes: STXBP1, SPTAN1, ENG, and TOR1A. Genetics in Medicine, 2012, 14, 868-876.	2.4	51
56	Deletions in chromosome 6p22.3-p24.3, including ATXN1, are associated with developmental delay and autism spectrum disorders. Molecular Cytogenetics, 2012, 5, 17.	0.9	43
57	Co-occurrence of recurrent duplications of the DiGeorge syndrome region on both chromosome 22 homologues due to inherited and de novo events. Journal of Medical Genetics, 2012, 49, 681-688.	3.2	11
58	Small genomic rearrangements involving FMR1 support the importance of its gene dosage for normal neurocognitive function. Neurogenetics, 2012, 13, 333-339.	1.4	21
59	Delineation of a deletion region critical for corpus callosal abnormalities in chromosome 1q43â€“q44. European Journal of Human Genetics, 2012, 20, 176-179.	2.8	42
60	Detection of â‰¥1â€‰Mb microdeletions and microduplications in a single cell using custom oligonucleotide arrays. Prenatal Diagnosis, 2012, 32, 10-20.	2.3	29
61	Phenotypic spectrum and genotypeâ€“phenotype correlations of NRXN1 exon deletions. European Journal of Human Genetics, 2012, 20, 1240-1247.	2.8	99
62	Int22h-1/int22h-2-mediated Xq28 rearrangements: intellectual disability associated with duplications and in utero male lethality with deletions. Journal of Medical Genetics, 2011, 48, 840-850.	3.2	43
63	Efficient Multiple Samples aCGH Analysis for Rare CNVs Detection. , 2011, , .		0
64	16q24.1 microdeletion in a premature newborn: Usefulness of array-based comparative genomic hybridization in persistent pulmonary hypertension of the newborn. Pediatric Critical Care Medicine, 2011, 12, e427-e432.	0.5	21
65	Disruption of the SCN2A and SCN3A genes in a patient with mental retardation, neurobehavioral and psychiatric abnormalities, and a history of infantile seizures. Clinical Genetics, 2011, 80, 191-195.	2.0	24
66	Exon deletions of the EP300 and CREBBP genes in two children with Rubinsteinâ€“Taybi syndrome detected by aCGH. European Journal of Human Genetics, 2011, 19, 43-49.	2.8	54
67	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
68	Phenotypic manifestations of copy number variation in chromosome 16p13.11. European Journal of Human Genetics, 2011, 19, 280-286.	2.8	97
69	The phenotype of recurrent 10q22q23 deletions and duplications. European Journal of Human Genetics, 2011, 19, 400-408.	2.8	63
70	Recurrent partial rhombencephalosynapsis and holoprosencephaly in siblings with a mutation of ZIC2. American Journal of Medical Genetics, Part A, 2011, 155, 1574-1580.	1.2	17
71	Complex genomic rearrangement of chromosome 16p13.3 detected by array comparative genomic hybridization in a patient with multiple congenital anomalies, dysmorphic craniofacial features, and developmental delay. American Journal of Medical Genetics, Part A, 2011, 155, 2589-2592.	1.2	1
72	Alveolar Capillary Dysplasia. American Journal of Respiratory and Critical Care Medicine, 2011, 184, 172-179.	5.6	194

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73	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. Human Molecular Genetics, 2011, 20, 1975-1988.	2.9	74
74	Use of array CGH to detect exonic copy number variants throughout the genome in autism families detects a novel deletion in TMLHE. Human Molecular Genetics, 2011, 20, 4360-4370.	2.9	101
75	Identification of a Recurrent Microdeletion at 17q23.1q23.2 Flanked by Segmental Duplications Associated with Heart Defects and Limb Abnormalities. American Journal of Human Genetics, 2010, 86, 454-461.	6.2	85
76	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. Human Mutation, 2010, 31, 840-850.	2.5	111
77	Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342.	2.5	225
78	Challenges in clinical interpretation of microduplications detected by array CGH analysis. American Journal of Medical Genetics, Part A, 2010, 152A, 1089-1100.	1.2	35
79	Insertional translocation detected using FISH confirmation of arrayâ€comparative genomic hybridization (aCGH) results. American Journal of Medical Genetics, Part A, 2010, 152A, 1111-1126.	1.2	85
80	HERVâ€mediated genomic rearrangement of EYA1 in an individual with branchioâ€otoâ€renal syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2854-2860.	1.2	32
81	A syndrome of short stature, microcephaly and speech delay is associated with duplications reciprocal to the common Sotos syndrome deletion. European Journal of Human Genetics, 2010, 18, 258-261.	2.8	41
82	Clinical spectrum associated with recurrent genomic rearrangements in chromosome 17q12. European Journal of Human Genetics, 2010, 18, 278-284.	2.8	114
83	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. Journal of Medical Genetics, 2010, 47, 332-341.	3.2	447
84	Whole-Genome Sequencing in a Patient with Charcotâ€“Marieâ€“Tooth Neuropathy. New England Journal of Medicine, 2010, 362, 1181-1191.	27.0	698
85	Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. Human Molecular Genetics, 2009, 18, 2188-2203.	2.9	165
86	Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. Human Molecular Genetics, 2009, 18, 3579-3593.	2.9	143
87	Redefined genomic architecture in 15q24 directed by patient deletion/duplication breakpoint mapping. Human Genetics, 2009, 126, 589-602.	3.8	65
88	Alu-specific microhomology-mediated deletions in CDKL5 in females with early-onset seizure disorder. Neurogenetics, 2009, 10, 363-369.	1.4	44
89	Interstitial deletion of 6q25.2â€“q25.3: a novel microdeletion syndrome associated with microcephaly, developmental delay, dysmorphic features and hearing loss. European Journal of Human Genetics, 2009, 17, 573-581.	2.8	45
90	A small recurrent deletion within 15q13.3 is associated with a range of neurodevelopmental phenotypes. Nature Genetics, 2009, 41, 1269-1271.	21.4	171

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91	Mosaicism for r(X) and der(X)del(X)(p11.23)dup(X)(p11.21p11.22) Provides Insight into the Possible Mechanism of Rearrangement. Molecular Cytogenetics, 2008, 1, 16.	0.9	12
92	Branchiootorenal syndrome and oculoauriculovertebral spectrum features associated with duplication ofSIX1,SIX6, andOTX2resulting from a complex chromosomal rearrangement. American Journal of Medical Genetics, Part A, 2008, 146A, 2480-2489.	1.2	42
93	Identification of chromosome abnormalities in subtelomeric regions by microarray analysis: A study of 5,380 cases. American Journal of Medical Genetics, Part A, 2008, 146A, 2242-2251.	1.2	113
94	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. Nature Genetics, 2008, 40, 1466-1471.	21.4	535
95	Genomic Imbalances in Neonates With Birth Defects: High Detection Rates by Using Chromosomal Microarray Analysis. Pediatrics, 2008, 122, 1310-1318.	2.1	137
96	Hominoid lineage specific amplification of low-copy repeats on 22q11.2 (LCR22s) associated with velo-cardio-facial/digeorge syndrome. Human Molecular Genetics, 2007, 16, 2560-2571.	2.9	32
97	SOX9cre1, a cis-acting regulatory element located 1.1ÂMb upstream of SOX9, mediates its enhancement through the SHH pathway. Human Molecular Genetics, 2007, 16, 1143-1156.	2.9	68
98	Use of array CGH in the evaluation of dysmorphology, malformations, developmental delay, and idiopathic mental retardation. Current Opinion in Genetics and Development, 2007, 17, 182-192.	3.3	293
99	Characterization of Potocki-Lupski Syndrome (dup(17)(p11.2p11.2)) and Delineation of a Dosage-Sensitive Critical Interval That Can Convey an Autism Phenotype. American Journal of Human Genetics, 2007, 80, 633-649.	6.2	340
100	Clinical Implementation of Chromosomal Microarray Analysis: Summary of 2513 Postnatal Cases. PLoS ONE, 2007, 2, e327.	2.5	191
101	Ovotestes and XY sex reversal in a female with an interstitial 9q33.3â€q34.1 deletion encompassing NR5A1 and LMX1B causing features of genitopatellar syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1071-1081.	1.2	43
102	Microarrayâ€based CGH detects chromosomal mosaicism not revealed by conventional cytogenetics. American Journal of Medical Genetics, Part A, 2007, 143A, 1679-1686.	1.2	158
103	Male-to-female sex reversal associated with an â¼250Âkb deletion upstream of NR0B1 (DAX1). Human Genetics, 2007, 122, 63-70.	3.8	59
104	Prenatal diagnosis of chromosomal abnormalities using array-based comparative genomic hybridization. Genetics in Medicine, 2006, 8, 719-727.	2.4	154
105	DNA sequence of human chromosome 17 and analysis of rearrangement in the human lineage. Nature, 2006, 440, 1045-1049.	27.8	130
106	Role of genomic architecture in PLP1 duplication causing Pelizaeus-Merzbacher disease. Human Molecular Genetics, 2006, 15, 2250-2265.	2.9	73
107	Smith-Magenis Syndrome Deletion, Reciprocal Duplication dup(17)(p11.2p11.2), and Other Proximal 17p Rearrangements. , 2006, , 179-191.		1
108	Position Effects. , 2006, , 357-369.		2

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109	Cryptic unbalanced translocation t(17;18)(p13.2;q22.3) identified by subtelomeric FISH and defined by array-based comparative genomic hybridization in a patient with mental retardation and dysmorphic features. American Journal of Medical Genetics, Part A, 2005, 137A, 88-93.	1.2	10
110	Trisomy 17p10-p12 due to mosaic supernumerary marker chromosome: Delineation of molecular breakpoints and clinical phenotype, and comparison to other proximal 17p segmental duplications. American Journal of Medical Genetics, Part A, 2005, 138A, 175-180.	1.2	20
111	Molecular cytogenetic characterization of a familial der(1)del(1)(p36.33)dup(1)(p36.33p36.22) with variable phenotype. American Journal of Medical Genetics, Part A, 2005, 139A, 136-140.	1.2	17
112	Genomic Disorders: Molecular Mechanisms for Rearrangements and Conveyed Phenotypes. PLoS Genetics, 2005, 1, e49.	3.5	496
113	Sotos syndrome common deletion is mediated by directly oriented subunits within inverted Sos-REP low-copy repeats. Human Molecular Genetics, 2005, 14, 535-542.	2.9	64
114	Development and validation of a CGH microarray for clinical cytogenetic diagnosis. Genetics in Medicine, 2005, 7, 422-432.	2.4	241
115	Position Effects Due to Chromosome Breakpoints that Map â^¼900 Kb Upstream and â^¼1.3 Mb Downstream of SOX9 in Two Patients with Campomelic Dysplasia. American Journal of Human Genetics, 2005, 76, 652-662.	6.2	178
116	Small marker chromosomes in two patients with segmental aneusomy for proximal 17p. Human Genetics, 2004, 115, 1-7.	3.8	24
117	A girl with duplication 17p10-p12 associated with a dicentric chromosome. American Journal of Medical Genetics Part A, 2004, 124A, 173-178.	2.4	9
118	The Breakpoint Region of the Most Common Isochromosome, i(17q), in Human Neoplasia Is Characterized by a Complex Genomic Architecture with Large, Palindromic, Low-Copy Repeats. American Journal of Human Genetics, 2004, 74, 1-10.	6.2	122
119	Variability in clinical phenotype despite common chromosomal deletion in Smith-Magenis syndrome [del(17)(p11.2p11.2)]. Genetics in Medicine, 2003, 5, 430-434.	2.4	104
120	Molecular-evolutionary mechanisms for genomic disorders. Current Opinion in Genetics and Development, 2002, 12, 312-319.	3.3	151
121	Genome architecture, rearrangements and genomic disorders. Trends in Genetics, 2002, 18, 74-82.	6.7	815