BartÅ‚omiej Budny

List of Publications by Year
in descending order

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38	1,050	567281 15	434195 31
papers	citations	h-index	g-index

39	39	39	2372
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. Molecular Psychiatry, 2016, 21, 133-148.	7.9	243
2	OFD1 Is Mutated in X-Linked Joubert Syndrome and Interacts with LCA5-Encoded Lebercilin. American Journal of Human Genetics, 2009, 85, 465-481.	6.2	180
3	A novel X-linked recessive mental retardation syndrome comprising macrocephaly and ciliary dysfunction is allelic to oralâ€“facialâ€“digital type I syndrome. Human Genetics, 2006, 120, 171-178.	3.8	166
4	A novel nonsense mutation in CUL4B gene in three brothers with Xâ€linked mental retardation syndrome. Clinical Genetics, 2010, 77, 141-144.	2.0	47
5	Novel missense mutations in the ubiquitinationâ€related gene UBE2A cause a recognizable Xâ€linked mental retardation syndrome. Clinical Genetics, 2010, 77, 541-551.	2.0	45
6	FOXE1 Polyalanine Tract Length Polymorphism in Patients with Thyroid Hemiagenesis and Subjects with Normal Thyroid. Hormone Research in Paediatrics, 2011, 75, 329-334.	1.8	30
7	The c.470ÂTâ€‰>â€‰C CHEK2 missense variant increases the risk of differentiated thyroid carcinoma in the Great Poland population. Hereditary Cancer in Clinical Practice, 2015, 13, 8.	1.5	30
8	The Role of Serum C-Reactive Protein Measured by High-Sensitive Method in Thyroid Disease. Archivum Immunologiae Et Therapiae Experimentalis, 2014, 62, 501-509.	2.3	29
9	IncreasedSTAG2dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Human Molecular Genetics, 2015, 24, 7171-7181.	2.9	28
10	Evaluation of 167 Gene Expression Classifier (GEC) and ThyroSeq v2 Diagnostic Accuracy in the Preoperative Assessment of Indeterminate Thyroid Nodules: Bivariate/HROC Meta-analysis. Endocrine Pathology, 2019, 30, 8-15.	9.0	27
11	Differences in Mutational Profile between Follicular Thyroid Carcinoma and Follicular Thyroid Adenoma Identified Using Next Generation Sequencing. International Journal of Molecular Sciences, 2019, 20, 3126.	4.1	25
12	Modifying impact of RET gene haplotypes on medullary thyroid carcinoma clinical course. Endocrine-Related Cancer, 2018, 25, 421-436.	3.1	20
13	PAX63â€² deletion in a family with aniridia. Ophthalmic Genetics, 2012, 33, 44-48.	1.2	18
14	Mutations in proteasome-related genes are associated with thyroid hemiagenesis. Endocrine, 2017, 56, 279-285.	2.3	18
15	Head and Neck Paragangliomasâ€”A Genetic Overview. International Journal of Molecular Sciences, 2020, 21, 7669.	4.1	17
16	A novelGJA1 missense mutation in a Polish child with oculodentodigital dysplasia. Journal of Applied Genetics, 2009, 50, 297-299.	1.9	15
17	SEMA3A and IGSF10 Are Novel Contributors to Combined Pituitary Hormone Deficiency (CPHD). Frontiers in Endocrinology, 2020, 11, 368.	3.5	13
18	Alternative 3' acceptor site in the exon 2 of human PAX8 gene resulting in the expression of unknown mRNA variant found in thyroid hemiagenesis and some types of cancers.. Acta Biochimica Polonica, 2013, 60, .	0.5	13

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19	Nicotinamide phosphoribosyltransferase leukocyte overexpression in Gravesâ€™ opthalmopathy. Endocrine, 2016, 53, 497-504.	2.3	12
20	Two coexisting heterozygous frameshift mutations in PROP1 are responsible for a different phenotype of combined pituitary hormone deficiency. Journal of Applied Genetics, 2016, 57, 373-381.	1.9	7
21	Genomic markers of ovarian adenocarcinoma and its relevancy to the effectiveness of chemotherapy. Oncology Letters, 2017, 14, 3401-3414.	1.8	7
22	Copy Number Variants Contributing to Combined Pituitary Hormone Deficiency. International Journal of Molecular Sciences, 2020, 21, 5757.	4.1	7
23	The genetic heterogeneity of indeterminate thyroid nodules assessed preoperatively with next generation sequencing reflects the diversity of the final histopathological diagnoses. Polish Archives of Internal Medicine, 2019, 129, 761-769.	0.4	7
24	Determinants of Visfatin/NAMPT Serum Concentration and its Leukocyte Expression in Hyperthyroidism. Hormone and Metabolic Research, 2018, 50, 653-660.	1.5	5
25	Defects in GnRH Neuron Migration/Development and Hypothalamic-Pituitary Signaling Impact Clinical Variability of Kallmann Syndrome. Genes, 2021, 12, 868.	2.4	5
26	Alternative 3' acceptor site in the exon 2 of human PAX8 gene resulting in the expression of unknown mRNA variant found in thyroid hemiagenesis and some types of cancers. Acta Biochimica Polonica, 2013, 60, 573-8.	0.5	5
27	CCND1 gene polymorphic variants in patients with differentiated thyroid carcinoma. Oncology Letters, 2015, 9, 442-448.	1.8	4
28	Pituitary Microsomal Autoantibodies in Patients with Childhood-Onset Combined Pituitary Hormone Deficiency: an Antigen Identification Attempt. Archivum Immunologiae Et Therapiae Experimentalis, 2016, 64, 485-495.	2.3	4
29	Elevated serum RANTES chemokine in autoimmune Addisonâ€™s disease. Polish Archives of Internal Medicine, 2018, 128, 216-221.	0.4	4
30	Genomic mapping of pathways in endometrial adenocarcinoma and a gastrointestinal stromal tumor located in Meckel's diverticulum. Oncology Letters, 2016, 11, 1007-1015.	1.8	3
31	Compound heterozygous GLI3 variants in siblings with thyroid hemiagenesis. Endocrine, 2021, 71, 514-519.	2.3	3
32	CDON gene contributes to pituitary stalk interruption syndrome associated with unilateral facial and abducens nerve palsy. Journal of Applied Genetics, 2021, 62, 621-629.	1.9	3
33	VEGF-C Is a Thyroid Marker of Malignancy Superior to VEGF-A in the Differential Diagnostics of Thyroid Lesions. PLoS ONE, 2016, 11, e0150124.	2.5	3
34	Homozygous microdeletion in the 11p13 region in the patient with isolated form of aniridia: New challenges in the genetic diagnostics of aniridia. American Journal of Medical Genetics, Part A, 2021, , .	1.2	2
35	High incidence of FLT3 mutations in follicular thyroid cancer: potential therapeutic target in patients with advanced disease stage. Therapeutic Advances in Medical Oncology, 2020, 12, 175883592090753.	3.2	1
36	Progressing Vulvar Melanoma Caused by Instability in cKIT Juxtamembrane Domain: A Case Report and Review of Literature. Current Oncology, 2022, 29, 3130-3137.	2.2	1

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37	â€œSlipped capital femoral epiphysis in a 25-year-old hypogonadic man with a large cranial chondroma: causality or coincidence? â€œ. BMC Endocrine Disorders, 2021, 21, 167.	2.2	0
38	NKX2-5 Variant in Two Siblings with Thyroid Hemiagenesis. International Journal of Molecular Sciences, 2022, 23, 3414.	4.1	0