

Bartłomiej Budny

List of Publications by Year in descending order

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Version: 2024-02-01

38
papers

1,050
citations

567281

15
h-index

434195

31
g-index

39
all docs

39
docs citations

39
times ranked

2372
citing authors

#	ARTICLE	IF	CITATIONS
1	NKX2-5 Variant in Two Siblings with Thyroid Hemiagenesis. <i>International Journal of Molecular Sciences</i> , 2022, 23, 3414.	4.1	0
2	Progressing Vulvar Melanoma Caused by Instability in cKIT Juxtamembrane Domain: A Case Report and Review of Literature. <i>Current Oncology</i> , 2022, 29, 3130-3137.	2.2	1
3	Compound heterozygous GLI3 variants in siblings with thyroid hemiagenesis. <i>Endocrine</i> , 2021, 71, 514-519.	2.3	3
4	Defects in GnRH Neuron Migration/Development and Hypothalamic-Pituitary Signaling Impact Clinical Variability of Kallmann Syndrome. <i>Genes</i> , 2021, 12, 868.	2.4	5
5	CDON gene contributes to pituitary stalk interruption syndrome associated with unilateral facial and abducens nerve palsy. <i>Journal of Applied Genetics</i> , 2021, 62, 621-629.	1.9	3
6	“Slipped capital femoral epiphysis in a 25-year-old hypogonadic man with a large cranial chondroma: causality or coincidence?” <i>BMC Endocrine Disorders</i> , 2021, 21, 167.	2.2	0
7	Homozygous microdeletion in the 11p13 region in the patient with isolated form of aniridia: New challenges in the genetic diagnostics of aniridia. <i>American Journal of Medical Genetics, Part A</i> , 2021, , .	1.2	2
8	Head and Neck Paragangliomas – A Genetic Overview. <i>International Journal of Molecular Sciences</i> , 2020, 21, 7669.	4.1	17
9	Copy Number Variants Contributing to Combined Pituitary Hormone Deficiency. <i>International Journal of Molecular Sciences</i> , 2020, 21, 5757.	4.1	7
10	SEMA3A and IGSF10 Are Novel Contributors to Combined Pituitary Hormone Deficiency (CPHD). <i>Frontiers in Endocrinology</i> , 2020, 11, 368.	3.5	13
11	High incidence of FLT3 mutations in follicular thyroid cancer: potential therapeutic target in patients with advanced disease stage. <i>Therapeutic Advances in Medical Oncology</i> , 2020, 12, 175883592090753.	3.2	1
12	Differences in Mutational Profile between Follicular Thyroid Carcinoma and Follicular Thyroid Adenoma Identified Using Next Generation Sequencing. <i>International Journal of Molecular Sciences</i> , 2019, 20, 3126.	4.1	25
13	Evaluation of 167 Gene Expression Classifier (GEC) and ThyroSeq v2 Diagnostic Accuracy in the Preoperative Assessment of Indeterminate Thyroid Nodules: Bivariate/HROC Meta-analysis. <i>Endocrine Pathology</i> , 2019, 30, 8-15.	9.0	27
14	The genetic heterogeneity of indeterminate thyroid nodules assessed preoperatively with next generation sequencing reflects the diversity of the final histopathological diagnoses. <i>Polish Archives of Internal Medicine</i> , 2019, 129, 761-769.	0.4	7
15	Modifying impact of RET gene haplotypes on medullary thyroid carcinoma clinical course. <i>Endocrine-Related Cancer</i> , 2018, 25, 421-436.	3.1	20
16	Determinants of Visfatin/NAMPT Serum Concentration and its Leukocyte Expression in Hyperthyroidism. <i>Hormone and Metabolic Research</i> , 2018, 50, 653-660.	1.5	5
17	Elevated serum RANTES chemokine in autoimmune Addison’s disease. <i>Polish Archives of Internal Medicine</i> , 2018, 128, 216-221.	0.4	4
18	Mutations in proteasome-related genes are associated with thyroid hemiagenesis. <i>Endocrine</i> , 2017, 56, 279-285.	2.3	18

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19	Genomic markers of ovarian adenocarcinoma and its relevancy to the effectiveness of chemotherapy. <i>Oncology Letters</i> , 2017, 14, 3401-3414.	1.8	7
20	Pituitary Microsomal Autoantibodies in Patients with Childhood-Onset Combined Pituitary Hormone Deficiency: an Antigen Identification Attempt. <i>Archivum Immunologiae Et Therapiae Experimentalis</i> , 2016, 64, 485-495.	2.3	4
21	Genomic mapping of pathways in endometrial adenocarcinoma and a gastrointestinal stromal tumor located in Meckel's diverticulum. <i>Oncology Letters</i> , 2016, 11, 1007-1015.	1.8	3
22	Two coexisting heterozygous frameshift mutations in PROP1 are responsible for a different phenotype of combined pituitary hormone deficiency. <i>Journal of Applied Genetics</i> , 2016, 57, 373-381.	1.9	7
23	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. <i>Molecular Psychiatry</i> , 2016, 21, 133-148.	7.9	243
24	Nicotinamide phosphoribosyltransferase leukocyte overexpression in Graves' ophthalmopathy. <i>Endocrine</i> , 2016, 53, 497-504.	2.3	12
25	VEGF-C Is a Thyroid Marker of Malignancy Superior to VEGF-A in the Differential Diagnostics of Thyroid Lesions. <i>PLoS ONE</i> , 2016, 11, e0150124.	2.5	3
26	Increased STAG2 dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. <i>Human Molecular Genetics</i> , 2015, 24, 7171-7181.	2.9	28
27	CCND1 gene polymorphic variants in patients with differentiated thyroid carcinoma. <i>Oncology Letters</i> , 2015, 9, 442-448.	1.8	4
28	The c.470C>G CHEK2 missense variant increases the risk of differentiated thyroid carcinoma in the Great Poland population. <i>Hereditary Cancer in Clinical Practice</i> , 2015, 13, 8.	1.5	30
29	The Role of Serum C-Reactive Protein Measured by High-Sensitive Method in Thyroid Disease. <i>Archivum Immunologiae Et Therapiae Experimentalis</i> , 2014, 62, 501-509.	2.3	29
30	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. <i>Acta Biochimica Polonica</i> , 2013, 60, .	0.5	13
31	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. <i>Acta Biochimica Polonica</i> , 2013, 60, 573-8.	0.5	5
32	PAX6 deletion in a family with aniridia. <i>Ophthalmic Genetics</i> , 2012, 33, 44-48.	1.2	18
33	FOXE1 Polyalanine Tract Length Polymorphism in Patients with Thyroid Hemiagenesis and Subjects with Normal Thyroid. <i>Hormone Research in Paediatrics</i> , 2011, 75, 329-334.	1.8	30
34	A novel nonsense mutation in CUL4B gene in three brothers with X-linked mental retardation syndrome. <i>Clinical Genetics</i> , 2010, 77, 141-144.	2.0	47
35	Novel missense mutations in the ubiquitination-related gene UBE2A cause a recognizable X-linked mental retardation syndrome. <i>Clinical Genetics</i> , 2010, 77, 541-551.	2.0	45
36	A novel GJA1 missense mutation in a Polish child with oculodentodigital dysplasia. <i>Journal of Applied Genetics</i> , 2009, 50, 297-299.	1.9	15

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37	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
38	A novel X-linked recessive mental retardation syndrome comprising macrocephaly and ciliary dysfunction is allelic to oral-facial-digital type I syndrome. Human Genetics, 2006, 120, 171-178.	3.8	166