

Jernej Kovac

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

42
papers

537
citations

13
h-index

22
g-index

53
ext. papers

781
ext. citations

3.5
avg, IF

3.63
L-index

#	Paper	IF	Citations
42	Clinical and genetic characteristics of two patients with tyrosinemia type 1 in Slovenia - A novel fumarylacetoacetate hydrolase () intronic disease-causing variant.. <i>Molecular Genetics and Metabolism Reports</i> , 2022 , 30, 100836	1.8	0
41	Long-Term Follow-Up of Three Family Members with a Novel NNT Pathogenic Variant Causing Primary Adrenal Insufficiency. <i>Genes</i> , 2022 , 13, 717	4.2	0
40	Heterozygous Genetic Variants in Autosomal Recessive Genes of the Leptin-Melanocortin Signalling Pathway Are Associated With the Development of Childhood Obesity.. <i>Frontiers in Endocrinology</i> , 2022 , 13, 832911	5.7	1
39	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency: High Incidence of Detected Patients With Expanded Newborn Screening Program. <i>Frontiers in Genetics</i> , 2021 , 12, 648493	4.5	0
38	Next-Generation Sequencing in Newborn Screening: A Review of Current State. <i>Frontiers in Genetics</i> , 2021 , 12, 662254	4.5	9
37	Two Cases With an Early Presented Proopiomelanocortin Deficiency-A Long-Term Follow-Up and Systematic Literature Review. <i>Frontiers in Endocrinology</i> , 2021 , 12, 689387	5.7	2
36	Focused peptide library screening as a route to a superior affinity ligand for antibody purification. <i>Scientific Reports</i> , 2021 , 11, 11650	4.9	1
35	Does intervention with GLP-1 receptor agonist semaglutide modulate perception of sweet taste in women with obesity: study protocol of a randomized, single-blinded, placebo-controlled clinical trial. <i>Trials</i> , 2021 , 22, 464	2.8	1
34	Technological Approaches in the Analysis of Extracellular Vesicle Nucleotide Sequences.. <i>Frontiers in Bioengineering and Biotechnology</i> , 2021 , 9, 787551	5.8	0
33	SPTB related spherocytosis in a three-generation family presenting with kidney failure in adulthood due to co-occurrence of UMOD disease causing variant. <i>Nefrologia</i> , 2020 , 40, 420-427	0.4	
32	SPTB related spherocytosis in a three-generation family presenting with kidney failure in adulthood due to co-occurrence of UMOD disease causing variant. <i>Nefrologia</i> , 2020 , 40, 421-428	1.5	1
31	Extracellular Vesicles Derived Human-miRNAs Modulate the Immune System in Type 1 Diabetes. <i>Frontiers in Cell and Developmental Biology</i> , 2020 , 8, 202	5.7	12
30	High frequency of pathogenic ACAN variants including an intragenic deletion in selected individuals with short stature. <i>European Journal of Endocrinology</i> , 2020 , 182, 243-253	6.5	14
29	Expanded Newborn Screening Program in Slovenia using Tandem Mass Spectrometry and Confirmatory Next Generation Sequencing Genetic Testing. <i>Zdravstveno Varstvo</i> , 2020 , 59, 256-263	1.3	7
28	Challenges in identifying large germline structural variants for clinical use by long read sequencing. <i>Computational and Structural Biotechnology Journal</i> , 2020 , 18, 83-92	6.8	8
27	Relevant Weight Reduction and Reversed Metabolic Co-morbidities Can Be Achieved by Duodenojejunal Bypass Liner in Adolescents with Morbid Obesity. <i>Obesity Surgery</i> , 2020 , 30, 1001-1010	3.7	3
26	Dataset on amelogenesis-related genes variants (and interacting genes) and on human leukocyte antigen alleles (DQ2 and DQ8) distribution in children with and without molar-incisor hypomineralisation (MIH). <i>Data in Brief</i> , 2020 , 32, 106224	1.2	1

25	The possible influence of genetic aetiological factors on molar-incisor hypomineralisation. <i>Archives of Oral Biology</i> , 2020 , 118, 104848	2.8	8
24	and Genes Are Differentially Methylated in Patients With Periodic Fever, Aphthous Stomatitis, Pharyngitis, and Adenitis (PFAPA) Syndrome. <i>Frontiers in Immunology</i> , 2020 , 11, 1322	8.4	7
23	Genetic and Clinical Characteristics of Patients With Homozygous and Compound Heterozygous Familial Hypercholesterolemia From Three Different Populations: Case Series. <i>Frontiers in Genetics</i> , 2020 , 11, 572176	4.5	0
22	Circulating levels of miR-122 and nonalcoholic fatty liver disease in pre-pubertal obese children. <i>Pediatric Obesity</i> , 2018 , 13, 175-182	4.6	28
21	Next generation sequencing as a follow-up test in an expanded newborn screening programme. <i>Clinical Biochemistry</i> , 2018 , 52, 48-55	3.5	23
20	Identification of novel alleles associated with insulin resistance in childhood obesity using pooled-DNA genome-wide association study approach. <i>International Journal of Obesity</i> , 2018 , 42, 686-695	5.5	7
19	Overview of the current status of familial hypercholesterolaemia care in over 60 countries - The EAS Familial Hypercholesterolaemia Studies Collaboration (FHSC). <i>Atherosclerosis</i> , 2018 , 277, 234-255	3.1	93
18	Universal screening for familial hypercholesterolemia in children: The Slovenian model and literature review. <i>Atherosclerosis</i> , 2018 , 277, 383-391	3.1	31
17	Association of Glycemic Control and Cell Stress With Telomere Attrition in Type 1 Diabetes. <i>JAMA Pediatrics</i> , 2018 , 172, 879-881	8.3	13
16	DEPTOR promoter genetic variants and insulin resistance in obese children and adolescents. <i>Pediatric Diabetes</i> , 2017 , 18, 152-158	3.6	5
15	Multifocal gastric adenocarcinoma in a patient with LRBA deficiency. <i>Orphanet Journal of Rare Diseases</i> , 2017 , 12, 131	4.2	21
14	Characterization of a de novo sSMC 17 detected in a girl with developmental delay and dysmorphic features. <i>Molecular Cytogenetics</i> , 2017 , 10, 10	2	3
13	Discovering the Unexpected with the Utilization of NGS in Diagnostics of Non-syndromic Hearing Loss Disorders: The Family Case of -Dependent Hearing Loss Disorder. <i>Frontiers in Genetics</i> , 2017 , 8, 95	4.5	1
12	TMPRSS3 mutations in autosomal recessive nonsyndromic hearing loss. <i>European Archives of Oto-Rhino-Laryngology</i> , 2016 , 273, 1151-4	3.5	15
11	Universal Screening for Familial Hypercholesterolemia in Children. <i>Journal of the American College of Cardiology</i> , 2015 , 66, 1250-1257	15.1	93
10	Comparison of tandem mass spectrometry and amino acid analyzer for phenylalanine and tyrosine monitoring--implications for clinical management of patients with hyperphenylalaninemia. <i>Clinical Biochemistry</i> , 2015 , 48, 14-8	3.5	28
9	Weak association of glyoxalase 1 (GLO1) variants with autism spectrum disorder. <i>European Child and Adolescent Psychiatry</i> , 2015 , 24, 75-82	5.5	11
8	Clinical and Molecular Cytogenetic Characterisation of Children with Developmental Delay and Dysmorphic Features. <i>Zdravstveno Varstvo</i> , 2015 , 54, 69-73	1.3	

7	Association of Average Telomere Length with Body-Mass Index and Vitamin D Status in Juvenile Population with Type 1 Diabetes. <i>Zdravstveno Varstvo</i> , 2015 , 54, 74-8	1.3	6
6	Cytogenetic and Molecular Genetic Characterization of Children with Short Stature. <i>Zdravstveno Varstvo</i> , 2015 , 54, 98-102	1.3	1
5	Specific and global coagulation tests in patients with mild haemophilia A with a double mutation (Glu113Asp, Arg593Cys). <i>Blood Transfusion</i> , 2015 , 13, 622-30	3.6	6
4	Rare single nucleotide polymorphisms in the regulatory regions of the superoxide dismutase genes in autism spectrum disorder. <i>Autism Research</i> , 2014 , 7, 138-44	5.1	19
3	Severe progressive obstructive cardiomyopathy and renal tubular dysfunction in Donohue syndrome with decreased insulin receptor autophosphorylation due to a novel INSR mutation. <i>European Journal of Pediatrics</i> , 2013 , 172, 1125-9	4.1	21
2	Five novel mutations and two large deletions in a population analysis of the phenylalanine hydroxylase gene. <i>Molecular Genetics and Metabolism</i> , 2012 , 106, 142-8	3.7	31
1	GPR143 gene mutation analysis in pediatric patients with albinism. <i>Ophthalmic Genetics</i> , 2012 , 33, 167-70.	0.2	4