Jernej Kovac

List of Publications by Citations

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	Universal Screening for Familial Hypercholesterolemia in Children. *Journal of the American College of Cardiology*, **2015**, 66, 1250-1257	15.1	93
41	Overview of the current status of familial hypercholesterolaemia care in over 60 countries - The EAS Familial Hypercholesterolaemia Studies Collaboration (FHSC). *Atherosclerosis*, **2018**, 277, 234-255	3.1	93
40	Five novel mutations and two large deletions in a population analysis of the phenylalanine hydroxylase gene. *Molecular Genetics and Metabolism*, **2012**, 106, 142-8	3.7	31
39	Universal screening for familial hypercholesterolemia in children: The Slovenian model and literature review. *Atherosclerosis*, **2018**, 277, 383-391	3.1	31
38	Comparison of tandem mass spectrometry and amino acid analyzer for phenylalanine and tyrosine monitoring--implications for clinical management of patients with hyperphenylalaninemia. *Clinical Biochemistry*, **2015**, 48, 14-8	3.5	28
37	Circulating levels of miR-122 and nonalcoholic fatty liver disease in pre-pubertal obese children. *Pediatric Obesity*, **2018**, 13, 175-182	4.6	28
36	Next generation sequencing as a follow-up test in an expanded newborn screening programme. *Clinical Biochemistry*, **2018**, 52, 48-55	3.5	23
35	Multifocal gastric adenocarcinoma in a patient with LRBA deficiency. *Orphanet Journal of Rare Diseases*, **2017**, 12, 131	4.2	21
34	Severe progressive obstructive cardiomyopathy and renal tubular dysfunction in Donohue syndrome with decreased insulin receptor autophosphorylation due to a novel INSR mutation. *European Journal of Pediatrics*, **2013**, 172, 1125-9	4.1	21
33	Rare single nucleotide polymorphisms in the regulatory regions of the superoxide dismutase genes in autism spectrum disorder. *Autism Research*, **2014**, 7, 138-44	5.1	19
32	TMPRSS3 mutations in autosomal recessive nonsyndromic hearing loss. *European Archives of Oto-Rhino-Laryngology*, **2016**, 273, 1151-4	3.5	15
31	High frequency of pathogenic ACAN variants including an intragenic deletion in selected individuals with short stature. *European Journal of Endocrinology*, **2020**, 182, 243-253	6.5	14
30	Association of Glycemic Control and Cell Stress With Telomere Attrition in Type 1 Diabetes. *JAMA Pediatrics*, **2018**, 172, 879-881	8.3	13
29	Extracellular Vesicles Derived Human-miRNAs Modulate the Immune System in Type 1 Diabetes. *Frontiers in Cell and Developmental Biology*, **2020**, 8, 202	5.7	12
28	Weak association of glyoxalase 1 (GLO1) variants with autism spectrum disorder. *European Child and Adolescent Psychiatry*, **2015**, 24, 75-82	5.5	11
27	Next-Generation Sequencing in Newborn Screening: A Review of Current State. *Frontiers in Genetics* , **2021**, 12, 662254	4.5	9
26	Challenges in identifying large germline structural variants for clinical use by long read sequencing. *Computational and Structural Biotechnology Journal*, **2020**, 18, 83-92	6.8	8

25	The possible influence of genetic aetiological factors on molar-incisor hypomineralisation. *Archives of Oral Biology*, **2020**, 118, 104848	2.8	8
24	Expanded Newborn Screening Program in Slovenia using Tandem Mass Spectrometry and Confirmatory Next Generation Sequencing Genetic Testing. *Zdravstveno Varstvo*, **2020**, 59, 256-263	1.3	7
23	and Genes Are Differentially Methylated in Patients With Periodic Fever, Aphthous Stomatitis, Pharyngitis, and Adenitis (PFAPA) Syndrome. *Frontiers in Immunology*, **2020**, 11, 1322	8.4	7
22	Identification of novel alleles associated with insulin resistance in childhood obesity using pooled-DNA genome-wide association study approach. *International Journal of Obesity*, **2018**, 42, 686-695	5.5	7
21	Association of Average Telomere Length with Body-Mass Index and Vitamin D Status in Juvenile Population with Type 1 Diabetes. *Zdravstveno Varstvo*, **2015**, 54, 74-8	1.3	6
20	Specific and global coagulation tests in patients with mild haemophilia A with a double mutation (Glu113Asp, Arg593Cys). *Blood Transfusion*, **2015**, 13, 622-30	3.6	6
19	DEPTOR promoter genetic variants and insulin resistance in obese children and adolescents. *Pediatric Diabetes*, **2017**, 18, 152-158	3.6	5
18	GPR143 gene mutation analysis in pediatric patients with albinism. *Ophthalmic Genetics*, **2012**, 33, 167-70	1.2	4
17	Characterization of a de novo sSMC 17 detected in a girl with developmental delay and dysmorphic features. *Molecular Cytogenetics*, **2017**, 10, 10	2	3
16	Relevant Weight Reduction and Reversed Metabolic Co-morbidities Can Be Achieved by Duodenojejunal Bypass Liner in Adolescents with Morbid Obesity. *Obesity Surgery*, **2020**, 30, 1001-1010	3.7	3
15	Two Cases With an Early Presented Proopiomelanocortin Deficiency-A Long-Term Follow-Up and Systematic Literature Review. *Frontiers in Endocrinology*, **2021**, 12, 689387	5.7	2
14	SPTB related spherocytosis in a three-generation family presenting with kidney failure in adulthood due to co-occurrence of UMOD disease causing variant. *Nefrologia*, **2020**, 40, 421-428	1.5	1
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. *Frontiers in Genetics*, **2017**, 8, 95	4.5	1
12	Cytogenetic and Molecular Genetic Characterization of Children with Short Stature. *Zdravstveno Varstvo*, **2015**, 54, 98-102	1.3	1
11	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). *Data in Brief*, **2020**, 32, 106224	1.2	1
10	Focused peptide library screening as a route to a superior affinity ligand for antibody purification. *Scientific Reports*, **2021**, 11, 11650	4.9	1
9	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. *Trials*, **2021**, 22, 464	2.8	1
8	Heterozygous Genetic Variants in Autosomal Recessive Genes of the Leptin-Melanocortin Signalling Pathway Are Associated With the Development of Childhood Obesity.. *Frontiers in Endocrinology*, **2022**, 13, 832911	5.7	1

7	Clinical and genetic characteristics of two patients with tyrosinemia type 1 in Slovenia - A novel fumarylacetoacetate hydrolase () intronic disease-causing variant.. *Molecular Genetics and Metabolism Reports*, **2022**, 30, 100836	1.8	0
6	Genetic and Clinical Characteristics of Patients With Homozygous and Compound Heterozygous Familial Hypercholesterolemia From Three Different Populations: Case Series. *Frontiers in Genetics*, **2020**, 11, 572176	4.5	0
5	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency: High Incidence of Detected Patients With Expanded Newborn Screening Program. *Frontiers in Genetics*, **2021**, 12, 648493	4.5	0
4	Technological Approaches in the Analysis of Extracellular Vesicle Nucleotide Sequences.. *Frontiers in Bioengineering and Biotechnology*, **2021**, 9, 787551	5.8	0
3	Long-Term Follow-Up of Three Family Members with a Novel NNT Pathogenic Variant Causing Primary Adrenal Insufficiency. *Genes*, **2022**, 13, 717	4.2	0
2	SPTB related spherocytosis in a three-generation family presenting with kidney failure in adulthood due to co-occurrence of UMOD disease causing variant. *Nefrologia*, **2020**, 40, 420-427	0.4	
1	Clinical and Molecular Cytogenetic Characterisation of Children with Developmental Delay and Dysmorphic Features. *Zdravstveno Varstvo*, **2015**, 54, 69-73	1.3	