

# Shadab Salehpour

## List of Publications by Year in descending order

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Version: 2024-02-01

27  
papers

248  
citations

1162889

8  
h-index

996849

15  
g-index

28  
all docs

28  
docs citations

28  
times ranked

409  
citing authors

#	ARTICLE	IF	CITATIONS
1	Identification of novel mutations among Iranian NPC1 patients: a bioinformatics approach to predict pathogenic mutations. <i>Hereditas</i> , 2022, 159, 8.	0.5	0
2	Identification of a novel de novo mutation in the CTNNB1 gene in an Iranian patient with intellectual disability. <i>Neurological Sciences</i> , 2022, 43, 2859.	0.9	4
3	Molecular characterization of a large cohort of mucopolysaccharidosis patients: Iran Mucopolysaccharidosis REâ€diagnosis study (IMPRESsion). <i>Human Mutation</i> , 2022, , .	1.1	1
4	A novelSRD5A2mutation in an Iranian family with sex development disorder. <i>Andrologia</i> , 2021, 53, e13847.	1.0	1
5	The treatment and clinical follow-up outcome in Iranian patients with tetrahydrobiopterin deficiency. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2021, 34, 1157-1167.	0.4	3
6	Analysis of the HEXA, HEXB, ARSA, and SMPD1 Genes in 68 Iranian Patients. <i>Journal of Molecular Neuroscience</i> , 2021, , 1.	1.1	1
7	Mutations in the VPS13B Gene in Iranian Patients with Different Phenotypes of Cohen Syndrome. <i>Journal of Molecular Neuroscience</i> , 2020, 70, 21-25.	1.1	9
8	The effect of omega-3 fatty acids and vitamin E co-supplementation on glycemic profile: A systematic review and meta-analysis of randomized controlled trials. <i>Obesity Medicine</i> , 2020, 17, 100180.	0.5	1
9	Effects of selenium supplementation on serum C reactive protein level: A systematic review and meta-analysis of randomized controlled clinical trials. <i>Obesity Medicine</i> , 2020, 17, 100182.	0.5	3
10	Whole-Exome Sequencing Uncovers Novel Causative Variants and Additional Findings in Three Patients Affected by Glycogen Storage Disease Type VI and Fanconiâ€™Bickel Syndrome. <i>Frontiers in Genetics</i> , 2020, 11, 601566.	1.1	7
11	ELMO Domain Containing 1 (ELMOD1) Gene Mutation Is Associated with Mental Retardation and Autism Spectrum Disorder. <i>Journal of Molecular Neuroscience</i> , 2019, 69, 312-315.	1.1	12
12	Next generation sequencing elucidated a clinically undiagnosed metabolic disorder - An Iranian family with hereditary orotic aciduria. <i>Gene Reports</i> , 2019, 16, 100457.	0.4	0
13	Neurodegeneration with brain iron accumulation 2A: Report of four independent cases. <i>Meta Gene</i> , 2018, 15, 87-89.	0.3	0
14	Molecular genetics of a cohort of 635 cases of phenylketonuria in a consanguineous population. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 1159-1167.	1.7	14
15	Long-Term Effect of Nephrocalcinosis on Renal Function and Body Growth Index in Children: A Retrospective Single Center Study. <i>Iranian Journal of Pediatrics</i> , 2018, 28, .	0.1	1
16	Clinical, biochemical and molecular features of Iranian families with mucopolysaccharidosis: A case series. <i>Clinica Chimica Acta</i> , 2017, 474, 88-95.	0.5	10
17	Association of a Novel Nonsense Mutation in KIAA1279 with Goldberg-Shprintzen Syndrome. <i>Iranian Journal of Child Neurology</i> , 2017, 11, 70-74.	0.2	7
18	A novel splice site mutation in the GNPTAB gene in an Iranian patient with mucopolisidosis II $\hat{1}\pm/\hat{1}^2$ . <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2016, 29, 991-3.	0.4	3

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19	Co-existence of phenylketonuria either with maple syrup urine disease or Sandhoff disease in two patients from Iran: emphasizing the role of consanguinity. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2016, 29, 1215-1219.	0.4	7
20	Four Years of Diagnostic Challenges with Tetrahydrobiopterin Deficiencies in Iranian Patients. <i>JIMD Reports</i> , 2016, 32, 7-14.	0.7	16
21	Molecular Characterization of QDPR Gene in Iranian Families with BH4 Deficiency: Reporting Novel and Recurrent Mutations. <i>JIMD Reports</i> , 2015, 21, 123-128.	0.7	8
22	Three novel mutations in Iranian patients with Tay-Sachs disease. <i>Iranian Biomedical Journal</i> , 2014, 18, 114-9.	0.4	5
23	Effects of Miglustat on Stabilization of Neurological Disorder in Niemann-Pick Disease Type C. <i>Journal of Child Neurology</i> , 2013, 28, 1599-1606.	0.7	27
24	Frequency of Pediatric Acute Respiratory Tract Infections in Iran; A Systematic Review. <i>Archives of Pediatric Infectious Diseases</i> , 2013, 1, 44-52.	0.1	3
25	A Double-Blind, Placebo-Controlled Comparison of Letrozole to Oxandrolone Effects upon Growth and Puberty of Children with Constitutional Delay of Puberty and Idiopathic Short Stature. <i>Hormone Research in Paediatrics</i> , 2010, 74, 428-435.	0.8	51
26	Cyclic Pamidronate Therapy in Children with Osteogenesis Imperfecta. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2010, 23, 73-80.	0.4	29
27	Craniosynostosis in a patient with 2q37.3 deletion 5q34 duplication: Association of extra copy of <i>MSX2</i> with craniosynostosis. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1544-1549.	0.7	24