

Natalia Semenova

List of Publications by Year in descending order

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Version: 2024-02-01

10
papers

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citations

2682572

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2272923

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docs citations

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#	ARTICLE	IF	CITATIONS
1	DIFFERENTIAL DIAGNOSIS OF THE FLOPPY INFANT SYNDROME. <i>Pediatrics</i> , 2022, 101, 171-178.	0.2	0
2	Clinical and genetic characteristics of skeletal dysplasias – short-rib thoracic dysplasia. <i>Pediatric Traumatology, Orthopaedics and Reconstructive Surgery</i> , 2022, 10, 43-56.	0.3	1
3	Mutation in <i>PHACTR1</i> associated with multifocal epilepsy with infantile spasms and hypsarrhythmia. <i>Clinical Genetics</i> , 2021, 99, 673-683.	2.0	6
4	CLINICAL GENETIC CHARACTERISTICS OF JANSEN-DE VRIES SYNDROME CAUSED BY HETEROZYGOUS MUTATIONS IN THE PPM1D GENE. <i>Pediatrics</i> , 2021, 100, 277-282.	0.2	0
5	A novel splice site mutation in OTC gene of a female with ornithine transcarbamylase deficiency and her asymptomatic mosaic father. <i>Journal of Genetics</i> , 2020, 99, 1.	0.7	2
6	Microcephaly-capillary malformation syndrome: the newly reported cases. <i>Bulletin of Russian State Medical University</i> , 2020, , 31-37.	0.2	0
7	Noonan-like syndrome with loose anagen hair associated with the heterozygous mutation c.4A>G (p.Ser2Gly) in the SHOC2 gene. <i>Voprosy Prakticheskoi Pediatrii</i> , 2020, 15, 93-98.	0.2	0
8	Identification of novel variants in the <i>LDLR</i> gene in Russian patients with familial hypercholesterolemia using targeted sequencing. <i>Biomedical Reports</i> , 2020, 14, 15.	2.0	13
9	CONGENITAL CENTRAL HYPOVENTILATION SYNDROME, CAUSED BY DE NOVO DELETION IN THE PHOX2B GENE. <i>Pediatrics</i> , 2019, 98, 235-238.	0.2	1
10	Clinical and genetic characteristics of ponto-cerebellar hypoplasia caused by mutations in the TSEN54 gene (OMIM: 277470). <i>Nervno-Myshechnye Bolezni</i> , 2019, 9, 30-36.	0.4	0