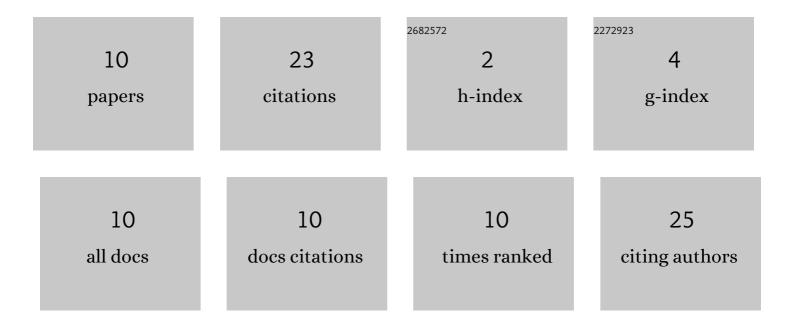
## Natalia Semenova

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/976747/publications.pdf Version: 2024-02-01



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