# Natalia Semenova

List of Publications by Year
in descending order



| 10 | 23 | 2682572<br>2 | 2272923<br>4 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 10 | 10 | 10 | 25 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Identification of novel variants in the LDLR gene in Russian patients with familial hypercholesterolemia using targeted sequencing. Biomedical Reports, 2020, 14, 15. | 2.0 | 13 |
| 2 | Mutation in <scp>PHACTR1</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 | Ð¡linical and genetic characteristics of skeletal cyliopathies â€“ 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 | 0 |
| 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 |