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List of Publications by Year in descending order

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1684188 1720034 7 161 5 7 citations h-index g-index papers 7 7 7 322 docs citations times ranked citing authors all docs

| # | Article | IF | CITATIONS |
|---|---|-----|-----------|
| 1 | Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in <i>POLR3A</i> , <i>POLR3B</i> , and <i>POLR1C</i> , Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674. | 3.6 | 26 |
| 2 | Overuse continues despite no new evidence on the efficacy of proton pump inhibitors for crying and irritability in infants. Acta Paediatrica, International Journal of Paediatrics, $2021, \ldots$ | 1.5 | 2 |
| 3 | Exome Sequencing Reveals Novel Variants and Expands the Genetic Landscape for Congenital Microcephaly. Genes, 2021, 12, 2014. | 2.4 | 8 |
| 4 | Comprehensive genomic analysis of patients with disorders of cerebral cortical development. European Journal of Human Genetics, 2018, 26, 1121-1131. | 2.8 | 35 |
| 5 | No Effect of Proton Pump Inhibitors on Crying and Irritability in Infants: Systematic Review of Randomized Controlled Trials. Journal of Pediatrics, 2015, 166, 767-770.e3. | 1.8 | 77 |
| 6 | Is diagnosing cardio-facio-cutaneous (CFC) syndrome still a challenge? Delineation of the phenotype in 15 Polish patients with proven mutations, including novel mutations in the BRAF gene. European Journal of Medical Genetics, 2015, 58, 14-20. | 1.3 | 11 |
| 7 | Oculocutaneous albinism in a patient with 17p13.2-pter duplication - a review on the molecular syndromology of 17p13 duplication. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2015, 159, 333-337. | 0.6 | 2 |