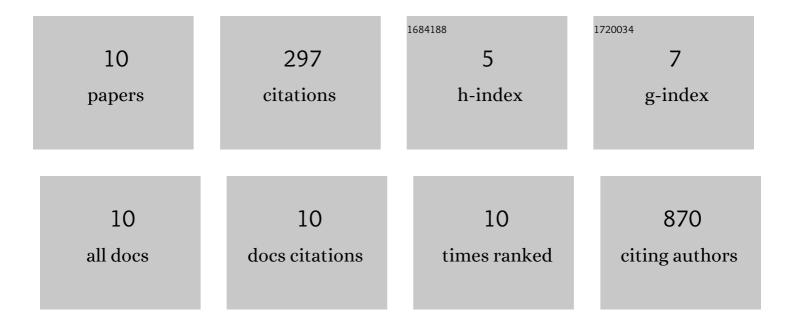
## Paul R Mark

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/2883931/publications.pdf Version: 2024-02-01



DALLI P. MADK

| #  | Article  | IF   | CITATIONS |
|----|--|------|-----------|
| 1  | <scp>NAD</scp> + deficiency in human congenital malformations and miscarriage: A new model of pleiotropy. American Journal of Medical Genetics, Part A, 2022, 188, 2834-2849.      | 1.2  | 5         |
| 2  | Further characterization of <scp>Borjesonâ€Forssman‣ehmann</scp> syndrome in females due to de<br>novo variants in <scp><i>PHF6</i></scp> . Clinical Genetics, 2022, 102, 182-190. | 2.0  | 5         |
| 3  | Two cases of different genetic variants of alveolar capillary dysplasia associated with left-sided obstructive CHDs. Cardiology in the Young, 2021, 31, 1368-1370.                 | 0.8  | 0         |
| 4  | Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. Genetics in Medicine, 2021, 23, 1234-1245.                                | 2.4  | 6         |
| 5  | Lethal renal anomalies in a fetus with 21q22.11â€q22.12 deletion. American Journal of Medical Genetics,<br>Part A, 2020, 182, 3060-3063.   | 1.2  | 0         |
| 6  | Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy.<br>American Journal of Human Genetics, 2019, 104, 1210-1222.                         | 6.2  | 56        |
| 7  | Mutation update for the <i>SATB2</i> gene. Human Mutation, 2019, 40, 1013-1029.  | 2.5  | 38        |
| 8  | NAD Deficiency, Congenital Malformations, and Niacin Supplementation. New England Journal of Medicine, 2017, 377, 544-552.   | 27.0 | 177       |
| 9  | Comment on critical region for talipes equinovarus in patients with 5q23 deletions. European Journal of Medical Genetics, 2015, 58, 243.   | 1.3  | 0         |
| 10 | Narrowing the Critical Region for Congenital Vertical Talus in Patients With Interstitial 18q<br>Deletions. American Journal of Medical Genetics, Part A, 2013, 161, 1117-1121.    | 1.2  | 10        |