# Daniela Barge-Schaapveld

List of Publications by Year
in descending order



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|---|---|---|---|
| 31 papers | 958 citations | 516710 16 h-index | 454955 30 g-index |
| 33 all docs | 33 docs citations | 33 times ranked | 2190 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Consolidation of the clinical and genetic definition of aSOX4-related neurodevelopmental syndrome. Journal of Medical Genetics, 2022, 59, 1058-1068. | 3.2 | 10 |
| 2 | Characterization of Degenerative Mitral Valve Disease: Differences between Fibroelastic Deficiency and Barlowâ€™s Disease. Journal of Cardiovascular Development and Disease, 2021, 8, 23. | 1.6 | 21 |
| 3 | Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137. | 2.4 | 16 |
| 4 | Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. American Journal of Human Genetics, 2021, 108, 1692-1709. | 6.2 | 18 |
| 5 | Case series, Chemotherapy-induced cardiomyopathy: mind the family history!. European Heart Journal - Case Reports, 2021, 5, ytab333. | 0.6 | 4 |
| 6 | Distinct Metabolomic Signatures in Preclinical and Obstructive Hypertrophic Cardiomyopathy. Cells, 2021, 10, 2950. | 4.1 | 5 |
| 7 | Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7. | 4.1 | 40 |
| 8 | Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. American Heart Journal, 2020, 225, 108-119. | 2.7 | 25 |
| 9 | A mutation update for the FLNC gene in myopathies and cardiomyopathies. Human Mutation, 2020, 41, 1091-1111. | 2.5 | 92 |
| 10 | Identification of known and unknown genes associated with mitral valve prolapse using an exome slice methodology. Journal of Medical Genetics, 2020, 57, 843-850. | 3.2 | 22 |
| 11 | GATA6 mutations: Characterization of two novel patients and a comprehensive overview of the GATA6 genotypic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2019, 179, 1836-1845. | 1.2 | 16 |
| 12 | Pathogenic effect of a TGFBR1 mutation in a family with Loeysâ€“Dietz syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e00943. | 1.2 | 3 |
| 13 | Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. American Journal of Human Genetics, 2019, 105, 493-508. | 6.2 | 48 |
| 14 | Panel-Based Exome Sequencing for Neuromuscular Disorders as a Diagnostic Service. Journal of Neuromuscular Diseases, 2019, 6, 241-258. | 2.6 | 32 |
| 15 | Mortality Risk Associated With Truncating Founder Mutations in Titin. Circulation Genomic and Precision Medicine, 2019, 12, e002436. | 3.6 | 5 |
| 16 | Autosomal dominant Marfan syndrome caused by a previously reported recessive FBN1 variant. Molecular Genetics & Genomic Medicine, 2019, 7, e00518. | 1.2 | 6 |
| 17 | Putting genome-wide sequencing in neonates into perspective. Genetics in Medicine, 2019, 21, 1074-1082. | 2.4 | 15 |
| 18 | Toward an effective exome-based genetic testing strategy in pediatric dilated cardiomyopathy. Genetics in Medicine, 2018, 20, 1374-1386. | 2.4 | 36 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | The first titin (c.59926 + 1G > A) founder mutation associated with dilated cardiomyopathy. European Journal of Heart Failure, 2018, 20, 803-806. | 7.1 | 16 |
| 20 | Possible hints and pitfalls in diagnosing Peutz-Jeghers syndrome. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 1381-1386. | 0.9 | 5 |
| 21 | The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46. | 1.8 | 75 |
| 22 | Isolated Subepicardial Right Ventricular Outflow Tract Scar in Athletes With VentricularÂTachycardia. Journal of the American College of Cardiology, 2017, 69, 497-507. | 2.8 | 56 |
| 23 | Prospective risk of cancer and the influence of tobacco use in carriers of the p16-Leiden germline variant. European Journal of Human Genetics, 2015, 23, 711-714. | 2.8 | 29 |
| 24 | Testing an mHealth Momentary Assessment Routine Outcome Monitoring Application: A Focus on Restoration of Daily Life Positive Mood States. PLoS ONE, 2014, 9, e115254. | 2.5 | 31 |
| 25 | Intellectual Disability and Hemizygous <scp>GPD</scp>2 Mutation. American Journal of Medical Genetics, Part A, 2013, 161, 1044-1050. | 1.2 | 16 |
| 26 | Complex genetics of radial ray deficiencies: screening of a cohort of 54 patients. Genetics in Medicine, 2013, 15, 195-202. | 2.4 | 15 |
| 27 | Beare-Stevenson Syndrome: Two Dutch Patients With Cerebral Abnormalities. Pediatric Neurology, 2011, 44, 303-307. | 2.1 | 12 |
| 28 | Early improvement in positive rather than negative emotion predicts remission from depression after pharmacotherapy. European Neuropsychopharmacology, 2011, 21, 241-247. | 0.7 | 94 |
| 29 | The atypical 16p11.2 deletion: A not so atypical microdeletion syndrome?. American Journal of Medical Genetics, Part A, 2011, 155, 1066-1072. | 1.2 | 46 |
| 30 | What Is New in Dilatation of the Ascending Aorta?. Circulation, 2011, 123, 924-928. | 1.6 | 72 |
| 31 | Effects of Antidepressant Treatment on the Quality of Daily Life. Journal of Clinical Psychiatry, 2002, 63, 477-485. | 2.2 | 73 |