# Christopher P Barnett

List of Publications by Year
in descending order



| 29 | 958 | 933410 10 | 501174 28 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 29 | 29 | 29 | 2367 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Gain-of-Function Mutations in RIT1 Cause Noonan Syndrome, a RAS/MAPK Pathway Syndrome. American Journal of Human Genetics, 2013, 93, 173-180. | 6.2 | 279 |
| 2 | Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System. JAMA - Journal of the American Medical Association, 2020, 323, 2503. | 7.4 | 160 |
| 3 | High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€“Phenotype Correlation. Human Mutation, 2015, 36, 1052-1063. | 2.5 | 143 |
| 4 | Next generation sequencing in a large cohort of patients presenting with neuromuscular disease before or at birth. Orphanet Journal of Rare Diseases, 2015, 10, 148. | 2.7 | 94 |
| 5 | Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. Lancet Neurology, The, 2015, 14, 1182-1195. | 10.2 | 74 |
| 6 | Attitudes of Australian health professionals towards rapid genomic testing in neonatal and paediatric intensive care. European Journal of Human Genetics, 2019, 27, 1493-1501. | 2.8 | 29 |
| 7 | Self-reverting mutations partially correct the blood phenotype in a Diamond Blackfan anemia patient. Haematologica, 2017, 102, e506-e509. | 3.5 | 26 |
| 8 | Genomic intensive care: should we perform genome testing in critically ill newborns?: TableÂ1. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2016, 101, F94-F98. | 2.8 | 23 |
| 9 | Recurrent chronic histiocytic intervillositis with intrauterine growth restriction, osteopenia, and fractures. American Journal of Medical Genetics, Part A, 2016, 170, 2960-2964. | 1.2 | 16 |
| 10 | Pathogenic variants in MDFIC cause recessive central conducting lymphatic anomaly with lymphedema. Science Translational Medicine, 2022, 14, eabm4869. | 12.4 | 14 |
| 11 | Paternal mosaicism for a novel <scp>PBX1</scp> mutation associated with recurrent perinatal death: Phenotypic expansion of the <scp>PBX1</scp>â€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1273-1277. | 1.2 | 12 |
| 12 | Genetic variation affecting DNA methylation and the human imprinting disorder, Beckwith-Wiedemann syndrome. Clinical Epigenetics, 2018, 10, 114. | 4.1 | 10 |
| 13 | Maternal uniparental isodisomy of chromosome 6 unmasks a novel variant in in a patient with early onset retinal dystrophy. Molecular Vision, 2018, 24, 478-484. | 1.1 | 9 |
| 14 | Pseudodiastrophic dysplasia expands the known phenotypic spectrum of defects in proteoglycan biosynthesis. Journal of Medical Genetics, 2020, 57, 454-460. | 3.2 | 8 |
| 15 | Geneâ€specific facial dysmorphism in Axenfeldâ€Rieger syndrome caused by FOXC1 and PITX2 variants. American Journal of Medical Genetics, Part A, 2021, 185, 434-439. | 1.2 | 7 |
| 16 | Novel de novo 2q14.3 deletion disrupting <scp>CNTNAP5</scp> in a girl with intellectual impairment, thin corpus callosum, and microcephaly. American Journal of Medical Genetics, Part A, 2020, 182, 1824-1828. | 1.2 | 6 |
| 17 | Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760. | 2.4 | 6 |
| 18 | Multisegment coloboma in a case of Marfan syndrome: another possible effect of increased TGFÎ² signaling. Journal of AAPOS, 2014, 18, 90-92. | 0.3 | 5 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | <scp>CDH1</scp>â€related blepharocheilodontic syndrome is associated with diffuse gastric cancer risk. American Journal of Medical Genetics, Part A, 2020, 182, 1780-1784. | 1.2 | 5 |
| 20 | Compound heterozygous variants in LAMC3 in association with posterior periventricular nodular heterotopia. BMC Medical Genomics, 2021, 14, 64. | 1.5 | 5 |
| 21 | A de novo missense variant in <scp>MED13</scp> in a patient with global developmental delay, marked facial dysmorphism, macroglossia, short stature, and macrocephaly. American Journal of Medical Genetics, Part A, 2021, 185, 2586-2592. | 1.2 | 4 |
| 22 | Recurrent de novo missense variants in GNB2 can cause syndromic intellectual disability. Journal of Medical Genetics, 2022, 59, 511-516. | 3.2 | 4 |
| 23 | Severe Cloverleaf Skull Deformity in c.1061C>G (p.Ser354Cys) Mutated Fibroblast Growth Factor Receptor 2 Gene in Crouzon Syndrome. Journal of Craniofacial Surgery, 2021, 32, 261-264. | 0.7 | 4 |
| 24 | Whole Genome Sequencing, Focused Assays and Functional Studies Increasing Understanding in Cryptic Inherited Retinal Dystrophies. International Journal of Molecular Sciences, 2022, 23, 3905. | 4.1 | 4 |
| 25 | 8q13.1-q13.2 Deletion Associated With Inferior Cerebellar Vermian Hypoplasia and Digital Anomalies: A New Syndrome?. Pediatric Neurology, 2015, 52, 230-234.e1. | 2.1 | 3 |
| 26 | New Pathogenic Mutations Associated with Diacylglycerol O-Acyltransferase 1 Deficiency. Journal of Pediatrics, 2021, 233, 268-272. | 1.8 | 3 |
| 27 | Case report: North Carolina macular dystrophy misdiagnosed as congenital ocular toxoplasmosis. Molecular Vision, 2019, 25, 731-733. | 1.1 | 3 |
| 28 | Novel KIT mutation presenting as marked lentiginosis. Pediatric Dermatology, 2019, 36, 922-925. | 0.9 | 2 |
| 29 | Cavitating Lung Disease Associated With Sudden Death. Chest, 2022, 161, e279-e285. | 0.8 | 0 |