

CITATION REPORT

List of articles citing

An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy

DOI: 10.1038/ncb3201

Nature Cell Biology, 2015, 17, 1074-1087.

Source: <https://exaly.com/paper-pdf/62026866/citation-report.pdf>

Version: 2024-04-28

This report has been generated based on the citations recorded by exaly.com for the above article. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

| # | Paper | IF | Citations |
|-----|---|-----|-----------|
| 196 | Specialized Cilia in Mammalian Sensory Systems. <i>Cells</i> , 2015 , 4, 500-19 | 7.9 | 59 |
| 195 | C21orf2 is mutated in recessive early-onset retinal dystrophy with macular staphyloma and encodes a protein that localises to the photoreceptor primary cilium. 2015 , 99, 1725-31 | | 24 |
| 194 | Screen-based identification and validation of four new ion channels as regulators of renal ciliogenesis. 2015 , 128, 4550-9 | | 12 |
| 193 | Identification of Novel Mutations in the LRR-Cap Domain of C21orf2 in Japanese Patients With Retinitis Pigmentosa and Cone-Rod Dystrophy. 2016 , 57, 4255-63 | | 16 |
| 192 | GEMC1 is a critical regulator of multiciliated cell differentiation. <i>EMBO Journal</i> , 2016 , 35, 942-60 | 13 | 59 |
| 191 | Characterizing the morbid genome of ciliopathies. 2016 , 17, 242 | | 89 |
| 190 | Sfr1, a Sfi1 Repeat Protein, Modulates the Production of Cortical Row Basal Bodies. 2016 , 1, | | 3 |
| 189 | Identification and Correction of Mechanisms Underlying Inherited Blindness in Human iPSC-Derived Optic Cups. 2016 , 18, 769-781 | | 193 |
| 188 | A primer on the mouse basal body. 2016 , 5, 17 | | 27 |
| 187 | Isolated and Syndromic Retinal Dystrophy Caused by Biallelic Mutations in RCBTB1, a Gene Implicated in Ubiquitination. 2016 , 99, 470-80 | | 32 |
| 186 | An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. 2016 , 7, 11491 | | 134 |
| 185 | A non-canonical function of Plk4 in centriolar satellite integrity and ciliogenesis through PCM1 phosphorylation. 2016 , 17, 326-37 | | 33 |
| 184 | Ciliogenesis and the DNA damage response: a stressful relationship. 2016 , 5, 19 | | 30 |
| 183 | Centrosome positioning in non-dividing cells. 2016 , 253, 1007-21 | | 16 |
| 182 | Mutations in PIH1D3 Cause X-Linked Primary Ciliary Dyskinesia with Outer and Inner Dynein Arm Defects. 2017 , 100, 160-168 | | 92 |
| 181 | Centriole splitting caused by loss of the centrosomal linker protein C-NAP1 reduces centriolar satellite density and impedes centrosome amplification. <i>Molecular Biology of the Cell</i> , 2017 , 28, 736-745 | 3.5 | 19 |
| 180 | Axial spondylometaphyseal dysplasia is also caused by NEK1 mutations. <i>Journal of Human Genetics</i> , 2017 , 62, 503-506 | 4.3 | 18 |

| | | | |
|-----|--|------|-----|
| 179 | Systematic high-content genome-wide RNAi screens of endothelial cell migration and morphology. 2017 , 4, 170009 | | 16 |
| 178 | Homozygous variant in C21orf2 in a case of Jeune syndrome with severe thoracic involvement: Extending the phenotypic spectrum. 2017 , 173, 1698-1704 | | 9 |
| 177 | Cilia - The sensory antennae in the eye. 2017 , 60, 144-180 | | 75 |
| 176 | Comparative Proteomics Reveals Timely Transport into Cilia of Regulators or Effectors as a Mechanism Underlying Ciliary Disassembly. 2017 , 16, 2410-2418 | | 9 |
| 175 | Role of primary cilia in non-dividing and post-mitotic cells. 2017 , 369, 11-25 | | 24 |
| 174 | Retinal Degeneration and Regeneration-Lessons From Fishes and Amphibians. 2017 , 5, 67-78 | | 29 |
| 173 | Spatially and temporally regulating translation via mRNA-binding proteins in cellular and neuronal function. 2017 , 591, 1508-1525 | | 19 |
| 172 | Fifteen years of research on oral-facial-digital syndromes: from 1 to 16 causal genes. <i>Journal of Medical Genetics</i> , 2017 , 54, 371-380 | 5.8 | 58 |
| 171 | Exome sequencing for the differential diagnosis of ciliary chondrodysplasias: Example of a WDR35 mutation case and review of the literature. 2017 , 60, 658-666 | | 9 |
| 170 | Microtubule stabilization drives 3D centrosome migration to initiate primary ciliogenesis. <i>Journal of Cell Biology</i> , 2017 , 216, 3713-3728 | 7.3 | 39 |
| 169 | Genes and molecular pathways underpinning ciliopathies. 2017 , 18, 533-547 | | 618 |
| 168 | 1700012B09Rik, a FOXJ1 effector gene active in ciliated tissues of the mouse but not essential for motile ciliogenesis. 2017 , 429, 186-199 | | 4 |
| 167 | Motile and non-motile cilia in human pathology: from function to phenotypes. 2017 , 241, 294-309 | | 225 |
| 166 | Regulation of centriolar satellite integrity and its physiology. 2017 , 74, 213-229 | | 80 |
| 165 | The Cilium: Cellular Antenna and Central Processing Unit. <i>Trends in Cell Biology</i> , 2017 , 27, 126-140 | 18.3 | 189 |
| 164 | Bardet-Biedl Syndrome as a Chaperonopathy: Dissecting the Major Role of Chaperonin-Like BBS Proteins (BBS6-BBS10-BBS12). 2017 , 4, 55 | | 34 |
| 163 | Novel Insights Into the Phenotypical Spectrum of KIF11-Associated Retinopathy, Including a New Form of Retinal Ciliopathy. 2017 , 58, 3950-3959 | | 34 |
| 162 | A biallelic 36-bp insertion in PIBF1 is associated with Joubert syndrome. <i>Journal of Human Genetics</i> , 2018 , 63, 935-939 | 4.3 | 7 |

| | | |
|-----|---|---------|
| 161 | A CRISPR-based screen for Hedgehog signaling provides insights into ciliary function and ciliopathies. 2018 , 50, 460-471 | 84 |
| 160 | RNA Biology in Retinal Development and Disease. 2018 , 34, 341-351 | 21 |
| 159 | Whole-exome sequencing reveals POC5 as a novel gene associated with autosomal recessive retinitis pigmentosa. 2018 , 27, 614-624 | 14 |
| 158 | Elsahy-Waters syndrome is caused by biallelic mutations in CDH11. 2018 , 176, 477-482 | 9 |
| 157 | Expanding the genetic architecture and phenotypic spectrum in the skeletal ciliopathies. 2018 , 39, 152-166 | 51 |
| 156 | Novel genes associated with amyotrophic lateral sclerosis: diagnostic and clinical implications. 2018 , 17, 94-102 | 256 |
| 155 | Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. 2019 , 40, 267-280 | 9 |
| 154 | Bexarotene - a novel modulator of AURKA and the primary cilium in -deficient cells. 2018 , 131, | 4 |
| 153 | ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. 2018 , 103, 612-620 | 41 |
| 152 | A MAPK/c-Jun-mediated switch regulates the initial adaptive and cell death responses to mitochondrial damage in a neuronal cell model. 2018 , 104, 73-86 | 5 |
| 151 | Disrupted alternative splicing for genes implicated in splicing and ciliogenesis causes PRPF31 retinitis pigmentosa. 2018 , 9, 4234 | 85 |
| 150 | M-Phase Phosphoprotein 9 regulates ciliogenesis by modulating CP110-CEP97 complex localization at the mother centriole. 2018 , 9, 4511 | 34 |
| 149 | Ciliary signalling in cancer. 2018 , 18, 511-524 | 67 |
| 148 | Targeted deletion of the AAA-ATPase Ruvbl1 in mice disrupts ciliary integrity and causes renal disease and hydrocephalus. <i>Experimental and Molecular Medicine</i> , 2018 , 50, 1-17 | 12.8 13 |
| 147 | Functional analyses of Pericentrin and Syne-2 interaction in ciliogenesis. 2018 , 131, | 5 |
| 146 | Ganetespib limits ciliation and cystogenesis in autosomal-dominant polycystic kidney disease (ADPKD). 2018 , 32, 2735-2746 | 22 |
| 145 | Severe skeletal abnormalities caused by defects in retrograde intraflagellar transport dyneins. 2018 , 356-401 | 3 |
| 144 | Mutations in LNPK, Encoding the Endoplasmic Reticulum Junction Stabilizer Lunapark, Cause a Recessive Neurodevelopmental Syndrome. 2018 , 103, 296-304 | 10 |

| | | | |
|-----|---|-----|-----|
| 143 | Signaling through the Primary Cilium. <i>Frontiers in Cell and Developmental Biology</i> , 2018 , 6, 8 | 5-7 | 181 |
| 142 | Autosomal dominant retinitis pigmentosa-associated gene PRPF8 is essential for hypoxia-induced mitophagy through regulating ULK1 mRNA splicing. 2018 , 14, 1818-1830 | | 20 |
| 141 | The E3 ubiquitin ligase UBR5 regulates centriolar satellite stability and primary cilia. <i>Molecular Biology of the Cell</i> , 2018 , 29, 1542-1554 | 3-5 | 20 |
| 140 | The complexity of the cilium: spatiotemporal diversity of an ancient organelle. 2018 , 55, 139-149 | | 13 |
| 139 | SPATA7 maintains a novel photoreceptor-specific zone in the distal connecting cilium. <i>Journal of Cell Biology</i> , 2018 , 217, 2851-2865 | 7-3 | 26 |
| 138 | Motor Neuron Susceptibility in ALS/FTD. 2019 , 13, 532 | | 77 |
| 137 | Eye in a Disk: eyeIntegration Human Pan-Eye and Body Transcriptome Database Version 1.0. 2019 , 60, 3236-3246 | | 14 |
| 136 | The molecular genetics of Joubert syndrome and related ciliopathies: The challenges of genetic and phenotypic heterogeneity. 2019 , 4, 25-49 | | 41 |
| 135 | Splicing in the pathogenesis, diagnosis and treatment of ciliopathies. 2019 , 1862, 194433 | | 12 |
| 134 | Rab35 controls cilium length, function and membrane composition. 2019 , 20, e47625 | | 16 |
| 133 | Gpr63 is a modifier of microcephaly in Ttc21b mouse mutants. <i>PLoS Genetics</i> , 2019 , 15, e1008467 | 6 | 5 |
| 132 | Interplay between primary cilia, ubiquitin-proteasome system and autophagy. 2019 , 166, 286-292 | | 13 |
| 131 | The Nuclear Arsenal of Cilia. 2019 , 49, 161-170 | | 15 |
| 130 | 661W Photoreceptor Cell Line as a Cell Model for Studying Retinal Ciliopathies. <i>Frontiers in Genetics</i> , 2019 , 10, 308 | 4-5 | 21 |
| 129 | Opportunities and Challenges for Molecular Understanding of Ciliopathies-The 100,000 Genomes Project. <i>Frontiers in Genetics</i> , 2019 , 10, 127 | 4-5 | 29 |
| 128 | The Frog as a Model to Study Joubert Syndrome: The Case of a Human Patient With Compound Heterozygous Variants in. 2019 , 10, 134 | | 8 |
| 127 | A Combined , and Clinical Approach to Characterize Novel Pathogenic Missense Variants in PRPF31 in Retinitis Pigmentosa. <i>Frontiers in Genetics</i> , 2019 , 10, 248 | 4-5 | 4 |
| 126 | The ciliary Frizzled-like receptor Tmem67 regulates canonical Wnt/βcatenin signalling in the developing cerebellum via Hoxb5. <i>Scientific Reports</i> , 2019 , 9, 5446 | 4-9 | 6 |

| | | |
|-----|---|-------|
| 125 | Leucine Rich Repeat Proteins: Sequences, Mutations, Structures and Diseases. 2019 , 26, 108-131 | 28 |
| 124 | Polo-like kinase 4 maintains centriolar satellite integrity by phosphorylation of centrosomal protein 131 (CEP131). 2019 , 294, 6531-6549 | 13 |
| 123 | Joubert syndrome with multiple pituitary hormone deficiency. 2019 , 12, | 2 |
| 122 | The role of ubiquitination in the regulation of primary cilia assembly and disassembly. 2019 , 93, 145-152 | 10 |
| 121 | Insights into photoreceptor ciliogenesis revealed by animal models. 2019 , 71, 26-56 | 17 |
| 120 | The deubiquitinating enzyme Usp14 controls ciliogenesis and Hedgehog signaling. 2019 , 28, 764-777 | 19 |
| 119 | Prediction of structural consequences for disease causing variants in C21orf2 protein using computational approaches. 2019 , 37, 465-480 | 4 |
| 118 | Healthcare recommendations for Joubert syndrome. 2020 , 182, 229-249 | 32 |
| 117 | Cilia in cystic kidney and other diseases. 2020 , 69, 109519 | 15 |
| 116 | RNA Splicing Factor Mutations That Cause Retinitis Pigmentosa Result in Circadian Dysregulation. 2020 , 35, 72-83 | 1 |
| 115 | Identification of two novel pathogenic variants of PIBF1 by whole exome sequencing in a 2-year-old boy with Joubert syndrome. 2020 , 21, 192 | 1 |
| 114 | ALS Genetics: Gains, Losses, and Implications for Future Therapies. 2020 , 108, 822-842 | 72 |
| 113 | An Amyotrophic Lateral Sclerosis-Associated Mutant of C21ORF2 Is Stabilized by NEK1-Mediated Hyperphosphorylation and the Inability to Bind FBXO3. 2020 , 23, 101491 | 6 |
| 112 | Targeting E3 Ubiquitin Ligases and Deubiquitinases in Ciliopathy and Cancer. <i>International Journal of Molecular Sciences</i> , 2020 , 21, | 6.3 2 |
| 111 | A novel variant in C5ORF42 gene is associated with Joubert syndrome. 2020 , 47, 4099-4103 | 0 |
| 110 | Disease Modeling To Understand the Pathomechanisms of Human Genetic Kidney Disorders. 2020 , 15, 855-872 | 7 |
| 109 | mRNA Editing, Processing and Quality Control in. 2020 , 215, 531-568 | 9 |
| 108 | Expanding the Clinical and Molecular Heterogeneity of Nonsyndromic Inherited Retinal Dystrophies. 2020 , 22, 532-543 | 15 |

| | | | |
|-----|--|------|----|
| 107 | Mutation spectrum of PRPF31, genotype-phenotype correlation in retinitis pigmentosa, and opportunities for therapy. 2020 , 192, 107950 | | 11 |
| 106 | - associated microcephaly and chorioretinopathy. 2020 , 41, 189-193 | | 7 |
| 105 | Microtubule-associated proteins and emerging links to primary cilium structure, assembly, maintenance, and disassembly. 2021 , 288, 786-798 | | 4 |
| 104 | Whole-genome screen identifies diverse pathways that negatively regulate ciliogenesis. <i>Molecular Biology of the Cell</i> , 2021 , 32, 169-185 | 3.5 | 4 |
| 103 | A CRISPR and high-content imaging assay compliant with ACMG/AMP guidelines for clinical variant interpretation in ciliopathies. 2021 , 140, 593-607 | | 2 |
| 102 | The Role of the U5 snRNP in Genetic Disorders and Cancer. <i>Frontiers in Genetics</i> , 2021 , 12, 636620 | 4.5 | 3 |
| 101 | Ciliopathies and the Kidney: A Review. 2021 , 77, 410-419 | | 35 |
| 100 | Mendelian pathway analysis of laboratory traits reveals distinct roles for ciliary subcompartments in common disease pathogenesis. 2021 , 108, 482-501 | | 1 |
| 99 | Let-7, Lin28 and Hmga2 Expression in Ciliary Epithelium and Retinal Progenitor Cells. 2021 , 62, 31 | | |
| 98 | A ciliopathy complex builds distal appendages to initiate ciliogenesis. | | 0 |
| 97 | Clinical heterogeneity and intrafamilial variability of Joubert syndrome in two siblings with CPLANE1 variants. 2021 , 9, e1682 | | 1 |
| 96 | Hedgehog signaling and the primary cilium: implications for spatial and temporal constraints on signaling. <i>Development (Cambridge)</i> , 2021 , 148, | 6.6 | 10 |
| 95 | The TBC1D31/praja2 complex controls primary ciliogenesis through PKA-directed OFD1 ubiquitylation. <i>EMBO Journal</i> , 2021 , 40, e106503 | 13 | 3 |
| 94 | SANS (USH1G) regulates pre-mRNA splicing by mediating the intra-nuclear transfer of tri-snRNP complexes. <i>Nucleic Acids Research</i> , 2021 , 49, 5845-5866 | 20.1 | 5 |
| 93 | The ciliary impact of nonciliary gene mutations. <i>Trends in Cell Biology</i> , 2021 , 31, 876-887 | 18.3 | 4 |
| 92 | Amyotrophic Lateral Sclerosis: Molecular Mechanisms, Biomarkers, and Therapeutic Strategies. <i>Antioxidants</i> , 2021 , 10, | 7.1 | 7 |
| 91 | Primary ciliogenesis is a crucial step for multiciliated cell determinism in the respiratory epithelium. <i>Journal of Cellular and Molecular Medicine</i> , 2021 , 25, 7575-7579 | 5.6 | 2 |
| 90 | The role of splicing factors in retinitis pigmentosa: links to cilia. <i>Biochemical Society Transactions</i> , 2021 , 49, 1221-1231 | 5.1 | 0 |

| | | | |
|----|--|------|----|
| 89 | Ciliary GPCR-based transcriptome as a key regulator of cilia length control. <i>FASEB BioAdvances</i> , 2021 , 3, 744-767 | 2.8 | 4 |
| 88 | The ternary complex CEP90, FOPNL and OFD1 specifies the future location of centriolar distal appendages, and promotes their assembly. | | |
| 87 | CEP78 functions downstream of CEP350 to control biogenesis of primary cilia by negatively regulating CP110 levels. <i>ELife</i> , 2021 , 10, | 8.9 | 11 |
| 86 | Pre-mRNA Processing Factors and Retinitis Pigmentosa: RNA Splicing and Beyond. <i>Frontiers in Cell and Developmental Biology</i> , 2021 , 9, 700276 | 5.7 | 2 |
| 85 | A ciliopathy complex builds distal appendages to initiate ciliogenesis. <i>Journal of Cell Biology</i> , 2021 , 220, | 7.3 | 5 |
| 84 | A genome-wide association study of quantitative computed tomographic emphysema in Korean populations. <i>Scientific Reports</i> , 2021 , 11, 16692 | 4.9 | |
| 83 | Zebrafish Models for Human Skeletal Disorders. <i>Frontiers in Genetics</i> , 2021 , 12, 675331 | 4.5 | 0 |
| 82 | Primary cilia in retinal pigment epithelium development and diseases. <i>Journal of Cellular and Molecular Medicine</i> , 2021 , 25, 9084-9088 | 5.6 | 3 |
| 81 | Cilia kinases in skeletal development and homeostasis. <i>Developmental Dynamics</i> , 2021 , | 2.9 | |
| 80 | Genetic analysis in Chinese patients with familial or young-onset amyotrophic lateral sclerosis. <i>Neurological Sciences</i> , 2021 , 1 | 3.5 | 0 |
| 79 | Differential barcoding of opioid receptors trafficking. <i>Journal of Neuroscience Research</i> , 2021 , | 4.4 | 1 |
| 78 | Limitations and opportunities in the pharmacotherapy of ciliopathies. <i>Pharmacology & Therapeutics</i> , 2021 , 225, 107841 | 13.9 | 3 |
| 77 | Prpf31 is essential for the survival and differentiation of retinal progenitor cells by modulating alternative splicing. <i>Nucleic Acids Research</i> , 2021 , 49, 2027-2043 | 20.1 | 2 |
| 76 | KIF14 controls ciliogenesis via regulation of Aurora A and is important for Hedgehog signaling. <i>Journal of Cell Biology</i> , 2020 , 219, | 7.3 | 10 |
| 75 | A comprehensive portrait of cilia and ciliopathies from a CRISPR-based screen for Hedgehog signaling. | | 2 |
| 74 | Time-resolved proteomic profiling of the ciliary Hedgehog response reveals that GPR161 and PKA undergo regulated co-exit from cilia. | | 5 |
| 73 | IFT81 as a Candidate Gene for Nonsyndromic Retinal Degeneration. 2017 , 58, 2483-2490 | | 7 |
| 72 | Genome-wide suppressor screen identifies USP35/USP38 as therapeutic candidates for ciliopathies. <i>JCI Insight</i> , 2019 , 4, | 9.9 | 4 |

| | | | |
|----|---|-----|----|
| 71 | Polycystin-1 regulates ARHGAP35-dependent centrosomal RhoA activation and ROCK signaling. <i>JCI Insight</i> , 2020 , 5, | 9.9 | 9 |
| 70 | Formation of 53BP1 foci and ATM activation under oxidative stress is facilitated by RNA:DNA hybrids and loss of ATM-53BP1 expression promotes photoreceptor cell survival in mice. <i>F1000Research</i> , 2018 , 7, 1233 | 3.6 | 10 |
| 69 | Structural and Functional Recovery of Sensory Cilia in <i>C. elegans</i> IFT Mutants upon Aging. <i>PLoS Genetics</i> , 2016 , 12, e1006325 | 6 | 9 |
| 68 | Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. <i>PLoS ONE</i> , 2016 , 11, e0150555 | 3.7 | 21 |
| 67 | Ocular Ciliopathies: Genetic and Mechanistic Insights into Developing Therapies. <i>Current Medicinal Chemistry</i> , 2019 , 26, 3120-3131 | 4.3 | 4 |
| 66 | Primary Cilia, Ciliogenesis and the Actin Cytoskeleton: A Little Less Resorption, A Little More Actin Please. <i>Frontiers in Cell and Developmental Biology</i> , 2020 , 8, 622822 | 5.7 | 11 |
| 65 | An updated SYSCILIA gold standard (SCGSv2) of known ciliary genes, revealing the vast progress that has been made in the cilia research field. <i>Molecular Biology of the Cell</i> , 2021 , 32, br13 | 3.5 | 4 |
| 64 | Microtubule stabilization drives 3D centrosome migration to initiate primary ciliogenesis. | | 1 |
| 63 | The E3 ubiquitin ligase UBR5 regulates centriolar satellite stability and primary cilia formation via ubiquitylation of CSPP-L. | | |
| 62 | Human iPSC-derived RPE and retinal organoids reveal impaired alternative splicing of genes involved in pre-mRNA splicing in PRPF31 autosomal dominant retinitis pigmentosa. | | |
| 61 | Eye in a Disk: eyeIntegrat ion human pan-eye and body transcriptome database version 1.0. | | 0 |
| 60 | A Unique Manifestation of Bardet-Biedl Syndrome with Otolaryngologic Symptoms and Bronchopneumonia in a One-year-old Girl. <i>Cureus</i> , 2019 , 11, e5717 | 1.2 | 2 |
| 59 | Characterization of primary cilia features reveal cell-type specific variability in in vitro models of osteogenic and chondrogenic differentiation. <i>PeerJ</i> , 2020 , 8, e9799 | 3.1 | 1 |
| 58 | Molecular diagnoses in the congenital malformations caused by ciliopathies cohort of the 100,000 Genomes Project. <i>Journal of Medical Genetics</i> , 2021 , | 5.8 | 3 |
| 57 | Regulation of canonical Wnt signalling by the ciliopathy protein MKS1 and the E2 ubiquitin-conjugating enzyme UBE2E1. | | |
| 56 | High-throughput PRPF31 variant characterisation pipeline consistent with ACMG/AMP clinical variant interpretation guidelines. | | |
| 55 | CEP78 functions downstream of CEP350 to control biogenesis of primary cilia by negatively regulating CP110 levels. | | |
| 54 | Mendelian pathway analysis of laboratory traits reveals distinct roles for ciliary subcompartments in common disease pathogenesis. | | 0 |

| | | | |
|----|---|------|---|
| 53 | SANS (USH1G) regulates pre-mRNA splicing by mediating the intra-nuclear transfer of tri-snRNP complexes. | | |
| 52 | Drug and siRNA screens identify ROCK2 as a therapeutic target for ciliopathies. | | |
| 51 | LUBAC regulates ciliogenesis by promoting CP110 removal from the mother centriole. <i>Journal of Cell Biology</i> , 2022 , 221, | 7.3 | 4 |
| 50 | The Role of Centrosome Distal Appendage Proteins (DAPs) in Nephronophthisis and Ciliogenesis. <i>International Journal of Molecular Sciences</i> , 2021 , 22, | 6.3 | 2 |
| 49 | 3D-Structured Illumination Microscopy of Centrosomes in Human Cell Lines.. <i>Bio-protocol</i> , 2022 , 12, e43609 | 6.0 | 1 |
| 48 | Regulation of canonical Wnt signalling by the ciliopathy protein MKS1 and the E2 ubiquitin-conjugating enzyme UBE2E1.. <i>ELife</i> , 2022 , 11, | 8.9 | 0 |
| 47 | Reduced expression of TAZ inhibits primary cilium formation in renal glomeruli.. <i>Experimental and Molecular Medicine</i> , 2022 , | 12.8 | |
| 46 | Post-transcriptional and Post-translational Modifications of Primary Cilia: How to Fine Tune Your Neuronal Antenna.. <i>Frontiers in Cellular Neuroscience</i> , 2022 , 16, 809917 | 6.1 | 0 |
| 45 | C21orf2 mutations found in ALS disrupt primary cilia function. | | |
| 44 | Primary Cilia and Their Role in Acquired Heart Disease.. <i>Cells</i> , 2022 , 11, | 7.9 | 0 |
| 43 | Genome-wide association meta-analysis identifies novel ancestry-specific primary open-angle glaucoma loci and shared biology with vascular mechanisms and cell proliferation. | | 3 |
| 42 | Update on the Phenotypic and Genotypic Spectrum of -Related Retinopathy.. <i>Genes</i> , 2022 , 13, | 4.2 | 1 |
| 41 | Image_1.tif. 2019 , | | |
| 40 | Image_2.tif. 2019 , | | |
| 39 | Image_3.tiff. 2019 , | | |
| 38 | Table_1.XLSX. 2019 , | | |
| 37 | Table_2.XLSX. 2019 , | | |
| 36 | Table_3.XLSX. 2019 , | | |

| | | | |
|----|---|-----|---|
| 35 | Table_4.XLSX. 2019, | | |
| 34 | Table_5.XLSX. 2019, | | |
| 33 | Table_1.DOCX. 2020, | | |
| 32 | Image_1.JPEG. 2019, | | |
| 31 | Video_1.AVI. 2019, | | |
| 30 | Data_Sheet_1.docx. 2019, | | |
| 29 | Nomenclature of Genetic Movement Disorders: Recommendations of the International Parkinson and Movement Disorder Society Task Force - An Update.. <i>Movement Disorders</i> , 2022, 37, 905-935 | 7 | 3 |
| 28 | The Joubert-Meckel-Nephronophthisis Spectrum of Ciliopathies. <i>Annual Review of Genomics and Human Genetics</i> , 2022, 23, | 9-7 | 0 |
| 27 | Genetics of amyotrophic lateral sclerosis: seeking therapeutic targets in the era of gene therapy. <i>Journal of Human Genetics</i> , | 4-3 | 1 |
| 26 | The centrosomal protein 83 (CEP83) regulates human pluripotent stem cell differentiation towards the kidney lineage. | | |
| 25 | Proteolytic control in ciliogenesis: Temporal restriction or early initiation?. <i>BioEssays</i> , 2200087 | 4-1 | 0 |
| 24 | Recent Updates on the Genetics of Amyotrophic Lateral Sclerosis and Frontotemporal Dementia. <i>Molecular Neurobiology</i> , | 6-2 | 0 |
| 23 | TTC30A and TTC30B Redundancy Protects IFT Complex B Integrity and Its Pivotal Role in Ciliogenesis. <i>Genes</i> , 2022, 13, 1191 | 4-2 | |
| 22 | Whole Exome Sequencing in a Population With Severe Congenital Anomalies of Kidney and Urinary Tract. 10, | | |
| 21 | Modeling PRPF31 retinitis pigmentosa using retinal pigment epithelium and organoids combined with gene augmentation rescue. 2022, 7, | | 1 |
| 20 | Microtubule modification defects underlie cilium degeneration in cell models of retinitis pigmentosa associated with pre-mRNA splicing factor mutations. 13, | | 0 |
| 19 | Genetics behind Cerebral Disease with Ocular Comorbidity: Finding Parallels between the Brain and Eye Molecular Pathology. 2022, 23, 9707 | | 0 |
| 18 | Interaction with C21ORF2 controls the cellular functioning of the NEK1 kinase. | | 0 |

- 17 Genetic treatment for autosomal dominant inherited retinal dystrophies: approaches, challenges and targeted genotypes. *bjophthalmol-2022-321903* ○
- 16 Retinal organoids provide unique insights into molecular signatures of inherited retinal disease throughout retinogenesis. ○
- 15 The evolutionary conserved proteins CEP90, FOPNL, and OFD1 recruit centriolar distal appendage proteins to initiate their assembly. **2022**, 20, e3001782 ○
- 14 Structural studies of cilia and flagella associated protein 410 (CFAP410) reveal its bimodular organization with an N-terminal LRR motif and a C-terminal tetrameric helical bundle. ○
- 13 Cilia regeneration requires an RNA splicing factor from the ciliary base. **2022**, 11, ○
- 12 The centrosomal protein 83 (CEP83) regulates human pluripotent stem cell differentiation toward the kidney lineage. 11, ○
- 11 Autophagy and the primary cilium in cell metabolism: What's upstream?. 10, ○
- 10 A case of siblings with juvenile retinitis pigmentosa associated with NEK1 gene variants. 1-6 ○
- 9 Gene augmentation prevents retinal degeneration in a CRISPR/Cas9-based mouse model of PRPF31 retinitis pigmentosa. **2022**, 13, 1
- 8 A Phylogenetic Profiling Approach Identifies Novel Ciliogenesis Genes In *Drosophila* And *C. elegans*. ○
- 7 New insights into the centrosome-associated spliceosome components as regulators of ciliogenesis and tissue identity. ○
- 6 A targeted multi-proteomics approach generates a blueprint of the ciliary ubiquitinome. 11, ○
- 5 Novel multi-allelic variants, two BBS2 and one PKD1 variant, of renal ciliopathies: A case report and literature review. **2023**, 66, 104753 ○
- 4 The phosphorylation of PHF5A by TrkA-ERK1/2-ABL1 cascade regulates centrosome separation. **2023**, 14, ○
- 3 Primary cilia sense glutamine availability and respond via asparagine synthetase. **2023**, 5, 385-397 ○
- 2 C21orf2 variants causing inherited retinal disease: A review of what we know and a report of two new suspected cases. **2023**, 11, ○
- 1 CilioGenics: an integrated method and database for predicting novel ciliary genes. ○