

List of Publications by Year in descending order

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CHAOLI

| # | Article | IF | CITATIONS |
|----|--|------|-----------|
| 1 | Leukemic IDH1 and IDH2 Mutations Result inÂa Hypermethylation Phenotype, Disrupt TET2 Function, and Impair Hematopoietic Differentiation. Cancer Cell, 2010, 18, 553-567. | 7.7 | 2,328 |
| 2 | IDH mutation impairs histone demethylation and results in a block to cell differentiation. Nature, 2012, 483, 474-478. | 13.7 | 1,693 |
| 3 | IDH1 mutation is sufficient to establish the glioma hypermethylator phenotype. Nature, 2012, 483, 479-483. | 13.7 | 1,668 |
| 4 | Metabolic Regulation of Epigenetics. Cell Metabolism, 2012, 16, 9-17. | 7.2 | 568 |
| 5 | Hypoxia Induces Production of L-2-Hydroxyglutarate. Cell Metabolism, 2015, 22, 304-311. | 7.2 | 374 |
| 6 | Serine Catabolism Regulates Mitochondrial Redox Control during Hypoxia. Cancer Discovery, 2014, 4, 1406-1417. | 7.7 | 342 |
| 7 | The histone mark H3K36me2 recruits DNMT3A and shapes the intergenic DNA methylation landscape. Nature, 2019, 573, 281-286. | 13.7 | 338 |
| 8 | Histone H3K36 mutations promote sarcomagenesis through altered histone methylation landscape. Science, 2016, 352, 844-849. | 6.0 | 327 |
| 9 | The hexosamine biosynthetic pathway couples growth factor-induced glutamine uptake to glucose metabolism. Genes and Development, 2010, 24, 2784-2799. | 2.7 | 315 |
| 10 | Ammonia-induced autophagy is independent of ULK1/ULK2 kinases. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 11121-11126. | 3.3 | 311 |
| 11 | Therapeutic targets in cancer cell metabolism and autophagy. Nature Biotechnology, 2012, 30, 671-678. | 9.4 | 310 |
| 12 | Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. Nature Genetics, 2015, 47, 717-726. | 9.4 | 310 |
| 13 | DNA Hydroxymethylation Profiling Reveals that WT1 Mutations Result in Loss of TET2 Function in Acute Myeloid Leukemia. Cell Reports, 2014, 9, 1841-1855. | 2.9 | 237 |
| 14 | Bad bones, absent smell, selfish testes: The pleiotropic consequences of human FGF receptor mutations. Cytokine and Growth Factor Reviews, 2005, 16, 187-203. | 3.2 | 223 |
| 15 | H3K27M induces defective chromatin spread of PRC2-mediated repressive H3K27me2/me3 and is essential for glioma tumorigenesis. Nature Communications, 2019, 10, 1262. | 5.8 | 215 |
| 16 | Impaired H3K36 methylation defines a subset of head and neck squamous cell carcinomas. Nature Genetics, 2017, 49, 180-185. | 9.4 | 195 |
| 17 | FGFs, their receptors, and human limb malformations: Clinical and molecular correlations. American Journal of Medical Genetics Part A, 2002, 112, 266-278. | 2.4 | 186 |
| 18 | Cancer-associated IDH2 mutants drive an acute myeloid leukemia that is susceptible to Brd4 inhibition. Genes and Development, 2013, 27, 1974-1985. | 2.7 | 165 |

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|----|---|------|-----------|
| 19 | A biallelic mutation in <i>IL6ST</i> encoding the GP130 co-receptor causes immunodeficiency and craniosynostosis. Journal of Experimental Medicine, 2017, 214, 2547-2562. | 4.2 | 158 |
| 20 | Functional interrogation of DNA damage response variants with base editing screens. Cell, 2021, 184, 1081-1097.e19. | 13.5 | 145 |
| 21 | Lowered H3K27me3 and DNA hypomethylation define poorly prognostic pediatric posterior fossa ependymomas. Science Translational Medicine, 2016, 8, 366ra161. | 5.8 | 144 |
| 22 | Clinical genetics of craniosynostosis. Current Opinion in Pediatrics, 2017, 29, 622-628. | 1.0 | 142 |
| 23 | The Potential for Isocitrate Dehydrogenase Mutations to Produce 2-Hydroxyglutarate Depends on Allele Specificity and Subcellular Compartmentalization. Journal of Biological Chemistry, 2013, 288, 3804-3815. | 1.6 | 141 |
| 24 | Induction of sarcomas by mutant IDH2. Genes and Development, 2013, 27, 1986-1998. | 2.7 | 135 |
| 25 | New insights into craniofacial malformations. Human Molecular Genetics, 2015, 24, R50-R59. | 1.4 | 122 |
| 26 | Targeting p53 for enhanced radio- and chemo-sensitivity. Apoptosis: an International Journal on Programmed Cell Death, 2009, 14, 597-606. | 2.2 | 116 |
| 27 | Specific detection of methionine 27 mutation in histone 3 variants (H3K27M) in fixed tissue from high-grade astrocytomas. Acta Neuropathologica, 2014, 128, 733-741. | 3.9 | 116 |
| 28 | Molecular basis for oncohistone H3 recognition by SETD2 methyltransferase. Genes and Development, 2016, 30, 1611-1616. | 2.7 | 111 |
| 29 | The interplay between DNA and histone methylation: molecular mechanisms and disease implications. EMBO Reports, 2021, 22, e51803. | 2.0 | 83 |
| 30 | SWI/SNF complex in cancer. Nature Genetics, 2017, 49, 178-179. | 9.4 | 82 |
| 31 | Clinical dividends from the molecular genetic diagnosis of craniosynostosisâ€. American Journal of Medical Genetics, Part A, 2007, 143A, 1941-1949. | 0.7 | 75 |
| 32 | DNMT1 Is Regulated by ATP-Citrate Lyase and Maintains Methylation Patterns during Adipocyte Differentiation. Molecular and Cellular Biology, 2013, 33, 3864-3878. | 1.1 | 75 |
| 33 | A Recurrent Mosaic Mutation in SMO , Encoding the Hedgehog Signal Transducer Smoothened, Is the Major Cause of Curry-Jones Syndrome. American Journal of Human Genetics, 2016, 98, 1256-1265. | 2.6 | 70 |
| 34 | Generation of functional lungs via conditional blastocyst complementation using pluripotent stem cells. Nature Medicine, 2019, 25, 1691-1698. | 15.2 | 69 |
| 35 | Genome analyses of the new model protist <i>Euplotes vannus</i> focusing on genome rearrangement and resistance to environmental stressors. Molecular Ecology Resources, 2019, 19, 1292-1308. | 2.2 | 69 |
| 36 | Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. American Journal of Human Genetics, 2018, 102, 468-479. | 2.6 | 63 |

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| 37 | Clinical dividends from the molecular genetic diagnosis of craniosynostosis. American Journal of Medical Genetics, Part A, 2006, 140A, 2631-2639. | 0.7 | 60 |
| 38 | The Mitochondrial Permeability Transition Regulates Cytochrome c Release for Apoptosis during Endoplasmic Reticulum Stress by Remodeling the Cristae Junction. Journal of Biological Chemistry, 2008, 283, 3476-3486. | 1.6 | 59 |
| 39 | Two competing mechanisms of DNMT3A recruitment regulate the dynamics of de novo DNA methylation at PRC1-targeted CpG islands. Nature Genetics, 2021, 53, 794-800. | 9.4 | 59 |
| 40 | Genomic aberrations frequently alter chromatin regulatory genes in chordoma. Genes Chromosomes and Cancer, 2016, 55, 591-600. | 1.5 | 58 |
| 41 | Histone H3.3 G34 mutations promote aberrant PRC2 activity and drive tumor progression. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 27354-27364. | 3.3 | 57 |
| 42 | Oncogenic Mechanisms of Histone H3 Mutations. Cold Spring Harbor Perspectives in Medicine, 2017, 7, a026443. | 2.9 | 56 |
| 43 | Selfish mutations dysregulating RAS-MAPK signaling are pervasive in aged human testes. Genome Research, 2018, 28, 1779-1790. | 2.4 | 56 |
| 44 | Histone 3 Lysine 9 Trimethylation Is Differentially Associated With Isocitrate Dehydrogenase Mutations in Oligodendrogliomas and High-Grade Astrocytomas. Journal of Neuropathology and Experimental Neurology, 2013, 72, 298-306. | 0.9 | 51 |
| 45 | Pparg promotes differentiation and regulates mitochondrial gene expression in bladder epithelial cells. Nature Communications, 2019, 10, 4589. | 5.8 | 50 |
| 46 | Role of calcium and cyclophilin D in the regulation of mitochondrial permeabilization induced by glutathione depletion. Biochemical and Biophysical Research Communications, 2007, 363, 572-577. | 1.0 | 47 |
| 47 | Resveratrol blocks high glucose–induced mitochondrial reactive oxygen species production in bovine aortic endothelial cells: role of phase 2 enzyme induction?. Diabetes, Obesity and Metabolism, 2008, 10, 347-349. | 2.2 | 46 |
| 48 | Chromatin dysregulation associated with NSD1 mutation in head and neck squamous cell carcinoma. Cell Reports, 2021, 34, 108769. | 2.9 | 42 |
| 49 | De Novo Missense Substitutions in the Gene Encoding CDK8, a Regulator of the Mediator Complex, Cause a Syndromic Developmental Disorder. American Journal of Human Genetics, 2019, 104, 709-720. | 2.6 | 41 |
| 50 | De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203. | 2.6 | 37 |
| 51 | Is Antioxidant Potential of the Mitochondrial Targeted Ubiquinone Derivative MitoQ Conserved in Cells Lacking mtDNA?. Antioxidants and Redox Signaling, 2008, 10, 651-660. | 2.5 | 36 |
| 52 | Non-genotoxic anti-neoplastic effects of ellipticine derivative NSC176327 in p53-deficient human colon carcinoma cells involve stimulation of p73. Cancer Biology and Therapy, 2008, 7, 2039-2046. | 1,5 | 32 |
| 53 | Depletion of H3K36me2 recapitulates epigenomic and phenotypic changes induced by the H3.3K36M oncohistone mutation. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, . | 3.3 | 31 |
| 54 | Targeting integrated epigenetic and metabolic pathways in lethal childhood PFA ependymomas. Science Translational Medicine, 2021, 13, eabc0497. | 5.8 | 29 |

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|----|--|-----|-----------|
| 55 | Pparg signaling controls bladder cancer subtype and immune exclusion. Nature Communications, 2021, 12, 6160. | 5.8 | 28 |
| 56 | Truncated SALL1 Impedes Primary Cilia Function in Townes-Brocks Syndrome. American Journal of Human Genetics, 2018, 102, 249-265. | 2.6 | 27 |
| 57 | TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459. | 1.1 | 26 |
| 58 | Histone H3.3 K27M and K36M mutations de-repress transposable elements through perturbation of antagonistic chromatin marks. Molecular Cell, 2021, 81, 4876-4890.e7. | 4.5 | 26 |
| 59 | Symplastic/pseudoanaplastic giant cell tumor of the bone. Skeletal Radiology, 2016, 45, 929-935. | 1.2 | 25 |
| 60 | Localized TWIST1 and TWIST2 basic domain substitutions cause four distinct human diseases that can be modeled in Caenorhabditis elegans. Human Molecular Genetics, 2017, 26, 2118-2132. | 1.4 | 21 |
| 61 | Identification of Intragenic Exon Deletions and Duplication of <i>TCF12</i> by Whole Genome or Targeted Sequencing as a Cause of <i>TCF12</i> Related Craniosynostosis. Human Mutation, 2016, 37, 732-736. | 1.1 | 19 |
| 62 | De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. American Journal of Human Genetics, 2020, 106, 830-845. | 2.6 | 17 |
| 63 | The INO80 Complex Regulates Epigenetic Inheritance of Heterochromatin. Cell Reports, 2020, 33, 108561. | 2.9 | 17 |
| 64 | Apparently synonymous substitutions in FGFR2affect splicing and result in mild Crouzon syndrome. BMC Medical Genetics, 2014, 15, 95. | 2.1 | 14 |
| 65 | Many faces of SMCHD1. Nature Genetics, 2017, 49, 176-178. | 9.4 | 14 |
| 66 | Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611. | 1.1 | 14 |
| 67 | Evaluating the performance of a clinical genome sequencing program for diagnosis of rare genetic disease, seen through the lens of craniosynostosis. Genetics in Medicine, 2021, 23, 2360-2368. | 1.1 | 13 |
| 68 | CHAF1A/B mediate silencing of unintegrated HIV-1 DNAs early in infection. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, . | 3.3 | 13 |
| 69 | Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758. | 2.6 | 13 |
| 70 | Association of mutations in FLNA with craniosynostosis. European Journal of Human Genetics, 2015, 23, 1684-1688. | 1.4 | 10 |
| 71 | Disruption of <i>TWIST1</i> translation by 5′ UTR variants in Saethre-Chotzen syndrome. Human Mutation, 2018, 39, 1360-1365. | 1.1 | 10 |
| 72 | <i>PIK3CA</i> and <i>p53</i> Mutations Promote 4NQO-Initated Head and Neck Tumor Progression and Metastasis in Mice. Molecular Cancer Research, 2020, 18, 822-834. | 1.5 | 10 |

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|----|--|-----|-----------|
| 73 | The histone H3K9M mutation synergizes with H3K14 ubiquitylation to selectively sequester histone H3K9 methyltransferase Clr4 at heterochromatin. Cell Reports, 2021, 35, 109137. | 2.9 | 8 |
| 74 | Oncohistones: Hijacking the Histone Code. Annual Review of Cancer Biology, 2022, 6, 293-312. | 2.3 | 8 |
| 75 | Feeding, Communication, Hydrocephalus, and Intracranial Hypertension in Patients With Severe FGFR2-Associated Pfeiffer Syndrome. Journal of Craniofacial Surgery, 2021, 32, 134-140. | 0.3 | 7 |
| 76 | The cAMP signaling pathway regulates Epe1 protein levels and heterochromatin assembly. PLoS Genetics, 2022, 18, e1010049. | 1.5 | 6 |
| 77 | Chromosome 3p loss of heterozygosity and reduced expression of H3K36me3 correlate with longer relapse-free survival in sacral conventional chordoma. Human Pathology, 2020, 104, 73-83. | 1.1 | 5 |
| 78 | Growth Inhibition and Induction of Innate Immune Signaling of Chondrosarcomas with Epigenetic Inhibitors. Molecular Cancer Therapeutics, 2021, 20, 2362-2371. | 1.9 | 4 |
| 79 | Pitfalls in the phylogenomic evaluation of human disease-causing mutations. Journal of Biology, 2009, 8, 26. | 2.7 | 3 |
| 80 | A custom-designed panel sequencing study in 201 Chinese patients with craniosynostosis revealed novel variants and distinct mutation spectra. Journal of Genetics and Genomics, 2021, 48, 167-171. | 1.7 | 3 |
| 81 | Decoding the function of an oncogenic transcription factor: finding the first responders. Molecular Cell, 2021, 81, 418-420. | 4.5 | 1 |