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List of Publications by Year
in descending order

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54 papers	1,913 citations	257101 24 h-index	276539 41 g-index
54 all docs	54 docs citations	54 times ranked	3069 citing authors

#	Article	IF	Citations
1	HRAS mutation analysis in Costello syndrome: Genotype and phenotype correlation. American Journal of Medical Genetics, Part A, 2006, 140A, 1-7.	0.7	164
2	Diamondâ€“Blackfan anemia with mandibulofacial dystostosis is heterogeneous, including the novel DBA genes TSR2 and RPS28. American Journal of Medical Genetics, Part A, 2014, 164, 2240-2249.	0.7	121
3	A novel rasopathy caused by recurrent de novo missense mutations in PPP1CB closely resembles Noonan syndrome with loose anagen hair. American Journal of Medical Genetics, Part A, 2016, 170, 2237-2247.	0.7	117
4	Mutations Impairing GSK3-Mediated MAF Phosphorylation Cause Cataract, Deafness, Intellectual Disability, Seizures, and a Down Syndrome-like Facies. American Journal of Human Genetics, 2015, 96, 816-825.	2.6	102
5	GGC Repeat Expansion and Exon 1 Methylation of XYLT1 Is a Common Pathogenic Variant in Baratela-Scott Syndrome. American Journal of Human Genetics, 2019, 104, 35-44.	2.6	81
6	Somatic mosaicism for anHRAS mutation causes Costello syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 2163-2169.	0.7	79
7	Further delineation of the phenotype resulting fromBRAForMEK1germline mutations helps differentiate cardio-facio-cutaneous syndrome from Costello syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1472-1480.	0.7	79
8	Paternal bias in parental origin ofHRASmutations in Costello syndrome. Human Mutation, 2006, 27, 736-741.	1.1	65
9	Maleâ€toâ€male transmission of Costello syndrome: G12S HRAS germline mutation inherited from a father with somatic mosaicism. American Journal of Medical Genetics, Part A, 2009, 149A, 315-321.	0.7	62
10	Costello syndrome associated with novel germline HRAS mutations: An attenuated phenotype?. American Journal of Medical Genetics, Part A, 2008, 146A, 683-690.	0.7	61
11	Truncating mutations in the last exon of NOTCH3 cause lateral meningocele syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 271-281.	0.7	59
12	SMN1 and SMN2 copy numbers in cell lines derived from patients with spinal muscular atrophy as measured by array digital PCR. Molecular Genetics & Genomic Medicine, 2015, 3, 248-257.	0.6	56
13	Evolution of placentally expressed cathepsins. Biochemical and Biophysical Research Communications, 2002, 293, 23-29.	1.0	55
14	Phenotypic analysis of individuals with Costello syndrome due to HRAS p.G13C. , 2011, 155, 706-716.		55
15	A new major histocompatibility complex class Iâ€Šb gene expressed in the mouse blastocyst and placenta. Immunogenetics, 1996, 45, 108-120.	1.2	47
16	A PLP splicing abnormality is associated with an unusual presentation of PMD. Annals of Neurology, 2002, 52, 477-488.	2.8	47
17	A phase I trial and viral clearance study of reovirus (Reolysin) in children with relapsed or refractory extraâ€cranial solid tumors: A Children's Oncology Group Phase I Consortium report. Pediatric Blood and Cancer, 2015, 62, 751-758.	0.8	47
18	Hepatoblastoma and heart transplantation in a patient with cardio-facio-cutaneous syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1481-1488.	0.7	39

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19	Univariate and bivariate variance component linkage analysis of a whole-genome scan for loci contributing to bone mineral density. European Journal of Human Genetics, 2005, 13, 781-788.	1.4	38
20	Expanding the SHOC2 mutation associated phenotype of noonan syndrome with loose anagen hair: Structural brain anomalies and myelofibrosis. American Journal of Medical Genetics, Part A, 2013, 161, 2420-2430.	0.7	38
21	Longitudinal assessment of cognitive characteristics in Costello syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 3185-3193.	0.7	36
22	Exploring whole genome amplification as a DNA recovery tool for molecular genetic studies. Journal of Biomolecular Techniques, 2005, 16, 125-33.	0.8	34
23	Plasma Membrane Ca2+-ATPase 4 in Murine Epididymis: Secretion of Splice Variants in the Luminal Fluid and a Role in Sperm Maturation1. Biology of Reproduction, 2013, 89, 6.	1.2	33
24	A new polymorphism in the proteolipid protein (PLP1) gene and its use for carrier detection ofPLP1 gene duplication in Pelizaeus-Merzbacher disease. Human Mutation, 2001, 17, 152-152.	1.1	28
25	Longitudinal course of cognitive, adaptive, and behavioral characteristics in Costello syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 2666-2672.	0.7	28
26	CNS imaging is a key diagnostic tool in the evaluation of patients with CFC syndrome: Two cases and literature review. American Journal of Medical Genetics, Part A, 2011, 155, 605-611.	0.7	24
27	Phenotypic spectrum of Costello syndrome individuals harboring the rare HRAS mutation p.Gly13Asp. , 2017, 173, 1309-1318.		24
28	Paternal uniparental disomy with segmental loss of heterozygosity of chromosome 11 are hallmark characteristics of syndromic and sporadic embryonal rhabdomyosarcoma. American Journal of Medical Genetics, Part A, 2016, 170, 3197-3206.	0.7	23
29	A novel HRAS substitution (c.266C>G; p.S89C) resulting in decreased downstream signaling suggests a new dimension of RAS pathway dysregulation in human development. American Journal of Medical Genetics, Part A, 2012, 158A, 2106-2118.	0.7	20
30	An attenuated phenotype of Costello syndrome in three unrelated individuals with a HRAS c.179G>A (p.Gly60Asp) mutation correlates with uncommon functional consequences. American Journal of Medical Genetics, Part A, 2015, 167, 2085-2097.	0.7	20
31	Normative growth charts for individuals with Costello syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2692-2699.	0.7	18
32	Assessing genotypeâ€“phenotype correlation in Costello syndrome using a severity score. Genetics in Medicine, 2013, 15, 554-557.	1.1	18
33	Molecular confirmation of HRAS p.G12S in siblings with Costello syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 2263-2268.	0.7	16
34	Evolution of Placental Proteases. Biological Chemistry, 2002, 383, 1113-8.	1.2	15
35	Establishing a reference dataset for the authentication of spinal muscular atrophy cell lines using STR profiling and digital PCR. Neuromuscular Disorders, 2017, 27, 439-446.	0.3	15
36	Detection of SMN1 to SMN2 gene conversion events and partial SMN1 gene deletions using array digital PCR. Neurogenetics, 2021, 22, 53-64.	0.7	14

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37	Transmission of the rare HRAS mutation (c. 173Câ€‰>â€‰T; p.T58I) further illustrates its attenuated phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 1095-1101.	0.7	13
38	Living with Costello syndrome: Quality of life issues in older individuals. American Journal of Medical Genetics, Part A, 2010, 152A, 84-90.	0.7	12
39	Paternal uniparental disomy 11p15.5 in the pancreatic nodule of an infant with Costello syndrome: Shared mechanism for hyperinsulinemic hypoglycemia in neonates with Costello and Beckwithâ€“Wiedemann syndrome and somatic loss of heterozygosity in Costello syndrome driving clonal expansion. American Journal of Medical Genetics, Part A, 2016, 170, 559-564.	0.7	11
40	Verbal memory functioning in adolescents and young adults with costello syndrome: Evidence for relative preservation in recognition memory. American Journal of Medical Genetics, Part A, 2013, 161, 2258-2265.	0.7	10
41	Ageâ€related differences in prevalence of autism spectrum disorder symptoms in children and adolescents with Costello syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1294-1300.	0.7	10
42	Pulmonary immune cell transcriptome changes in double-hit model of BPD induced by chorioamnionitis and postnatal hyperoxia. Pediatric Research, 2021, 90, 565-575.	1.1	10
43	Failure of Shortening and Inversion of the Perinatal Gubernaculum in the Cryptorchid Long-Evans orl Rat. Journal of Urology, 2006, 176, 1612-1617.	0.2	9
44	A newly recognized syndrome with characteristic facial features, skeletal dysplasia, and developmental delay. American Journal of Medical Genetics, Part A, 2012, 158A, 1815-1822.	0.7	9
45	An Integrated Approach for Analyzing Clinical Genomic Variant Data from Next-Generation Sequencing. Journal of Biomolecular Techniques, 2015, 26, 19-28.	0.8	9
46	Early-Lethal Costello Syndrome Due to Rare HRAS Tandem Base Substitution (c.35_36GC>AA;) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 38 421-430.	0.5	8
47	Cytotoxicity of Zardaverine in Embryonal Rhabdomyosarcoma from a Costello Syndrome Patient. Frontiers in Oncology, 2017, 7, 42.	1.3	7
48	MurineSpam1 mRNA: Involvement of AU-rich elements in the 3â€²UTR and antisense RNA in its tight post-transcriptional regulation in spermatids. Molecular Reproduction and Development, 2006, 73, 247-255.	1.0	6
49	Medically actionable comorbidities in adults with Costello syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 130-136.	0.7	6
50	The novel duplication HRAS c.186_206dup p.(Glu62_Arg68dup): clinical and functional aspects. European Journal of Human Genetics, 2020, 28, 1548-1554.	1.4	6
51	PCR identification of class I major histocompatibility complex genes transcribed in mouse blastocyst and placenta. Journal of Reproductive Immunology, 1997, 33, 31-43.	0.8	5
52	Genomic copy number variation association study in Caucasian patients with nonsyndromic cryptorchidism. BMC Urology, 2016, 16, 62.	0.6	4
53	Expression of PMCA4a in the Extratesticular Pathway and Accessory Organs of the Mouse. FASEB Journal, 2012, 26, 602.1.	0.2	0
54	Plasma Membrane Ca2+â€ATPase 4 Splice Variants are Acquired by Murine Sperm During Epididymal Transit via Epididymosomes. FASEB Journal, 2013, 27, 590.7.	0.2	0