Martina Rinelli

List of Publications by Year
in descending order

23 papers	220 citations	1305906 8 h-index	1255698 13 g-index
23 all docs	23 docs citations	23 times ranked	355 citing authors

#	Article	IF	Citations
1	A Novel CHEK2 Variant Identified by Next-Generation Sequencing in an Italian Family with Li-Fraumeni Syndrome: Case Report. SN Comprehensive Clinical Medicine, 2022, 4, 1.	0.3	0
2	Liquid Biopsy with Detection of NRASQ61K Mutation in Cerebrospinal Fluid: An Alternative Tool for the Diagnosis of Primary Pediatric Leptomeningeal Melanoma. Diagnostics, 2022, 12, 1609.	1.3	2
3	Effectiveness of emicizumab in preventing lifeâ€threatening bleeding complications in type 3 von Willebrand disease with inhibitors: A paediatric report. Haemophilia, 2021, 27, e495-e497.	1.0	7
4	Infantile myofibromatosis: a rare cause of subcutaneous nodules in an infant. Archives of Disease in Childhood, 2021, 106, 428-428.	1.0	1
5	Rhabdoid Tumor Predisposition Syndrome: From Clinical Suspicion to General Management. Frontiers in Oncology, 2021, 11, 586288.	1.3	20
6	Medulloblastoma Associated with Down Syndrome: From a Rare Event Leading to a Pathogenic Hypothesis. Diagnostics, 2021, 11, 254.	1.3	3
7	Molecular Characterization of Medulloblastoma in a Patient with Neurofibromatosis Type 1: Case Report and Literature Review. Diagnostics, 2021, 11, 647.	1.3	4
8	Expansion of the clinical and molecular spectrum of an <scp>XPD</scp>â€related disorder linked to biallelic mutations in <scp>ERCC2</scp> gene. Clinical Genetics, 2021, 99, 842-848.	1.0	4
9	Gastric cancer, inflammatory bowel disease and polyautoimmunity in a 17-year-old boy. European Journal of Gastroenterology and Hepatology, 2021, Publish Ahead of Print, .	0.8	5
10	Ectodermal Dysplasia-Syndactyly Syndrome with Toe-Only Minimal Syndactyly Due to a Novel Mutation in NECTIN4: A Case Report and Literature Review. Genes, 2021, 12, 748.	1.0	3
11	Establishment and Characterization of a Cell Line (S-RMS1) Derived from an Infantile Spindle Cell Rhabdomyosarcoma with SRF-NCOA2 Fusion Transcript. International Journal of Molecular Sciences, 2021, 22, 5484.	1.8	4
12	Pediatric gastrointestinal stromal tumor: Report of two novel patients harboring germline variants in SDHB and SDHC genes. Cancer Genetics, 2020, 241, 61-65.	0.2	4
13	Cancer Predisposition Syndromes Associated With Pediatric High-Grade Gliomas. Frontiers in Pediatrics, 2020, 8, 561487.	0.9	8
14	Cancer Predisposition Syndromes and Medulloblastoma in the Molecular Era. Frontiers in Oncology, 2020, 10, 566822.	1.3	17
15	Low-Grade Gliomas in Patients with Noonan Syndrome: Case-Based Review of the Literature. Diagnostics, 2020, 10, 582.	1.3	21
16	KBG syndrome: Common and uncommon clinical features based on 31 new patients. American Journal of Medical Genetics, Part A, 2020, 182, 1073-1083.	0.7	27
17	Targeting Epidermal Growth Factor Receptor (EGFR) in Pediatric Colorectal Cancer. Cancers, 2020, 12, 414.	1.7	2
18	DICER1 Syndrome and Cancer Predisposition: From a Rare Pediatric Tumor to Lifetime Risk. Frontiers in Oncology, 2020, 10, 614541.	1.3	30

#	Article	IF	Citations
19	Structural modeling of a novel TERC variant in a patient with aplastic anemia and short telomeres. Annals of Hematology, 2019, 98, 805-807.	0.8	1
20	Novel clinical features associated with Clouston syndrome. International Journal of Dermatology, 2019, 58, e143-e146.	0.5	8
21	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	2.6	23
22	Proliferative vasculopathy and hydranencephalyâ€“hydrocephaly syndrome or Fowler syndrome: Report of a family and insight into the disease's mechanism. Molecular Genetics & Genomic Medicine, 2018, 6, 446-451.	0.6	11
23	Paroxysmal features responding to flunarizine in a child with rapid-onset dystonia-parkinsonism. Neurology, 2014, 82, 2037-2038.	1.5	15