Manuela Morleo

List of Publications by Year in descending order

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28 papers	955 citations	623734 14 h-index	526287 27 g-index
30 all docs	30 docs citations	30 times ranked	2343 citing authors

#	Article	IF	Citations
1	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	12.8	207
2	Mutations of the Mitochondrial Holocytochrome câ€“Type Synthase in X-Linked Dominant Microphthalmia with Linear Skin Defects Syndrome. American Journal of Human Genetics, 2006, 79, 878-889.	6.2	110
3	Mutations in COX7B Cause Microphthalmia with Linear Skin Lesions, an Unconventional Mitochondrial Disease. American Journal of Human Genetics, 2012, 91, 942-949.	6.2	104
4	Fifteen years of research on oralâ€“facialâ€“digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	3.2	85
5	Ciliopathy proteins regulate paracrine signaling by modulating proteasomal degradation of mediators. Journal of Clinical Investigation, 2014, 124, 2059-2070.	8.2	79
6	Dosage compensation of the mammalian X chromosome influences the phenotypic variability of X-linked dominant male-lethal disorders. Journal of Medical Genetics, 2008, 45, 401-408.	3.2	50
7	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	2.8	34
8	Microphthalmia with linear skin defects (MLS) syndrome: Clinical, cytogenetic, and molecular characterization of 11 cases. American Journal of Medical Genetics, Part A, 2005, 137A, 190-198.	1.2	32
9	The Autophagy-Cilia Axis: An Intricate Relationship. Cells, 2019, 8, 905.	4.1	27
10	The deubiquitinating enzyme Usp14 controls ciliogenesis and Hedgehog signaling. Human Molecular Genetics, 2019, 28, 764-777.	2.9	25
11	Regulation of autophagosome biogenesis by OFD1â€mediated selective autophagy. EMBO Journal, 2021, 40, e105120.	7.8	25
12	HDAC6-dependent ciliophagy is involved in ciliary loss and cholangiocarcinoma growth in human cells and murine models. American Journal of Physiology - Renal Physiology, 2020, 318, G1022-G1033.	3.4	24
13	Molecular characterization of the human PLC Î²1 gene. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2002, 1584, 46-54.	2.4	21
14	A network-based approach to dissect the cilia/centrosome complex interactome. BMC Genomics, 2014, 15, 658.	2.8	19
15	The HOPS complex subunit VPS39 controls ciliogenesis through autophagy. Human Molecular Genetics, 2020, 29, 1018-1029.	2.9	16
16	The TBC1D31/praja2 complex controls primary ciliogenesis through PKAâ€directed OFD1 ubiquitylation. EMBO Journal, 2021, 40, e106503.	7.8	15
17	OFD Type I syndrome: lessons learned from a rare ciliopathy. Biochemical Society Transactions, 2020, 48, 1929-1939.	3.4	13
18	Terminal osseous dysplasia with pigmentary defects: Clinical description of a new family. American Journal of Medical Genetics, Part A, 2007, 143A, 51-57.	1.2	11

#	Article	IF	Citations
19	A ZFYVE19 gene mutation associated with neonatal cholestasis and cilia dysfunction: case report with a novel pathogenic variant. Orphanet Journal of Rare Diseases, 2021, 16, 179.	2.7	11
20	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	6.2	11
21	Crosstalk between cilia and autophagy: implication for human diseases. Autophagy, 2023, 19, 24-43.	9.1	10
22	Disruption of the IQSEC2 transcript in a female with X;autosome translocation t(X;20)(p11.2;q11.2) and a phenotype resembling X-linked infantile spasms (ISSX) syndrome. Molecular Medicine Reports, 2008, , .	2.4	8
23	The OFD1 protein is a novel player in selective autophagy: another tile to the cilia/autophagy puzzle. Cell Stress, 2021, 5, 33-36.	3.2	6
24	The role of OFD1 in selective autophagy. Molecular and Cellular Oncology, 2021, 8, 1903291.	0.7	4
25	Solving unsolved rare neurological diseasesâ€"a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	2.8	4
26	Identification of an Identical de Novo SCAMP5 Missense Variant in Four Unrelated Patients With Seizures and Severe Neurodevelopmental Delay. Frontiers in Pharmacology, 2020, 11, 599191.	3.5	2
27	Drug Repurposing to Target the Apoptosome in MAPKi-Resistant Melanoma. SSRN Electronic Journal, 0, , .	0.4	1
28	A network-based approach to dissect the cilia/centrosome complex interactome. Cilia, 2015, 4, .	1.8	0