Marco Henneke

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	RNASET2-deficient cystic leukoencephalopathy resembles congenital cytomegalovirus brain infection. Nature Genetics, 2009, 41, 773-775.	21.4	124
2	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114
3	<i>rnaset2</i> mutant zebrafish model familial cystic leukoencephalopathy and reveal a role for RNase T2 in degrading ribosomal RNA. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 1099-1103.	7.1	91
4	Immune Sensing of Synthetic, Bacterial, and Protozoan RNA by Toll-like Receptor 8 Requires Coordinated Processing by RNase T2 and RNase 2. Immunity, 2020, 52, 591-605.e6.	14.3	83
5	Complex Genomic Rearrangements at the PLP1 Locus Include Triplication and Quadruplication. PLoS Genetics, 2015, 11, e1005050.	3.5	57
6	Identification of twelve novel mutations in patients with classic and variant forms of maple syrup urine disease. Human Mutation, 2003, 22, 417-417.	2.5	37
7	The failure of microglia to digest developmental apoptotic cells contributes to the pathology of RNASET2â€deficient leukoencephalopathy. Glia, 2020, 68, 1531-1545.	4.9	35
8	Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-Goutià res syndrome. European Journal of Paediatric Neurology, 2016, 20, 604-610.	1.6	29
9	The Phenotypic Spectrum of PRRT2-Associated Paroxysmal Neurologic Disorders in Childhood. Biomedicines, 2020, 8, 456.	3.2	23
10	Compound heterozygous variants in PGAP1 causing severe psychomotor retardation, brain atrophy, recurrent apneas and delayed myelination: a case report and literature review. BMC Neurology, 2016, 16, 74.	1.8	19
11	Interferon-driven brain phenotype in a mouse model of RNaseT2 deficient leukoencephalopathy. Nature Communications, 2021, 12, 6530.	12.8	16
12	Mutation analysis of the M6bgene in patients with Pelizaeus-Merzbacher-like syndrome., 2004, 128A, 156-158.		7
13	Zebrafish disease model of human RNASET2 deficient cystic leukoencephalopathy displays abnormalities in early microglia. Biology Open, 2020, 9, .	1.2	4
14	A novel remitting leukodystrophy associated with a variant in FBP2. Brain Communications, 2021, 3, fcab036.	3.3	2