

# Anna Duarri

## List of Publications by Year in descending order

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21  
papers

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citations

566801

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676716

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23  
docs citations

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1324  
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#	ARTICLE	IF	CITATIONS
1	Mutant GlialCAM Causes Megalencephalic Leukoencephalopathy with Subcortical Cysts, Benign Familial Macrocephaly, and Macrocephaly with Retardation and Autism. <i>American Journal of Human Genetics</i> , 2011, 88, 422-432.	2.6	148
2	Mutations in potassium channel <i>KCNK3</i> cause spinocerebellar ataxia type 19. <i>Annals of Neurology</i> , 2012, 72, 870-880.	2.8	121
3	Exome sequencing and network analysis identifies shared mechanisms underlying spinocerebellar ataxia. <i>Brain</i> , 2017, 140, 2860-2878.	3.7	98
4	Megalencephalic leukoencephalopathy with cysts: defect in chloride currents and cell volume regulation. <i>Brain</i> , 2011, 134, 3342-3354.	3.7	63
5	Molecular pathogenesis of megalencephalic leukoencephalopathy with subcortical cysts: mutations in MLC1 cause folding defects. <i>Human Molecular Genetics</i> , 2008, 17, 3728-3739.	1.4	60
6	Knockdown of MLC1 in primary astrocytes causes cell vacuolation: A MLC disease cell model. <i>Neurobiology of Disease</i> , 2011, 43, 228-238.	2.1	60
7	First de novo KCND3 mutation causes severe Kv4.3 channel dysfunction leading to early onset cerebellar ataxia, intellectual disability, oral apraxia and epilepsy. <i>BMC Medical Genetics</i> , 2015, 16, 51.	2.1	46
8	Elevated mutant dynorphin A causes Purkinje cell loss and motor dysfunction in spinocerebellar ataxia type 23. <i>Brain</i> , 2015, 138, 2537-2552.	3.7	34
9	Functional Analysis Helps to Define KCNC3 Mutational Spectrum in Dutch Ataxia Cases. <i>PLoS ONE</i> , 2015, 10, e0116599.	1.1	26
10	Spinocerebellar ataxia type 19/22 mutations alter heterocomplex Kv4.3 channel function and gating in a dominant manner. <i>Cellular and Molecular Life Sciences</i> , 2015, 72, 3387-3399.	2.4	24
11	The N-terminal domain of the human eIF2 $\beta$ subunit and the CK2 phosphorylation sites are required for its function. <i>Biochemical Journal</i> , 2006, 394, 227-236.	1.7	23
12	Cell therapy with hiPSC-derived RPE cells and RPCs prevents visual function loss in a rat model of retinal degeneration. <i>Molecular Therapy - Methods and Clinical Development</i> , 2021, 20, 688-702.	1.8	22
13	Multiocular organoids from human induced pluripotent stem cells displayed retinal, corneal, and retinal pigment epithelium lineages. <i>Stem Cell Research and Therapy</i> , 2021, 12, 581.	2.4	20
14	Megalencephalic leukoencephalopathy with subcortical cysts: A personal biochemical retrospective. <i>European Journal of Medical Genetics</i> , 2018, 61, 50-60.	0.7	19
15	The L450P mutation in KCND3 brings spinocerebellar ataxia and Brugada syndrome closer together. <i>Neurogenetics</i> , 2013, 14, 257-258.	0.7	15
16	Impaired proteostasis in rare neurological diseases. <i>Seminars in Cell and Developmental Biology</i> , 2019, 93, 164-177.	2.3	14
17	Repopulation of decellularized retinas with hiPSC-derived retinal pigment epithelial and ocular progenitor cells shows cell engraftment, organization and differentiation. <i>Biomaterials</i> , 2021, 276, 121049.	5.7	13
18	Generation of six multiple sclerosis patient-derived induced pluripotent stem cell lines. <i>Stem Cell Research</i> , 2017, 24, 155-159.	0.3	10

#	ARTICLE	IF	CITATIONS
19	Transcriptomics analysis of Ccl2/Cx3cr1/Crb1rd8 deficient mice provides new insights into the pathophysiology of progressive retinal degeneration. <i>Experimental Eye Research</i> , 2021, 203, 108424.	1.2	10
20	Transplantation of Human Induced Pluripotent Stem Cell-Derived Retinal Pigment Epithelium in a Swine Model of Geographic Atrophy. <i>International Journal of Molecular Sciences</i> , 2021, 22, 10497.	1.8	10
21	Cross talk between protein kinase CK2 and eukaryotic translation initiation factor eIF2 $\hat{1}$ <sup>2</sup> subunit. <i>Molecular and Cellular Biochemistry</i> , 2005, 274, 53-61.	1.4	6