Chiara Pescucci

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/11276413/publications.pdf

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12	839	759233 12	1199594 12
papers	citations	h-index	g-index

13	13	13	1526
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	CDKL5 belongs to the same molecular pathway of MeCP2 and it is responsible for the early-onset seizure variant of Rett syndrome. Human Molecular Genetics, 2005, 14, 1935-1946.	2.9	279
2	Autosomal-dominant Alport syndrome: Natural history of a disease due to COL4A3 or COL4A4 gene. Kidney International, 2004, 65, 1598-1603.	5.2	124
3	Real-time quantitative PCR as a routine method for screening large rearrangements in Rett syndrome: Report of one case of MECP2 deletion and one case of MECP2 duplication. Human Mutation, 2004, 24, 172-177.	2.5	96
4	Rett syndrome: the complex nature of a monogenic disease. Journal of Molecular Medicine, 2003, 81, 346-354.	3.9	80
5	Novel Î±-Actinin 2 Variant Associated With Familial Hypertrophic Cardiomyopathy and Juvenile Atrial Arrhythmias. Circulation: Cardiovascular Genetics, 2014, 7, 741-750.	5.1	74
6	Autosomal recessive Alport syndrome: an in-depth clinical and molecular analysis of five families. Nephrology Dialysis Transplantation, 2006, 21, 665-671.	0.7	40
7	Clinical and molecular characterization of a patient with a 2q31.2-32.3 deletion identified by array-CGH. American Journal of Medical Genetics, Part A, 2007, 143A, 858-865.	1.2	37
8	Italian Rett database and biobank. Human Mutation, 2007, 28, 329-335.	2.5	27
9	Thin glomerular basement membrane disease: clinical significance of a morphological diagnosis--a collaborative study of the Italian Renal Immunopathology Group. Nephrology Dialysis Transplantation, 2005, 20, 545-551.	0.7	26
10	Clinical and molecular characterization of Italian patients affected by Cohen syndrome. Journal of Human Genetics, 2007, 52, 1011-1017.	2.3	25
11	Type-IV collagen related diseases. Journal of Nephrology, 2003, 16, 314-6.	2.0	17
12	Is Rett syndrome a loss-of-imprinting disorder?. Nature Genetics, 2005, 37, 10-11.	21.4	14