Linda Berna

List of Publications by Year
in descending order

9	414	1163117 8	1588992 8
papers	citations	h-index	g-index

9	9	9	594
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The birth prevalence of lysosomal storage disorders in the Czech Republic: comparison with data in different populations. Journal of Inherited Metabolic Disease, 2010, 33, 387-396.	3.6	157
2	Prosaposin deficiency and saposin B deficiency (activatorâ€deficient metachromatic leukodystrophy): Report on two patients detected by analysis of urinary sphingolipids and carrying novel PSAP gene mutations. American Journal of Medical Genetics, Part A, 2009, 149A, 613-621.	1.2	79
3	Determination of Urinary Sulfatides and Other Lipids by Combination of Reversed-Phase and Thin-Layer Chromatographies. Analytical Biochemistry, 1999, 269, 304-311.	2.4	32
4	Replacement of Î±-galactosidase A in Fabry disease: effect on fibroblast cultures compared with biopsied tissues of treated patients. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2008, 452, 651-665.	2.8	29
5	Mutations c.459+1G>A and p.P426L in the ARSA gene: Prevalence in metachromatic leukodystrophy patients from European countries. Molecular Genetics and Metabolism, 2005, 86, 353-359.	1.1	27
6	Mucopolysaccharidosis type I in 21 Czech and Slovak patients: Mutation analysis suggests a functional importance of Câ€terminus of the IDUA protein. American Journal of Medical Genetics, Part A, 2009, 149A, 965-974.	1.2	27
7	Missense mutations as a cause of metachromatic leukodystrophy. FEBS Journal, 2005, 272, 1179-1188.	4.7	25
8	Abnormal expression and processing of uromodulin in Fabry disease reflects tubular cell storage alteration and is reversible by enzyme replacement therapy. Journal of Inherited Metabolic Disease, 2008, 31, 508-517.	3.6	25
9	Novel mutations associated with metachromatic leukodystrophy: Phenotype and expression studies in nine Czech and Slovak patients. , 2004, 129A, 277-281.		13