

# Linda Berna

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6355010/publications.pdf>

Version: 2024-02-01

9  
papers

414  
citations

1163117  
8  
h-index

1588992  
8  
g-index

9  
all docs

9  
docs citations

9  
times ranked

594  
citing authors

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