Etienne Mornet

List of Publications by Year
in descending order

32 papers	1,968 citations	257450 24 h-index	454955 30 g-index
32 all docs	32 docs citations	32 times ranked	1180 citing authors

#	Article	IF	Citations
1	Hypophosphatasia: a genetic-based nosology and new insights in genotype-phenotype correlation. European Journal of Human Genetics, 2021, 29, 289-299.	2.8	43
2	Utility of genetic testing for prenatal presentations of hypophosphatasia. Molecular Genetics and Metabolism, 2021, 132, 198-203.	1.1	2
3	Largeâ€scale in vitro functional testing and novel variant scoring via protein modeling provide insights into alkaline phosphatase activity in hypophosphatasia. Human Mutation, 2020, 41, 1250-1262.	2.5	32
4	Diagnostic gÃ©nÃ©tique dâ€™une maladie osseuse rareÂ: lâ€™apport du NGS. Revue Du Rhumatisme Monographies, 2019, 86, 8-13.	0.0	0
5	Hypophosphatasia. Metabolism: Clinical and Experimental, 2018, 82, 142-155.	3.4	101
6	Genetic analysis of adults heterozygous for ALPL mutations. Journal of Bone and Mineral Metabolism, 2018, 36, 723-733.	2.7	29
7	Identification of altered brain metabolites associated with <scp>TNAP</scp> activity in a mouse model of hypophosphatasia using untargeted <scp>NMR</scp>â€based metabolomics analysis. Journal of Neurochemistry, 2017, 140, 919-940.	3.9	34
8	Identification of a p.Arg708Gln variant in COL1A2 in atypical femoral fractures. Joint Bone Spine, 2017, 84, 715-718.	1.6	17
9	Molecular Genetics of Hypophosphatasia and Phenotype-Genotype Correlations. Sub-Cellular Biochemistry, 2015, 76, 25-43.	2.4	40
10	Molecular diagnosis of hypophosphatasia and differential diagnosis by targeted Next Generation Sequencing. Molecular Genetics and Metabolism, 2015, 116, 215-220.	1.1	54
11	Clinical utility gene card for: Hypophosphatasia â€“ update 2013. European Journal of Human Genetics, 2014, 22, 572-572.	2.8	52
12	Unexpected high intrafamilial phenotypic variability observed in hypophosphatasia. European Journal of Human Genetics, 2014, 22, 1160-1164.	2.8	63
13	Molecular Evolution of the Tissue-nonspecific Alkaline Phosphatase Allows Prediction and Validation of Missense Mutations Responsible for Hypophosphatasia. Journal of Biological Chemistry, 2014, 289, 24168-24179.	3.4	52
14	Genetics of Hypophosphatasia. Clinical Reviews in Bone and Mineral Metabolism, 2013, 11, 71-77.	0.8	13
15	A Molecularâ€Based Estimation of the Prevalence of Hypophosphatasia in the European Population. Annals of Human Genetics, 2011, 75, 439-445.	0.8	174
16	Clinical utility gene card for: hypophosphatasia. European Journal of Human Genetics, 2011, 19, 4-5.	2.8	19
17	Differential expression of the bone and the liver tissue non-specific alkaline phosphatase isoforms in brain tissues. Cell and Tissue Research, 2011, 343, 521-536.	2.9	51
18	Hypophosphatasia may lead to bone fragility: donâ€™t miss it. European Journal of Pediatrics, 2009, 168, 783-788.	2.7	38

#	Article	IF	Citations
19	Mild forms of hypophosphatasia mostly result from dominant negative effect of severe alleles or from compound heterozygosity for severe and moderate alleles. BMC Medical Genetics, 2009, 10, 51.	2.1	118
20	Orodental phenotype and genotype findings in all subtypes of hypophosphatasia. Orphanet Journal of Rare Diseases, 2009, 4, 6.	2.7	98
21	Hypophosphatasia: phenotypic variability and possible Croatian origin of the c.1402g>A mutation of TNSALP gene. Collegium Antropologicum, 2009, 33, 1255-8.	0.2	5
22	Hypophosphatasia. Best Practice and Research in Clinical Rheumatology, 2008, 22, 113-127.	3.3	118
23	Delayed transport of tissue-nonspecific alkaline phosphatase with missense mutations causing hypophosphatasia. European Journal of Medical Genetics, 2007, 50, 367-378.	1.3	34
24	Hypophosphatasia. Orphanet Journal of Rare Diseases, 2007, 2, 40.	2.7	231
25	Characterization of Missense Mutations and Large Deletions in the ALPL Gene by Sequencing and Quantitative Multiplex PCR of Short Fragments. Genetic Testing and Molecular Biomarkers, 2006, 10, 252-257.	1.7	24
26	Characterization of 11 novel mutations in the tissue non-specific alkaline phosphatase gene responsible for hypophosphatasia and genotypeâ€“phenotype correlations. Molecular Genetics and Metabolism, 2005, 84, 273-277.	1.1	37
27	Evidence of a founder effect for the tissue-nonspecific alkaline phosphatase (TNSALP) gene E174K mutation in hypophosphatasia patients. European Journal of Human Genetics, 2002, 10, 666-668.	2.8	28
28	Severe cleidocranial dysplasia can mimic hypophosphatasia. European Journal of Pediatrics, 2002, 161, 623-626.	2.7	46
29	Structural Evidence for a Functional Role of Human Tissue Nonspecific Alkaline Phosphatase in Bone Mineralization. Journal of Biological Chemistry, 2001, 276, 31171-31178.	3.4	182
30	Hypophosphatasia: The mutations in the tissue-nonspecific alkaline phosphatase gene. Human Mutation, 2000, 15, 309-315.	2.5	129
31	Characterization of a family with dominant hypophosphatasia. European Journal of Oral Sciences, 2000, 108, 189-194.	1.5	74
32	Correlation of alkaline phosphatase (ALP) determination and analysis of the tissue non-specific ALP gene in prenatal diagnosis of severe hypophosphatasia. , 1999, 19, 755-757.		30