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. Orphanet Journal of Rare Diseases, 2007, 2, 40.	2.7	231
2	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
3	A Molecularâ€Based Estimation of the Prevalence of Hypophosphatasia in the European Population. Annals of Human Genetics, 2011, 75, 439-445.	0.8	174
4	Hypophosphatasia: The mutations in the tissue-nonspecific alkaline phosphatase gene. Human Mutation, 2000, 15, 309-315.	2.5	129
5	Hypophosphatasia. Best Practice and Research in Clinical Rheumatology, 2008, 22, 113-127.	3.3	118
6	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
7	Hypophosphatasia. Metabolism: Clinical and Experimental, 2018, 82, 142-155.	3.4	101
8	Orodental phenotype and genotype findings in all subtypes of hypophosphatasia. Orphanet Journal of Rare Diseases, 2009, 4, 6.	2.7	98
9	Characterization of a family with dominant hypophosphatasia. European Journal of Oral Sciences, 2000, 108, 189-194.	1.5	74
10	Unexpected high intrafamilial phenotypic variability observed in hypophosphatasia. European Journal of Human Genetics, 2014, 22, 1160-1164.	2.8	63
11	Molecular diagnosis of hypophosphatasia and differential diagnosis by targeted Next Generation Sequencing. Molecular Genetics and Metabolism, 2015, 116, 215-220.	1.1	54
12	Clinical utility gene card for: Hypophosphatasia â€“ update 2013. European Journal of Human Genetics, 2014, 22, 572-572.	2.8	52
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	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
15	Severe cleidocranial dysplasia can mimic hypophosphatasia. European Journal of Pediatrics, 2002, 161, 623-626.	2.7	46
16	Hypophosphatasia: a genetic-based nosology and new insights in genotype-phenotype correlation. European Journal of Human Genetics, 2021, 29, 289-299.	2.8	43
17	Molecular Genetics of Hypophosphatasia and Phenotype-Genotype Correlations. Sub-Cellular Biochemistry, 2015, 76, 25-43.	2.4	40
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	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
20	Delayed transport of tissue-nonspecific alkaline phosphatase with missense mutations causing hypophosphatasia. European Journal of Medical Genetics, 2007, 50, 367-378.	1.3	34
21	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
22	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
23	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
24	Genetic analysis of adults heterozygous for ALPL mutations. Journal of Bone and Mineral Metabolism, 2018, 36, 723-733.	2.7	29
25	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
26	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
27	Clinical utility gene card for: hypophosphatasia. European Journal of Human Genetics, 2011, 19, 4-5.	2.8	19
28	Identification of a p.Arg708Gln variant in COL1A2 in atypical femoral fractures. Joint Bone Spine, 2017, 84, 715-718.	1.6	17
29	Genetics of Hypophosphatasia. Clinical Reviews in Bone and Mineral Metabolism, 2013, 11, 71-77.	0.8	13
30	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
31	Utility of genetic testing for prenatal presentations of hypophosphatasia. Molecular Genetics and Metabolism, 2021, 132, 198-203.	1.1	2
32	Diagnostic gÃ©nÃ©tique dâ€™une maladie osseuse rareÂ: lâ€™apport du NGS. Revue Du Rhumatisme Monographies, 2019, 86, 8-13.	0.0	0