# Ana Maria Fortuna

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3016053/publications.pdf

Version: 2024-02-01

| 9 | 371 | 1163117 8 | 1474206 9 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 10 | 10 | 10 | 725 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659. | 2.8 | 108 |
| 2 | Genotype-phenotype correlations in L1 syndrome: a guide for genetic counselling and mutation analysis. Journal of Medical Genetics, 2010, 47, 169-175. | 3.2 | 82 |
| 3 | A Novel Domain-Specific Mutation in a Sclerosteosis Patient Suggests a Role of LRP4 as an Anchor for Sclerostin in Human Bone. Journal of Bone and Mineral Research, 2016, 31, 874-881. | 2.8 | 65 |
| 4 | T Cell Numbers Relate to Bone Involvement in Gaucher Disease. Blood Cells, Molecules, and Diseases, 1999, 25, 130-138. | 1.4 | 47 |
| 5 | New splicing mutation in the choline kinase beta (CHKB) gene causing a muscular dystrophy detected by whole-exome sequencing. Journal of Human Genetics, 2015, 60, 305-312. | 2.3 | 33 |
| 6 | Private dysferlin exon skipping mutation (c.5492G>A) with a founder effect reveals further alternative splicing involving exons 49â€“51. Journal of Human Genetics, 2010, 55, 546-549. | 2.3 | 12 |
| 7 | A NONSENSEPORCNMUTATION IN SEVERE FOCAL DERMAL HYPOPLASIA WITH NATAL TEETH. Fetal and Pediatric Pathology, 2010, 29, 305-313. | 0.7 | 11 |
| 8 | Galactosialidosis presenting as nonimmune fetal hydrops: a case report. Prenatal Diagnosis, 2009, 29, 895-896. | 2.3 | 10 |
| 9 | Usher syndrome and Nebulinâ€associated myopathy in a single patient due to variants in MYO7A and NEB. Clinical Case Reports (discontinued), 2020, 8, 2476-2482. | 0.5 | 0 |