

Thuong Ha

List of Publications by Year in descending order

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6
papers

73
citations

1937685
4
h-index

1872680
6
g-index

8
all docs

8
docs citations

8
times ranked

110
citing authors

| # | ARTICLE | IF | CITATIONS |
|---|---|-----|-----------|
| 1 | Compound heterozygous variants in LAMC3 in association with posterior periventricular nodular heterotopia. BMC Medical Genomics, 2021, 14, 64. | 1.5 | 5 |
| 2 | The RUNX1 database (RUNX1db): establishment of an expert curated RUNX1 registry and genomics database as a public resource for familial platelet disorder with myeloid malignancy. Haematologica, 2021, 106, 3004-3007. | 3.5 | 29 |
| 3 | OUP accepted manuscript. Human Molecular Genetics, 2021, 30, 2068-2081. | 2.9 | 7 |
| 4 | Paternal mosaicism for a novel <i>PBX1</i> mutation associated with recurrent perinatal death: Phenotypic expansion of the <i>PBX1</i> -related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1273-1277. | 1.2 | 12 |
| 5 | Familial epilepsy with anterior polymicrogyria as a presentation of COL18A1 mutations. European Journal of Medical Genetics, 2017, 60, 437-443. | 1.3 | 10 |
| 6 | A non-coding variant in the 5' UTR of DLG3 attenuates protein translation to cause non-syndromic intellectual disability. European Journal of Human Genetics, 2016, 24, 1612-1616. | 2.8 | 10 |