# Sarah F Smithson

List of Publications by Year
in descending order



| 14 | 134 | 1307594 7 | 1281871 11 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 15 | 15 | 15 | 239 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Non-collagen pathogenic variants resulting in the osteogenesis imperfecta phenotype in children: a single-country observational cohort study. Archives of Disease in Childhood, 2022, 107, 486-490. | 1.9 | 2 |
| 2 | Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758. | 6.2 | 13 |
| 3 | The third case of TNFRSF11A-associated dysosteosclerosis with a mutation producing elongating proteins. Journal of Human Genetics, 2021, 66, 371-377. | 2.3 | 8 |
| 4 | <scp>AIFM1</scp>â€associated Xâ€linked spondylometaphyseal dysplasia with cerebral hypomyelination. American Journal of Medical Genetics, Part A, 2021, 185, 1228-1235. | 1.2 | 5 |
| 5 | Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150. | 6.2 | 17 |
| 6 | Challenges in long-term control of hypercalcaemia with denosumab after haematopoietic stem cell transplantation for TNFRSF11A osteoclast-poor autosomal recessive osteopetrosis. Bone Reports, 2021, 14, 100738. | 0.4 | 4 |
| 7 | Expanding the phenotypic spectrum of IFT81 : Associated ciliopathy syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2403-2408. | 1.2 | 3 |
| 8 | Post-mortem histology in transient receptor potential cation channel subfamily V member 6 (TRPV6) under-mineralising skeletal dysplasia suggests postnatal skeletal recovery: a case report. BMC Medical Genetics, 2020, 21, 64. | 2.1 | 9 |
| 9 | PAPSS2â€related brachyolmia: Clinical and radiological phenotype in 18 new cases. American Journal of Medical Genetics, Part A, 2019, 179, 1884-1894. | 1.2 | 9 |
| 10 | Deep phenotyping of 14 new patients with IQSEC2 variants, including monozygotic twins of discordant phenotype. Clinical Genetics, 2019, 95, 496-506. | 2.0 | 20 |
| 11 | Cover Image, Volume 176A, Number 9, September 2018. American Journal of Medical Genetics, Part A, 2018, 176, . | 1.2 | 0 |
| 12 | Disruption ofTWIST1translation by 5â€² UTR variants in Saethre-Chotzen syndrome. Human Mutation, 2018, 39, 1360-1365. | 2.5 | 10 |
| 13 | TRPV6 compound heterozygous variants result in impaired placental calcium transport and severe undermineralization and dysplasia of the fetal skeleton. American Journal of Medical Genetics, Part A, 2018, 176, 1950-1955. | 1.2 | 31 |
| 14 | Prenatal diagnosis and postnatal outcome of massive abdominal aortic aneurysmsâ€”a case report. Prenatal Diagnosis, 2015, 35, 923-925. | 2.3 | 0 |