Anna SowiÅ„ska-Seidler

List of Publications by Year
in descending order

Version: 2024-02-01

26	379	840776 11	839539 18
papers	citations	h-index	g-index

28	28	28	660
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Split-hand/foot malformation - molecular cause and implications in genetic counseling. Journal of Applied Genetics, 2014, 55, 105-115.	1.9	66
2	Deletions of exons with regulatory activity at the DYNC1I1 locus are associated with split-hand/split-foot malformation: array CGH screening of 134 unrelated families. Orphanet Journal of Rare Diseases, 2014, 9, 108.	2.7	43
3	Noncoding copy-number variations are associated with congenital limb malformation. Genetics in Medicine, 2018, 20, 599-607.	2.4	42
4	Mutational screening of EXT1 and EXT2 genes in Polish patients with hereditary multiple exostoses. Journal of Applied Genetics, 2014, 55, 183-188.	1.9	28
5	Three novel GJA1 missense substitutions resulting in oculo-dento-digital dysplasia (ODDD) â€" Further extension of the mutational spectrum. Gene, 2014, 539, 157-161.	2.2	19
6	Exome sequencing reveals two novel compound heterozygous XYLT1 mutations in a Polish patient with Desbuquois dysplasia type 2 and growth hormone deficiency. Journal of Human Genetics, 2016, 61, 577-583.	2.3	18
7	Functional analysis of novel RUNX2 mutations identified in patients with cleidocranial dysplasia. Clinical Genetics, 2019, 96, 429-438.	2.0	17
8	Heterozygous DLX5 nonsense mutation associated with isolated splitâ€hand/foot malformation with reduced penetrance and variable expressivity in two unrelated families. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 764-771.	1.6	16
9	Variable expressivity of the phenotype in two families with brachydactyly type E, craniofacial dysmorphism, short stature and delayed bone age caused by novel heterozygous mutations in the PTHLH gene. Journal of Human Genetics, 2016, 61, 457-461.	2.3	16
10	Adapting SureSelect enrichment protocol to the Ion Torrent S5 platform in molecular diagnostics of craniosynostosis. Scientific Reports, 2020, 10, 4159.	3.3	14
11	Duplication of 10q24 locus: broadening the clinical and radiological spectrum. European Journal of Human Genetics, 2019, 27, 525-534.	2.8	13
12	Hyperosmia, ectrodactyly, mild intellectual disability, and other defects in a male patient with an X-linked partial microduplication and overexpression of the KAL1 gene. Journal of Applied Genetics, 2015, 56, 177-184.	1.9	12
13	Novel synonymous and missense variants in FGFR1 causing Hartsfield syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2447-2453.	1.2	10
14	Novel 1q22-q23.1 duplication in a patient with lambdoid and metopic craniosynostosis, muscular hypotonia, and psychomotor retardation. Journal of Applied Genetics, 2018, 59, 281-289.	1.9	9
15	X-Linked Adrenal Hypoplasia Congenita in a Boy due to a Novel Deletion of the EntireNR0B1 (DAX1)andMAGEB1â€“4Genes. International Journal of Endocrinology, 2016, 2016, 1-7.	1.5	8
16	Duplication of PTHLH causes osteochondroplasia with a combined brachydactyly type E/A1 phenotype with disturbed bone maturation and rhizomelia. European Journal of Human Genetics, 2016, 24, 1132-1136.	2.8	8
17	The First Report of Biallelic Missense Mutations in the SFRP4 Gene Causing Pyle Disease in Two Siblings. Frontiers in Genetics, 2020, 11, 593407.	2.3	8
18	Further phenotypic delineation of the auriculocondylar syndrome type 2 with literature review. Journal of Applied Genetics, 2021, 62, 107-113.	1.9	6

#	Article	IF	Citations
19	Prenatal diagnosis of Fraser syndrome using routine ultrasound examination, confirmed by exome sequencing: Report of a novel homozygous missense FRAS1 mutation. Congenital Anomalies (discontinued), 2017, 57, 37-38.	0.6	5
20	A novel biallelic spliceâ€site variant in the LRP4 gene causes sclerosteosis 2. Birth Defects Research, 2020, 112, 652-659.	1.5	5
21	Position effects at the FGF8 locus are associated with femoral hypoplasia. American Journal of Human Genetics, 2021, 108, 1725-1734.	6.2	4
22	Results from Genetic Studies in Patients Affected with Craniosynostosis: Clinical and Molecular Aspects. Frontiers in Molecular Biosciences, 2022, 9, 865494.	3.5	4
23	Identification of a molecular defect in a stillborn fetus with perinatal lethal hypophosphatasia using a disease-associated genome sequencing approach. Polish Journal of Pathology, 2016, 1, 78-83.	0.3	3
24	Clinical expression of Holt-Oram syndrome on the basis of own clinical experience considering prenatal diagnosis. Ginekologia Polska, 2016, 87, 706-710.	0.7	2
25	Homozygous microdeletion in the 11p13 region in the patient with isolated form of aniridia: New challenges in the genetic diagnostics of aniridia. American Journal of Medical Genetics, Part A, 2021, , .	1.2	2
26	The pZRS non-coding regulatory mutation resulting in triphalangeal thumbâ€“polysyndactyly syndrome changes the pattern of local interactions. Molecular Genetics and Genomics, 2022, 297, 1343-1352.	2.1	1