Arti Pandya

List of Publications by Year
in descending order

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25 papers	1,372 citations	623734 14 h-index	713466 21 g-index
25 all docs	25 docs citations	25 times ranked	1540 citing authors

#	Article	IF	Citations
1	Clinical evaluation and etiologic diagnosis of hearing loss: A clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2022, 24, 1392-1406.	2.4	18
2	Confirmation of COL4A6 variants in X-linked nonsyndromic hearing loss and its clinical implications. European Journal of Human Genetics, 2021, , .	2.8	5
3	Analyses of del(GJB6â€D13S1830) and del(GJB6â€D13S1834) deletions in a large cohort with hearing loss: Caveats to interpretation of molecular test results in multiplex families. Molecular Genetics & Genomic Medicine, 2020, 8, e1171.	1.2	10
4	Oculodentodigital Dysplasia: A Case Report and Major Review of the Eye and Ocular Adnexa Features of 295 Reported Cases. Case Reports in Ophthalmological Medicine, 2020, 2020, 1-16.	0.5	4
5	Cancer Risk in Klippelâ€“Trenaunay Syndrome. Lymphatic Research and Biology, 2019, 17, 630-636.	1.1	20
6	Analysis of risk factors associated with unilateral hearing loss in children who initially passed newborn hearing screening. International Journal of Pediatric Otorhinolaryngology, 2018, 106, 100-104.	1.0	15
7	Natural history and genotypeâ€phenotype correlations in 72 individuals with SATB2â€associated syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 925-935.	1.2	57
8	Cover Image, Volume 176A, Number 4, April 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
9	Co-occurring Down syndrome and SUCLA2 -related mitochondrial depletion syndrome. , 2017, 173, 2720-2724.		2
10	Genetic hearing loss: the journey of discovery to destination â€“ how close are we to therapy?. Molecular Genetics & Genomic Medicine, 2016, 4, 583-587.	1.2	15
11	American College of Medical Genetics and Genomics guideline for the clinical evaluation and etiologic diagnosis of hearing loss. Genetics in Medicine, 2014, 16, 347-355.	2.4	207
12	A Comparative Analysis of the Genetic Epidemiology of Deafness in the United States in Two Sets of Pedigrees Collected More than a Century Apart. American Journal of Human Genetics, 2008, 83, 200-207.	6.2	34
13	Does Universal Newborn Hearing Screening Identify All Children with GJB2 (Connexin 26) Deafness? Penetrance of GJB2 Deafness. Ear and Hearing, 2006, 27, 732-741.	2.1	69
14	Genetic Evaluation and Counseling in the Context of Early Hearing Detection and Intervention. Seminars in Hearing, 2006, 27, 205-212.	1.2	5
15	Prevalence and Evolutionary Origins of the del(GJB6-D13S1830) Mutation in the DFNB1 Locus in Hearing-Impaired Subjects: a Multicenter Study. American Journal of Human Genetics, 2003, 73, 1452-1458.	6.2	269
16	Frequency and distribution of GJB2 (connexin 26) and GJB6 (connexin 30) mutations in a large North American repository of deaf probands. Genetics in Medicine, 2003, 5, 295-303.	2.4	138
17	Genetic Epidemiology of Deafness. , 2002, , 67-91.		6
18	Advances in hereditary deafness. Lancet, The, 2001, 358, 1082-1090.	13.7	136

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19	Coffin-Siris syndrome: Review and presentation of new cases from a questionnaire study. American Journal of Medical Genetics Part A, 2001, 99, 1-7.	2.4	71
20	Fine mapping of a gene responsible for regulating dietary cholesterol absorption; founder effects underlie cases of phytosterolaemia in multiple communities. European Journal of Human Genetics, 2001, 9, 375-384.	2.8	38
21	Age-related language characteristics of children and adolescents with fragile X syndrome. American Journal of Medical Genetics Part A, 1999, 83, 253-256.	2.4	29
22	Longitudinal assessment of adaptive and maladaptive behaviors in fragile X males: Growth, development, and profiles. , 1999, 83, 257-263.		38
23	Heterogenous Point Mutations in the Mitochondrial tRNA Ser(UCN) Precursor Coexisting with the A1555G Mutation in Deaf Students from Mongolia. American Journal of Human Genetics, 1999, 65, 1803-1806.	6.2	78
24	Correlation between Waardenburg syndrome phenotype and genotype in a population of individuals with identified PAX3 mutations. Human Genetics, 1998, 102, 499-506.	3.8	69
25	Phenotypic variation in Waardenburg syndrome: mutational heterogeneity, modifier genes or polygenic background?. Human Molecular Genetics, 1996, 5, 497-502.	2.9	39