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List of Publications by Year in descending order

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Version: 2024-02-01

516710 752698 1,376 21 16 20 h-index citations g-index papers 21 21 21 2967 all docs docs citations times ranked citing authors

#	Article	IF	CITATIONS
1	Guidelines for diagnostic next-generation sequencing. European Journal of Human Genetics, 2016, 24, 2-5.	2.8	389
2	TRIDENT-2: National Implementation of Genome-wide Non-invasive Prenatal Testing as a First-Tier Screening Test in the Netherlands. American Journal of Human Genetics, 2019, 105, 1091-1101.	6.2	222
3	The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands. European Journal of Human Genetics, 2017, 25, 308-314.	2.8	90
4	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586.	3.8	85
5	Origin and clinical relevance of chromosomal aberrations other than the common trisomies detected by genome-wide NIPS: results of the TRIDENT study. Genetics in Medicine, 2018, 20, 480-485.	2.4	85
6	1 in 38 individuals at risk of a dominant medically actionable disease. European Journal of Human Genetics, 2019, 27, 325-330.	2.8	56
7	The consequences of implementing non-invasive prenatal testing in Dutch national health care: a cost-effectiveness analysis. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2014, 182, 53-61.	1.1	53
8	The effect of a decision aid on informed decision-making in the era of non-invasive prenatal testing: a randomised controlled trial. European Journal of Human Genetics, 2016, 24, 1409-1416.	2.8	53
9	Rapid whole exome sequencing in pregnancies to identify the underlying genetic cause in fetuses with congenital anomalies detected by ultrasound imaging. Prenatal Diagnosis, 2020, 40, 972-983.	2.3	49
10	Balanced into array: genome-wide array analysis in 54 patients with an apparently balanced de novo chromosome rearrangement and a meta-analysis. European Journal of Human Genetics, 2011, 19, 1152-1160.	2.8	41
11	Disruption of Teashirt Zinc Finger Homeobox 1 Is Associated with Congenital Aural Atresia in Humans. American Journal of Human Genetics, 2011, 89, 813-819.	6.2	38
12	Women's and healthcare professionals' preferences for prenatal testing: a discrete choice experiment. Prenatal Diagnosis, 2015, 35, 549-557.	2.3	38
13	MPZL2, Encoding the Epithelial Junctional Protein Myelin Protein Zero-like 2, Is Essential for Hearing in Man and Mouse. American Journal of Human Genetics, 2018, 103, 74-88.	6.2	34
14	Heterozygous missense variants of LMX1A lead to nonsyndromic hearing impairment and vestibular dysfunction. Human Genetics, 2018, 137, 389-400.	3.8	32
15	De novo and inherited loss-of-function variants of ATP2B2 are associated with rapidly progressive hearing impairment. Human Genetics, 2019, 138, 61-72.	3.8	27
16	Noninvasive prenatal diagnosis of Huntington disease: detection of the paternally inherited expanded CAG repeat in maternal plasma. Prenatal Diagnosis, 2015, 35, 945-949.	2.3	23
17	Broadening the phenotype of DFNB28: Mutations in TRIOBP are associated with moderate, stable hereditary hearing impairment. Hearing Research, 2017, 347, 56-62.	2.0	17
18	ALG13-CDG with Infantile Spasms in a Male Patient Due to a De Novo ALG13 Gene Mutation. JIMD Reports, 2017, 40, 11-16.	1.5	15

#	Article	IF	CITATION
19	Lessons learned from unsolicited findings in clinical exome sequencing of 16,482 individuals. European Journal of Human Genetics, 2022, 30, 170-177.	2.8	15
20	The identification of a RNA splice variant in TULP1 in two siblings with earlyâ€onset photoreceptor dystrophy. Molecular Genetics & Enomic Medicine, 2019, 7, e660.	1.2	14
21	Trends in genetic diagnostics of hereditary hearing loss. Journal of Laryngology and Otology, 2016, 130, S27-S27.	0.8	0