Imen Chakchouk

List of Publications by Year
in descending order

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20 papers	484 citations	623734 14 h-index	839539 18 g-index
20 all docs	20 docs citations	20 times ranked	1094 citing authors

#	Article	IF	Citations
1	Reproductive Outcomes from Maternal Loss of Nlrp2 Are Not Improved by IVF or Embryo Transfer Consistent with Oocyte-Specific Defect. Reproductive Sciences, 2021, 28, 1850-1865.	2.5	3
2	Overview and recent developments in cellâ€based noninvasive prenatal testing. Prenatal Diagnosis, 2021, 41, 1202-1214.	2.3	22
3	Molecular insights into MYO3A kinase domain variants explain variability in both severity and progression of DFNB30 hearing impairment. Journal of Biomolecular Structure and Dynamics, 2021, , 1-12.	3.5	1
4	DNA Methylation Dynamics in the Female Germline and Maternal-Effect Mutations That Disrupt Genomic Imprinting. Genes, 2021, 12, 1214.	2.4	24
5	Disparities in discovery of pathogenic variants for autosomal recessive non-syndromic hearing impairment by ancestry. European Journal of Human Genetics, 2019, 27, 1456-1465.	2.8	19
6	Hearing impairment locus heterogeneity and identification of PLS1 as a new autosomal dominant gene in Hungarian Roma. European Journal of Human Genetics, 2019, 27, 869-878.	2.8	10
7	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. Human Genetics, 2019, 138, 593-600.	3.8	16
8	FAM92A Underlies Nonsyndromic Postaxial Polydactyly in Humans and an Abnormal Limb and Digit Skeletal Phenotype in Mice. Journal of Bone and Mineral Research, 2019, 34, 375-386.	2.8	27
9	Identification of CACNA1D variants associated with sinoatrial node dysfunction and deafness in additional Pakistani families reveals a clinical significance. Journal of Human Genetics, 2019, 64, 153-160.	2.3	32
10	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. Human Mutation, 2019, 40, 53-72.	2.5	48
11	Confirmation of the Role ofDHX38in the Etiology of Early-Onset Retinitis Pigmentosa. , 2018, 59, 4552.		16
12	Novel missense and 3â€²-UTR splice site variants in LHFPL5 cause autosomal recessive nonsyndromic hearing impairment. Journal of Human Genetics, 2018, 63, 1099-1107.	2.3	3
13	Novel candidate genes and variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability. Human Genetics, 2018, 137, 735-752.	3.8	42
14	A variant in LMX1A causes autosomal recessive severe-to-profound hearing impairment. Human Genetics, 2018, 137, 471-478.	3.8	18
15	Novel digenic inheritance of PCDH15 and USH1G underlies profound non-syndromic hearing impairment. BMC Medical Genetics, 2018, 19, 122.	2.1	18
16	MYO3A Causes Human Dominant Deafness and Interacts with Protocadherin 15-CD2 Isoform. Human Mutation, 2016, 37, 481-487.	2.5	27
17	A mutation in SLC22A4 encoding an organic cation transporter expressed in the cochlea strial endothelium causes human recessive non-syndromic hearing loss DFNB60. Human Genetics, 2016, 135, 513-524.	3.8	26
18	Novel mutations confirm that COL11A2 is responsible for autosomal recessive non-syndromic hearing loss DFNB53. Molecular Genetics and Genomics, 2015, 290, 1327-1334.	2.1	25

#	Article	IF	Citations
19	A missense mutation in DCDC2 causes human recessive deafness DFNB66, likely by interfering with sensory hair cell and supporting cell cilia length regulation. Human Molecular Genetics, 2015, 24, 2482-2491.	2.9	87
20	NADf Chip, a Two-Color Microarray for Simultaneous Screening of Multigene Mutations Associated with Hearing Impairment in North African Mediterranean Countries. Journal of Molecular Diagnostics, 2015, 17, 155-161.	2.8	20