

# Imen Chakchouk

## List of Publications by Year in descending order

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Version: 2024-02-01

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	A missense mutation in DCDC2 causes human recessive deafness DFNB66, likely by interfering with sensory hair cell and supporting cell cilia length regulation. <i>Human Molecular Genetics</i> , 2015, 24, 2482-2491.	2.9	87
2	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. <i>Human Mutation</i> , 2019, 40, 53-72.	2.5	48
3	Novel candidate genes and variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability. <i>Human Genetics</i> , 2018, 137, 735-752.	3.8	42
4	Identification of CACNA1D variants associated with sinoatrial node dysfunction and deafness in additional Pakistani families reveals a clinical significance. <i>Journal of Human Genetics</i> , 2019, 64, 153-160.	2.3	32
5	MYO3A Causes Human Dominant Deafness and Interacts with Protocadherin 15-CD2 Isoform. <i>Human Mutation</i> , 2016, 37, 481-487.	2.5	27
6	FAM92A Underlies Nonsyndromic Postaxial Polydactyly in Humans and an Abnormal Limb and Digit Skeletal Phenotype in Mice. <i>Journal of Bone and Mineral Research</i> , 2019, 34, 375-386.	2.8	27
7	A mutation in SLC22A4 encoding an organic cation transporter expressed in the cochlea stria endothelium causes human recessive non-syndromic hearing loss DFNB60. <i>Human Genetics</i> , 2016, 135, 513-524.	3.8	26
8	Novel mutations confirm that COL11A2 is responsible for autosomal recessive non-syndromic hearing loss DFNB53. <i>Molecular Genetics and Genomics</i> , 2015, 290, 1327-1334.	2.1	25
9	DNA Methylation Dynamics in the Female Germline and Maternal-Effect Mutations That Disrupt Genomic Imprinting. <i>Genes</i> , 2021, 12, 1214.	2.4	24
10	Overview and recent developments in cell-based noninvasive prenatal testing. <i>Prenatal Diagnosis</i> , 2021, 41, 1202-1214.	2.3	22
11	NADf Chip, a Two-Color Microarray for Simultaneous Screening of Multigene Mutations Associated with Hearing Impairment in North African Mediterranean Countries. <i>Journal of Molecular Diagnostics</i> , 2015, 17, 155-161.	2.8	20
12	Disparities in discovery of pathogenic variants for autosomal recessive non-syndromic hearing impairment by ancestry. <i>European Journal of Human Genetics</i> , 2019, 27, 1456-1465.	2.8	19
13	A variant in LMX1A causes autosomal recessive severe-to-profound hearing impairment. <i>Human Genetics</i> , 2018, 137, 471-478.	3.8	18
14	Novel digenic inheritance of PCDH15 and USH1G underlies profound non-syndromic hearing impairment. <i>BMC Medical Genetics</i> , 2018, 19, 122.	2.1	18
15	Confirmation of the Role of <i>DHX38</i> in the Etiology of Early-Onset Retinitis Pigmentosa. , 2018, 59, 4552.		16
16	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. <i>Human Genetics</i> , 2019, 138, 593-600.	3.8	16
17	Hearing impairment locus heterogeneity and identification of PLS1 as a new autosomal dominant gene in Hungarian Roma. <i>European Journal of Human Genetics</i> , 2019, 27, 869-878.	2.8	10
18	Novel missense and 5'-UTR splice site variants in LHFPL5 cause autosomal recessive nonsyndromic hearing impairment. <i>Journal of Human Genetics</i> , 2018, 63, 1099-1107.	2.3	3

#	ARTICLE	IF	CITATIONS
19	Reproductive Outcomes from Maternal Loss of Nlrp2 Are Not Improved by IVF or Embryo Transfer Consistent with Oocyte-Specific Defect. <i>Reproductive Sciences</i> , 2021, 28, 1850-1865.	2.5	3
20	Molecular insights into MYO3A kinase domain variants explain variability in both severity and progression of DFNB30 hearing impairment. <i>Journal of Biomolecular Structure and Dynamics</i> , 2021, , 1-12.	3.5	1