Masmoudi Saber

List of Publications by Year
in descending order

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63 papers	1,941 citations	304743 22 h-index	265206 42 g-index
63 all docs	63 docs citations	63 times ranked	2601 citing authors

#	Article	IF	Citations
1	Genetics and meta-analysis of recessive non-syndromic hearing impairment and Usher syndrome in Maghreb population: lessons from the past, contemporary actualities and future challenges. Human Genetics, 2022, 141, 583-593.	3.8	2
2	Custom Nextâ€Generation Sequencing Identifies Novel Mutations Expanding the Molecular and clinical spectrum of isolated Hearing Impairment or along with defects of the retina, the thyroid, and the kidneys. Molecular Genetics & Genomic Medicine, 2022, 10, e1868.	1.2	2
3	Evidence of SARS-CoV-2 symptomatic reinfection in four healthcare professionals from the same hospital despite the presence of antibodies. International Journal of Infectious Diseases, 2022, 117, 146-154.	3.3	10
4	Genetic diagnosis in Sudanese and Tunisian families with syndromic intellectual disability through exome sequencing. Annals of Human Genetics, 2022, 86, 181-194.	0.8	7
5	Genetic Association of rs1021188 and DNA Methylation Signatures of TNFSF11 in the Risk of Conductive Hearing Loss. Frontiers in Medicine, 2022, 9, 870244.	2.6	0
6	Cupressus sempervirens Essential Oil: Exploring the Antibacterial Multitarget Mechanisms, Chemcomputational Toxicity Prediction, and Safety Assessment in Zebrafish Embryos. Molecules, 2022, 27, 2630.	3.8	10
7	The risks of RELN polymorphisms and its expression in the development of otosclerosis. PLoS ONE, 2022, 17, e0269558.	2.5	3
8	A potential oral microbiome signature associated with coronary artery disease in Tunisia. Bioscience Reports, 2022, 42, .	2.4	6
9	<scp>SRD5A3â€CDG</scp>: <scp>3D</scp> structure modeling, clinical spectrum, and <scp>computerâ€based</scp> dysmorphic facial recognition. American Journal of Medical Genetics, Part A, 2021, 185, 1081-1090.	1.2	12
10	SARS-CoV-2 tracking in Tunisia through next-generation sequencing: lessons for the future. Euro-Mediterranean Journal for Environmental Integration, 2021, 6, 40.	1.3	2
11	Novel pathogenic mutations and further evidence for clinical relevance of genes and variants causing hearing impairment in Tunisian population. Journal of Advanced Research, 2021, 31, 13-24.	9.5	20
12	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
13	Expanding the Clinical and Molecular Spectrum of HARS2-Perrault Syndrome: Identification of a Novel Homozygous Missense Variant in the HARS2 gene. Genetic Testing and Molecular Biomarkers, 2021, 25, 528-539.	0.7	3
14	Deep analysis of the LRTOMTc.242G>A variant in nonâ€syndromic hearing loss North African patients and the Berber population: Implications for genetic diagnosis and genealogical studies. Molecular Genetics & Genomic Medicine, 2021, 9, e1810.	1.2	3
15	8q21.11 microdeletion syndrome: Delineation of HEY1 as a candidate gene in neurodevelopmental and cardiac defects. Molecular Genetics & Genomic Medicine, 2021, 9, e1811.	1.2	4
16	A year of genomic surveillance reveals how the SARS-CoV-2 pandemic unfolded in Africa. Science, 2021, 374, 423-431.	12.6	144
17	Further insights into the spectrum phenotype of TRAPPC9 and CDK5RAP2 genes, segregating independently in a large Tunisian family with intellectual disability and microcephaly. European Journal of Medical Genetics, 2021, 64, 104373.	1.3	4
18	Evaluation of the Genetic Association and mRNA Expression of the COL1A1, BMP2, and BMP4 Genes in the Development of Otosclerosis. Genetic Testing and Molecular Biomarkers, 2020, 24, 343-351.	0.7	5

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19	Osteoprotegerin gene polymorphisms and otosclerosis: an additional genetic association study, multilocus interaction and meta-analysis. BMC Medical Genetics, 2020, 21, 122.	2.1	8
20	Analysis of p.Gly12Valfs*2, p.Trp24* and p.Trp77Arg mutations in GJB2 and p.Arg81Gln variant in LRTOMT among non syndromic hearing loss Egyptian patients: implications for genetic diagnosis. Molecular Biology Reports, 2019, 46, 2139-2145.	2.3	15
21	Down-expression of P2RX2, KCNQ5, ERBB3 and SOCS3 through DNA hypermethylation in elderly women with presbycusis. Biomarkers, 2018, 23, 347-356.	1.9	31
22	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. Human Molecular Genetics, 2018, 27, 780-798.	2.9	49
23	CDH23 Methylation Status and Presbycusis Risk in Elderly Women. Frontiers in Aging Neuroscience, 2018, 10, 241.	3.4	21
24	FDXR Mutations Cause Sensorial Neuropathies and Expand the Spectrum of Mitochondrial Fe-S-Synthesis Diseases. American Journal of Human Genetics, 2017, 101, 630-637.	6.2	52
25	MYO3A Causes Human Dominant Deafness and Interacts with Protocadherin 15-CD2 Isoform. Human Mutation, 2016, 37, 481-487.	2.5	27
26	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
27	Long-term clinical follow-up and molecular testing for diagnosis of the first Tunisian family with AlstrÃ¶m syndrome. European Journal of Medical Genetics, 2016, 59, 444-451.	1.3	6
28	Spectrum of DNA variants for non-syndromic deafness in a large cohort from multiple continents. Human Genetics, 2016, 135, 953-961.	3.8	102
29	Molecular Analysis of Libyan Families with Allgrove Syndrome: Geographic Expansion of the Ancestral Mutation c.1331+1G>A in North Africa. Hormone Research in Paediatrics, 2016, 85, 18-21.	1.8	7
30	Genetic diversity and haplotype structure of 21 Yâ€STRs, including nine noncore loci, in South Tunisian Population: Forensic relevance. Electrophoresis, 2015, 36, 2908-2913.	2.4	1
31	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
32	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
33	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
34	Genome-wide analysis reveals a novel autosomal-recessive hearing loss locus DFNB80 on chromosome 2p16.1-p21. Journal of Human Genetics, 2013, 58, 98-101.	2.3	3
35	Segregation of a new mutation in SLC26A4 and p.E47X mutation in GJB2 within a consanguineous Tunisian family affected with Pendred syndrome. International Journal of Pediatric Otorhinolaryngology, 2012, 76, 832-836.	1.0	13
36	Genome wide analysis in a family with sensorineural hearing loss, autism and mental retardation. Gene, 2012, 510, 102-106.	2.2	5

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37	Transcriptomic Analysis of the Developing and Adult Mouse Cochlear Sensory Epithelia. PLoS ONE, 2012, 7, e42987.	2.5	24
38	COL1A1 association and otosclerosis: A metaâ€analysis. American Journal of Medical Genetics, Part A, 2012, 158A, 1066-1070.	1.2	20
39	A novel dominant mutation in SIX1, affecting a highly conserved residue, result in only auditory defects in humans. European Journal of Medical Genetics, 2011, 54, e484-e488.	1.3	26
40	A novel missense mutation in the ESRRB gene causes DFNB35 hearing loss in a Tunisian family. European Journal of Medical Genetics, 2011, 54, e535-e541.	1.3	22
41	DFNB66 and DFNB67 loci are non allelic and rarely contribute to autosomal recessive nonsyndromic hearing loss. European Journal of Medical Genetics, 2011, 54, e565-e569.	1.3	10
42	Association of COL1A1 and TGFB1 Polymorphisms with Otosclerosis in a Tunisian Population. Annals of Human Genetics, 2011, 75, 598-604.	0.8	24
43	Genetic variants in RELN are associated with otosclerosis in a nonâ€European population from Tunisia. Annals of Human Genetics, 2010, 74, 399-405.	0.8	18
44	High Frequency of the p.R34X Mutation in the TMC1 Gene Associated with Nonsyndromic Hearing Loss Is Due to Founder Effects. Genetic Testing and Molecular Biomarkers, 2010, 14, 307-311.	0.7	41
45	Whole mitochondrial genome screening in two families with hearing loss: detection of a novel mutation in the 12S rRNA gene. Bioscience Reports, 2010, 30, 405-411.	2.4	7
46	Screening of the DFNB3 Locus: Identification of Three Novel Mutations of MYO15A Associated with Hearing Loss and Further Suggestion for Two Distinctive Genes on This Locus. Genetic Testing and Molecular Biomarkers, 2009, 13, 147-151.	0.7	39
47	Re-assigning the DFNB33 locus to chromosome 10p11.23â€“q21.1. European Journal of Human Genetics, 2009, 17, 122-124.	2.8	4
48	Mutation in gap and tight junctions in patients with non-syndromic hearing loss. Biochemical and Biophysical Research Communications, 2009, 385, 1-5.	2.1	20
49	Mutations of LRTOMT, a fusion gene with alternative reading frames, cause nonsyndromic deafness in humans. Nature Genetics, 2008, 40, 1335-1340.	21.4	65
50	New polymorphic mtDNA restriction site in the 12S rRNA gene detected in Tunisian patients with non-syndromic hearing loss. Biochemical and Biophysical Research Communications, 2008, 369, 849-852.	2.1	14
51	<i>TMC1</i> but Not <i>TMC2</i> Is Responsible for Autosomal Recessive Nonsyndromic Hearing Impairment in Tunisian Families. Audiology and Neuro-Otology, 2008, 13, 213-218.	1.3	54
52	Mutational analysis of the mitochondrial tRNALeu(UUR) gene in Tunisian patients with mitochondrial diseases. Biochemical and Biophysical Research Communications, 2007, 355, 1031-1037.	2.1	21
53	GJB2 (connexin 26) gene mutations in Moroccan patients with autosomal recessive non-syndromic hearing loss and carrier frequency of the common GJB2â€“35delG mutation. International Journal of Pediatric Otorhinolaryngology, 2007, 71, 1239-1245.	1.0	36
54	Mutational analysis of the mitochondrial 12S rRNA and tRNASer(UCN) genes in Tunisian patients with nonsyndromic hearing loss. Biochemical and Biophysical Research Communications, 2006, 340, 1251-1258.	2.1	22

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55	Identification of a novel frameshift mutation in the DFNB31/WHRN gene in a Tunisian consanguineous family with hereditary non-syndromic recessive hearing loss. Human Mutation, 2005, 25, 503-503.	2.5	51
56	A Novel Autosomal Recessive Non-Syndromic Deafness Locus, DFNB66, Maps to Chromosome 6p21.2-22.3 in a Large Tunisian Consanguineous Family. Human Heredity, 2005, 60, 123-128.	0.8	26
57	Consanguinity and endogamy in Northern Tunisia and its impact on nonâ€syndromic deafness. Genetic Epidemiology, 2004, 27, 74-79.	1.3	64
58	Mapping of a new autosomal recessive nonsyndromic hearing loss locus (DFNB32) to chromosome 1p13.3-22.1. European Journal of Human Genetics, 2003, 11, 185-188.	2.8	20
59	PDS Is a New Susceptibility Gene to Autoimmune Thyroid Diseases: Association and Linkage Study. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 2274-2280.	3.6	39
60	Usher syndrome type I G (USH1G) is caused by mutations in the gene encoding SANS, a protein that associates with the USH1C protein, harmonin. Human Molecular Genetics, 2003, 12, 463-471.	2.9	262
61	From DFNB2 to Usher syndrome: Variable expressivity of the same disease. American Journal of Medical Genetics Part A, 2001, 101, 181-183.	2.4	49
62	Mutations in a new gene encoding a protein of the hair bundle cause non-syndromic deafness at the DFNB16 locus. Nature Genetics, 2001, 29, 345-349.	21.4	159
63	Pendred syndrome: Phenotypic variability in two families carrying the samePDS missense mutation. American Journal of Medical Genetics Part A, 2000, 90, 38-44.	2.4	68