Leila Ammar-Keskes

List of Publications by Year
in descending order

72	1,170	516561 16	454834 30
papers	citations	h-index	g-index

73	73	73	1994
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Autosomal dominant polycystic kidney disease (ADPKD) in Tunisia: From molecular genetics to the development of prognostic tools. Gene, 2022, 817, 146174.	1.0	0
2	Involvement of C677T MTHFR variant but not A1298C in methotrexate-induced toxicity in acute lymphoblastic leukemia. Journal of Oncology Pharmacy Practice, 2021, 27, 1382-1387.	0.5	5
3	<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.	0.7	12
4	Can leukocytospermia predict prostate cancer via its effects on mitochondrial DNA?. Andrologia, 2021, 53, e14129.	1.0	0
5	Relationship between nuclear DNA fragmentation, mitochondrial DNA damage and standard sperm parameters in spermatozoa of infertile patients with leukocytospermia. Journal of Gynecology Obstetrics and Human Reproduction, 2021, 50, 102101.	0.6	14
6	The first concurrent detection of mitochondrial DNA m.3243A>G mutation, deletion, and depletion in a family with mitochondrial diabetes. Molecular Genetics & Genomic Medicine, 2020, 8, e1292.	0.6	3
7	Detection of a novel mutation in a Tunisian child with polycystic kidney disease. IUBMB Life, 2020, 72, 1799-1806.	1.5	1
8	Renal angiomyolipoma: Clinico-pathologic study of 17 cases with emphasis on the epithelioid histology and p53 gene abnormalities. Annals of Diagnostic Pathology, 2020, 47, 151538.	0.6	5
9	Mutations in GAA Gene in Tunisian Families with Infantile Onset Pompe Disease: Novel Mutation and Structural Modeling Investigations. Journal of Molecular Neuroscience, 2020, 70, 1100-1109.	1.1	3
10	A novel disease-causing mutation in the Renin gene in a Tunisian family with autosomal dominant tubulointerstitial kidney disease. International Journal of Biochemistry and Cell Biology, 2019, 117, 105625.	1.2	1
11	Thyroid involvement in Chanarin-Dorfman syndrome in adults in the largest series of patients carrying the same founder mutation in ABHD5 gene. Orphanet Journal of Rare Diseases, 2019, 14, 112.	1.2	9
12	A homozygous ABCB4 mutation causing an LPAC syndrome evolves into cholangiocarcinoma. Clinica Chimica Acta, 2019, 495, 598-605.	0.5	8
13	A novel Câ€terminal truncated mutation in hCDKL5 protein causing a severe West syndrome: Comparison with previous truncated mutations and genotype/phenotype correlation. International Journal of Developmental Neuroscience, 2019, 72, 22-30.	0.7	6
14	Next generation sequencing in family with MNGIE syndrome associated to optic atrophy: Novel homozygous POLG mutation in the C-terminal sub-domain leading to mtDNA depletion. Clinica Chimica Acta, 2019, 488, 104-110.	0.5	12
15	Altered three-dimensional organization of sperm genome in DPY19L2-deficient globozoospermic patients. Journal of Assisted Reproduction and Genetics, 2019, 36, 69-77.	1.2	7
16	First description of a novel mitochondrial mutation in the MT-TI gene associated with multiple mitochondrial DNA deletion and depletion in family with severe dilated mitochondrial cardiomyopathy. Biochemical and Biophysical Research Communications, 2018, 497, 1049-1054.	1.0	6
17	Cytochrome C oxydase deficiency: SURF1 gene investigation in patients with Leigh syndrome. Biochemical and Biophysical Research Communications, 2018, 497, 1043-1048.	1.0	7
18	First report of an unusual novel double mutation affecting the transcription repression domain of MeCP2 and causing a severe phenotype of Rett syndrome: Molecular analyses and computational investigation. Biochemical and Biophysical Research Communications, 2018, 497, 93-101.	1.0	2

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19	A duplex polymerase chain reactionâ€restriction fragment length polymorphism for rapid screening of methylenetetrahydrofolate reductase gene variants: Genotyping in acute leukemia. Journal of Clinical Laboratory Analysis, 2018, 32, .	0.9	5
20	Measurement of absolute copy number variation of Glutathione Sâ€Transferase M1 gene by digital droplet <scp>PCR</scp> and association analysis in Tunisian Rheumatoid Arthritis population. Journal of Clinical Laboratory Analysis, 2018, 32, .	0.9	3
21	Clinical, Molecular, and Computational Analysis in two cases with mitochondrial encephalomyopathy associated with SUCLG1 mutation in a consanguineous family. Biochemical and Biophysical Research Communications, 2018, 495, 1730-1737.	1.0	9
22	Do GSTM1 and GSTT1 polymorphisms influence the risk of developing mitochondrial diseases in a Tunisian population?. Environmental Science and Pollution Research, 2018, 25, 5779-5787.	2.7	4
23	Association of hyperhomocysteinemia with genetic variants in key enzymes of homocysteine metabolism and methotrexate toxicity in rheumatoid arthritis patients. Inflammation Research, 2018, 67, 703-710.	1.6	9
24	Clinical, Molecular, and Computational Analysis Showed a Novel Homozygous Mutation Among the Substrate-Binding Site of ARSA Protein in Consanguineous Family with Late-Infantile MLD. Journal of Molecular Neuroscience, 2018, 66, 17-25.	1.1	3
25	Novel cases of Tunisian patients with mutations in the gene encoding 17Î²-hydroxysteroid dehydrogenase type 3 and a founder effect. Journal of Steroid Biochemistry and Molecular Biology, 2017, 165, 86-94.	1.2	11
26	Co segregation of the m.1555A>G mutation in the MT-RNR1 gene and mutations in MT-ATP6 gene in a family with dilated mitochondrial cardiomyopathy and hearing loss: A whole mitochondrial genome screening. Biochemical and Biophysical Research Communications, 2017, 484, 71-78.	1.0	12
27	Original tandem duplication in FXIIIA gene with splicing site modification and four amino acids insertion causes factor XIII deficiency. Blood Coagulation and Fibrinolysis, 2017, 28, 237-243.	0.5	0
28	Clinical, Molecular, and Computational Analysis in Patients With a Novel Double Mutation and a New Synonymous Variant in MeCP2: Report of the First Missense Mutation Within the AT-hook1 Cluster in Rett Syndrome. Journal of Child Neurology, 2017, 32, 694-703.	0.7	8
29	Oxidative stress and glutathioneÂS-transferaseÂgenetic polymorphisms in medical staff professionally exposed to ionizing radiation. International Journal of Radiation Biology, 2017, 93, 697-704.	1.0	16
30	Cytogenetic monitoring of hospital staff exposed to ionizing radiation: optimize protocol considering DNA repair genes variability. International Journal of Radiation Biology, 2017, 93, 1283-1288.	1.0	13
31	Novel mutations in the CDKL5 gene in complex genotypes associated with West syndrome with variable phenotype: First description of somatic mosaic state. Clinica Chimica Acta, 2017, 473, 51-59.	0.5	9
32	Analysis of two susceptibility SNPs in HLA region and evidence of interaction between rs6457617 in HLA-DQB1 and HLA-DRB1*04 locus on Tunisian rheumatoid arthritis. Journal of Genetics, 2017, 96, 911-918.	0.4	8
33	Phenotypic variability in two infants sharing the same MECP2 mutation: evidence of chromosomal rearrangements and high sister-chromatid exchange levels in Rett syndrome. Acta Neurologica Belgica, 2017, 117, 251-258.	0.5	3
34	Mitochondrial DNA triplication and punctual mutations in patients with mitochondrial neuromuscular disorders. Biochemical and Biophysical Research Communications, 2016, 473, 578-585.	1.0	4
35	Mutational screening in patients with profound sensorineural hearing loss and neurodevelopmental delay: Description of a novel m.3861AÂ>ÂC mitochondrial mutation in the MT-ND1 gene. Biochemical and Biophysical Research Communications, 2016, 474, 702-708.	1.0	4
36	Clinical and Genetic Characterization of 26 Tunisian Patients with Allgrove Syndrome. Archives of Medical Research, 2016, 47, 105-110.	1.5	15

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37	Splicing Defects in the <i>AAAS</i> Gene Leading to both Exon Skipping and Partial Intron Retention in a Tunisian Patient with Allgrove Syndrome. Hormone Research in Paediatrics, 2016, 86, 90-93.	0.8	5
38	gr/gr- DAZ2-DAZ4-CDY1b deletion is a high-risk factor for male infertility in Tunisian population. Gene, 2016, 592, 29-35.	1.0	9
39	In silico investigation of the impact of synonymous variants in ABCB4 gene on mRNA stability/structure, splicing accuracy and codon usage: Potential contribution to PFIC3 disease. Computational Biology and Chemistry, 2016, 65, 103-109.	1.1	6
40	Mutational analysis in patients with neuromuscular disorders: Detection of mitochondrial deletion and double mutations in the MT-ATP6 gene. Biochemical and Biophysical Research Communications, 2016, 473, 61-66.	1.0	9
41	A de-novo large deletion of 2.8 kb produced in the ABCD1 gene causing adrenoleukodystrophy disease. Biochemistry and Cell Biology, 2016, 94, 265-269.	0.9	0
42	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.	0.8	7
43	Phenotypic variability in a Tunisian family with X-linked adrenoleukodystrophy caused by the p.Gln316Pro novel mutation. Clinica Chimica Acta, 2016, 453, 141-146.	0.5	4
44	Biochemical analyses and molecular modeling explain the functional loss of 17Î²-hydroxysteroid dehydrogenase 3 mutant G133R in three Tunisian patients with 46, XY Disorders of Sex Development. Journal of Steroid Biochemistry and Molecular Biology, 2016, 155, 147-154.	1.2	14
45	Genetic Determinants of Methotrexate Toxicity in Tunisian Patients with Rheumatoid Arthritis: A Study of Polymorphisms Involved in the MTX Metabolic Pathway. European Journal of Drug Metabolism and Pharmacokinetics, 2016, 41, 385-393.	0.6	27
46	Splicing defects in ABCD1 gene leading to both exon skipping and partial intron retention in X-linked adrenoleukodystrophy Tunisian patient. Neuroscience Research, 2015, 97, 7-12.	1.0	8
47	Identification of a novel m.9588Gâ€‰>â€‰A missense mutation in the mitochondrial COIII gene in asthenozoospermic Tunisian infertile men. Journal of Assisted Reproduction and Genetics, 2014, 31, 595-600.	1.2	21
48	Deletion of CDY1b copy of Y chromosome CDY1 gene is a risk factor of male infertility in Tunisian men. Gene, 2014, 548, 251-255.	1.0	31
49	Combined deletion of DAZ2 and DAZ4 copies of Y chromosome DAZ gene is associated with male infertility in Tunisian men. Gene, 2014, 547, 191-194.	1.0	9
50	Molecular Detection of Chlamydia trachomatis and Other Sexually Transmitted Bacteria in Semen of Male Partners of Infertile Couples in Tunisia: The Effect on Semen Parameters and Spermatozoa Apoptosis Markers. PLoS ONE, 2014, 9, e98903.	1.1	48
51	Quercetin attenuates lambda cyhalothrinâ€induced reproductive toxicity in male rats. Environmental Toxicology, 2013, 28, 673-680.	2.1	42
52	Mitochondrial DNA mutations and polymorphisms in asthenospermic infertile men. Molecular Biology Reports, 2013, 40, 4705-4712.	1.0	31
53	Pericentric inversion of chromosom 12 [Inv (12) (p12q12)] associated with idiopathic azoospermia in one infertile Tunisian man. Biochemical and Biophysical Research Communications, 2013, 432, 472-474.	1.0	6
54	A novel m.6307A>G mutation in the mitochondrial COXI gene in asthenozoospermic infertile men. Molecular Reproduction and Development, 2013, 80, 581-587.	1.0	19

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55	Assessment of Chromatin Maturity in Human Spermatozoa: Useful Aniline Blue Assay for Routine Diagnosis of Male Infertility. Advances in Urology, 2013, 2013, 1-8.	0.6	51
56	Identification of a new recurrent Aurora kinase C mutation in both European and African men with macrozoospermia. Human Reproduction, 2012, 27, 3337-3346.	0.4	52
57	Partial Microdeletions in the Y-Chromosome AZFc Region Are Not a Significant Risk Factor for Spermatogenic Impairment in Tunisian Infertile Men. Genetic Testing and Molecular Biomarkers, 2012, 16, 775-779.	0.3	13
58	Protective role of caffeic acid on lambda cyhalothrin-induced changes in sperm characteristics and testicular oxidative damage in rats. Toxicology and Industrial Health, 2012, 28, 639-647.	0.6	29
59	Possible Association of a Novel Missense Mutation A6375G in the MitochondrialCytochrome C Oxidase IGene with Asthenospermia in the Tunisian Population. Genetic Testing and Molecular Biomarkers, 2012, 16, 1298-1302.	0.3	8
60	Chromosomal defects in infertile men with poor semen quality. Journal of Assisted Reproduction and Genetics, 2012, 29, 451-456.	1.2	39
61	A Proposed Mouse Model to Study Male Infertility Provoked by Genital Serovar E, Chlamydia trachomatis. Journal of Andrology, 2011, 32, 86-94.	2.0	10
62	Sperm DNA fragmentation and oxidation are independent of malondialdheyde. Reproductive Biology and Endocrinology, 2011, 9, 47.	1.4	76
63	Dimethoateâ€induced oxidative stress in human erythrocytes and the protective effect of Vitamins C and E in vitro. Environmental Toxicology, 2011, 26, 287-291.	2.1	16
64	Lipid peroxidation and antioxidant enzyme activities in infertile men: correlation with semen parameter. Journal of Clinical Laboratory Analysis, 2009, 23, 99-104.	0.9	61
65	Genetic association between AZF region polymorphism and Klinefelter syndrome. Reproductive BioMedicine Online, 2009, 19, 547-551.	1.1	18
66	Tag STS in the AZF Region Associated With Azoospermia in a Tunisian Population. Journal of Andrology, 2007, 28, 652-658.	2.0	8
67	Ureaplasma urealyticum, Ureaplasma parvum, Mycoplasma hominis and Mycoplasma genitalium infections and semen quality of infertile men. BMC Infectious Diseases, 2007, 7, 129.	1.3	150
68	Combining Autosomal and Y-Chromosomal Short Tandem Repeat Data in Paternity Testing with Male Child: Methods and Application. Journal of Forensic Sciences, 2007, 52, 1068-1072.	0.9	13
69	Date seed oil limit oxidative injuries induced by hydrogen peroxide in human skin organ culture. BioFactors, 2007, 29, 137-145.	2.6	14
70	Haplotypes for 13 Y-chromosomal STR loci in South Tunisian population (Sfax region). Forensic Science International, 2006, 164, 249-253.	1.3	18
71	Androgen receptor gene CAG repeats length in fertile and infertile Tunisian men. Annales De GÃ©nÃ©tique, 2004, 47, 217-224.	0.4	17
72	Skewed X-chromosome inactivation pattern in SRY positive XX maleness: a case report and review of literature. Annales De GÃ©nÃ©tique, 2003, 46, 11-18.	0.4	40