# Hala T Bassyouni

## List of Publications by Year in Descending Order



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| 67 papers | 309 citations | 10 h-index | 15 g-index |
| 81 ext. papers | 400 ext. citations | 2 avg, IF | 3.17 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 67 | Dynamic disequilibrium-based pathogenicity model in mutated pyrin B30.2 domain-Casp1/p20 complex.. *Journal of Genetic Engineering and Biotechnology*, **2022**, 20, 31 | 3.1 | 0 |
| 66 | Serum homocysteine, lipid profile and BMI as atherosclerotic risk factors in children with numerical chromosomal aberrations.. *World Journal of Pediatrics*, **2022**, 1 | 4.6 | 1 |
| 65 | The Effect of Anti-inflammatory Diet and Vitamin D Supplementation on the Amelioration of the Clinical Status and Cognitive functions of Familial Mediterranean Fever Patients. *Kobe Journal of Medical Sciences*, **2021**, 66, E159-E165 | 0.6 | |
| 64 | Differential Expression of micro RNAs and their Association with the Inflammatory Markers in Familial Mediterranean Fever Patients. *Biomedical and Pharmacology Journal*, **2021**, 14, 1351-1358 | 0.9 | |
| 63 | The correlation of estrogen receptor 1 and progesterone receptor genes polymorphisms with recurrent pregnancy loss in a cohort of Egyptian women. *Molecular Biology Reports*, **2021**, 48, 4413-4420 | 2.8 | 2 |
| 62 | Clinical, Biochemical, and Molecular Characterization of Metachromatic Leukodystrophy Among Egyptian Pediatric Patients: Expansion of the ARSA Mutational Spectrum. *Journal of Molecular Neuroscience*, **2021**, 71, 1112-1130 | 3.3 | |
| 61 | IGF1R, IGFALS, and IGFBP3 gene copy number variations in a group of non-syndromic Egyptian short children. *Journal of Genetic Engineering and Biotechnology*, **2021**, 19, 109 | 3.1 | |
| 60 | Clinical Implications of S100A12 and Resolvin D1 Serum Levels, and Related Genes in Children with Familial Mediterranean Fever. *Journal of Child Science*, **2021**, 11, e163-e169 | 0.2 | |
| 59 | A descriptive study of NPHS1 and NPHS2 mutations in children with congenital nephrotic syndrome. *Gene Reports*, **2020**, 20, 100722 | 1.4 | 1 |
| 58 | Mutation in the Gene in an Egyptian Patient with H Syndrome: A Case Report and Review of Literature. *Journal of Pediatric Genetics*, **2020**, 9, 109-113 | 0.7 | 3 |
| 57 | Assessment of Multiplex Ligation-Dependent Probe Amplification (MLPA) as a diagnostic test for Egyptian patients with Williams-Beuren syndrome. *Gene Reports*, **2020**, 20, 100767 | 1.4 | |
| 56 | Detection of low-grade mosaicism and its correlation with hormonal profile, testicular volume, and semen quality in a cohort of Egyptian Klinefelter and Klinefelter-like patients. *Reproductive Biology*, **2020**, 20, 259-263 | 2.3 | 0 |
| 55 | Microcephalic osteodysplastic primordial dwarfism type II: Additional nine patients with implications on phenotype and genotype correlation. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1407-1420 | 2.5 | 6 |
| 54 | Screening of the SHOX/PAR1 region using MLPA and miRNA expression profiling in a group of Egyptian children with non-syndromic short stature. *Egyptian Journal of Medical Human Genetics*, **2020**, 21, | 2 | 1 |
| 53 | Measurement of Serum Chemerin, Oxidized LDL, and Vitamin D Levels in Prader-Willi Syndrome: A Cross-Sectional Study in Pediatric Egyptian Patients. *Journal of Child Science*, **2020**, 10, e187-e195 | 0.2 | 0 |
| 52 | Inflammatory and endothelial dysfunction indices among Egyptian females with obesity classes I-III. *Bioscience Reports*, **2020**, 40, | 4.1 | 3 |
| 51 | Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. *Brain*, **2020**, 143, 2388-2397 | 11.2 | 10 |

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| 50 | Carotid intima-media thickness, lipid profile, serum amyloid A and vitamin D status in children with familial Mediterranean fever. *Egyptian Rheumatologist*, **2020**, 42, 237-240 | 1 | 4 |
| 49 | Genetic assessment of ten Egyptian patients with Sjögren-Larsson syndrome: expanding the clinical spectrum and reporting a novel ALDH3A2 mutation. *Archives of Dermatological Research*, **2019**, 311, 721-730 | 2.3 | 4 |
| 48 | Oxidative Stress, Neutrophil Elastase and Vascular Endothelial Growth Factor in Obese Pregnant Women with Preeclampsia. *Biomedical and Pharmacology Journal*, **2019**, 12, 1887-1891 | 0.9 | |
| 47 | DNA Damage and Neutrophil Elastase in Children with Prader-Willi Syndrome. *Biomedical and Pharmacology Journal*, **2019**, 12, 1967-1974 | 0.9 | 1 |
| 46 | Early Detection and Management of Prader-Willi Syndrome in Egyptian Patients. *Journal of Pediatric Genetics*, **2019**, 8, 179-186 | 0.7 | 1 |
| 45 | The association of +1150A polymorphism with low GH level in isolated growth hormone deficiency (IGHD) patients. *Gene Reports*, **2019**, 14, 118-123 | 1.4 | |
| 44 | Apoptosis, reactive oxygen species and DNA damage in Familial Mediterranean Fever patients. *Gene Reports*, **2019**, 14, 76-80 | 1.4 | 1 |
| 43 | Evaluation of DNA damage profile in obese women and its association to risk of metabolic syndrome, polycystic ovary syndrome and recurrent preeclampsia. *Genes and Diseases*, **2018**, 5, 367-373 | 6.6 | 13 |
| 42 | Screening of the most common MEFV mutations in a large cohort of Egyptian patients with Familial Mediterranean fever. *Gene Reports*, **2018**, 11, 23-28 | 1.4 | 4 |
| 41 | Assessment of DNA damage in obese premenopausal women with metabolic syndrome. *Gene Reports*, **2018**, 10, 42-46 | 1.4 | 4 |
| 40 | Assessment of physical growth, some oxidative stress biomarkers and vitamin D status in children with Familial Mediterranean Fever. *Meta Gene*, **2018**, 17, 212-215 | 0.7 | 2 |
| 39 | Immunological Evaluation in Patients with Familial Mediterranean fever. *Open Access Macedonian Journal of Medical Sciences*, **2018**, 6, 310-313 | 1 | 5 |
| 38 | Dysregulation of tumor necrosis factor- and interleukin-6 as predictors of gestational disorders. *Middle East Journal of Medical Genetics*, **2018**, 7, 112 | | 2 |
| 37 | Cytogenomic characterization of 1q43q44 deletion associated with 4q32.1q35.2 duplication and phenotype correlation. *Molecular Cytogenetics*, **2018**, 11, 57 | 2 | 6 |
| 36 | Coenzyme Q10 and pro-inflammatory markers in children with Down syndrome: clinical and biochemical aspects. *Jornal De Pediatria (Versão Em Português)*, **2017**, 93, 100-104 | 0.2 | 1 |
| 35 | Registry of ocular anomalies among patients with genetic disorders attending the clinical genetics department at the National Research Center in Egypt. *Ophthalmic Genetics*, **2017**, 38, 418-421 | 1.2 | 1 |
| 34 | Aicardi-Goutières syndrome: unusual neuro-radiological manifestations. *Metabolic Brain Disease*, **2017**, 32, 679-683 | 3.9 | 7 |
| 33 | Association of vitamin D receptor gene polymorphism (VDR) with vitamin D deficiency, metabolic and inflammatory markers in Egyptian obese women. *Genes and Diseases*, **2017**, 4, 176-182 | 6.6 | 29 |

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| 32 | Coenzyme Q10 and pro-inflammatory markers in children with Down syndrome: clinical and biochemical aspects. *Jornal De Pediatria*, **2017**, 93, 100-104 | 2.6 | 24 |
| 31 | Serum soluble receptor of advanced glycation end products and risk of metabolic syndrome in Egyptian obese women. *EXCLI Journal*, **2017**, 16, 973-980 | 2.4 | 6 |
| 30 | Association of the Pro12Ala Polymorphism with the Metabolic Parameters in Women with Polycystic Ovary Syndrome. *Open Access Macedonian Journal of Medical Sciences*, **2017**, 5, 275-280 | 1 | 6 |
| 29 | Mercury toxicity and DNA damage in patients with Down syndrome. *Medical Research Journal*, **2016** , 15, 22-26 | | 7 |
| 28 | Molybdenum cofactor and isolated sulphite oxidase deficiencies: Clinical and molecular spectrum among Egyptian patients. *European Journal of Paediatric Neurology*, **2016**, 20, 714-22 | 3.8 | 21 |
| 27 | Indicators of the metabolic syndrome in obese adolescents. *Archives of Medical Science*, **2015**, 11, 92-8 | 2.9 | 21 |
| 26 | The role of H. pylori infection in gall bladder cancer: clinicopathological study. *Tumor Biology*, **2015**, 36, 7093-8 | 2.9 | 14 |
| 25 | Metabolic abnormalities in young Egyptian women with polycystic ovary syndrome and their relation to ADIPOQ gene variants and body fat phenotype. *Egyptian Journal of Medical Human Genetics*, **2015**, 16, 367-374 | 2 | 1 |
| 24 | Cross-sectional analysis of long bones in a sample of ancient Egyptians. *Egyptian Journal of Radiology and Nuclear Medicine*, **2015**, 46, 675-681 | 1.4 | 3 |
| 23 | Obesity in relation to inflammatory biomarkers, adiponectin gene variability, and insulin resistance among middle-aged Egyptian women. *Middle East Journal of Medical Genetics*, **2015**, 4, 70-76 | | |
| 22 | Behavioral problems, biochemical, and anthropometric characteristics of patients with Prader Willi syndrome. *Middle East Journal of Medical Genetics*, **2015**, 4, 63-69 | | 3 |
| 21 | Oxidative Stress -a Phenotypic Hallmark of Fanconi Anemia and Down Syndrome: The Effect of Antioxidants. *Annals of Medical and Health Sciences Research*, **2015**, 5, 205-12 | | 4 |
| 20 | Detection and Quantification of Free Radicals in Peroxisomal Disorders: A Comparative Study with Oxidative Stress Parameters. *Journal of Clinical and Diagnostic Research JCDR*, **2015**, 9, SC17-20 | 0 | |
| 19 | Association of serum paraoxonase enzyme activity and oxidative stress markers with dyslipidemia in obese adolescents. *Indian Journal of Endocrinology and Metabolism*, **2014**, 18, 340-4 | 1.7 | 18 |
| 18 | Mutation analysis of the GJB2 and GJB6 genes in Egyptian patients with autosomal recessive sensorineural nonsyndromic hearing loss. *Middle East Journal of Medical Genetics*, **2014**, 3, 11-15 | | 2 |
| 17 | Screening for common mutations in four FANCA gene exons in Egyptian Fanconi anemia patients. *Middle East Journal of Medical Genetics*, **2014**, 3, 24-30 | | 2 |
| 16 | Osteoporosis in Chronic Hepatitis C Virus with Advanced Liver Fibrosis. *Journal of Gastroenterology and Hepatology Research*, **2014**, 3, 1392-1395 | 0.9 | 2 |
| 15 | Anti-diuretic hormone and genetic study in primary nocturnal enuresis. *Journal of Pediatric Urology*, **2013**, 9, 831-7 | 1.5 | 10 |

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| 14 | Clinical and molecular findings in eight Egyptian patients with suspected mitochondrial disorders and optic atrophy. *Egyptian Journal of Medical Human Genetics*, **2013**, 14, 37-47 | 2 | 0 |
| 13 | Distinct ocular expression in infants and children with Down syndrome in Cairo, Egypt: myopia and heart disease. *JAMA Ophthalmology*, **2013**, 131, 1057-66 | 3.9 | 18 |
| 12 | Screening seven common mitochondrial mutations in 28 Egyptian patients with suspected mitochondrial disease. *Middle East Journal of Medical Genetics*, **2013**, 2, 28-37 | | |
| 11 | Assessment of metal content and oxidative stress in autistic Egyptian patients. *Middle East Journal of Medical Genetics*, **2013**, 2, 23-27 | | |
| 10 | Growth curves of Egyptian patients with Turner syndrome. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2687-91 | 2.5 | 5 |
| 9 | A cephalometric study of skulls from the Bahriyah oasis. *Journal of Forensic Dental Sciences*, **2012**, 4, 88-92 | 2 | |
| 8 | Evidence of oxidative stress in peroxisomal disorders. *Singapore Medical Journal*, **2012**, 53, 608-14 | 1.9 | 2 |
| 7 | P53 protein and Ki-67 expression in chronic gastritis patients with positive Helicobacter pylori infection. *Journal of Genetic Engineering and Biotechnology*, **2011**, 9, 73-76 | 3.1 | 2 |
| 6 | Sexual dysfunction in males with hepatitis C virus: relevance to histopathologic changes and peginterferon treatment. *Saudi Journal of Gastroenterology*, **2011**, 17, 406-10 | 3 | 7 |
| 5 | Variable associations of Klinefelter syndrome in children. *Journal of Pediatric Endocrinology and Metabolism*, **2010**, 23, 985-9 | 1.6 | 4 |
| 4 | Management of rare side effects of peginterferon and ribavirin therapy during hepatitis C treatment: a case report. *Cases Journal*, **2009**, 2, 7429 | | 3 |
| 3 | Polyploidy in chronic lymphocytic leukemia with p53 deletion detected by fish: a case report. *Cases Journal*, **2009**, 2, 8872 | | |
| 2 | The Effect of Diet on Antioxidant Status in Patients with Galactosemia. *Journal of Medical Sciences (Faisalabad, Pakistan)*, **2006**, 6, 452-457 | 0.5 | 2 |
| 1 | Lipocalin-2 is an inflammatory biomarker associated with metabolic abnormalities in Egyptian obese children. *Journal of Applied Pharmaceutical Science*,007-012 | 2 | 2 |