Inas Mazen

List of Publications by Citations

45 papers	723 citations	12 h-index	26 g-index
46 ext. papers	883 ext. citations	3 avg, IF	3.07 L-index

#	Paper	IF	Citations
45	Phenotypical, biological, and molecular heterogeneity of 5□-reductase deficiency: an extensive international experience of 55 patients. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, 296-307	5.6	156
44	The spectrum of phenotypes associated with mutations in steroidogenic factor 1 (SF-1, NR5A1, Ad4BP) includes severe penoscrotal hypospadias in 46,XY males without adrenal insufficiency. *European Journal of Endocrinology*, **2009**, 161, 237-42	6.5	97
43	Changes over time in sex assignment for disorders of sex development. *Pediatrics*, **2014**, 134, e710-5	7.4	81
42	Severe Early-Onset Obesity Due to Bioinactive Leptin Caused by a p.N103K Mutation in the Leptin Gene. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, 3227-30	5.6	44
41	Mutations involving the SRY-related gene SOX8 are associated with a spectrum of human reproductive anomalies. *Human Molecular Genetics*, **2018**, 27, 1228-1240	5.6	40
40	Molecular analysis of 5alpha-reductase type 2 gene in eight unrelated egyptian children with suspected 5alpha-reductase deficiency: prevalence of the G34R mutation. *Clinical Endocrinology*, **2003**, 58, 627-31	3.4	36
39	Identification of NR5A1 Mutations and Possible Digenic Inheritance in 46,XY Gonadal Dysgenesis. *Sexual Development*, **2016**, 10, 147-51	1.6	27
38	Analysis of the gene coding for steroidogenic factor 1 (SF1, NR5A1) in a cohort of 50 Egyptian patients with 46,XY disorders of sex development. *European Journal of Endocrinology*, **2014**, 170, 759-67	6.5	22
37	Testis formation in XX individuals resulting from novel pathogenic variants in Wilms tumor 1 () gene. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 13680-13688	11.5	20
36	5 alpha-reductase deficiency in patients with micropenis. *Journal of Inherited Metabolic Disease*, **1997**, 20, 95-101	5.4	17
35	Pathogenic variants in the DEAH-box RNA helicase DHX37 are a frequent cause of 46,XY gonadal dysgenesis and 46,XY testicular regression syndrome. *Genetics in Medicine*, **2020**, 22, 150-159	8.1	16
34	Aromatase Deficiency due to a Homozygous CYP19A1 Mutation in a 46,XX Egyptian Patient with Ambiguous Genitalia. *Sexual Development*, **2017**, 11, 275-279	1.6	12
33	A new mutation of 5-alpha-reductase type 2 (A62E) in a large Egyptian kindred. *Hormone Research in Paediatrics*, **2003**, 59, 281-4	3.3	11
32	Transposition of external genitalia and associated malformations. *Clinical Dysmorphology*, **2003**, 12, 59-62	2.9	10
31	Homozygous Mutation of the FGFR1 Gene Associated with Congenital Heart Disease and 46,XY Disorder of Sex Development. *Sexual Development*, **2016**, 10, 16-22	1.6	10
30	Isodicentric Y chromosomes in Egyptian patients with disorders of sex development (DSD). *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1594-603	2.5	9
29	Tissue-specific mosaicism for tetrasomy 9p uncovered by array CGH. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2496-2500	2.5	9

28	AMH gene mutations in two Egyptian families with persistent m□lerian duct syndrome. *Sexual Development*, **2011**, 5, 277-80	1.6	9
27	Detection of the G34R mutation in the 5 alpha reductase 2 gene by allele specific PCR and its linkage to the 89L allele among Egyptian cases. *Sexual Development*, **2007**, 1, 293-6	1.6	9
26	Biallelic novel missense HHAT variant causes syndromic microcephaly and cerebellar-vermis hypoplasia. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1053-1057	2.5	8
25	A novel point mutation of the androgen receptor (F804L) in an Egyptian newborn with complete androgen insensitivity associated with congenital glaucoma and hypertrophic pyloric stenosis. *Clinical Genetics*, **2003**, 63, 59-63	4	8
24	Clinical and molecular characterization of seven Egyptian families with autosomal recessive robinow syndrome: Identification of four novel ROR2 gene mutations. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3054-61	2.5	7
23	Hereditary 1,25-dihydroxyvitamin D-resistant rickets with alopecia in four Egyptian families: report of three novel mutations in the vitamin D receptor gene. *Journal of Pediatric Endocrinology and Metabolism*, **2014**, 27, 873-8	1.6	6
22	Identification of a novel homozygous ALX4 mutation in two unrelated patients with frontonasal dysplasia type-2. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1190-1194	2.5	5
21	A novel mutation in the leptin gene (W121X) in an Egyptian family. *Molecular Genetics and Metabolism Reports*, **2014**, 1, 474-476	1.8	5
20	Biochemical Analysis of Four Missense Mutations in the HSD17B3 Gene Associated With 46,XY Disorders of Sex Development in Egyptian Patients. *Journal of Sexual Medicine*, **2017**, 14, 1165-1174	1.1	5
19	GAPO syndrome in seven new patients: Identification of five novel ANTXR1 mutations including the first large intragenic deletion. *American Journal of Medical Genetics, Part A*, **2019**, 179, 237-242	2.5	5
18	Achieving diagnostic certainty in resource-limited settings. *Endocrine Development*, **2014**, 27, 257-67		4
17	Variable associations of Klinefelter syndrome in children. *Journal of Pediatric Endocrinology and Metabolism*, **2010**, 23, 985-9	1.6	4
16	Advances in genomic diagnosis of a large cohort of Egyptian patients with disorders of sex development. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1666-1677	2.5	4
15	Novel AMH and AMHR2 Mutations in Two Egyptian Families with Persistent M□lerian Duct Syndrome. *Sexual Development*, **2017**, 11, 29-33	1.6	3
14	Unique karyotype: mos 46,X,dic(X;Y)(p22.33;p11.32)/ 45,X/45,dic(X;Y)(p22.33;p11.32) in an Egyptian patient with Ovotesticular disorder of sexual development. *Sexual Development*, **2013**, 7, 235-43	1.6	3
13	WT1 Gene Mutation, p.R462W, in a 46,XY DSD Patient from Egypt with Gonadoblastoma and Review of the Literature. *Sexual Development*, **2017**, 11, 280-283	1.6	3
12	Mutational pattern in the 5□reductase 2 (SRD5A2) gene in 46,XY Egyptian DSD patients. *Middle East Journal of Medical Genetics*, **2015**, 4, 77-82		3
11	A novel mutation (c.2735_2736delTC) in the androgen receptor gene in 46,XY females with complete androgen insensitivity syndrome in an Egyptian family. *Hormone Research in Paediatrics*, **2014**, 82, 411-4	3.3	3

10	A novel nonsense mutation in exon 1 of HSD17B3 gene in an Egyptian 46,XY adult female presenting with primary amenorrhea. *Sexual Development*, **2013**, 7, 277-81	1.6	3
9	Unusual association of simplified gyral pattern and sparse hair in an Egyptian patient with microcephaly-lymphoedema. *Clinical Dysmorphology*, **2006**, 15, 245-247	0.9	2
8	Chromosome 9p terminal deletion in nine Egyptian patients and narrowing of the critical region for trigonocephaly. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1829	2.3	2
7	A Homozygous Missense Variant in Hedgehog Acyltransferase (HHAT) Gene Associated with 46,XY Gonadal Dysgenesis.. *Sexual Development*, **2022**, 1-5	1.6	1
6	Gonadectomy in conditions affecting sex development: a registry-based cohort study. *European Journal of Endocrinology*, **2021**, 184, 791-801	6.5	1
5	Testosterone Therapy and Its Monitoring in Adolescent Boys with Hypogonadism: Results of an International Survey from the I-DSD Registry. *Sexual Development*, **2021**, 15, 236-243	1.6	1
4	Expanding DSD Phenotypes Associated with Variants in the DEAH-Box RNA Helicase DHX37. *Sexual Development*, **2021**, 15, 244-252	1.6	1
3	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
2	Assessment of the most common CYP21A2 point mutations in a cohort of congenital adrenal hyperplasia patients from Egypt. *Journal of Pediatric Endocrinology and Metabolism*, **2020**, 33, 893-900	1.6	
1	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	