

Inas Mazen

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	A Homozygous Missense Variant in Hedgehog Acyltransferase (HHAT) Gene Associated with 46,XY Gonadal Dysgenesis.. <i>Sexual Development</i> , 2022 , 1-5	1.6	1
44	Chromosome 9p terminal deletion in nine Egyptian patients and narrowing of the critical region for trigonocephaly. <i>Molecular Genetics & Genomic Medicine</i> , 2021 , 9, e1829	2.3	2
43	Advances in genomic diagnosis of a large cohort of Egyptian patients with disorders of sex development. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 1666-1677	2.5	4
42	Gonadectomy in conditions affecting sex development: a registry-based cohort study. <i>European Journal of Endocrinology</i> , 2021 , 184, 791-801	6.5	1
41	Testosterone Therapy and Its Monitoring in Adolescent Boys with Hypogonadism: Results of an International Survey from the I-DSD Registry. <i>Sexual Development</i> , 2021 , 15, 236-243	1.6	1
40	IGF1R, IGFALS, and IGFBP3 gene copy number variations in a group of non-syndromic Egyptian short children. <i>Journal of Genetic Engineering and Biotechnology</i> , 2021 , 19, 109	3.1	
39	Expanding DSD Phenotypes Associated with Variants in the DEAH-Box RNA Helicase DHX37. <i>Sexual Development</i> , 2021 , 15, 244-252	1.6	1
38	Testis formation in XX individuals resulting from novel pathogenic variants in Wilms tumor 1 () gene. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020 , 117, 13680-13688	11.5	20
37	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. <i>Reproductive Biology</i> , 2020 , 20, 259-263	2.3	0
36	Assessment of the most common CYP21A2 point mutations in a cohort of congenital adrenal hyperplasia patients from Egypt. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2020 , 33, 893-900	1.6	
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. <i>Genetics in Medicine</i> , 2020 , 22, 150-159	8.1	16
34	Biallelic novel missense HHAT variant causes syndromic microcephaly and cerebellar-vermis hypoplasia. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 1053-1057	2.5	8
33	GAPO syndrome in seven new patients: Identification of five novel ANTXR1 mutations including the first large intragenic deletion. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 237-242	2.5	5
32	Identification of a novel homozygous ALX4 mutation in two unrelated patients with frontonasal dysplasia type-2. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 1190-1194	2.5	5
31	Mutations involving the SRY-related gene SOX8 are associated with a spectrum of human reproductive anomalies. <i>Human Molecular Genetics</i> , 2018 , 27, 1228-1240	5.6	40
30	Novel AMH and AMHR2 Mutations in Two Egyptian Families with Persistent Mullerian Duct Syndrome. <i>Sexual Development</i> , 2017 , 11, 29-33	1.6	3
29	WT1 Gene Mutation, p.R462W, in a 46,XY DSD Patient from Egypt with Gonadoblastoma and Review of the Literature. <i>Sexual Development</i> , 2017 , 11, 280-283	1.6	3

28	Aromatase Deficiency due to a Homozygous CYP19A1 Mutation in a 46,XX Egyptian Patient with Ambiguous Genitalia. <i>Sexual Development</i> , 2017 , 11, 275-279	1.6	12
27	Biochemical Analysis of Four Missense Mutations in the HSD17B3 Gene Associated With 46,XY Disorders of Sex Development in Egyptian Patients. <i>Journal of Sexual Medicine</i> , 2017 , 14, 1165-1174	1.1	5
26	Homozygous Mutation of the FGFR1 Gene Associated with Congenital Heart Disease and 46,XY Disorder of Sex Development. <i>Sexual Development</i> , 2016 , 10, 16-22	1.6	10
25	Identification of NR5A1 Mutations and Possible Digenic Inheritance in 46,XY Gonadal Dysgenesis. <i>Sexual Development</i> , 2016 , 10, 147-51	1.6	27
24	Severe Early-Onset Obesity Due to Bioinactive Leptin Caused by a p.N103K Mutation in the Leptin Gene. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015 , 100, 3227-30	5.6	44
23	Clinical and molecular characterization of seven Egyptian families with autosomal recessive robinow syndrome: Identification of four novel ROR2 gene mutations. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 3054-61	2.5	7
22	Mutational pattern in the 5 α -reductase 2 (SRD5A2) gene in 46,XY Egyptian DSD patients. <i>Middle East Journal of Medical Genetics</i> , 2015 , 4, 77-82		3
21	Changes over time in sex assignment for disorders of sex development. <i>Pediatrics</i> , 2014 , 134, e710-5	7.4	81
20	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. <i>European Journal of Endocrinology</i> , 2014 , 170, 759-67	6.5	22
19	A novel mutation in the leptin gene (W121X) in an Egyptian family. <i>Molecular Genetics and Metabolism Reports</i> , 2014 , 1, 474-476	1.8	5
18	Hereditary 1,25-dihydroxyvitamin D-resistant rickets with alopecia in four Egyptian families: report of three novel mutations in the vitamin D receptor gene. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2014 , 27, 873-8	1.6	6
17	Achieving diagnostic certainty in resource-limited settings. <i>Endocrine Development</i> , 2014 , 27, 257-67		4
16	A novel mutation (c.2735_2736delTC) in the androgen receptor gene in 46,XY females with complete androgen insensitivity syndrome in an Egyptian family. <i>Hormone Research in Paediatrics</i> , 2014 , 82, 411-4	3.3	3
15	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. <i>Sexual Development</i> , 2013 , 7, 235-43	1.6	3
14	A novel nonsense mutation in exon 1 of HSD17B3 gene in an Egyptian 46,XY adult female presenting with primary amenorrhea. <i>Sexual Development</i> , 2013 , 7, 277-81	1.6	3
13	Isodicentric Y chromosomes in Egyptian patients with disorders of sex development (DSD). <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 1594-603	2.5	9
12	Phenotypical, biological, and molecular heterogeneity of 5 α -reductase deficiency: an extensive international experience of 55 patients. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2011 , 96, 296-307	5.6	156
11	Tissue-specific mosaicism for tetrasomy 9p uncovered by array CGH. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 2496-2500	2.5	9

10	AMH gene mutations in two Egyptian families with persistent müllerian duct syndrome. <i>Sexual Development</i> , 2011 , 5, 277-80	1.6	9
9	Variable associations of Klinefelter syndrome in children. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2010 , 23, 985-9	1.6	4
8	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. <i>European Journal of Endocrinology</i> , 2009 , 161, 237-42	6.5	97
7	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. <i>Sexual Development</i> , 2007 , 1, 293-6	1.6	9
6	Unusual association of simplified gyral pattern and sparse hair in an Egyptian patient with microcephaly-lymphoedema. <i>Clinical Dysmorphology</i> , 2006 , 15, 245-247	0.9	2
5	Transposition of external genitalia and associated malformations. <i>Clinical Dysmorphology</i> , 2003 , 12, 59-62.	0.9	10
4	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. <i>Clinical Genetics</i> , 2003 , 63, 59-63	4	8
3	Molecular analysis of 5alpha-reductase type 2 gene in eight unrelated egyptian children with suspected 5alpha-reductase deficiency: prevalence of the G34R mutation. <i>Clinical Endocrinology</i> , 2003 , 58, 627-31	3.4	36
2	A new mutation of 5-alpha-reductase type 2 (A62E) in a large Egyptian kindred. <i>Hormone Research in Paediatrics</i> , 2003 , 59, 281-4	3.3	11
1	5 alpha-reductase deficiency in patients with micropenis. <i>Journal of Inherited Metabolic Disease</i> , 1997 , 20, 95-101	5.4	17