Inas Mazen

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/7820420/publications.pdf

Version: 2024-02-01

45 papers	1,027 citations	686830 13 h-index	433756 31 g-index
46 all docs	46 docs citations	46 times ranked	1206 citing authors

#	Article	IF	Citations
1	Phenotypical, Biological, and Molecular Heterogeneity of 5Î±-Reductase Deficiency: An Extensive International Experience of 55 Patients. Journal of Clinical Endocrinology and Metabolism, 2011, 96, 296-307.	1.8	192
2	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, 237-242.	1.9	115
3	Changes Over Time in Sex Assignment for Disorders of Sex Development. Pediatrics, 2014, 134, e710-e715.	1.0	98
4	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, 3227-3230.	1.8	71
5	Mutations involving the SRY-related gene SOX8 are associated with a spectrum of human reproductive anomalies. Human Molecular Genetics, 2018, 27, 1228-1240.	1.4	64
6	Molecular analysis of 5Î±-reductase type 2 gene in eight unrelated egyptian children with suspected 5Î±-reductase deficiency: prevalence of the G34R mutation. Clinical Endocrinology, 2003, 58, 627-631.	1.2	56
7	Testis formation in XX individuals resulting from novel pathogenic variants in Wilmsâ€™ tumor 1 () Tj ETQq1 1 0.784314 rgBT /Overlo 2020, 117, 13680-13688.	3.3	42
8	Identification of <i>NR5A1</i> Mutations and Possible Digenic Inheritance in 46,XY Gonadal Dysgenesis. Sexual Development, 2016, 10, 147-151.	1.1	35
9	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.	1.1	34
10	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, 759-767.	1.9	32
11	5Î±-Reductase deficiency in patients with micropenis. Journal of Inherited Metabolic Disease, 1997, 20, 95-101.	1.7	23
12	A New Mutation of 5-Alpha-Reductase Type 2 (A62E) in a Large Egyptian Kindred. Hormone Research in Paediatrics, 2003, 59, 281-284.	0.8	17
13	Aromatase Deficiency due to a Homozygous <i>CYP19A1</i> Mutation in a 46,XX Egyptian Patient with Ambiguous Genitalia. Sexual Development, 2017, 11, 275-279.	1.1	17
14	Biallelic novel missense HHAT variant causes syndromic microcephaly and cerebellarâ€vermis hypoplasia. American Journal of Medical Genetics, Part A, 2019, 179, 1053-1057.	0.7	15
15	AMH Gene Mutations in Two Egyptian Families with Persistent MÃ¼llerian Duct Syndrome. Sexual Development, 2011, 5, 277-280.	1.1	14
16	Transposition of external genitalia and associated malformations. Clinical Dysmorphology, 2003, 12, 59-62.	0.1	13
17	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, 293-296.	1.1	11
18	Homozygous Mutation of the <i>FGFR1</i> Gene Associated with Congenital Heart Disease and 46,XY Disorder of Sex Development. Sexual Development, 2016, 10, 16-22.	1.1	11

#	Article	IF	Citations
19	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.	0.7	11
20	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, 2002, 63, 59-63.	1.0	10
21	Tissueâ€specific mosaicism for tetrasomy 9p uncovered by array CGH. American Journal of Medical Genetics, Part A, 2011, 155, 2496-2500.	0.7	10
22	Isodicentric Y chromosomes in Egyptian patients with disorders of sex development (DSD). American Journal of Medical Genetics, Part A, 2012, 158A, 1594-1603.	0.7	10
23	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, 167, 3054-3061.	0.7	10
24	Gonadectomy in conditions affecting sex development: a registry-based cohort study. European Journal of Endocrinology, 2021, 184, 791-801.	1.9	9
25	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.	0.3	9
26	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.	0.4	8
27	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.	0.7	8
28	Expanding DSD Phenotypes Associated with Variants in the DEAH-Box RNA Helicase DHX37. Sexual Development, 2021, 15, 244-252.	1.1	8
29	Variable Associations of Klinefelter Syndrome in Children. Journal of Pediatric Endocrinology and Metabolism, 2010, 23, 985-9.	0.4	7
30	A novel mutation in the leptin gene (W121X) in an Egyptian family. Molecular Genetics and Metabolism Reports, 2014, 1, 474-476.	0.4	7
31	Novel <i>AMH</i> and <i>AMHR2</i> Mutations in Two Egyptian Families with Persistent MÃ¼llerian Duct Syndrome. Sexual Development, 2017, 11, 29-33.	1.1	7
32	Identification of a novel homozygous ALX4 mutation in two unrelated patients with frontonasal dysplasia typeâ€2. American Journal of Medical Genetics, Part A, 2018, 176, 1190-1194.	0.7	7
33	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, 277-281.	1.1	6
34	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, 235-243.	1.1	5
35	Achieving Diagnostic Certainty in Resource-Limited Settings. Endocrine Development, 2014, 27, 257-267.	1.3	5
36	A Homozygous Missense Variant in Hedgehog Acyltransferase (HHAT) Gene Associated with 46,XY Gonadal Dysgenesis. Sexual Development, 2022, 16, 261-265.	1.1	5

#	Article	IF	Citations
37	<i>WT1</i> 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.	1.1	4
38	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.	1.1	4
39	Chromosome 9p terminal deletion in nine Egyptian patients and narrowing of the critical region for trigonocephaly. Molecular Genetics & Genomic Medicine, 2021, 9, e1829.	0.6	4
40	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, 411-414.	0.8	3
41	Mutational pattern in the 5Î± reductase 2 (SRD5A2) gene in 46,XY Egyptian DSD patients. Middle East Journal of Medical Genetics, 2015, 4, 77-82.	0.0	3
42	Unusual association of simplified gyral pattern and sparse hair in an Egyptian patient with microcephalyâ€“lymphoedema. Clinical Dysmorphology, 2006, 15, 245-247.	0.1	2
43	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.	0.9	1
44	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.	1.5	0
45	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.	0.4	0