FatÄºh GÃœrbÃœz

List of Publications by Year
in descending order

50 papers	1,291 citations	516710 16 h-index	377865 34 g-index
50 all docs	50 docs citations	50 times ranked	2204 citing authors

#	Article	IF	Citations
1	Inactivating NHLH2 variants cause idiopathic hypogonadotropic hypogonadism and obesity in humans. Human Genetics, 2022, 141, 295-304.	3.8	5
2	PLXNB1 mutations in the etiology of idiopathic hypogonadotropic hypogonadism. Journal of Neuroendocrinology, 2022, 34, e13103.	2.6	5
3	Pathogenic Variants in ZSWIM7 Cause Primary Ovarian Insufficiency. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e2359-e2364.	3.6	6
4	The utility of annual growth velocity standard deviation scores and measurements of biochemical parameters in long-term treatment monitoring of children with 21-hydroxylase deficiency. Hormones, 2022, , 1.	1.9	2
5	Loss-of-function variants in SEMA3F and PLXNA3 encoding semaphorin-3F and its receptor plexin-A3 respectively cause idiopathic hypogonadotropic hypogonadism. Genetics in Medicine, 2021, 23, 1008-1016.	2.4	19
6	Experience with the targeted next-generation sequencing in the diagnosis of hereditary hypophosphatemic rickets. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 639-648.	0.9	5
7	Changes in the presentation of newly diagnosed type 1 diabetes in children during the COVID-19 pandemic in a tertiary center in Southern Turkey. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 1303-1309.	0.9	25
8	<i>DLG2</i> Mutations in the Etiology of Pubertal Delay and Idiopathic Hypogonadotropic Hypogonadism. Hormone Research in Paediatrics, 2021, 94, 364-368.	1.8	2
9	Clinical Characteristics of 46,XX Males with Congenital Adrenal Hyperplasia. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2021, 13, 180-186.	0.9	0
10	21-Hydroxylase deficiency: Mutational spectrum and Genotypeâ€“Phenotype relations analyses by next-generation sequencing and multiplex ligation-dependent probe amplification. European Journal of Medical Genetics, 2020, 63, 103782.	1.3	12
11	Neonatal Screening for Congenital Adrenal Hyperplasia in Turkey: Outcomes of Extended Pilot Study in 241,083 Infants. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2020, 12, 287-294.	0.9	12
12	Mutations Within the Transcription Factor <i>PROP1</i> in a Cohort of Turkish Patients with Combined Pituitary Hormone Deficiency. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2020, 12, 261-268.	0.9	4
13	Coexistence of Type 1 Diabetes Mellitus and Periventricular Heterotopia in a Child: A Case Report. Journal of Pediatric Neurosciences, 2020, 15, 67-68.	0.3	0
14	Gender Identity and Assignment Recommendations in Disorders of Sex Development Patients: 20 Yearsâ€™ Experience and Challenges. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2020, 12, 347-357.	0.9	2
15	Gender Identity and Assignment Recommendations in Disorders of Sex Development Patients: 20 Yearsâ€™ Experience and Challenges. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2020, 12, 347-357.	0.9	3
16	Hyperphosphatemic Familial Tumoral Calcinosis in Two Siblings with a Novel Mutation in <i>GALNT3</i> Gene: Experience from Southern Turkey. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 94-99.	0.9	8
17	Prevalence and associated phenotypes of PLXNA1 variants in normosmic and anosmic idiopathic hypogonadotropic hypogonadism. Clinical Genetics, 2019, 95, 320-324.	2.0	29
18	Predicted Benign and Synonymous Variants in CYP11A1 Cause Primary Adrenal Insufficiency Through Missplicing. Journal of the Endocrine Society, 2019, 3, 201-221.	0.2	27

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19	Neonatal Screening for Congenital Adrenal Hyperplasia in Turkey: A Pilot Study with 38,935 Infants. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 13-23.	0.9	18
20	Efficiency of Single Dose of Tolvaptan Treatment During the Triphasic Episode After Surgery for Craniopharyngioma. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 202-206.	0.9	9
21	Molecular genetic studies in a case series of isolated hypoaldosteronism due to biosynthesis defects or aldosterone resistance. Clinical Endocrinology, 2018, 88, 799-805.	2.4	14
22	Prevalence of ZnT8 Antibody in Turkish Children and Adolescents with New Onset Type 1 Diabetes. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2018, 10, 108-112.	0.9	11
23	CCDC141 Mutations in Idiopathic Hypogonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1816-1825.	3.6	33
24	Hypogonadotropic Hypogonadism due to Novel FGFR1 Mutations. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 95-100.	0.9	10
25	Wolcott-Rallison Syndrome with Novel EIF2AK3 Gene Mutation. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2016, 8, 496-497.	0.9	4
26	An emerging, recognizable facial phenotype in association with mutations in GLIâ€similar 3 (GLIS3). American Journal of Medical Genetics, Part A, 2016, 170, 1918-1923.	1.2	16
27	Complete Idiopathic Hypogonadotropic Hypogonadism due to Homozygous <i>GNRH1</i> Mutations in the Mutational Hot Spots in the Region Encoding the Decapeptide. Hormone Research in Paediatrics, 2016, 85, 107-111.	1.8	18
28	Sertoli cell only syndrome with ambiguous genitalia. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 849-52.	0.9	3
29	Anthropometric findings from birth to adulthood and their relation with karyotpye distribution in Turkish girls with Turner syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 942-948.	1.2	7
30	CCDC141 Mutation Identified in Anosmic Hypogonadotropic Hypogonadism (Kallmann Syndrome) Alters GnRH Neuronal Migration. Endocrinology, 2016, 157, 1956-1966.	2.8	47
31	Effects of methylphenidate on appetite and growth in children diagnosed with attention deficit and hyperactivity disorder. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 85-92.	0.9	25
32	Coexistence of Kabuki Syndrome and Autoimmune Thyroiditis. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2016, 8, 105-106.	0.9	6
33	A Novel Homozygous Mutation in the KCNJ11 Gene of a Neonate with Congenital Hyperinsulinism and Successful Management with Sirolimus. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2016, 8, 478-481.	0.9	17
34	Idiopathic Hypogonadotropic Hypogonadism Caused by Inactivating Mutations in SRA1. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2016, 8, 125-134.	0.9	30
35	Crouzonodermoskeletal Syndrome with Hypoplasia of Corpus Callosum and Inferior Vermis. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2016, 8, 373-374.	0.9	2
36	Turner Syndrome and Associated Problems in Turkish Children: A Multicenter Study. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2015, 7, 27-36.	0.9	42

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37	The effect of lifestyle change and metformin therapy on serum arylesterase and paraoxonase activity in obese children. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 551-6.	0.9	6
38	Normosmic idiopathic hypogonadotropic hypogonadism due to a novel homozygous nonsense c.C969A (p.Y323X) mutation in the <scp>KISS</scp>1R gene in three unrelated families. Clinical Endocrinology, 2015, 82, 429-438.	2.4	16
39	Growth curves for Turkish Girls with Turner Syndrome: Results of the Turkish Turner Syndrome Study Group. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2015, 7, 183-191.	0.9	4
40	The Investigation of Plasma Glucagon-like Peptide-1 (glp-1) Levels in Newly Diagnosed Type 1 Diabetic Children. West Indian Medical Journal, 2015, 65, 141-146.	0.4	0
41	The first report of cabergoline-induced immune hemolytic anemia in an adolescent with prolactinoma. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 159-63.	0.9	6
42	MCM9 Mutations Are Associated with Ovarian Failure, Short Stature, and Chromosomal Instability. American Journal of Human Genetics, 2014, 95, 754-762.	6.2	172
43	Etiological Evaluation of Patients Presenting with Isolated Micropenis to an Academic Health Care Center. Indian Journal of Pediatrics, 2014, 81, 775-779.	0.8	4
44	Loss-of-Function Mutations inPNPLA6Encoding Neuropathy Target Esterase Underlie Pubertal Failure and Neurological Deficits in Gordon Holmes Syndrome. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E2067-E2075.	3.6	92
45	Mutations in FEZF1 Cause Kallmann Syndrome. American Journal of Human Genetics, 2014, 95, 326-331.	6.2	69
46	The Novel Mutation p.Trp147Arg of the Steroidogenic Acute Regulatory Protein Causes Classic Lipoid Congenital Adrenal Hyperplasia with Adrenal Insufficiency and 46,XY Disorder of Sex Development. Hormone Research in Paediatrics, 2013, 80, 163-169.	1.8	6
47	Distribution of Gene Mutations Associated with Familial Normosmic Idiopathic Hypogonadotropic Hypogonadism. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2012, 4, 121-126.	0.9	28
48	Unilateral exudative retinal detachment as the sole presentation of relapsing acute lymphoblastic leukemia. Turkish Journal of Haematology, 2012, 29, 181-184.	0.5	9
49	InactivatingKISS1Mutation and Hypogonadotropic Hypogonadism. New England Journal of Medicine, 2012, 366, 629-635.	27.0	394
50	Relationship between metabolic control and neurocognitive functions in children diagnosed with type I diabetes mellitus before and after 5 years of age. Turkish Journal of Pediatrics, 2012, 54, 352-61.	0.6	7