Serdar Ceylaner

List of Publications by Year
in descending order

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175	1,523	471061	476904
papers	citations	17	29
		h-index	g-index

179	179	179	2696
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in SLC29A3, Encoding an Equilibrative Nucleoside Transporter ENT3, Cause a Familial Histiocytosis Syndrome (Faisalabad Histiocytosis) and Familial Rosai-Dorfman Disease. PLoS Genetics, 2010, 6, e1000833.	1.5	174
2	SALL4mutations in Okihiro syndrome (Duane-radial ray syndrome), acro-renal-ocular syndrome, and related disorders. Human Mutation, 2005, 26, 176-183.	1.1	103
3	Extended clinical and immunological phenotype and transplant outcome in CD27 and CD70 deficiency. Blood, 2020, 136, 2638-2655.	0.6	64
4	Excellent response to deep brain stimulation in a young girl with GNAO1-related progressive choreoathetosis. Child's Nervous System, 2016, 32, 1567-1568.	0.6	40
5	Y chromosome azoospermia factor region microdeletions and recurrent pregnancy loss. American Journal of Obstetrics and Gynecology, 2008, 199, 662.e1-662.e5.	0.7	32
6	Empty follicle syndrome in two sisters with three cycles: Case report. Human Reproduction, 2003, 18, 1864-1867.	0.4	31
7	Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. Human Genetics, 2015, 134, 123-126.	1.8	30
8	Reversible pulmonary arterial hypertension in cobalamin-dependent cobalamin C disease due to a novel mutation in the MMACHC gene. European Journal of Pediatrics, 2014, 173, 1707-1710.	1.3	28
9	Molecular diagnosis of maturity-onset diabetes of the young (MODY) in Turkish children by using targeted next-generation sequencing. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1265-71.	0.4	28
10	Maturity onset diabetes of youth (MODY) in Turkish children: sequence analysis of 11 causative genes by next generation sequencing. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 487-96.	0.4	28
11	Genetic abnormalities in Turkish women with premature ovarian failure. International Journal of Gynecology and Obstetrics, 2010, 110, 122-124.	1.0	26
12	Vascular endothelial growth factor +405 C/G polymorphism is highly associated with an increased risk of endometriosis in Turkish women. Archives of Gynecology and Obstetrics, 2011, 283, 267-272.	0.8	26
13	The variable clinical phenotype of three patients with hepatic glycogen synthase deficiency. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 459-462.	0.4	23
14	Two Frameshift Mutations in MKRN3 in Turkish Patients with Familial Central Precocious Puberty. Hormone Research in Paediatrics, 2017, 87, 405-411.	0.8	21
15	Testotoxicosis: Report of Two Cases, One with a Novel Mutation in LHCGR Gene. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2015, 7, 242-248.	0.4	21
16	More than meets the eye: Expanding and reviewing the clinical and mutational spectrum of brittle cornea syndrome. Human Mutation, 2021, 42, 711-730.	1.1	19
17	Extremely skewed X-chromosome inactivation patterns in women with recurrent spontaneous abortion. Australian and New Zealand Journal of Obstetrics and Gynaecology, 2006, 46, 384-387.	0.4	17
18	GJB2 and mitochondrial A1555G gene mutations in nonsyndromic profound hearing loss and carrier frequencies in healthy individuals. Journal of Genetics, 2008, 87, 53-57.	0.4	17

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19	Clinical, immunological features and follow up of 20 patients with dedicator of cytokinesis 8 (DOCK8) deficiency. Pediatric Allergy and Immunology, 2020, 31, 515-527.	1.1	17
20	A Novel Mutation in the Lysyl Hydroxylase 1 Gene Causes Decreased Lysyl Hydroxylase Activity in an Ehlersâ€“Danlos VIA Patient. Journal of Investigative Dermatology, 2005, 124, 914-918.	0.3	16
21	Histopathological analysis of the placental lesions in pregnancies complicated with IUGR and stillbirths in comparison with noncomplicated pregnancies. Journal of the Turkish German Gynecology Association, 2011, 12, 75-79.	0.2	16
22	A Novel Heterozygous Mutation in Steroidogenic Factor-1 in Pubertal Virilization of a 46,XY Female Adolescent. Journal of Pediatric and Adolescent Gynecology, 2014, 27, 98-101.	0.3	16
23	Vici syndrome in siblings born to consanguineous parents. American Journal of Medical Genetics, Part A, 2016, 170, 220-225.	0.7	16
24	Postmortem evaluation of 220 prenatally diagnosed fetuses with neural tube defects: detection of associated anomalies in a Turkish population. Prenatal Diagnosis, 2006, 26, 147-153.	1.1	15
25	A rare case of primary coenzyme Q10 deficiency due to COQ9 mutation. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 165-170.	0.4	15
26	Genetic evaluation of severe male factor infertility in Turkey: A cross-sectional study. Human Fertility, 2012, 15, 100-106.	0.7	13
27	Early-Onset Mild Type Leukoencephalopathy Caused by a Homozygous EARS2 Mutation. Journal of Child Neurology, 2016, 31, 938-941.	0.7	13
28	Lethal neonatal rigidity and multifocal seizure syndrome with a new mutation in BRAT1. Epilepsy & Behavior Case Reports, 2017, 8, 31-32.	1.5	13
29	Coexistence of borderline ovarian epithelial tumor, primary pelvic hydatid cyst, and lymphoepithelioma-like gastric carcinoma. Taiwanese Journal of Obstetrics and Gynecology, 2011, 50, 201-204.	0.5	12
30	Nephron-sparing Surgery for Renal Cell Carcinoma of the Allograft After Renal Transplantation: Report of Two Cases. Transplantation Proceedings, 2013, 45, 958-960.	0.3	12
31	Twenty-seven mutations with three novel pathologenic variants causing biotinidase deficiency: a report of 203 patients from the southeastern part of Turkey. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 339-343.	0.4	12
32	MaFOS-GDM trial: Maternal fish oil supplementation in women with gestational diabetes and cord blood DNA methylation at insulin like growth factor-1 (IGF-1) gene. Clinical Nutrition ESPEN, 2018, 23, 73-78.	0.5	12
33	Monogenic Diabetes Not Caused By Mutations in Mody Genes: A Very Heterogenous Group of Diabetes. Experimental and Clinical Endocrinology and Diabetes, 2018, 126, 612-618.	0.6	12
34	A Novel Homozygous <i>CYP19A1</i> Gene Mutation: Aromatase Deficiency Mimicking Congenital Adrenal Hyperplasia in an Infant without Obvious Maternal Virilisation. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 196-201.	0.4	12
35	Evaluation of 2407 fetuses in a Turkish population. Prenatal Diagnosis, 2007, 27, 800-807.	1.1	11
36	Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. American Journal of Human Genetics, 2021, 108, 1301-1317.	2.6	11

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37	Three new cases of disorganizationlike syndrome: One with accessory extrophia vesicalis. Journal of Pediatric Surgery, 2004, 39, e6-e8.	0.8	10
38	Clinical, Histochemical, and Molecular Study of Three Turkish Siblings Diagnosed with H Syndrome, and Literature Review. Hormone Research in Paediatrics, 2019, 91, 346-355.	0.8	10
39	Genotypes and estimated prevalence of phosphomannomutase 2 deficiency in Turkey differ significantly from those in Europe. American Journal of Medical Genetics, Part A, 2020, 182, 705-712.	0.7	10
40	A case of fetal anticonvulsant syndrome with severe bilateral upper limb defect. Journal of Maternal-Fetal and Neonatal Medicine, 2006, 19, 115-117.	0.7	9
41	Sporadic Nonautoimmune Neonatal Hyperthyroidism Due to A623V Germline Mutation in the Thyrotropin Receptor Gene-Case Report. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2010, 2, 168-172.	0.4	9
42	Chronic lymphocytic leukemia in a child: A challenging diagnosis in pediatric oncology practice. Pediatric Blood and Cancer, 2014, 61, 933-935.	0.8	9
43	Cytokine Concentrations In Pediatric Patients With Crimean-Congo Hemorrhagic Fever. Pediatric Infectious Disease Journal, 2014, 33, 1185-1187.	1.1	9
44	A novel missense mutation in HSD17B3 gene in a 46, XY adolescent presenting with primary amenorrhea and virilization at puberty. Clinica Chimica Acta, 2015, 438, 154-156.	0.5	9
45	Novel mutation in MASP1 gene in a new family with 3MC syndrome. Clinical Dysmorphology, 2019, 28, 91-93.	0.1	9
46	Early-onset progressive encephalopathy associated with NAXE gene variants: a case report of a Turkish child. Acta Neurologica Belgica, 2020, 120, 733-735.	0.5	9
47	Congenital myasthenic syndrome in Turkey: clinical and genetic features in the long-term follow-up of patients. Acta Neurologica Belgica, 2021, 121, 529-534.	0.5	9
48	Novel CRLF1 gene mutation in a newborn infant diagnosed with Crisponi syndrome. Congenital Anomalies (discontinued), 2012, 52, 216-218.	0.3	8
49	A novel frameshift mutation of malonylâ€CoA decarboxylase deficiency: clinical signs and therapy response of a lateâ€diagnosed case. Clinical Case Reports (discontinued), 2017, 5, 1284-1288.	0.2	8
50	Pyruvate dehydrogenase-E1Î± deficiency presenting as recurrent acute proximal muscle weakness of upper and lower extremities in an 8-year-old boy. Neuromuscular Disorders, 2017, 27, 94-97.	0.3	8
51	Nonketotic hyperglycinemia: Clinical range and outcome of a rare neurometabolic disease in a single-center. Brain and Development, 2018, 40, 865-875.	0.6	8
52	Identification of a New de Novo Mutation Underlying Regressive Episodic Ataxia Type I. Frontiers in Neurology, 2018, 9, 587.	1.1	8
53	Clinical findings in five Turkish patients with citrin deficiency and identification of a novel mutation on SLC25A13. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 157-163.	0.4	8
54	Prenatal and Postnatal Findings in a Case with the Autosomal Recessive Type of Robinow Syndrome. Fetal Diagnosis and Therapy, 2006, 21, 386-389.	0.6	7

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55	Mitochondrial Membrane Proteinâ€“Associated Neurodegeneration. Pediatric Neurology, 2015, 53, 373-374.	1.0	7
56	Brown-Vialetto-Van Laere syndrome: two siblings with a new mutation and dramatic therapeutic effect of high-dose riboflavin. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 227-31.	0.4	7
57	Early-onset severe obesity due to complete deletion of the leptin gene in a boy. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 1227-1230.	0.4	7
58	Association of vitamin D receptor gene polymorphisms with osteosarcoma risk and prognosis. Journal of Bone Oncology, 2019, 14, 100208.	1.0	7
59	Episodic psychosis, ataxia, motor neuropathy with pyramidal signs (PAMP syndrome)Âcaused by a novel mutation in ADPRHL2 (AHR3). Neurological Sciences, 2021, 42, 3871-3878.	0.9	7
60	Clinical Features and Outcomes of 23 Patients with Wiskott- Aldrich Syndrome: A Single-Center Experience. Turkish Journal of Haematology, 2020, 37, 271-281.	0.2	7
61	Neu-Laxova syndrome, grossly appearing normal on 20Âweeks ultrasonographic scan, that manifested late in pregnancy: a case report. Archives of Gynecology and Obstetrics, 2007, 276, 367-370.	0.8	6
62	A boy with trisomy 13 presenting with a subtle clinical picture and metopic synostosis. American Journal of Medical Genetics, Part A, 2009, 149A, 1608-1609.	0.7	6
63	Recurrent proximal 18p monosomy and 18q trisomy in a family due to a pericentric inversion. American Journal of Medical Genetics, Part A, 2014, 164, 1239-1244.	0.7	6
64	Giant axonal disease: Report of eight cases. Brain and Development, 2015, 37, 803-807.	0.6	6
65	A rare cause of fatal pulmonary alveolar proteinosis: Niemann-Pick disease type C2 and a novel mutation. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1163-7.	0.4	6
66	Improved metabolic control in tetrahydrobiopterin (BH4), responsive phenylketonuria with sapropterin administered in two divided doses vs. a single daily dose. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 713-718.	0.4	6
67	Four Gaucher disease type II patients with three novel mutations: a single centre experience from Turkey. Metabolic Brain Disease, 2018, 33, 1223-1227.	1.4	6
68	The Second Case of Saposin A Deficiency and Altered Autophagy. JIMD Reports, 2018, 44, 43-54.	0.7	6
69	A rare case of prolidase deficiency with situs inversus totalis, identified by a novel mutation in the PEPD gene. JAAD Case Reports, 2019, 5, 436-438.	0.4	6
70	A Very Rare Etiology of Hypotonia and Seizures: Congenital Glutamine Synthetase Deficiency. Neuropediatrics, 2019, 50, 051-053.	0.3	6
71	Cytochrome P450 oxidoreductase deficiency caused by a novel mutation in the POR gene in two siblings: case report and literature review. Hormones, 2021, 20, 293-298.	0.9	6
72	Merosin-negative congenital muscular dystrophy: Report of five cases. Journal of Pediatric Neurosciences, 2015, 10, 346.	0.2	6

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73	The Prevalence of Fabry Disease Among Turkish Patients with Non-obstructive Hypertrophic Cardiomyopathy: Insights from a Screening Study. Balkan Medical Journal, 2019, 36, 354-358.	0.3	6
74	Coexistence of Kabuki Syndrome and Autoimmune Thyroiditis. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2016, 8, 105-106.	0.4	6
75	A Mutation in INSR in a Child Presenting with Severe Acanthosis Nigricans. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 371-374.	0.4	6
76	Clinical features of 27 turkish propionic acidemia patients with 12 novel mutations. Turkish Journal of Pediatrics, 2019, 61, 330.	0.3	6
77	Hypomelanosis of Ito and Sturge-Weber Syndrome Without Facial Nevus: An Association or a New Syndrome?. Pediatric Neurology, 2009, 40, 395-397.	1.0	5
78	Lumbocostovertebral syndrome in an infant of a diabetic mother. American Journal of Medical Genetics, Part A, 2010, 152A, 1374-1377.	0.7	5
79	Primary Adrenal Insufficiency Caused by a Novel Mutation in DAX1 Gene. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2013, 5, 55-57.	0.4	5
80	Two Novel Missense Mutations in Nonketotic Hyperglycinemia. Journal of Child Neurology, 2015, 30, 789-792.	0.7	5
81	Primary systemic carnitine deficiency: a Turkish case with a novel homozygous SLC22A5 mutation and 14 years follow-up. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1179-81.	0.4	5
82	Cystinosis in Eastern Turkey. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 965-969.	0.4	5
83	Congenital Glucoseâ€“Galactose Malabsorption in a Turkish Newborn: A Novel Mutation of Na+/Glucose Cotransporter Gene. Digestive Diseases and Sciences, 2017, 62, 280-281.	1.1	5
84	A novel homozygous HOXB1 mutation in a Turkish family with hereditary congenital facial paresis. Brain and Development, 2017, 39, 166-170.	0.6	5
85	Delayed Diagnosis of a 17-Hydroxylase/17,20-Lyase Deficient Patient Presenting as a 46,XY Female: A Low Normal Potassium Level Can Be an Alerting Diagnostic Sign. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 163-167.	0.4	5
86	Mild hypotonia and recurrent seizures in an 8-month-old boy: Answers. Pediatric Nephrology, 2019, 34, 1729-1731.	0.9	5
87	A rare cause of delayed puberty in two cases with 46,XX and 46,XY karyotype: 17 Î±-hydroxylase deficiency due to a novel variant in CYP17A1 gene. Gynecological Endocrinology, 2020, 36, 739-742.	0.7	5
88	Aceruloplasminemia Presenting with Asymmetric Chorea Due to a Novel Frameshift Mutation. Movement Disorders Clinical Practice, 2020, 7, S67-S70.	0.8	5
89	LRBA deficiency: a rare cause of type 1 diabetes, colitis, and severe immunodeficiency. Hormones, 2021, 20, 389-394.	0.9	5
90	Camptodactyly-Arthropathy-Coxa Vara-Pericarditis Syndrome Resembling Juvenile Idiopathic Arthritis: A Single-Center Experience from Southern Turkey. Molecular Syndromology, 2021, 12, 112-117.	0.3	5

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91	Treatment Difficulties in Hypomagnesemia Secondary to the Transient Receptor Potential Melastatin 6 Gene: A Case Report with Novel Mutation. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2021, 13, 114-118.	0.4	5
92	Genotypic and phenotypic features in Turkish patients with classic nonketotic hyperglycinemia. Metabolic Brain Disease, 2021, 36, 1213-1222.	1.4	5
93	A Case of Vitamin D-Dependent Rickets Type 1A with a Novel Mutation in the Uzbek Population. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2016, 8, 484-489.	0.4	5
94	Clinical and Genetic Characteristics of Patients with Corticosterone Methyloxidase Deficiency Type 2: Novel Mutations in <i>CYP11B2</i>. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2021, 13, 232-238.	0.4	5
95	A novel mutation in the desmoplakin gene in two female siblings with a rare form of dilated cardiomyopathy: Carvajal syndrome. Anatolian Journal of Cardiology, 2017, 18, 435-436.	0.5	5
96	Ectopic Posterior Pituitary, Polydactyly, Midfacial Hypoplasia and Multiple Pituitary Hormone Deficiency due to a Novel Heterozygous IVS11-2A>C(c.1957-2A>C) Mutation in the <i>GLI2</i> Gene. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2020, 12, 319-328.	0.4	5
97	An uncommon complementary isochromosome of 46,XY, i(9)(p10),i(9)(q10) in an infertile oligoasthenoteratozoospermic man. Fertility and Sterility, 2011, 95, 290.e5-290.e8.	0.5	4
98	Are low maternal estriol levels a predictor for proâ€opiomelanocortin (POMC) deficiency caused by POMC mutation during pregnancy?. Prenatal Diagnosis, 2013, 33, 1297-1298.	1.1	4
99	Three cases of Wolfram syndrome with different clinical aspects. Journal of Pediatric Endocrinology and Metabolism, 2014, 28, 433-8.	0.4	4
100	A Rare Cause of Elevated Chitotriosidase Activity: Glycogen Storage Disease Type IV. JIMD Reports, 2014, 17, 63-66.	0.7	4
101	A case of Riley Ruvalcaba syndrome with a novel PTEN mutation accompanied by diffuse testicular microlithiasis and precocious puberty. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 95-99.	0.4	4
102	A Rare Cause of Adrenal Insufficiency â€“ Isolated ACTH Deficiency Due to TBX19 Mutation: Long-Term Follow-Up of Two Cases and Review of the Literature. Hormone Research in Paediatrics, 2019, 92, 395-403.	0.8	4
103	A novel mutation in the GP1BA gene in Bernardâ€“Soulier syndrome. Blood Coagulation and Fibrinolysis, 2020, 31, 83-86.	0.5	4
104	Molecular and clinical findings of Turkish patients with hereditary fructose intolerance. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 1017-1022.	0.4	4
105	Isolated congenital diaphragm hernia associated with homozygous SLIT3 gene variant in dizygous twins. European Journal of Medical Genetics, 2021, 64, 104215.	0.7	4
106	An infant with glutaric aciduria type IIc diagnosed with a novel mutation. Turkish Journal of Pediatrics, 2017, 59, 315-317.	0.3	4
107	Microcephaly and developmental delay caused by short-chain acyl-coa dehydrogenase deficiency. Turkish Journal of Pediatrics, 2017, 59, 708.	0.3	4
108	Neonatal form of biotin-thiamine-responsive basal ganglia disease. clues to diagnosis. Turkish Journal of Pediatrics, 2019, 61, 261.	0.3	4

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109	Prenatal diagnosis of a Turkish Bartsocasâ€“Papas syndrome case with upper limb pterigia. Prenatal Diagnosis, 2007, 27, 563-565.	1.1	3
110	Carmi syndrome with congenital heart defects. American Journal of Medical Genetics, Part A, 2010, 152A, 2120-2122.	0.7	3
111	High frequency of p.Thr93Met in Smithâ€Lemliâ€Opitz syndrome patients in Turkey. Clinical Genetics, 2012, 81, 598-601.	1.0	3
112	Stimulus-induced myoclonus treated effectively with clonazepam in genetically confirmed Coffinâ€“Lowry syndrome. Epilepsy & Behavior Case Reports, 2014, 2, 196-198.	1.5	3
113	Sertoli cell only syndrome with ambiguous genitalia. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 849-52.	0.4	3
114	Unexplained cyanosis caused by hepatopulmonary syndrome in a girl with APECED syndrome. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 365-369.	0.4	3
115	A rare mutation in the EPG5 gene causes Vici syndrome. Clinical Dysmorphology, 2018, 27, 145-147.	0.1	3
116	Cardiac Tamponade in Gorham-Stout Syndrome Associated with GATA2 Mutation. Indian Journal of Pediatrics, 2020, 87, 239-240.	0.3	3
117	Brown vialetto van laere syndrome: presenting with left ventricular non-compaction and mimicking mitochondrial disorders. Turkish Journal of Pediatrics, 2021, 63, 314.	0.3	3
118	Novel mutations in TRPM6 gene associated with primary hypomagnesemia with secondary hypocalcemia. Case report. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2021, 165, 454-457.	0.2	3
119	Importance of pedigree in patients with familial epilepsy and intellectual disability. Sudanese Journal of Paediatrics, 2019, 19, 52-56.	0.6	3
120	The earlier described mutation (c.307C>T [p.R103X]) in the SRD5A2 gene causing a 46,XY female phenotype. Journal of Pediatric Endocrinology and Metabolism, 2012, 25, 543-5.	0.4	2
121	Rodriguez lethal acrofacial dysostosis syndrome with ambiguous genitalia. Taiwanese Journal of Obstetrics and Gynecology, 2016, 55, 613-615.	0.5	2
122	Recurrent hepatic failure and status epilepticus: an uncommon presentation of hyperargininemia. Metabolic Brain Disease, 2018, 33, 1775-1778.	1.4	2
123	A Case of Shwachmanâ€“Diamond Syndrome who Presented with Hypotonia. Journal of Pediatric Genetics, 2018, 07, 117-121.	0.3	2
124	Mild hypotonia and recurrent seizures in an 8-month-old boy: Questions. Pediatric Nephrology, 2019, 34, 1727-1728.	0.9	2
125	Hypokalemia and hearing loss in a 3-year-old boy: Answers. Pediatric Nephrology, 2020, 35, 617-618.	0.9	2
126	Genetic Management Algorithm in High-Risk Fabry Disease Cases; Especially in Female Indexes with Mutations. Endocrine, Metabolic and Immune Disorders - Drug Targets, 2021, 21, 324-337.	0.6	2

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127	Horizontal Gaze Palsy with Progressive Scoliosis in an Infant Diagnosed Before Developing Scoliosis: MRI and DTI Findings. Klinische Padiatrie, 2022, 234, 52-55.	0.2	2
128	P.val452ile mutation of the slc25a13 gene in a turkish patient with citrin deficiency. Turkish Journal of Pediatrics, 2017, 59, 311.	0.3	2
129	Ataxia, tremor, intellectual disability: a case of stxbp1 encephalopathy with a new mutation. Turkish Journal of Pediatrics, 2019, 61, 757.	0.3	2
130	Mitochondrial membrane protein-associated neurodegeneration in a Turkish patient. Journal of Pediatric Neurosciences, 2016, 11, 288.	0.2	2
131	Crouzonodermoskeletal Syndrome with Hypoplasia of Corpus Callosum and Inferior Vermis. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2016, 8, 373-374.	0.4	2
132	Niemannâ€“Pick type C disease with a novel intronic mutation: three Turkish cases from the same family. Journal of Pediatric Endocrinology and Metabolism, 2021, .	0.4	2
133	A rare cause of hydrops fetalis in two Gaucher disease type 2 patients with a novel mutation. Metabolic Brain Disease, 2022, , .	1.4	2
134	Craniosynostosis and ectopia lentis in a propositus whose parents are cousins. , 2005, 134A, 231-231.		1
135	Chromosomal heteromorphisms may help for the diagnosis of uniparental disomy (UPD): a case report. Prenatal Diagnosis, 2007, 27, 1072-1074.	1.1	1
136	A child with L-2 hydroxyglutaric aciduria presenting with dilated cardiomyopathy: Coincidence or a new syndrome?. Anatolian Journal of Cardiology, 2013, 14, 92-3.	0.4	1
137	Combination of two different homozygote mutations in Pompe disease. Pediatrics International, 2016, 58, 241-243.	0.2	1
138	A novel genetic mutation in a Turkish family with GCK-MODY. International Journal of Diabetes in Developing Countries, 2017, 37, 323-326.	0.3	1
139	Analysis of TP53 gene in uterine myomas: No mutations but P72R polymorphism is associated with myoma development. Journal of Obstetrics and Gynaecology Research, 2019, 45, 2088-2094.	0.6	1
140	Prenatal Diagnosis of Osteogenesis Imperfecta Type III. Journal of Obstetrics and Gynecology of India, 2019, 69, 374-376.	0.3	1
141	Hypokalemia and hearing loss in a 3-year-old boy: Questions. Pediatric Nephrology, 2020, 35, 615-615.	0.9	1
142	Mild lamellar ichthyosis with a truncated homozygous TGM1 mutation in a pediatric patient from Turkey. Dermatologic Therapy, 2020, 33, e14152.	0.8	1
143	A newborn case of adenylosuccinate lyase deficiency with a novel heterozygous mutation diagnosed by whole exome sequencing. Clinical Neurology and Neurosurgery, 2021, 202, 106506.	0.6	1
144	A Novel mRNA Modification Mutation in a Patient With Ligneous Conjunctivitis Coexisting With Heterozygous Familial Mediterranean Fever Mutation. Cornea, 2021, 40, 764-768.	0.9	1

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145	Glycogen storage disease type XII; an ultra rare cause of hemolytic anemia and rhabdomyolysis: one new case report. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 1335-1339.	0.4	1
146	Retrospective evaluation of patients with X-linked adrenoleukodystrophy with a wide range of clinical presentations: a single center experience. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 1169-1179.	0.4	1
147	Clinical and molecular findings in 37 Turkish patients with isolated methylmalonic acidemia. Turkish Journal of Medical Sciences, 2021, 51, 1220-1228.	0.4	1
148	Further observation of Hemoglobin Fontainebleau (a21(B2) Ala-Pro) in a Turkish family. Egyptian Journal of Medical Human Genetics, 2020, 21, .	0.5	1
149	First observation of hemoglobin G-Norfolk in the Turkish population. Turkish Journal of Biochemistry, 2021, 46, 97-102.	0.3	1
150	A rare structural myopathy: nemaline myopathy. Turk Pediatri Arsivi, 2019, 54, 49-52.	0.9	1
151	Evaluation of MSX1 gene as the common candidate gene of nonsyndromic congenital hypodontia and cleft lip and palate. Journal of Cleft Lip Palate and Craniofacial Anomalies, 2017, 4, 31.	0.1	1
152	Is Bioavailable Vitamin D Better Than Total Vitamin D to Evaluate Vitamin D Status in Obese Children?. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2021, 13, 391-399.	0.4	1
153	X-linked adrenoleukodystrophy in a 6-year-old boy initially presenting with psychiatric symptoms. Turkish Journal of Pediatrics, 2014, 56, 651-3.	0.3	1
154	Turkish cases of early infantile epileptic encephalopathy: two novel mutations in the cyclin-dependent kinase-like 5 (CDKL5) gene. Turkish Journal of Pediatrics, 2015, 57, 272-6.	0.3	1
155	Glucose-6-phosphate dehydrogenase gene Ala365Thr mutation in an Iraqi family with confusing clinical differences. Biyokimya Dergisi, 2021, 46, 729-731.	0.1	1
156	Correspondence: Is it a Proteus syndrome?. Journal of Pediatric Orthopaedics Part B, 2007, 16, 385.	0.3	0
157	A new variant of a known mutation in two siblings with permanent neonatal diabetes mellitus. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 373-5.	0.4	0
158	Hydranencephaly, pituitary hypoplasia, and anophthalmia in a male infant. Clinical Dysmorphology, 2012, 21, 155-156.	0.1	0
159	A child with XYY karyotype and epilepsy. Journal of Pediatric Neurology, 2015, 09, 255-258.	0.0	0
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