

## Prof. SERDAR CEYLANER

### Personal Information

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### International Researcher IDs

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### Education Information

Expertise In Medicine, Erciyes University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Turkey 1993 - 1996

### Academic and Administrative Experience

University Of Higher Specialization, 2015 - 2015

University Of Higher Specialization, 2015 - 2015

### Advising Theses

CEYLANER S., Sendromik olmayan mandibular retrognati olgularında igf-1 gen mutasyonu ve polimorfizmlerinin kodlandığı bölgelerin ve intron-ekzon birleşim bölgelerinin dizi analizi ile araştırılması, Doctorate, D.ÖZTÜRK(Student), 2017

### Published journal articles indexed by SCI, SSCI, and AHCI

- I. **Phenotypic and molecular characterization of five patients with PIK3CA-related overgrowth spectrum (PROS)**  
Gökpinar İli E., Taşdelen E., Durmaz C. D., ALTINER Ş., TUNCALI T., Martinez-Glez V., KARABULUT H. G., Vural S., CEYLANER S., ACAR M. O., et al.  
American Journal of Medical Genetics, Part A, vol.188, no.6, pp.1792-1800, 2022 (SCI-Expanded)
- II. **A rare cause of hydrops fetalis in two Gaucher disease type 2 patients with a novel mutation**  
Kilavuz S., Basaranoglu M., Epcacan S., Bako D., Ozer A., Donmez Y. N., Ceylan E. I., Tukun A., Ceylaner S., Geylani H., et al.  
METABOLIC BRAIN DISEASE, vol.37, no.4, pp.1283-1287, 2022 (SCI-Expanded)
- III. **Niemann-Pick type C disease with a novel intronic mutation: Three Turkish cases from the same family**  
Killıç Yıldırım G., YARAR C., Şeker Yılmaz B., CEYLANER S.  
Journal of Pediatric Endocrinology and Metabolism, vol.35, pp.535-541, 2022 (SCI-Expanded)
- IV. **Successful therapeutic plasma exchange in a case with extremely severe hypertriglyceridemia secondary to diabetic ketoacidosis concomitant with type IX glycogen storage disease**  
KİŞİOĞLU M., YEŞİLBAŞ O., GÜVEN B., CEYLANER S., KARAGÜZEL G.

Transfusion and Apheresis Science, vol.61, no.1, 2022 (SCI-Expanded)

- V. **First Case of MELAS Syndrome Presenting with Local Brain Edema Requiring Decompressive Craniectomy**  
YEŞİLBAŞ O., Sengenc E., Olbak M. E., Bako D., Nizam O. G., SEYİTHANOĞLU M. H., Pehlivan D., CEYLANER S., Icagasioglu D., Aydin K.  
Turkish Neurosurgery, vol.32, no.1, pp.155-159, 2022 (SCI-Expanded)
- VI. **Glucose-6-phosphate dehydrogenase gene Ala365Thr mutation in an Iraqi family with confusing clinical differences**  
Akisin Y. A., Arslan Z., CEYLANER S., Akar N.  
Turkish Journal of Biochemistry, vol.46, no.6, pp.729-731, 2021 (SCI-Expanded)
- VII. **Rare slow channel congenital myasthenic syndromes without repetitive compound muscle action potential and dramatic response to low dose fluoxetine**  
Durmus H., Sticht H., CEYLANER S., Hashemolhosseini S., Deymeer F.  
Acta Neurologica Belgica, vol.121, no.6, pp.1755-1760, 2021 (SCI-Expanded)
- VIII. **Glycogen storage disease type XII; An ultra rare cause of hemolytic anemia and rhabdomyolysis: One new case report**  
Kara E., KOR D., Bulut F. D., Hergüner Ö., CEYLANER S., Köşeci B., BURGAÇ E., Mungan N. Ö.  
Journal of Pediatric Endocrinology and Metabolism, vol.34, no.10, pp.1335-1339, 2021 (SCI-Expanded)
- IX. **Episodic psychosis, ataxia, motor neuropathy with pyramidal signs (PAMP syndrome) caused by a novel mutation in ADPRHL2 (AHR3)**  
Durmus H., Mertoğlu E., Sticht H., CEYLANER S., Kulaksızoğlu I. B., Hashemolhosseini S., Uçar E. Ö., Parman Y.  
Neurological Sciences, vol.42, no.9, pp.3871-3878, 2021 (SCI-Expanded)
- X. **Retrospective evaluation of patients with X-linked adrenoleukodystrophy with a wide range of clinical presentations: A single center experience**  
Olgac A., Kasapkara Ç. S., Derinkuyu B., Yüksel D., Çetinkaya S., Aksoy A., CEYLANER S., Güleray N., Yeşilipek A., Aydin H. I., et al.  
Journal of Pediatric Endocrinology and Metabolism, vol.34, no.9, pp.1169-1179, 2021 (SCI-Expanded)
- XI. **Molecular and clinical findings of Turkish patients with hereditary fructose intolerance**  
Gunduz M., Ünal-Uzun Ö., Koç N., CEYLANER S., Özaydin E., Kasapkara Ç. S.  
Journal of Pediatric Endocrinology and Metabolism, vol.34, no.8, pp.1017-1022, 2021 (SCI-Expanded)
- XII. **Genotypic and phenotypic features in Turkish patients with classic nonketotic hyperglycinemia**  
Bayrak H., YILDIZ Y., Olgaç A., Kasapkara Ç. S., Küçükcongür A., Zenciroğlu A., Yüksel D., CEYLANER S., Kılıç M.  
Metabolic Brain Disease, vol.36, no.6, pp.1213-1222, 2021 (SCI-Expanded)
- XIII. **Isolated congenital diaphragm hernia associated with homozygous SLIT3 gene variant in dizygous twins**  
Kaya T. B., AYDEMİR Ö., CEYLANER S., Ceylaner G., TEKİN A. N.  
European Journal of Medical Genetics, vol.64, no.7, 2021 (SCI-Expanded)
- XIV. **Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy**  
Wong H. H., Seet S. H., Maier M., Gurel A., Traspas R. M., Lee C., Zhang S., Talim B., Loh A. Y., Chia C. Y., et al.  
American Journal of Human Genetics, vol.108, no.7, pp.1301-1317, 2021 (SCI-Expanded)
- XV. **Cytochrome P450 oxidoreductase deficiency caused by a novel mutation in the POR gene in two siblings: case report and literature review**  
Unal E., Demiral M., Yıldırım R., Taş F. F., CEYLANER S., Özbek M. N.  
Hormones, vol.20, no.2, pp.293-298, 2021 (SCI-Expanded)
- XVI. **Antioxidant Therapy in a Patient with Hyperprolinemia Type 1 Presenting with Mild Neuromotor Retardation and Speech Disturbance**  
Ersoy M., Yılmaz S., CEYLANER S.  
Indian Journal of Pediatrics, vol.88, no.6, pp.601, 2021 (SCI-Expanded)
- XVII. **A Novel mRNA Modification Mutation in a Patient With Ligneous Conjunctivitis Coexisting With Heterozygous Familial Mediterranean Fever Mutation**

- Koseoglu N. D., CEYLANER S., YILDIRIM N.  
Cornea, vol.40, no.6, pp.764-768, 2021 (SCI-Expanded)
- XVIII. LRBA deficiency: a rare cause of type 1 diabetes, colitis, and severe immunodeficiency**  
Kardelen A. D., Kara M., Güller D., KARAKILIÇ ÖZTURAN E., Abalı Z. Y., CEYLANER S., KIYKIM A., Cantez S., HANÇERLİ TÖRÜN S., Poyrazoglu S., et al.  
Hormones, vol.20, no.2, pp.389-394, 2021 (SCI-Expanded)
- XIX. More than meets the eye: Expanding and reviewing the clinical and mutational spectrum of brittle cornea syndrome**  
Dhooge T., Van Damme T., Syx D., Mosquera L. M., Nampoothiri S., Radhakrishnan A., Simsek-Kiper P. O., Utine G. E., Bonduelle M., Migeotte I., et al.  
Human Mutation, vol.42, no.6, pp.711-730, 2021 (SCI-Expanded)
- XX. Congenital myasthenic syndrome in Turkey: clinical and genetic features in the long-term follow-up of patients**  
GÜL MERT G., ÖZCAN N., Hergüner Ö., ALTUNBAŞAK Ş., İNCECİK F., BİŞGİN A., CEYLANER S.  
Acta Neurologica Belgica, vol.121, no.2, pp.529-534, 2021 (SCI-Expanded)
- XXI. Camptodactyly-Arthropathy-Coxa Vara-Pericarditis Syndrome Resembling Juvenile Idiopathic Arthritis: A Single-Center Experience from Southern Turkey**  
Ekinci R. M. K., Balci S., Dogan H., CEYLANER S., Varan C., Erdem S., Coban F., BİŞGİN A.  
Molecular Syndromology, vol.12, no.2, pp.112-117, 2021 (SCI-Expanded)
- XXII. A newborn case of adenylosuccinate lyase deficiency with a novel heterozygous mutation diagnosed by whole exome sequencing**  
Cakmak Celik F., Ozlu M. M., CEYLANER S.  
Clinical Neurology and Neurosurgery, vol.202, 2021 (SCI-Expanded)
- XXIII. Is Bioavailable Vitamin D Better Than Total Vitamin D to Evaluate Vitamin D Status in Obese Children?**  
Karacan Küçükali G., GÜLBAHAR Ö., Özalkak Ş., Dağlı H., CEYLANER S., AYCAN Z., Savaş Erdeve Ş.  
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.13, no.4, pp.391-399, 2021 (SCI-Expanded)
- XXIV. An Atypical Presentation of Mevalonate Kinase Deficiency in Response to Colchicine Treatment**  
Koç Yekedüz M., DOĞULU N., Öncül Ü., KÖSE E., CEYLANER S., EMİNOĞLU F. T.  
Molecular Syndromology, 2021 (SCI-Expanded)
- XXV. Novel mutations in trpm6 gene associated with primary hypomagnesemia with secondary hypocalcemia. Case report**  
Papez J., Starha J., Slaba K., Hubacek J. A., Pecl J., Aulicka S., Urik M., CEYLANER S., Vesela P., Slaby O., et al.  
Biomedical Papers, vol.165, no.4, pp.454-457, 2021 (SCI-Expanded)
- XXVI. Clinical and genetic characteristics of patients with corticosterone methyl oxidase deficiency type 2: Novel mutations in cyp11b2**  
Turan H., Çakır A. D., Özer Y., Tarçın G., Özcabi B., CEYLANER S., ERCAN O., EVLİYAĞLU S. O.  
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.13, no.2, pp.232-238, 2021 (SCI-Expanded)
- XXVII. Clinical and molecular findings in 37 Turkish patients with isolated methylmalonic acidemia**  
Şeker Yılmaz B., Kor D., Bulut F. D., Kilavuz S., CEYLANER S., Önenli Mungan H. N.  
Turkish Journal of Medical Sciences, vol.51, no.3, pp.1220-1228, 2021 (SCI-Expanded)
- XXVIII. Brown Vialetto Van Laere syndrome: Presenting with left ventricular non-compaction and mimicking mitochondrial disorders**  
Yılmaz B. Ş., CEYLANER S., Mungan N. Ö.  
Turkish Journal of Pediatrics, vol.63, no.2, pp.314-318, 2021 (SCI-Expanded)
- XXIX. Treatment difficulties in hypomagnesemia secondary to the transient receptor potential melastatin 6 gene: A case report with novel mutation**  
Yücel H., Sel Ç. G., Kasapkar Ç. S., Küçükali G. K., Savas-Erdeve S., Öztoprak Ü., CEYLANER S., Şenel S., Akçaboy M.  
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.13, no.1, pp.114-118, 2021 (SCI-Expanded)
- XXX. Genetic management algorithm in high-risk fabry disease cases; especially in female indexes with mutations**

Sezer O., CEYLANER S.

Endocrine, Metabolic and Immune Disorders - Drug Targets, vol.21, no.2, pp.324-337, 2021 (SCI-Expanded)

- XXXI. **Extended clinical and immunological phenotype and transplant outcome in CD27 and CD70 deficiency**  
Ghosh S., Bal S. K., Edwards E. S., Pillay B., Heredia R. J., Cipe F. E., Rao G., Salzer E., Zoghi S., Abolhassani H., et al.  
Blood, vol.136, no.23, pp.2638-2655, 2020 (SCI-Expanded)
- XXXII. **Coinheritance of novel mutations in NAGLU causing mucopolysaccharidosis type IIIB and in DDHD2 causing spastic paraplegia54 in a Turkish family**  
Gun Bilgic D., Gerik Celebi H. B., Aydin Gumus A., Bilgic A., YAZICI H., CEYLANER S., Yilmaz C., Polat M., Akbal Sahin M., Dereli F., et al.  
Journal of Clinical Neuroscience, vol.82, pp.214-218, 2020 (SCI-Expanded)
- XXXIII. **Mild lamellar ichthyosis with a truncated homozygous TGM1 mutation in a pediatric patient from Turkey**  
İmren I. G., Tanacan E., CEYLANER S., Sumer G., Eksioğlu M.  
Dermatologic Therapy, vol.33, no.6, 2020 (SCI-Expanded)
- XXXIV. **Aceruloplasminemia Presenting with Asymmetric Chorea Due to a Novel Frameshift Mutation**  
Aydemir S. T., BULUT O., CEYLANER S., AKBOSTANCI M. C.  
Movement Disorders Clinical Practice, vol.7, no.3, 2020 (SCI-Expanded)
- XXXV. **A rare cause of delayed puberty in two cases with 46,XX and 46,XY karyotype: 17  $\alpha$ -hydroxylase deficiency due to a novel variant in CYP17A1 gene**  
Unal E., Yıldırım R., Taş F. F., Tekin S., CEYLANER S., Haspolat Y. K.  
Gynecological Endocrinology, vol.36, no.8, pp.739-742, 2020 (SCI-Expanded)
- XXXVI. **A novel mutation which causes a frameshift in the PHOX2B gene causes Haddad syndrome**  
Guzoglu N., Aslan M. K., Gunay Y. D., Atasoy P., CEYLANER S., Aliefendioglu D.  
Clinical Dysmorphology, vol.29, pp.152-154, 2020 (SCI-Expanded)
- XXXVII. **Clinical, immunological features and follow up of 20 patients with dedicator of cytokinesis 8 (DOCK8) deficiency**  
Haskoğlu S., Kostel Bal S., Islamoğlu C., Aytekin C., Guner S., Sevinc S., Keles S., KENDİRLİ T., CEYLANER S., DOĞU E. F., et al.  
Pediatric Allergy and Immunology, vol.31, no.5, pp.515-527, 2020 (SCI-Expanded)
- XXXVIII. **Two Novel Variants and One Previously Reported Variant in the Insulin Receptor Gene in Two Cases with Severe Insulin Resistance Syndrome**  
Dagdeviren Cakir A., Saidov S., Turan H., CEYLANER S., Özer Y., KUTLU H. T., ERCAN O., EVLİAYOĞLU S. O.  
Molecular Syndromology, vol.11, no.2, pp.90-96, 2020 (SCI-Expanded)
- XXXIX. **A Rare Cause of Adrenal Insufficiency - Isolated ACTH Deficiency Due to TBX19 Mutation: Long-Term Follow-Up of Two Cases and Review of the Literature**  
Kardelen Al A. D., Poyrazoğlu Ş., Aslanger A., YEŞİL SAYIN G., CEYLANER S., BAŞ F., Darendeliler F.  
Hormone Research in Paediatrics, vol.92, no.6, pp.395-403, 2020 (SCI-Expanded)
- XL. **Early-onset progressive encephalopathy associated with NAXE gene variants: a case report of a Turkish child**  
İNCECİK F., CEYLANER S.  
Acta Neurologica Belgica, vol.120, no.3, pp.733-735, 2020 (SCI-Expanded)
- XLI. **Genotypes and estimated prevalence of phosphomannomutase 2 deficiency in Turkey differ significantly from those in Europe**  
YILDIZ Y., Arslan M., Çelik G., Kasapkara Ç. S., CEYLANER S., DURSUN A., SİVRİ H. S., Coşkun T., TOKATLI A.  
American Journal of Medical Genetics, Part A, vol.182, no.4, pp.705-712, 2020 (SCI-Expanded)
- XLII. **Hypokalemia and hearing loss in a 3-year-old boy: Questions**  
Aksoy O. Y., Cayci F. S., CEYLANER S., Tokgoz S. A., Kaplan G., Bayrakci U. S.  
Pediatric Nephrology, vol.35, no.4, pp.615, 2020 (SCI-Expanded)
- XLIII. **Cardiac Tamponade in Gorham-Stout Syndrome Associated with GATA2 Mutation**  
Oguz M. M., OĞUZ B., Dogan V., Aydin B., Eyuboglu T. S., Yesil S., CEYLANER S., Senel S.

Indian Journal of Pediatrics, vol.87, no.3, pp.239-240, 2020 (SCI-Expanded)

- XLIV. **First observation of hemoglobin G-Norfolk in the Turkish population**  
Ünal H., Atay A., Yücel M., Narin F., CEYLANER S., Canatan D.  
Turkish Journal of Biochemistry, vol.46, no.1, pp.95-100, 2020 (SCI-Expanded)
- XLV. **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 GLI2 gene**  
Demiral M., DEMİR BİLEK H., Unal E., Durmaz C. D., CEYLANER S., Özbek M. N.  
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.12, no.3, pp.319-328, 2020 (SCI-Expanded)
- XLVI. **A rare case of primary coenzyme Q10 deficiency due to COQ9 mutation**  
Olgac A., Öztoprak Ü., Kasapkara Ç. S., Kılıç M., Yüksel D., Derinkuyu E. B., Taşçı Yıldız Y., CEYLANER S., EZGÜ F. S.  
Journal of Pediatric Endocrinology and Metabolism, vol.33, no.1, pp.165-170, 2020 (SCI-Expanded)
- XLVII. **Importance of dna sequencing for abnormal hemoglobins detected by hplc screening Hplc ile tanımlanan anormal hemoglobinler için dna dizilemenin önemi**  
Canatan D., Çim A., Delibaş S., Altunsoy E., CEYLANER S.  
Turkish Journal of Hematology, vol.37, no.2, pp.134-135, 2020 (SCI-Expanded)
- XLVIII. **A novel mutation in the GP1BA gene in Bernard-Soulier syndrome**  
ÖZDEMİR Z. C., Düzenli Kar Y., CEYLANER S., BÖR Ö.  
Blood Coagulation and Fibrinolysis, vol.31, no.1, pp.83-86, 2020 (SCI-Expanded)
- XLIX. **Clinical findings in five Turkish patients with citrin deficiency and identification of a novel mutation on SLC25A13**  
Köse M. D., Kagnici M., Özdemir T. R., Erdur C. B., Erdemir G., Karakoyun M., Guzin Y., CEYLANER S., Genel F.  
Journal of Pediatric Endocrinology and Metabolism, vol.33, no.1, pp.157-163, 2020 (SCI-Expanded)
- L. **Clinical features and outcomes of 23 patients with wiskottaldrich syndrome: A single-center experience Wiskott-aldrich sendromlu 23 hastanın klinik özellikleri ve sonuçları: Tek merkez deneyimi**  
Haskoğlu Ş., Öztürk A., Öztürk G., Bal S. K., İslamoğlu C., Baskın K., CEYLANER S., Satiroğlu L. T., DOĞU E. F., İkinciğulları A.  
Turkish Journal of Hematology, vol.37, no.4, pp.271-281, 2020 (SCI-Expanded)
- LI. **The prevalence of fabry disease among turkish patients with non-obstructive hypertrophic cardiomyopathy: Insights from a screening study**  
BARMAN H. A., İKİTİMUR B., Avcı B. K., DURMAZ E., Atıcı A., Aslan S., CEYLANER S., KARPUZ M. H.  
Balkan Medical Journal, vol.36, no.6, pp.354-358, 2019 (SCI-Expanded)
- LII. **Analysis of TP53 gene in uterine myomas: No mutations but P72R polymorphism is associated with myoma development**  
Altinkaya S. O., Avcioglu S. N., Sezer S. D., CEYLANER S.  
Journal of Obstetrics and Gynaecology Research, vol.45, no.10, pp.2088-2094, 2019 (SCI-Expanded)
- LIII. **Clinical, Histochemical, and Molecular Study of Three Turkish Siblings Diagnosed with H Syndrome, and Literature Review**  
ŞİMŞEK E., ŞİMŞEK T., Eren M., YILMAZ E., ARIK D., ÇİLİNGİR O., CEYLANER S., Harmanlı K.  
Hormone Research in Paediatrics, vol.91, no.5, pp.346-355, 2019 (SCI-Expanded)
- LIV. **A novel homozygous cyp19a1 gene mutation: Aromatase deficiency mimicking congenital adrenal hyperplasia in an infant without obvious maternal virilisation**  
Dursun F., CEYLANER S.  
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.11, no.2, pp.196-201, 2019 (SCI-Expanded)
- LV. **Novel mutation in MASP1 gene in a new family with 3MC syndrome**  
Basdemirci M., Sen A., CEYLANER S.  
Clinical Dysmorphology, vol.28, no.2, pp.91-93, 2019 (SCI-Expanded)
- LVI. **Association of vitamin D receptor gene polymorphisms with osteosarcoma risk and prognosis**  
KURUCU N., Şahin G., Sarı N., CEYLANER S., İlhan İ. E.  
Journal of Bone Oncology, vol.14, 2019 (SCI-Expanded)
- LVII. **A Very Rare Etiology of Hypotonia and Seizures: Congenital Glutamine Synthetase Deficiency**

- Ünal O., CEYLANER S., Akln R.  
Neuropediatrics, vol.50, no.1, pp.51-53, 2019 (SCI-Expanded)
- LVIII. **Neonatal form of biotin-thiamine-responsive basal ganglia disease. Clues to diagnosis**  
Değerliyurt A., Gündüz M., CEYLANER S., Ünal Ö., Ünal S.  
Turkish Journal of Pediatrics, vol.61, no.2, pp.261-266, 2019 (SCI-Expanded)
- LIX. **Clinical features of 27 Turkish propionic acidemia patients with 12 novel mutations**  
KOR D., Şeker-Yılmaz B., Bulut F. D., Kılavuz S., Öktem M., CEYLANER S., Yıldızdaş D., Önenli-Mungan N.  
Turkish Journal of Pediatrics, vol.61, no.3, pp.330-336, 2019 (SCI-Expanded)
- LX. **Ataxia, tremor, intellectual disability: A case of STXBP1 encephalopathy with a new mutation**  
Değerliyurt A., Kesen G. G., CEYLANER S.  
Turkish Journal of Pediatrics, vol.61, no.5, pp.757-759, 2019 (SCI-Expanded)
- LXI. **Nonketotic hyperglycinemia: Clinical range and outcome of a rare neurometabolic disease in a single-center**  
Genç Sel Ç., Kılıç M., Yüksel D., Aksoy A., Kasapkara Ç. S., CEYLANER S., Oğuz K. K.  
Brain and Development, vol.40, no.10, pp.865-875, 2018 (SCI-Expanded)
- LXII. **A rare mutation in the EPG5 gene causes Vici syndrome**  
Demiral E., Sen A., Esener Z., CEYLANER S., TEKEDERELİ İ.  
Clinical Dysmorphology, vol.27, no.4, pp.145-147, 2018 (SCI-Expanded)
- LXIII. **A rare cause of fever of unknown origin: Hypohidrotic ectodermal dysplasia with a splice site mutation**  
Oguz M. M., Akcaboy M., Gurkan A., Acoglu E. A., Zorlu P., CEYLANER S., Senel S.  
Minerva Pediatrica, vol.70, no.5, pp.493-495, 2018 (SCI-Expanded)
- LXIV. **Recurrent hepatic failure and status epilepticus: an uncommon presentation of hyperargininemia**  
Yucel H., Kasapkara Ç. S., Akcaboy M., Aksoy E., Sahin G. E., Derinkuyu B. E., Senel S., CEYLANER S.  
Metabolic Brain Disease, vol.33, no.5, pp.1775-1778, 2018 (SCI-Expanded)
- LXV. **Four Gaucher disease type II patients with three novel mutations: a single centre experience from Turkey**  
BULUT F. D., KOR D., Şeker-Yılmaz B., Hergüner Ö., CEYLANER S., Özknay F., Kılavuz S., Önenli-Mungan N.  
Metabolic Brain Disease, vol.33, no.4, pp.1223-1227, 2018 (SCI-Expanded)
- LXVI. **Identification of a new de novo mutation underlying regressive episodic ataxia type I**  
Karalok Z. S., Megaro A., Cenciarini M., Guven A., Hasan S. M., Taskin B. D., Imbrici P., CEYLANER S., Pessia M., D'Adamo M. C.  
Frontiers in Neurology, vol.9, no.JUL, 2018 (SCI-Expanded)
- LXVII. **Twenty-seven mutations with three novel pathogenic variants causing biotinidase deficiency: A report of 203 patients from the southeastern part of Turkey**  
Yilmaz B. S., Mungan N. O., Kor D., Bulut D., Seydaoglu G., Öktem M., CEYLANER S.  
Journal of Pediatric Endocrinology and Metabolism, vol.31, no.3, pp.339-343, 2018 (SCI-Expanded)
- LXVIII. **A case of Riley Ruvulcaba syndrome with a novel PTEN mutation accompanied by diffuse testicular microlithiasis and precocious puberty**  
Ozsu E., Sen A., CEYLANER S.  
Journal of Pediatric Endocrinology and Metabolism, vol.31, no.1, pp.95-99, 2018 (SCI-Expanded)
- LXIX. **A mutation in INSR in a child presenting with severe acanthosis nigricans**  
Tuhan H., CEYLANER S., Nalbantoğlu Ö., Acar S., ABACI A., BÖBER E., DEMİR K.  
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.9, no.4, pp.371-374, 2017 (SCI-Expanded)
- LXX. **Early-onset severe obesity due to complete deletion of the leptin gene in a boy**  
Ozsu E., CEYLANER S., Onay H.  
Journal of Pediatric Endocrinology and Metabolism, vol.30, no.11, pp.1227-1230, 2017 (SCI-Expanded)
- LXXI. **A novel genetic mutation in a Turkish family with GCK-MODY**  
Ahmet Ucakturk S., Gunindi F., CEYLANER S., Mengen E., Elmaogulları S., YÜKSEL B.  
International Journal of Diabetes in Developing Countries, vol.37, no.3, pp.323-326, 2017 (SCI-Expanded)
- LXXII. **Type 1 rhizomelic chondrodysplasia punctata with a homozygous PEX7 mutation**

- Muratoğlu Sahin N, Bilici M. E., Kurnaz E., Pala Akdoğan M., CEYLANER S., AYCAN Z.  
Journal of Pediatric Endocrinology and Metabolism, vol.30, no.8, pp.889-892, 2017 (SCI-Expanded)
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