Prof. SERDAR CEYLANER

Personal Information

Email: serdar.ceylaner@lokmanhekim.edu.tr
Web: https://avesis.lokmanhekim.edu.tr/serdar.ceylaner

International Researcher IDs
ScholarID: g1xg4D8AAAAJ
ORCID: 0000-0003-2786-1911
ScopusID: 6508114123
Yoksis Researcher ID: 228432

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 ıgf-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)**
Gökpınar 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**
Kllç Yldlrlm G., YARAR C., Şeker Yllmaz 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., Aydln 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., Özaydln 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., Küçükcongar 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 methyloxidase deficiency type 2: Novel mutations in cyp11b2**
Turan H., Çakır A. D., Özer Y., Tarçın G., Özcabi B., CEYLANER S., ERCAN O., EVLİYAOĞLU 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 Yilmaz 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., Kasapkara 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., Eksioglu 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.S3, 2020 (SCI-Expanded)

XXXV. **A rare cause of delayed puberty in two cases with 46,XX and 46,XY karyotype: 17 α-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**
Haskologlu S., Kostel Bal S., Islamoglu 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., EVLİYAOĞLU 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., DEMİRBİLEK 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., Taşçl 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**
Haskoloğlu Ş., Öztürk A., Öztürk G., Bal S. K., İslamoğlu C., Baskın K., CEYLANER S., Satıroğlu L. T., DOĞU E. F., İkincioğulları 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., Harmancl 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. **Arare 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., Özkınay 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 pathologenic 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 Ruvalcaba 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)

LXXIII. **Improved metabolic control in tetrahydrobiopterin (BH4), responsive phenylketonuria with sapropterin administered in two divided doses vs. a single daily dose**
KOR D., Yilmaz B. Ş., BULUT F. D., CEYLANER S., Mungan N. Ö.
Journal of Pediatric Endocrinology and Metabolism, vol.30, no.7, pp.713-718, 2017 (SCI-Expanded)

LXXIV. **Congenital Glucose–Galactose Malabsorption in a Turkish Newborn: A Novel Mutation of Na+/Glucose Cotransporter Gene**
Atay F. Y., Derme T., Uras N., CEYLANER G., CEYLANER S., Sari F. N., Oguz S. S.
Digestive Diseases and Sciences, vol.62, no.1, pp.280-281, 2017 (SCI-Expanded)

LXXV. **A case of vitamin d-dependent rickets type 1a with a novel mutation in the uzbek population**
Özcabı B., Bucak F. T., Jaferova S., Oruç Ç., Adrovic A., CEYLANER S., ERCAN O., EVLİYAOĞLU S. O.
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.8, no.4, pp.484-489, 2016 (SCI-Expanded)

LXXVI. **Crouzonodermoskeletal syndrome with hypoplasia of corpus callosum and inferior vermis**
Gürbüz F., CEYLANER S., TOPALOĞLU A. K., YÜKSEL B.
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.8, no.3, pp.373-374, 2016 (SCI-Expanded)

LXXVII. **Excellent response to deep brain stimulation in a young girl with GNAO1-related progressive choreoathetosis**
YILMAZ SÜSLÜER S., Turhan T., CEYLANER S., GÖKBEN S., TEKGÜL H., Serdaroglu G.
Child's Nervous System, vol.32, no.9, pp.1567-1568, 2016 (SCI-Expanded)

LXXVIII. **Rodriguez lethal acrofacial dysostosis syndrome with ambiguous genitalia**
Ural U. M., CEYLANER S.
Taiwanese Journal of Obstetrics and Gynecology, vol.55, no.4, pp.613-615, 2016 (SCI-Expanded)

LXXIX. **Cystinosis in Eastern Turkey**
Doğan M., Bulan K., Kaba S., Cesur Y., CEYLANER S., Ustyol L.
Journal of Pediatric Endocrinology and Metabolism, vol.29, no.8, pp.965-969, 2016 (SCI-Expanded)

LXXX. **Sertoli cell only syndrome with ambiguous genitalia**
Gurbuz F., CEYLANER S., ERDOĞAN Ş., TOPALOĞLU A. K., YÜKSEL B.
Journal of Pediatric Endocrinology and Metabolism, vol.29, no.7, pp.849-852, 2016 (SCI-Expanded)

LXXXI. **Early-Onset Mild Type Leukoencephalopathy Caused by a Homozygous EARS2 Mutation**
Taskin B. D., Karalok Z. S., Gurkas E., Aydin K., Aydogmus U., CEYLANER S., Karaer K., Yilmaz C., Pearl P. L.
Journal of Child Neurology, vol.31, no.7, pp.938-941, 2016 (SCI-Expanded)

LXXXII. **Maturity onset diabetes of youth (MODY) in Turkish children: Sequence analysis of 11 causative genes by next generation sequencing**
Ağladioğlu S. Y., AYCAN Z., Çetinkaya S., Baş V. N., Önder A., Peltek Kendirci H. N., Doğan H., CEYLANER S.
Journal of Pediatric Endocrinology and Metabolism, vol.29, no.4, pp.487-496, 2016 (SCI-Expanded)

LXXXIII. **Combination of two different homozygote mutations in Pompe disease**
Arslan A., Poyrazoǧlu H. G., Kiraz A., ÖZCAN A., Işik H., Ergul A. B., Mungan N. Ö., Streubel B., CEYLANER S., Altuner Torun Y.
Pediatrics International, vol.58, no.3, pp.241-243, 2016 (SCI-Expanded)

LXXXIV. **Coexistence of kabuki syndrome and autoimmune thyroiditis**
Gürbüz F., Yüreğir Ö. Ö., CEYLANER S., TOPALOĞLU A. K., YÜKSEL B.
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.8, no.1, pp.105-106, 2016 (SCI-Expanded)

LXXXV. **Brown-Vialetto-Van Laere syndrome: Two siblings with a new mutation and dramatic therapeutic effect of high-dose riboflavin**
Horoz O. O., Mungan N. O., Yildizdas D., Hergüner Ö., CEYLANER S., KOR D., Waterham H., Coskun T.
Journal of Pediatric Endocrinology and Metabolism, vol.29, no.2, pp.227-231, 2016 (SCI-Expanded)

LXXXVI. **KEPT in mind infantile neuroaxonal dystrophy**
Karalok Z., Taskin B., Aydogmus U., CEYLANER S., Karaer K., Yilmaz C.
Genetic Counseling, vol.27, no.2, pp.279-282, 2016 (SCI-Expanded)

LXXXVII. **Vici syndrome in siblings born to consanguineous parents**

Tasdemir S., ŞAHİN İ., Cayir A., Yuce I., CEYLANER S., TATAR A.
American Journal of Medical Genetics, Part A, vol.170, no.1, pp.220-225, 2016 (SCI-Expanded)

LXXXVIII. **Molecular diagnosis of maturity-onset diabetes of the young (MODY) in Turkish children by using targeted next-generation sequencing**
Anlk A., Cątll G., Abacl A., Sarl E., Yeşilkaya E., Korkmaz H. A., Demir K., Altlnclk A., Tuhan H. Ü., Klzlldaą S., et al.
Journal of Pediatric Endocrinology and Metabolism, vol.28, no.11-12, pp.1265-1271, 2015 (SCI-Expanded)

LXXXIX. **Mitochondrial Membrane Protein-Associated Neurodegeneration**
YILMAZ SÜSLÜER S., GÖKBEN S., CEYLANER S.
Pediatric Neurology, vol.53, no.4, pp.373-374, 2015 (SCI-Expanded)

XC. **Primary systemic carnitine deficiency: A Turkish case with a novel homozygous SLC22A5 mutation and 14 years follow-up**
Yilmaz B. S., KOR D., Mungan N. O., Erdem S., CEYLANER S.
Journal of Pediatric Endocrinology and Metabolism, vol.28, no.9-10, pp.1179-1181, 2015 (SCI-Expanded)

XCI. **Testotoxicosis: Report of two cases, one with a novel mutation in LHCGR gene**
Özcabı B., Bucak F. T., CEYLANER S., ÖZCAN R., Büyükünal C., ERCAN O., TÜYSÜZ B., EVLİYAOĞLU S. O.
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.7, no.3, pp.242-248, 2015 (SCI-Expanded)

XCII. **A rare cause of fatal pulmonary alveolar proteinosis: Niemann-Pick disease type C2 and a novel mutation**
Yaman A., Eminoąlu F. T., KENDİRLİ T., Ödek Ç., CEYLANER S., KANSU TANCA A., İNCE E., Deda G.
Journal of Pediatric Endocrinology and Metabolism, vol.28, no.9-10, pp.1163-1167, 2015 (SCI-Expanded)

XCIII. **Giant axonal disease: Report of eight cases**
İNCECİK F., Herguner O. M., CEYLANER S., ZORLUDEMİR S., ALTUNBAŞAK Ş.
Brain and Development, vol.37, no.8, pp.803-807, 2015 (SCI-Expanded)

XCIV. **Pyridoxine-dependent epilepsy in two Turkish patients in Turkey and review of the literature**
Mert G. G., İNCECİK F., Özlem Hergüner M., CEYLANER S., ALTUNBAŞAK Ş.
Turkish Journal of Pediatrics, vol.57, no.4, pp.394-397, 2015 (SCI-Expanded)

XCV. **Two Novel Missense Mutations in Nonketotic Hyperglycinemia**
Yilmaz B. S., KOR D., CEYLANER S., Mert G. G., İNCECİK F., Kartal E., Mungan N. O.
Journal of Child Neurology, vol.30, no.6, pp.789-792, 2015 (SCI-Expanded)

XCVI. **Three cases of Wolfram syndrome with different clinical aspects**
Çamtosun E., ŞIKLAR Z., Kocaay P., CEYLANER S., Flanagan S. E., Ellard S., Berberoąlu M.
Journal of Pediatric Endocrinology and Metabolism, vol.28, no.3-4, pp.433-438, 2015 (SCI-Expanded)

XCVII. **Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?**
Romani M., Mancini F., Micalizzi A., Poretti A., Miccinilli E., Accorsi P., Avola E., Bertini E., Borgatti R., Romaniello R., et al.
Human Genetics, vol.134, no.1, pp.123-126, 2015 (SCI-Expanded)

XCVIII. **A familial case of Coffin-Lowry syndrome caused by RPS6KA3 C.898C>T mutation associated with multiple abnormal brain imaging findings**
Tos T., Alp M., Aksoy A., CEYLANER S., Uer A.
Genetic Counseling, vol.26, no.1, pp.47-52, 2015 (SCI-Expanded)

XCIX. **Turkish cases of early infantile epileptic encephalopathy: Two novel mutations in the cyclin-dependent kinase-like 5 (CDKL5) gene**
GÖKBEN S., Serdaroğlu G., Yılmaz S., Bienvenu T., CEYLANER S.
Turkish Journal of Pediatrics, vol.57, no.3, pp.272-276, 2015 (SCI-Expanded)

C. **Reversible clinical and magnetic resonance imaging findings in late-onset cobalamin C defect**
Gurkas E., Kartal A., Aydin K., Kucukçongar A., Dilber C., CEYLANER S.
Genetic Counseling, vol.26, no.4, pp.425-430, 2015 (SCI-Expanded)

CI. **Unilateral ectrodactyly in a newborn with trisomy 18 syndrome: An unusual association**
Kislal F. M., Altuntas N., Ozdemir O., CEYLANER S., Kislal M. H., Andiran N.
Journal of the College of Physicians and Surgeons Pakistan, vol.25, no.8, pp.619-620, 2015 (SCI-Expanded)

CII. **Citrin deficiency: An infant incidentally detected by phenylketonuria screening with a novel mutation**

in SLC25A 13 gene
AKTUĞLU ZEYBEK A. Ç., KIYKIM E., ZÜBARİOĞLU T., CANSEVER M. Ş., CEYLANER S., Erkan T.
Genetic Counseling, vol.26, no.4, pp.409-413, 2015 (SCI-Expanded)

CIII. **Single median maxillary central incisor syndrome and its clinical importance**
Aydemr H., Aydemr S., BEZGİN T., CEYLANER S., Senel S.
Genetic Counseling, vol.25, no.4, pp.453-455, 2015 (SCI-Expanded)

CIV. **Reversible pulmonary arterial hypertension in cobalamin-dependent cobalamin C disease due to a novel mutation in the MMACHC gene**
Gündüz M., Ekici F., Özaydın E., CEYLANER S., Perez B.
European Journal of Pediatrics, vol.173, no.12, pp.1707-1710, 2014 (SCI-Expanded)

CV. **X-linked adrenoleukodystrophy in a 6-year-old boy initially presenting with psychiatric symptoms**
İNCECİK F., HERGÜNER M., Mert G., Önenli-Mungan N., CEYLANER S., KOR D., Altunbaçak Ç.
Turkish Journal of Pediatrics, vol.56, no.6, pp.651-653, 2014 (SCI-Expanded)

CVI. **A novel missense mutation in HSD17B3 gene in a 46, XY adolescent presenting with primary amenorrhea and virilization at puberty**
Tuhan H. U., Anik A., Catli G., CEYLANER S., Dundar B., BÖBER E., ABACI A.
Clinica Chimica Acta, vol.438, pp.154-156, 2014 (SCI-Expanded)

CVII. **Recurrent proximal 18p monosomy and 18q trisomy in a family due to a pericentric inversion**
Zamani A. G., Acar A., Durakbasi-Dursun G., Yildirim M. S., CEYLANER S., Tuncez E.
American Journal of Medical Genetics, Part A, vol.164, no.5, pp.1239-1244, 2014 (SCI-Expanded)

CVIII. **Chronic lymphocytic leukemia in a child: A challenging diagnosis in pediatric oncology practice**
Demir H. A., Bayhan T., ÜNER A., KURTULAN O., Karakuş E., Emir S., Özyörük D., CEYLANER S.
Pediatric Blood and Cancer, vol.61, no.5, pp.933-935, 2014 (SCI-Expanded)

CIX. **A novel heterozygous mutation in steroidogenic factor-1 in pubertal virilization of a 46,XY female adolescent**
ŞIKLAR Z., BERBEROĞLU M., CEYLANER S., Çamtosun E., Kocaay P., GÖLLÜ BAHADIR G., Sertçelik A., Öcal G.
Journal of Pediatric and Adolescent Gynecology, vol.27, no.2, pp.98-101, 2014 (SCI-Expanded)

CX. **Cytokine concentrations in pediatric patients with Crimean-Congo hemorrhagic fever**
TEZER H., Ozkaya-Parlakay A., Kizilgün M., Kaya A., Gulhan B., Yüksek S. K., Kara S. S., Sonmezer M. C., CEYLANER S.
Pediatric Infectious Disease Journal, vol.33, no.11, pp.1185-1187, 2014 (SCI-Expanded)

CXI. **Are low maternal estriol levels a predictor for pro-opiomelanocortin (POMC) deficiency caused by POMC mutation during pregnancy?**
Aldemir O., ÖZEN S., Sanlialp C., CEYLANER S.
Prenatal Diagnosis, vol.33, no.13, pp.1297-1298, 2013 (SCI-Expanded)

CXII. **Prenatal diagnosis of goldenhar syndrome with unusual features by 3D ultrasonography**
Guzelmansur I., CEYLANER G., CEYLANER S., CEYLAN N., Daplan T.
Genetic Counseling, vol.24, no.3, pp.319-325, 2013 (SCI-Expanded)

CXIII. **Acromesomelic dysplasia with cardiac and neurologic abnormalities: An association by chance, new features of Maroteaux type or a new syndrome?**
Kurt F., CEYLANER S., Yakut H.
Genetic Counseling, vol.24, no.1, pp.75-80, 2013 (SCI-Expanded)

CXIV. **Primary adrenal insufficiency caused by a novel mutation in DAX1 gene**
EVLİYAOĞLU S. O., Dokurel I., Bucak F., Özcabi B., ERCAN O., CEYLANER S.
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.5, no.1, pp.55-57, 2013 (SCI-Expanded)

CXV. **Nephron-sparing surgery for renal cell carcinoma of the allograft after renal transplantation: Report of two cases**
TÜZÜNER A., Çakir F., AKYOL C., Çelebi Z., CEYLANER S., CEYLANER G., ŞENGÜL Ş., KEVEN K.
Transplantation Proceedings, vol.45, no.3, pp.958-960, 2013 (SCI-Expanded)

CXVI. **Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders**
Travaglini L., Brancati F., Silhavy J., Iannicelli M., Nickerson E., Elkhartoufi N., Scott E., Spencer E., Gabriel S., Thomas

S., et al.
European Journal of Human Genetics, vol.21, no.10, pp.1074-1078, 2013 (SCI-Expanded)

CXVII. **Novel CRLF1 gene mutation in a newborn infant diagnosed with Crisponi syndrome**
Hakan N., Eminoglu F. T., Aydin M., Zenciroglu A., Karadag N. N., Dursun A., Okumus N., CEYLANER S.
Congenital Anomalies, vol.52, no.4, pp.216-218, 2012 (SCI-Expanded)

CXVIII. **A 7q11.23 microduplication patient with cerebral palsy and facial dysmorphism**
Değerliyurt A., CEYLANER S., ÖZDAĞ SEVGİLİ H.
Genetic Counseling, vol.23, no.2, pp.263-267, 2012 (SCI-Expanded)

CXIX. **A new family with autosomal dominant porencephaly with a novel COL4A1 mutation. Are arachnoid cysts related to COL4A1 mutations?**
Değerliyurt A., CEYLANER G., Koçak H., Bilginer Gürbüz B., Cihan B., Rizzu P., CEYLANER S.
Genetic Counseling, vol.23, no.2, pp.185-193, 2012 (SCI-Expanded)

CXX. **Hydranencephaly, pituitary hypoplasia, and anophthalmia in a male infant**
Köstü M., Tuncer O., CEYLANER S., Çaksen H.
Clinical Dysmorphology, vol.21, no.3, pp.155-156, 2012 (SCI-Expanded)

CXXI. **High frequency of p.Thr93Met in Smith-Lemli-Opitz syndrome patients in Turkey**
Kalb S., ÇAĞLAYAN A. O., Degerliyurt A., Schmid S., CEYLANER S., HATİPOĞLU N., Hinderhofer K., Rehder H., Kurtoglu S., CEYLANER G., et al.
Clinical Genetics, vol.81, no.6, pp.598-601, 2012 (SCI-Expanded)

CXXII. **The earlier described mutation (c.307C> T [p.R103X]) in the SRD5A2 gene causing a 46,XY female phenotype**
ŞİMŞEK E., Binay C., CEYLANER S.
Journal of Pediatric Endocrinology and Metabolism, vol.25, no.5-6, pp.543-545, 2012 (SCI-Expanded)

CXXIII. **Genetic evaluation of severe male factor infertility in Turkey: A cross-sectional study**
Cavkaytar S., Batioglu S., Gunel M., CEYLANER S., KARAER A.
Human Fertility, vol.15, no.2, pp.100-106, 2012 (SCI-Expanded)

CXXIV. **Ovulation induction with gonadotropins causes increased sister chromatid exchanges**
Tonguç E., CEYLANER G., Var T., Zülfikaroğlu E., CEYLANER S.
Genetic Counseling, vol.22, no.2, pp.193-198, 2011 (SCI-Expanded)

CXXV. **A new variant of a known mutation in two siblings with permanent neonatal diabetes mellitus**
AYCAN Z., Çetinkaya S., Oğuz Ş. S., CEYLANER S.
Journal of Pediatric Endocrinology and Metabolism, vol.24, no.5-6, pp.373-375, 2011 (SCI-Expanded)

CXXVI. **Coexistence of borderline ovarian epithelial tumor, primary pelvic hydatid cyst, and lymphoepithelioma-like gastric carcinoma**
Gungor T., Altinkaya S. O., Sirvan L., Lafuente R. A., CEYLANER S.
Taiwanese Journal of Obstetrics and Gynecology, vol.50, no.2, pp.201-204, 2011 (SCI-Expanded)

CXXVII. **A novel mutation in the RPS6KA3 gene in a patient with Coffin-Lowry syndrome**
Senel S., CEYLANER S., CEYLANER G., Hanli Sahin A., Andrieux J., Delaunoy J.
Genetic Counseling, vol.22, no.1, pp.21-24, 2011 (SCI-Expanded)

CXXVIII. **Vascular endothelial growth factor +405 C/G polymorphism is highly associated with an increased risk of endometriosis in Turkish women**
Altinkaya S. O., Ugur M., CEYLANER G., Ozat M., Gungor T., CEYLANER S.
Archives of Gynecology and Obstetrics, vol.283, no.2, pp.267-272, 2011 (SCI-Expanded)

CXXIX. **An uncommon complementary isochromosome of 46,XY, i(9)(p10),i(9)(q10) in an infertile oligoasthenoteratozoospermic man**
Guvendag Guven E. S., Dilbaz S., CEYLANER S., Acar H., Cinar O., Ozdegirmenci O., Karcaaltincaba D.
Fertility and Sterility, vol.95, no.1, 2011 (SCI-Expanded)

CXXX. **Sporadic nonautoimmune neonatal hyperthyroidism due to A623V germline mutation in the thyrotropin receptor gene**
AYCAN Z., Ağladioğlu S. Y., CEYLANER S., Çetinkaya S., Baş V. N., Kendirci H. N. P.
JCRPE Journal of Clinical Research in Pediatric Endocrinology, vol.2, no.4, pp.168-172, 2010 (SCI-Expanded)

CXXXI. **A case of lower mesodermal defects sequence**
Tos T., Aktas S., Ikbal M., Avci M., Senel S., CEYLANER S.
Genetic Counseling, vol.21, no.4, pp.381-384, 2010 (SCI-Expanded)

CXXXII. **A case of otocephaly with anencephaly and meningomyelocele**
Tos T., CEYLANER S., Senel S., Aktas S., Alp Y.
Genetic Counseling, vol.21, no.3, pp.325-328, 2010 (SCI-Expanded)

CXXXIII. **Carmi syndrome with congenital heart defects**
Aydin M., Zenciroglu A., Yaman A., Orun U. A., Arda N., Colak A. G., Okumus N., Ipek M. S., CEYLANER S.
American Journal of Medical Genetics, Part A, vol.152, no.8, pp.2120-2122, 2010 (SCI-Expanded)

CXXXIV. **Lumbocostovertebral syndrome in an infant of a diabetic mother**
Okumus N., Bas A. Y., Demirel N., Zenciroglu A., Surmeli S., CEYLANER S.
American Journal of Medical Genetics, Part A, vol.152, no.6, pp.1374-1377, 2010 (SCI-Expanded)

CXXXV. **Partial trisomy due to a de novo duplication 22q11.1-22q13.1: A cat-eye syndrome variant with brain anomalies**
Karcaaltincaba D., CEYLANER S., CEYLANER G., Dalkilic S., Karli-Oguz K., Kandemr O.
Genetic Counseling, vol.21, no.1, pp.19-24, 2010 (SCI-Expanded)

CXXXVI. **Mutations in SLC29a3, encoding an equilibrative nucleoside transporter ENT3, cause a familial histiocytosis syndrome (Faisalabad histiocytosis) and familial Rosai-Dorfman disease**
Morgan N. V., Morris M. R., Cangul H., Gleeson D., Straatman-Iwanowska A., Davies N., Keenan S., Pasha S., Rahman F., Gentle D., et al.
PLoS Genetics, vol.6, no.2, 2010 (SCI-Expanded)

CXXXVII. **Genetic abnormalities in Turkish women with premature ovarian failure**
CEYLANER G., Altinkaya S. O., Mollamahmutoglu L., CEYLANER S.
International Journal of Gynecology and Obstetrics, vol.110, no.2, pp.122-124, 2010 (SCI-Expanded)

CXXXVIII. **Multiple pterygium syndrome: A case report, comparison with fetal akinesia sequence and pterygium syndrome Multiple pterygium sendromu: Olgu sunumu, fetal akinezi sekansi ve pterygium sendromu ile karşilaştirilmasi**
Kişlal F. M., Pinar R., CEYLANER S., Dilmen U., Cörüt N.
Guncel Pediatri, vol.7, no.2, pp.101-103, 2009 (SCI-Expanded)

CXXXIX. **A Feingold syndrome case with previously undescribed features and a new mutation**
Koçak H., Özaydin E., Köse G., Marcelis C., Kamsteeg E., CEYLANER S.
Genetic Counseling, vol.20, no.3, pp.261-267, 2009 (SCI-Expanded)

CXL. **Prenatal diagnosis of a case with anencephaly-omphalocele-unilateral absent radial ray**
CEYLANER S., CEYLANER G., Altun M., Coşkun A., Danişman N.
Genetic Counseling, vol.20, no.2, pp.189-193, 2009 (SCI-Expanded)

CXLI. **Prenatal diagnosis of a case probably with oral-facial-digital syndrome - Gabrielli type**
Güven M., CEYLANER G., CEYLANER S., ÜZEL M., Coskun A.
Genetic Counseling, vol.20, no.2, pp.167-172, 2009 (SCI-Expanded)

CXLII. **A boy with trisomy 13 presenting with a subtle clinical picture and metopic synostosis**
ÜNAL S., Celik F. C., Soy D., CEYLANER S., Ceylaner G.
American Journal of Medical Genetics, Part A, vol.149, no.7, pp.1608-1609, 2009 (SCI-Expanded)

CXLIII. **Hypomelanosis of Ito and Sturge-Weber Syndrome Without Facial Nevus: An Association or a New Syndrome?**
Değerliyurt A., Kantar A., CEYLANER S., Aysun S.
Pediatric Neurology, vol.40, no.5, pp.395-397, 2009 (SCI-Expanded)

CXLIV. **Results of ICSI in severe oligozoospermic and azoospermic patients with AZF microdeletions**
Kilic S., Yuksel B., Yilmaz N., Ozdemir E., Ozturk U., CEYLANER S., Dogan M.
Iranian Journal of Reproductive Medicine, vol.7, no.2, pp.79-84, 2009 (SCI-Expanded)

CXLV. **A prenatally diagnosed case of sirenomelia with polydactyly and vestigial tail**
Guven M., ÜZEL M., CEYLANER S., Coskun A., CEYLANER G., Gungoren A.
Genetic Counseling, vol.19, no.4, pp.419-424, 2008 (SCI-Expanded)

CXLVI. **Goldenhar syndrome associated with growth hormone deficiency**
Yusufoğlu A., Çetinkaya E., CEYLANER S., AYCAN Z., Kibar E., Ekici F., Kizilgün M.
Genetic Counseling, vol.19, no.2, pp.173-176, 2008 (SCI-Expanded)

CXLVII. **Apparent Lenz microphthalmia syndrome: A patient with unusual manifestations**
Okumus N., Zenciroglu A., Demrel N., Bas A., CEYLANER S.
Genetic Counseling, vol.19, no.2, pp.177-182, 2008 (SCI-Expanded)

CXLVIII. **GJB2 and mitochondrial A1555G gene mutations in nonsyndromic profound hearing loss and carrier frequencies in healthy individuals**
Baysal E., Bayazit Y., CEYLANER S., Alatas N., Donmez B., CEYLANER G., San I., Korkmaz B., Yilmaz A., Menevse A., et al.
Journal of Genetics, vol.87, no.1, pp.53-57, 2008 (SCI-Expanded)

CXLIX. **Y chromosome azoospermia factor region microdeletions and recurrent pregnancy loss**
Karaer A., Karaer K., Ozaksit G., CEYLANER S., PERÇİN F. E.
American Journal of Obstetrics and Gynecology, vol.199, no.6, 2008 (SCI-Expanded)

CL. **Autosomal dominant inheritance of congenital dislocation of the hip in 16 members of a family Bir ailenin 16 üyesinde otozomal dominant kalıtım gösteren gelişimsel kalça çıkığı**
CEYLANER G., CEYLANER S., Ustunkan F., Inan M.
Acta Orthopaedica et Traumatologica Turcica, vol.42, no.4, pp.289-291, 2008 (SCI-Expanded)

CLI. **Chromosomal heteromorphisms may help for the diagnosis of uniparental disomy (UPD): A case report**
CEYLANER G., CEYLANER S., Danişman N., Ergün A., Ekici E., Schinzel A., Baumer A.
Prenatal Diagnosis, vol.27, no.11, pp.1072-1074, 2007 (SCI-Expanded)

CLII. **Neu-Laxova syndrome, grossly appearing normal on 20 weeks ultrasonographic scan, that manifested late in pregnancy: A case report**
Kahyaoglu S., Turgay I., Ertas I. E., CEYLANER S., Danisman N.
Archives of Gynecology and Obstetrics, vol.276, no.4, pp.367-370, 2007 (SCI-Expanded)

CLIII. **Evaluation of 2407 fetuses in a Turkish population**
CEYLANER G., CEYLANER S., Günyeli I., Ekici E., Celasun B., Danişman N.
Prenatal Diagnosis, vol.27, no.9, pp.800-807, 2007 (SCI-Expanded)

CLIV. **Correspondence: Is it a Proteus syndrome? [3]**
Senel S., Senel E., CEYLANER S.
Journal of Pediatric Orthopaedics Part B, vol.16, no.5, pp.385, 2007 (SCI-Expanded)

CLV. **Proteus Syndrome in the aetiology of carpal tunnel syndrome**
Senel S., Okumus N., CEYLANER S.
Acta Orthopaedica Belgica, vol.73, no.4, pp.548, 2007 (SCI-Expanded)

CLVI. **Prenatal diagnosis of a Turkish Bartsocas-Papas syndrome case with upper limb pterigia**
CEYLANER G., Güven M. A., CEYLANER S., ÜZEL M., Müftüoğlu K.
Prenatal Diagnosis, vol.27, no.6, pp.563-565, 2007 (SCI-Expanded)

CLVII. **Prenatal diagnosis of Meckel Gruber syndrome presenting with renal agenesis: report of a case.**
Guven M., CEYLANER S., CEYLANER G., Gul D., Ertas L.
Genetic counseling (Geneva, Switzerland), vol.17, no.1, pp.65-68, 2006 (SCI-Expanded)

CLVIII. **Extremely skewed X-chromosome inactivation patterns in women with recurrent spontaneous abortion**
Bagislar S., Ustuner I., Cengiz B., Soylemez F., Akyerli C. B., CEYLANER S., CEYLANER G., Acar A., Ozcelik T.
Australian and New Zealand Journal of Obstetrics and Gynaecology, vol.46, no.5, pp.384-387, 2006 (SCI-Expanded)

CLIX. **An infant with diaphragmatic hernia, anophthalmia and cardiac defect: Evaluation by magnetic resonance imaging autopsy**
CEYLANER S., Gozer H., CEYLANER G., Ertas I., Kizilates S., Edguer T.
Genetic Counseling, vol.17, no.2, pp.231-236, 2006 (SCI-Expanded)

CLX. **Prenatal and postnatal findings in a case with the autosomal recessive type of Robinow syndrome**
Guven M. A., Batukan C., CEYLANER S., ÜZEL M., Ozbek A., Demirpolat G.

Fetal Diagnosis and Therapy, vol.21, no.4, pp.386-389, 2006 (SCI-Expanded)

CLXI. **Postmortem evaluation of 220 prenatally diagnosed fetuses with neural tube defects: Detection of associated anomalies in a Turkish population**
CEYLANER S., CEYLANER G., Günyeli I., Ekici E., Tuğ M., Taner D., Ekerbicer H., Mollamahmutoğlu L., Danişman N.
Prenatal Diagnosis, vol.26, no.2, pp.147-153, 2006 (SCI-Expanded)

CLXII. **A case of fetal anticonvulsant syndrome with severe bilateral upper limb defect**
Guven M. A., Batukan C., CEYLANER S., CEYLANER G., ÜZEL M.
Journal of Maternal-Fetal and Neonatal Medicine, vol.19, no.2, pp.115-117, 2006 (SCI-Expanded)

CLXIII. **SALL4 mutations in Okihiro syndrome (Duane-radial ray syndrome), acro-renal-ocular syndrome, and related disorders**
Kohlhase J., Chitayat D., Kotzot D., CEYLANER S., Froster U. G., Fuchs S., Montgomery T., Rösler B.
Human Mutation, vol.26, no.3, pp.176-183, 2005 (SCI-Expanded)

CLXIV. **Craniosynostosis and ectopia lentis in a propositus whose parents are cousins [4]**
Güven D., Kalayci D., Hasiripi H., CEYLANER S., CEYLANER G.
American Journal of Medical Genetics, vol.134 A, no.2, pp.231, 2005 (SCI-Expanded)

CLXV. **A novel mutation in the lysyl hydroxylase 1 gene causes decreased lysyl hydroxylase activity in an ehlers-danlos VIA patient**
Walker L. C., Overstreet M. A., Siddiqui A., De Paepe A., CEYLANER G., Malfait F., Symoens S., Atsawasuwan P., Yamauchi M., CEYLANER S., et al.
Journal of Investigative Dermatology, vol.124, no.5, pp.914-918, 2005 (SCI-Expanded)

CLXVI. **Prenatal sonographic findings in a case of Varadi-Papp syndrome**
Guven M. A., CEYLANER S., Prefumo F., ÜZEL M.
Prenatal Diagnosis, vol.24, no.12, pp.989-991, 2004 (SCI-Expanded)

CLXVII. **Three new cases of disorganizationlike syndrome: One with accessory extrophia vesicalis**
Ilkehan H., ÇOBAN Y. K., Guven M. A., CEYLANER S.
Journal of Pediatric Surgery, vol.39, no.9, 2004 (SCI-Expanded)

CLXVIII. **Empty follicle syndrome in two sisters with three cycles: Case report**
Önalan G., Pabuçcu R., Önalan R., CEYLANER S., Selam B.
Human Reproduction, vol.18, no.9, pp.1864-1867, 2003 (SCI-Expanded)

CLXIX. **Evaluation of segregation patterns of 21;21 Robertsonian translocation along with sex chromosomes and interchromosomal effects in sperm nuclei of carrier by FISH technique**
Acar H., Yildirim M. S., Çora T., CEYLANER S.
Molecular Reproduction and Development, vol.63, no.2, pp.232-236, 2002 (SCI-Expanded)

CLXX. **Cutis laxa with growth and developmental delay [2]**
Karakurt C., Sipahi T., CEYLANER S., Şenocak F., Karademir S., Becer M.
Clinical Pediatrics, vol.40, no.7, pp.422-423, 2001 (SCI-Expanded)

CLXXI. **A large Turkish kindred with syndactyly type II (synpolydactyly). 1 Field investigation, clinical and pedigree data**
Sayli B. S., Akarsu A. N., Sayli U., AKHAN O., CEYLANER S., Sarfarazi M.
Journal of Medical Genetics, vol.32, no.6, pp.421-434, 1995 (SCI-Expanded)

Articles Published in Other Journals

I. **Further observation of Hemoglobin Fontainebleau (a21(B2) Ala-Pro) in a Turkish family**
Akar N., CEYLANER S., Akisin Y. A., Tastan G. S.
Egyptian Journal of Medical Human Genetics, vol.21, no.1, 2020 (ESCI)

II. **Prenatal Diagnosis of Osteogenesis Imperfecta Type III**
Canda M. T., CEYLANER S., Doganay Caglayan L., Demir A. B., Demir N.
Journal of Obstetrics and Gynecology of India, vol.69, no.4, pp.374-376, 2019 (ESCI)

III. **A rare case of prolidase deficiency with situs inversus totalis, identified by a novel mutation in the**

PEPD gene
Kiratli Nalbant E., Karaosmanoglu N., Kutlu O., CEYLANER S., Eksioglu H. M.
JAAD Case Reports, vol.5, no.5, pp.436-438, 2019 (Scopus)

IV. **A rare structural myopathy: Nemaline myopathy Nadir bir yapisal miyopati: Nemalin miyopatisi**
Yeşilbaş O., Şevketoğlu E., Kihtir H. S., Ersoy M., Petmezci M. T., Akkuş C. H., Şahin Ö., CEYLANER S.
Turk Pediatri Arsivi, vol.54, no.1, pp.49-52, 2019 (ESCI)

V. **When a common symptom of a neonate become an unusual diagnosis: A case report of HMG-CoA lyase deficiency with a novel mutation**
Kasapkara C. S., Akar M., Ozbek M. N., Tuzun H., Akcaboy M., CEYLANER S.
Gazi Medical Journal, vol.30, no.3, pp.298-299, 2019 (ESCI)

VI. **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**
Dilli D., Doğan N. N., İpek M. Ş., Çavuş Y., CEYLANER S., Doğan H., Dursun A., Küçüközkan T., Zenciroğlu A.
Clinical Nutrition ESPEN, vol.23, pp.73-78, 2018 (ESCI)

VII. **Mitochondrial membrane protein-associated neurodegeneration in a Turkish patient**
İNCECİK F., Hergüner O., Besen S., CEYLANER S.
Journal of Pediatric Neurosciences, vol.11, no.3, pp.288-289, 2016 (ESCI)

VIII. **Merosin-negative congenital muscular dystrophy: Report of five cases**
İNCECİK F., Herguner O. M., CEYLANER S., ALTUNBAŞAK Ş.
Journal of Pediatric Neurosciences, vol.10, no.4, pp.346-349, 2015 (ESCI)

IX. **Stimulus-induced myoclonus treated effectively with clonazepam in genetically confirmed Coffin-Lowry syndrome**
Arslan E. A., CEYLANER S., Turanli G.
Epilepsy and Behavior Case Reports, vol.2, pp.196-198, 2014 (ESCI)

X. **A child with XYY karyotype and epilepsy**
Değerliyurt A., CEYLANER G., CEYLANER S.
Journal of Pediatric Neurology, vol.9, no.2, pp.255-258, 2011 (ESCI)

XI. **Histopathological analysis of the placental lesions in pregnancies complicated with iugr and stillbirths in comparison with noncomplicated pregnancies Iugr ve ölü doğumlarla komplike olmuş gebeliklerde plasental lezyonların histopatolojik analizi ve nonkomplike gebeliklerle karşılaştırılması**
GÜNYELİ İ., ERDEMOĞLU E., CEYLANER S., Zergeroğlu S., Mungan T.
Journal of the Turkish German Gynecology Association, vol.12, no.2, pp.75-79, 2011 (ESCI)

XII. **A Smith Lemli Opitz syndrome patient diagnosed with mild symptoms Hafif klinik belirtilerle tani alan bir Smith Lemli Opitz sendromu olgusu**
Değerliyurt A., KILIÇ YILDIRIM G., AYCAN Z., CEYLANER S.
Turkiye Klinikleri Pediatri, vol.19, no.2, pp.191-194, 2010 (Scopus)

XIII. **A prenatally diagnosed pentalogy of cantrell case with encephaloce: A rare variant Prenatal tanisi konmuş Cantrell Pentalojisiolgusu: Ensefaloselin eşlik ettiğinadir bir varyant**
Güven M. A., CEYLANER G., CEYLANER S., Coşkun A., Bayazit H.
Turk Jinekoloji ve Obstetrik Dernegi Dergisi, vol.6, no.2, pp.123-127, 2009 (Scopus)

XIV. **A case of rhizomelic chondrodysplasia punctata complicated with fetal arrhythmia**
Dilli D., Yasar H., Baydar Z., Dilmen U., CEYLANER S., Altuğ N., Karadeniz S.
Erciyes Tip Dergisi, vol.30, no.4, pp.278-283, 2008 (ESCI)

XV. **A case with dextrocardia and multiple cardiac anomalies in a family with congenital heart malformations**
CEYLANER S., Güven M. A., CEYLANER G., Çiragil G., Tuğ M., Ertaş I. E.
Journal of the Turkish German Gynecology Association, vol.6, no.2, pp.158-160, 2005 (ESCI)

XVI. **CHARGE association: A case report CHARGE asosiasyonu: Bir olgu sunumu**
Dilli D., CEYLANER S., Bostanci I., Tök Ö., Dallar Y.
Gulhane Medical Journal, vol.46, no.3, pp.260-263, 2004 (Scopus)

XVII. **Asymmetric crying facies associated with hemihypertrophy: Report of one case**

Çaksen H., Patiroğlu T., Çiftçi A., Çikrikçi V., CEYLANER S.
Acta Paediatrica Taiwanica, vol.44, no.2, pp.98-100, 2003 (Scopus)

Metrics

Publication: 188
Citation (WoS): 1027
Citation (Scopus): 1141
H-Index (WoS): 16
H-Index (Scopus): 16