Expert PhD Umut ALTUNOĞLU

Personal Information

Office Phone: +90 212 534 8440
Office Phone: +90 212 414 2000 Extension: 32564
Fax Phone: +90 212 534 8440
Email: umut.altunoglu@istanbul.edu.tr
Address: İstanbul Tıp Fakültesi, Monoblok Poliklinikler Binası, Tıbbi Genetik Polikliniği Millet Cad., 34093, Fatih - İstanbul

Biography

Dr. Umut Altunoğlu, tıp eğitimini İstanbul Üniversitesi, İstanbul Tıp Fakültesinde 2006 yılında, tıbbi genetik uzmanlık eğitimini ise aynı fakültenin Tıbbi Genetik Anabilim Dalında 2012 yılında tamamlamıştır. 2010 yılında Nijmegen Radboud Üniversitesi, İnsan Genetiği Bölümü'nde Walker-Warburg sendromu ve Herediter konjenital fasiyal paralizi genetik etiyolojisinin aydınlatılmasına yönelik araştırma projelerine 3 ay süreyle dahil olmuştur. 2013-2014 yılları arasında İstanbul Tıp Fakültesi Tıbbi Genetik Anabilim Dalı'nda zorunlu hizmetini tamamlamıştır. 2014 yılından bu yana İstanbul Tıp Fakültesi Tıbbi Genetik Anabilim Dalında uzman doktor olarak görev yapmaktadır. Hasta yoğunluğu fazla olan bir referans merkezinde klinik çalışmalara ağırlık verdiğinden, nadir dismorfik sendromlar ve prenatal genetik konusunda özellikle deneyim kazanmıştır. Akademik çalışmalarını uzmanlık tezini tamamladığı frontonazal displazi grubu hastalıklar üzerinde yoğunlaştırmıştır. SCI kapsamında yabancı dergilerde yayınlanmış 20'den fazla makalesi bulunmaktadır. Güncel ilgi alanı lenfödem ile seyreden dismorfik sendromlardır.

Education Information

Expertise In Medicine, Istanbul University, Istanbul Medical Faculty, Tıbbi Genetik Anabilim Dalı, Turkey 2006 - 2012
Undergraduate, Istanbul University, Istanbul Medical Faculty, Tıp Bölümü, Turkey 2000 - 2006

Dissertations

Expertise In Medicine, Frontonazal Displazi Olgularında Klinik Sınıflandırma ve Nörokristopati ile İlişkilendirilmiş Genlerin Araştırılması, Istanbul University, Istanbul Medical Faculty, Tıbbi Genetik Anabilim Dalı, 2012

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Medical Genetics, Life Sciences, Molecular Biology and Genetics, Genetic Disorders, Cytogenetic, Natural Sciences

Academic Titles / Tasks

Expert, Istanbul University, Istanbul Medical Faculty, Dahili Bilimler, 2013 - Continues

Courses

Genetik Danışma, Postgraduate, 2015 - 2016

Published journal articles indexed by SCI, SSCI, and AHCI

I. **The first case of Dyssegmental Dysplasia Rolland-Desbuquois type with a variant in HSPG2**
KALAYCI T., Balanda N., Ferreira C. R., Altunoglu U.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, no.SUPPL 1, pp.237, 2020 (SCI-Expanded)

II. **Heterozygous pathogenic variants in GLI1 are a common finding in isolated postaxial polydactyly A/B**
Palencia-Campos A., Martinez-Fernandez M., Altunoglu U., Soto-Bielicka P., Torres A., Marin P., Aller E., Senturk L., Berkoz O., Yildiran M., et al.
HUMAN MUTATION, vol.41, no.1, pp.265-276, 2020 (SCI-Expanded)

III. **Primary coenzyme Q10 Deficiency-6 (COQ10D6): Two siblings with variable expressivity of the renal phenotype**
Yildirim Z. N., Toksoy G., Uyguner O., Nayir A., Yavuz S., Altunoglu U., Turkkan O. N., Sevinc B., Gokcay G. F., Gunes D. K., et al.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.63, no.1, 2020 (SCI-Expanded)

IV. **Genetic Evaluation of Idiopathic Short Stature**
Karaman B., Bas F., Najafli A., Avci S., Al A. D. K., Toksoy G., Altunoglu U., Poyrazoglu S., Uyguner Z. O., Darendeliler F. F., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.91, pp.323, 2019 (SCI-Expanded)

V. **The Clinical Features and Effect of Growth Hormone Treatment in 3-M Syndrome Cases with Severe Growth Retardation**
Ozturk A. P., Altunoglu U., Ozturan E. K., Toksoy G., Poyrazoglu S., Bas F., Uyguner O., Darendeliler F. F.
HORMONE RESEARCH IN PAEDIATRICS, vol.91, pp.452, 2019 (SCI-Expanded)

VI. **Targeted Panel Gene Sequencing for Identification of Genetic Etiology of 46, XY Disorders of Sex Development**
Poyrazoglu S., Toksoy G., Aghayev A., Karaman B., Avci S., Altunoglu U., Yildiz M., Abali Z. Y., Bas F., Basaran S., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.91, pp.193, 2019 (SCI-Expanded)

VII. **MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctive cerebellar, ocular, craniofacial and genital features (COFG syndrome)**
Rad A., Rad A., Altunoglu U., Altunoglu U., Miller R., Miller R., Maroofian R., Maroofian R., James K. N., James K. N., et al.
JOURNAL OF MEDICAL GENETICS, vol.56, no.5, pp.332-339, 2019 (SCI-Expanded)

VIII. **Variants affecting diverse domains of MEPE are associated with two distinct bone disorders, a craniofacial bone defect and otosclerosis**
Schrauwen I., Valgaeren H., Tomas-Roca L., Sommen M., Altunoglu U., Wesdorp M., Beyens M., Fransen E., Nasir A., Vandeweyer G., et al.
GENETICS IN MEDICINE, vol.21, no.5, pp.1199-1208, 2019 (SCI-Expanded)

IX. **Prenatal Diagnosis and Management of Ectopia Cordis: Varied Presentation Spectrum.**
Turkyilmaz G., Avci S., Sivrikoz T., Erturk E., Altunoglu U., Turkyilmazlmaz S. E., Kalelioglu I. H., Has R., Yuksel A.
Fetal and pediatric pathology, vol.38, no.2, pp.127-137, 2019 (SCI-Expanded)

X. **Turkish Ectodermal Dysplasia Cohort: From Phenotype to Genotype in 17 Families.**
Güven Y., Bal E., Altunoglu U., Yücel E., Hadj-Rabıa S., Koruyucu M., Bahar T., Çıldır Ş., Aktören O., Bodemer C., et al.
Cytogenetic and genome research, vol.157, pp.189-196, 2019 (SCI-Expanded)

XI. **Manufacture of custom-made spectacles using three-dimensional printing technology**
Altinkurt E., Ceylan N., Altunoglu U., Turgut G. T.
Clinical and Experimental Optometry, 2019 (SCI-Expanded)

XII. **The oculoauriculofrontonasal syndrome: Further clinical characterization and additional evidence suggesting a nontraditional mode of inheritance**

Lehalle D., Altunoglu U., Bruel A., Assoum M., Duffourd Y., Masurel A., Baujat G., Bessieres B., Captier G., Edery P., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.176, no.12, pp.2740-2750, 2018 (SCI-Expanded)

XIII. **Prevalence, clinical characteristics and long-term outcomes of classical 11 β-hydroxylase deficiency (11BOHD) in Turkish population and novel mutations in CYP11B1 gene.**
Baş F., Toksoy G., Ergun-Longmire B., Uyguner Z. O., Abalı Z., Poyrazoğlu Ş., Karaman V., Avcı Ş., Altunoglu U., Bundak R., et al.
The Journal of steroid biochemistry and molecular biology, vol.181, pp.88-97, 2018 (SCI-Expanded)

XIV. **Osteoporosis-Pseudoglioma Syndrome (OPPG): Improvement of Osteoporosis on Biphosphonate Therapy**
Ozturan E. K., Altunoglu U., Kardelen A. D., Abali Z. Y., Avci S., Karabey H. K., Poyrazoglu S., Bas F., Darendeliler F.
HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.181-182, 2018 (SCI-Expanded)

XV. **Evaluation of Genetic Etiology in Patients with 46,XY Disorders of Sex Development: One Center Experience**
Aghayev A., Toksoy G., Poyrazoglu S., Karaman B., Avci S., Yildiz M., Abali Z. Y., Altunoglu U., Bas F., Darendeliler F., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.542, 2018 (SCI-Expanded)

XVI. **Clinical, Laboratory and Molecular Genetic Findings of Patients with 17 beta-Hydroxysteroid Dehydrogenase 3 Deficiency**
Poyrazoglu S., Toksoy G., Aghayev A., Karaman B., Avci S., Altunoglu U., Kardelen A. A. D., Ozturan E. K., Bas F., Basaran S., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.562, 2018 (SCI-Expanded)

XVII. **PROKR2 Mutations in Patients With Growth Hormone Deficiency and Multiple Pituitary Hormone Deficiency**
Najafli A., Bas F., Karaman B., Kardelen Al A. D., Toksoy G., Poyrazoglu S., Uyguner O., Avci S., Altunoglu U., Ozturan E. K., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.500, 2018 (SCI-Expanded)

XVIII. **Evaluation of Three Patients with 46,XY Gonadal Dysgenesis due to Desert Hedgehog Gene Mutations**
Poyrazoglu S., Aghayev A., Toksoy G., Karaman B., Avci S., Kardelen A. A. D., Ozturan E. K., Altunoglu U., Bas F., Basaran S., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.558-559, 2018 (SCI-Expanded)

XIX. **Clinical delineation of a subtype of frontonasal dysplasia with creased nasal ridge and upper limb anomalies: Report of six unrelated patients**
Lehalle D., Altunoglu U., Bruel A., Arnaud E., Blanchet P., Choi J., Desir J., Kilic E., Lederer D., Pinson L., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.173, no.12, pp.3136-3142, 2017 (SCI-Expanded)

XX. **CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays**
Windpassinger C., Piard J., Bonnard C., Alfadhel M., Lim S., Bisteau X., Blouin S., Ali N. B., Ng A. Y. J., Lu H., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.101, no.3, pp.391-403, 2017 (SCI-Expanded)

XXI. **PRIMARY COENZYME Q10 DEFICIENCY-6 (COQ10D6): CASE REPORT**
Yildirim Z. Y., Nayir A., Uyguner O., Toksoy G., Yavuz S., Altunoglu U., Turkkan O. N., Sevinc B., Gokcay G. F., Gunes D. K., et al.
PEDIATRIC NEPHROLOGY, vol.32, no.9, pp.1763, 2017 (SCI-Expanded)

XXII. **The Application of array CGH for Monogenic Disorders; Clinical and Molecular Cytogenetic Characterization of Twenty Patients**
Karaman B., Kayserili H., Najafli A., Altunoglu U., Kumbasar G., Avci S., Heidargholizadeh S., Uyguner Z. O., Satkin B. N., Toksoy G., et al.
MOLECULAR CYTOGENETICS, vol.10, 2017 (SCI-Expanded)

XXIII. **Homozygous mutation in NUP107 leads to microcephaly with steroid-resistant nephrotic condition similar to Galloway-Mowat syndrome**
Rosti R. O., Sotak B. N., Bielas S. L., Bhat G., Silhavy J. L., Aslanger A. D., Altunoglu U., Bilge I., Tasdemir M., Yzaguirrem

A. D., et al.
JOURNAL OF MEDICAL GENETICS, vol.54, no.6, pp.399-403, 2017 (SCI-Expanded)

XXIV. **Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies**
Acuna-Hidalgo R., Deriziotis P., Steehouwer M., Gilissen C., Graham S. A., van Dam S., Hoover-Fong J., Telegrafi A. B., Destree A., Smigiel R., et al.
PLOS GENETICS, vol.13, no.3, 2017 (SCI-Expanded)

XXV. **Cleidocranial dysplasia: Clinical, endocrinologic and molecular findings in 15 patients from 11 families.**
Dinçsoy B., DINÇKAN N., GÜVEN Y., BAŞ F., ALTUNOĞLU U., KUVVETLI S., Poyrazoğlu Ş., TOKSOY G., KAYSERILI H., UYGUNER Z. O.
European journal of medical genetics, vol.60, pp.163-168, 2017 (SCI-Expanded)

XXVI. **Biallelic mutations in the 3 ' exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing**
Lardelli R. M., Schaffer A. E., Eggens V. R. C., Zaki M. S., Grainger S., Sathe S., Van Nostrand E. L., Schlachetzki Z., Rosti B., Akizu N., et al.
NATURE GENETICS, vol.49, no.3, pp.457-464, 2017 (SCI-Expanded)

XXVII. **An Unusual Presentation of Kabuki Syndrome with Orbital Cysts, Microphthalmia, and Cholestasis with Bile Duct Paucity**
Boegershausen N., Altunoglu U., Beleggia F., Yigit G., Kayserili H., Nuernberg P., Li Y., Altmueller J., Wollnik B.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.170, no.12, pp.3282-3288, 2016 (SCI-Expanded)

XXVIII. **Holt-Oram syndrome because of the novel TBX5 mutation c.481A>C.**
Koçak E., ALTUNOĞLU U., Toksoy G., KAYSERILI H.
Clinical dysmorphology, vol.25, pp.192-4, 2016 (SCI-Expanded)

XXIX. **Mutation Update for Kabuki Syndrome Genes KMT2D and KDM6A and Further Delineation of X-Linked Kabuki Syndrome Subtype 2.**
BÖGERSHAUSEN N., GATINOIS V., RIEHMER V., KAYSERILI H., BECKER J., THOENES M., SIMSEK-KIPER P., BARAT-HOUARI M., ELCIOGLU N., WIECZOREK D., et al.
Human mutation, vol.37, pp.847-64, 2016 (SCI-Expanded)

XXX. **Mutations in CEP120 cause Joubert syndrome as well as complex ciliopathy phenotypes.**
ROOSING S., ROMANI M., ISRIE M., ROSTI R. Ö., MICALIZZI A., MUSAEV D., MAZZA T., AL-GAZALI L., Altunoglu U., BOLTSHAUSER E., et al.
Journal of medical genetics, vol.53, pp.608-15, 2016 (SCI-Expanded)

XXXI. **ALX4 related parietal foramina mimicking encephalocele in prenatal period.**
Saraç S., Altunoglu U., KALELIOGLU İ. H., YÜKSEL A., UYGUNER O., HAS R., KAYSERILI H.
Prenatal diagnosis, vol.36, pp.591-3, 2016 (SCI-Expanded)

XXXII. **Microcephaly, Dysmorphic Features, Corneal Dystrophy, Hairy Nipples, Underdeveloped Labioscrotal Folds, and Small Cerebellum in Four Patients**
Kayserili H., Kayserili H., Altunoglu U., Altunoglu U., YEŞİL G., Yesil G., Rosti R. O., Rosti R. O.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.170, no.6, pp.1391-1399, 2016 (SCI-Expanded)

XXXIII. **Genotypic and phenotypic presentation of TTR-FAP in Turkey**
Durmus H., ÇAKAR A., Atmaca M. M., Matur Z., Altunoglu U., PODA M., Ulas U. H., Oflazer P., Deymeer F., Parman Y.
EUROPEAN JOURNAL OF NEUROLOGY, vol.23, pp.341, 2016 (SCI-Expanded)

XXXIV. **The 3M Syndrome: A Cause of Pre- and Post-Natal Severe Growth Retardation**
Genens M., Altunoglu U., Bas F., Poyrazoglu S., Abali Z. Y., Bundak R., Darendeliler F. F.
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.465-466, 2016 (SCI-Expanded)

XXXV. **Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome.**
ATIK T., KOPARIR A., BADEMCI G., FOSTER J. 2., Altunoglu U., MUTLU G., BOWDIN S., ELCIOGLU N., TAYFUN G., ATIK S., et al.
Orphanet journal of rare diseases, vol.10, pp.128, 2015 (SCI-Expanded)

XXXVI. **Mutations in CDK5RAP2 cause Seckel syndrome.**

YIGIT G., BROWN K., KAYSERILI H., POHL E., CALIEBE A., ZAHNLEITER D., ROSSER E., BÖGERSHAUSEN N., UYGUNER Z. O., Altunoglu U., et al.
Molecular genetics & genomic medicine, vol.3, pp.467-80, 2015 (SCI-Expanded)

XXXVII. **De novo mutations in PLXND1 and REV3L cause Möbius syndrome.**
TOMAS-ROCA L., TSAALBI-SHTYLIK A., JANSEN J., SINGH M., EPSTEIN J., Altunoglu U., VERZIJL H., SORIA L., van B., ROSCIOLI T., et al.
Nature communications, vol.6, pp.7199, 2015 (SCI-Expanded)

XXXVIII. **Fraser syndrome due to mutations in GRIP1--clinical phenotype in two families and expansion of the mutation spectrum.**
SCHANZE D., KAYSERILI H., SATKıN B., Altunoglu U., ZENKER M.
American journal of medical genetics. Part A, pp.837-40, 2014 (SCI-Expanded)

XXXIX. **Mutations in WNT1 cause different forms of bone fragility.**
KEUPP K., BELEGGIA F., KAYSERILI H., BARNES A., STEINER M., SEMLER O., FISCHER B., YIGIT G., JANDA C., BECKER J., et al.
American journal of human genetics, vol.92, pp.565-74, 2013 (SCI-Expanded)

XL. **Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of α-dystroglycan.**
ROSCIOLI T., KAMSTEEG E., BUYSSE K., MAYSTADT I., van R., van d., van B., RIEMERSMA M., PFUNDT R., VISSERS L., et al.
Nature genetics, vol.44, pp.581-5, 2012 (SCI-Expanded)

Articles Published in Other Journals

I. **Clinical and Molecular Findings of Nine Cases with Tay- Sachs Disease From Turkiye**
ASLANGER A. D., GÜLEÇ Ç., KALAYCI T., Sengenc E., Avci S., Altunoglu U., KARAMAN V., TOKSOY G., KARACA M., Iscan A., et al.
MEDICAL JOURNAL OF BAKIRKOY, vol.19, no.2, pp.222-228, 2023 (ESCI)

II. **MOLECULAR ANALYSIS OF FGFR1-3, TWIST1, MSX2, POR, FREM1 AND RAB23 GENES IN SYNDROMIC AND NON-SYNDROMIC CRANIOSYNOSTOSIS CASES**
Karaman V., Toksoy G., Karaman B., Kayserili Karabey H., Basaran S., Altunoglu U., Avci S., Uyguner Z. O.
JOURNAL OF ISTANBUL FACULTY OF MEDICINE-ISTANBUL TIP FAKULTESI DERGISI, vol.82, no.2, pp.116-122, 2019 (ESCI)

III. **SENDROMİK VE NON-SENDROMİK KRANİYOSİNOSTOZ OLGULARINDA FGFR1-3, TWIST1, MSX2, POR, FREM1 VE RAB23 GENLERİNİN MOLEKÜLER ANALİZİ**
Karaman V., TOKSOY G., KARAMAN B., KAYSERİLİ KARABEY H., BAŞARAN S., ALTUNOĞLU U., UYGUNER Z. O.
İSTANBUL TIP FAKÜLTESİ DERGİSİ, vol.82, no.2, pp.9-10, 2019 (Peer-Reviewed Journal)

IV. **FETAL BRAIN SHRINKAGE: A RARE, MYSTIFYING ANOMALY**
Turkyilmaz G., Avci S., Altunoglu U., Erturk E., Canturk M., Sivrikoz T., Kalelioglu I., Has R., Yuksel A.
JOURNAL OF ISTANBUL FACULTY OF MEDICINE-ISTANBUL TIP FAKULTESI DERGISI, vol.82, no.2, pp.123-126, 2019 (ESCI)

V. **PRENATAL DIAGNOSIS OF ISOLATED SPLIT HAND/FOOT MALFORMATION**
Turkyilmaz G., Avci S., Erturk E., Sarac Sivrikoz T., Altunoglu U., Kalelioglu I., Has R., Yuksel A.
JOURNAL OF ISTANBUL FACULTY OF MEDICINE-ISTANBUL TIP FAKULTESI DERGISI, vol.81, no.1, pp.37-41, 2018 (ESCI)

VI. **CLINICAL CLASSIFICATION OF RADIAL RAY DEFECTS AND RESEARCH INTO ETIOPATHOGENESIS**
Avci S., Toksoy G., Bagirova G., Altunoglu U., Karaman B., Basaran S., Kayserili H., Uyguner Z. O.
JOURNAL OF ISTANBUL FACULTY OF MEDICINE-ISTANBUL TIP FAKULTESI DERGISI, vol.81, no.4, pp.127-138, 2018 (ESCI)

VII. **A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome.**
OUD M., BONNARD C., MANS D., Altunoglu U., TOHARI S., Ng A., ESKIN A., LEE H., RUPAR C., de W., et al.

Cilia, vol.5, pp.8, 2016 (Scopus)

VIII. **A classical phenotype of Duchenne muscular dystrophy in a girl with X; autosome translocation.**
UZUNHAN T., Altunoğlu U., YıLDıZ E., AYDıNLı N.
Journal of pediatric neurosciences, vol.9, pp.290-1, 2014 (ESCI)

Books & Book Chapters

I. **Tıbbi Genetikte Semiyoloji**
ALTUNOĞLU U.
in: Pediatrik Semiyoloji, Oğuz F., Editor, İstanbul Tıp Kitabevi, İstanbul, pp.321-348, 2016

Refereed Congress / Symposium Publications in Proceedings

I. **Genotype-Phenotype Correlation and Clinical Findings in 145 Patients with Congenital Adrenal Hyperplasia: Single Centre Experience**
Çilsaat G., Toksoy G., Baş F., Karaman B., Poyrazoğlu Ş., Uyguner Z., Başaran S., Altunoğlu U., Darendeliler F.
58 th Annual Meeting European Society for Paediatric Endocrinology (ESPE), Vienna, Austria, 20 - 22 September 2019, vol.1, no.1, pp.282

II. **Genetic Evaluation of Idiopathic Short Stature**
Karaman B., Baş F., Najaflı A., Şahin A., Toksoy G., Darendeliler F., Başaran S., Poyrazoğlu Ş., Altunoğlu U., Uyguner Z. O.
European Society for Paediatric Endocrinology (ESPE), Vienna, Austria, 19 - 21 September 2019, pp.323

III. **Targeted Panel Gene Sequencing for Identification of Genetic Etiology of 46, XY Disorders of Sex Development**
Poyrazoğlu Ş., TOKSOY G., Aghayev A., KARAMAN B., Şahin A., ALTUNOĞLU U., YAVAŞ A. Z., BAŞ F., BAŞARAN S., UYGUNER Z. O., et al.
European Society for Paediatric Endocrinology (ESPE), Basel, Switzerland, 20 - 22 September 2019, pp.193

IV. **Evaluation of Three Patients with 46,XY Gonadal Dysgenesis due to Desert Hedgehog Gene Mutations**
POYRAZOĞLU Ş., KARAMAN B., BAŞ F., Darendeliler F., TOKSOY G., BAŞARAN S., ALTUNOĞLU U., UYGUNER Z. O., Darendeliler F., TOKSOY G., et al.
57th Annual Meeting of the European Society for Paediatric Endocrinology, Atina, Greece, 27 - 29 September 2018, pp.558

V. **Copy-Number Variations of the Human Olfactory Receptor Gene Family in Patients with Macromastia and Prepubertal Gynecomastia**
BAŞ F., KARAMAN B., KARDELEN A., DARENDELILER F., TOKSOY G., BAŞARAN S., POYRAZOĞLU Ş., ALTUNOĞLU U., UYGUNER Z. O.
57th Annual Meeting of the European Society for Paediatric Endocrinology, Atina, Greece, 27 - 29 September 2018, pp.562

VI. **PrimerKoenzim Q10 eksikliği-6 (COQ10D6), Olgu Sunumu**
Yürük Yıldırım Z. N., Nayır A. N., Uyguner Z. O., Toksoy G., Yavuz S., Altunoğlu U., Türkkan Ö. N., Sevinç B., Gökçay G. F., Kürkçü D., et al.
4. Çocuk Nefroloji Olgu Panayırı, İzmir, Turkey, 3 - 04 November 2017, pp.4

VII. **Loss of function mutations in Carboxypeptidase D cause a new syndrome with recognizable dysmorphisms, lymphedema and sensorineural hearing loss.**
ALTUNOĞLU U.
17TH MANCHESTER DYSMORPHOLOGY CONFERENCE, Manchester, United Kingdom, 7 - 11 November 2016, pp.1

VIII. **Step by Step, Formation of Complex Chromosomal Rearrangements**
Satkın B. N., KARAMAN B., Yılmaz K., Altunoğlu U., BAŞARAN S.
European Human Genetic Congress, Germany, pp.110

IX. **Girl with left hemiatrophy reveals confined mosaicisms for r(13)in fibroblasts**
Altunoğlu U., KARAMAN B., BAŞARAN S., KAYSERİLİ H.
European Human Genetic Congress, Austria, pp.125

Supported Projects

KARAMAN B., BAŞARAN S., UYGUNER Z. O., ALTUNOĞLU U., AVCI Ş., TOKSOY G., KAYSERİLİ H., HEİDARGHOLİZADEH S., KUMBASAR G., SATKIN B. N., et al., Project Supported by Higher Education Institutions, The Application of array CGH for Monogenic Disorders Clinical and Molecular Cytogenetic Characterizations of Twenty Patients, 2017 - 2017
TOKSOY G., BAGİROVA G., ALTUNOĞLU U., PARMAN F. Y., UYGUNER Z. O., OFLAZER Z. P., AVCI Ş., YAPICI Z., AGHAYEV A., DURMUŞ TEKÇE H., et al., Project Supported by Higher Education Institutions, 32 novel pathogenic sequence variants in 253 DMD/BMD patients from Turkey, 2017 - 2017

Metrics

Publication: 58
Citation (WoS): 620
Citation (Scopus): 666
H-Index (WoS): 12
H-Index (Scopus): 13