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Education Information

Doctorate, Westfaelische Wilhelms-Universitaet Münster, Medizinische Fakultaet, Institute für Humangenetik, Germany 1983 - 1988
Postgraduate, Westfaelische Wilhelms-Universitaet Münster, Tıp Fakültesi, Humangenetik Enstitütüsü, Germany 1980 - 1983
Undergraduate, Istanbul University, Faculty Of Science, Depertment of Biology, Turkey 1974 - 1979

Foreign Languages

German, C1 Advanced
English, B2 Upper Intermediate

Dissertations

Doctorate, Sebebi Açıklanamayan İnfertilite ile İlişkili Genlerin Araştırılması, Istanbul University, Health Sciences Institute, İstanbul Tıp Fakültesi Bölümü, 2021
Expertise In Medicine, FETAL İSKELET DİSPLAZİLERİNİN KLİNİK, RADYOLOJİK BULGULAR ve MOLEKÜLER ETİYOPATOGENEZE GÖRE SINIFLANDIRILMASI, Istanbul University, Dahili Tıp Bilimleri, Tıbbi Genetik, 2016
Doctorate, Görünürde dengeli yapısal kromozom anomalilerinde submikroskobik dengesizliklerin a_CGH yöntemi ile araştırılması, Istanbul University, İstanbul Üniversitesi Sağlık Bilimleri Enstitüsü, Genetik Ad, 2014

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Medical Genetics

Academic Titles / Tasks

Professor, Istanbul University, Istanbul Medical Faculty, Division of Medical Sciences , 1996 - Continues

Academic and Administrative Experience

Head of Department, Istanbul University, Istanbul Medical Faculty, Division Of Medical Sciences , 2004 - Continues
Head of Department, Istanbul University, Istanbul Medical Faculty, Division Of Medical Sciences , 2004 - Continues

Jury Memberships

Appointment to Academic Staff-Assistant Professorship, Appointment Academic Staff, İstanbul Üniversitesi, August, 2022
Appointment to Academic Staff-Assistant Professorship, Appointment Academic Staff, İstanbul Üniversitesi, July, 2022
Appointment to Academic Staff-Assistant Professorship, Appointment Academic Staff, İstanbul Üniversitesi, October, 2021
Expertise In Medicine, Expertise In Medicine, İstanbul Üniversitesi, January, 2021
Appointment to Academic Staff-Assistant Professorship, Appointment Academic Staff, İstanbul Üniversitesi, August, 2020
Appointment to Academic Staff-Assistant Professorship, Appointment Academic Staff, Biruni Üniversitesi, February, 2020
Appointment to Academic Staff-Assistant Professorship, Appointment Academic Staff, İstanbul Üniversitesi, November, 2019
Appointment to Academic Staff-Assistant Professorship, Appointment Academic Staff, İstanbul Üniversitesi, November, 2019
Appointment to Academic Staff-Assistant Professorship, Appointment Academic Staff, Sanko Üniversitesi, September, 2019
Associate Professor Exam, Associate Professor Exam, Istanbul University, July, 2015
Associate Professor Exam, Associate Professor Exam, Istanbul University, July, 2015
Associate Professor Exam, Associate Professor Exam, Istanbul University, July, 2015
Associate Professor Exam, Associate Professor Exam, Istanbul University, January, 2015
Associate Professor Exam, Associate Professor Exam, Istanbul University, January, 2015
Associate Professor Exam, Associate Professor Exam, Istanbul University, January, 2015
Associate Professor Exam, Associate Professor Exam, Istanbul University, April, 2014
Associate Professor Exam, Associate Professor Exam, Istanbul University, April, 2014
Associate Professor Exam, Associate Professor Exam, Istanbul University, March, 2014
Associate Professor Exam, Associate Professor Exam, Istanbul University, March, 2014
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Associate Professor Exam, Associate Professor Exam, Istanbul University, January, 2010
Associate Professor Exam, Associate Professor Exam, Istanbul University, January, 2010
Associate Professor Exam, Associate Professor Exam, Istanbul University, January, 2010
Associate Professor Exam, Associate Professor Exam, Istanbul University, January, 2010
Associate Professor Exam, Associate Professor Exam, Istanbul University, September, 2008
Associate Professor Exam, Associate Professor Exam, Istanbul University, October, 2005
Associate Professor Exam, Associate Professor Exam, Istanbul University, October, 2005

Published journal articles indexed by SCI, SSCI, and AHCI

I. **Clinical and molecular characteristics of 26 fetuses with lethal multiple congenital contractures**
Turgut G. T., Altunoglu U., Gulec Ç., Sarac Sivrikoz T., Kalaycı T., Toksoy G., Avcı Ş., Yıldırım B. T., Sayın G. Y., Kalelioglu I. H., et al.
CLINICAL GENETICS, vol.105, no.6, pp.596-610, 2024 (SCI-Expanded)

II. **Association of antenatal evaluations with postmortem and genetic findings in the series of fetal osteogenesis imperfecta.**
Senturk L., Gulec Ç., Sarac Sivrikoz T., Kayserili H., Kalelioglu I. H., Avci S., Has R., Coucke P., Kalayci T., Wollnik B., et al.
Fetal diagnosis and therapy, 2024 (SCI-Expanded)

III. **Novel GALT variations and genetic spectrum in Turkish population with the correlation of genotype and phenotype.**
Kalay I., Gulec C., Balcı M. C., Toksoy G., Gokcay G., Basaran S., Demirkol M., Uyguner Z. O.
Annals of human genetics, vol.87, no.6, pp.285-294, 2023 (SCI-Expanded)

IV. **Predictive value of ultrasound in prenatal diagnosis of hypospadias: Hints for accurate diagnosis**
Uygur L., Sivrikoz T. S., Kalelioglu I. H., HAS R., Isguder C. K., Oktar T., Basaran S., Yuksel A.
Journal of Perinatal Medicine, vol.51, no.7, pp.932-939, 2023 (SCI-Expanded)

V. **Revisiting TOP2B-related phenotypes: Three new cases and literature review**
Çepni E., Börklü E., Avcı Ş., Kalaycı T., Eraslan S., Kayserili H.
Clinical Genetics, vol.104, no.2, pp.251-258, 2023 (SCI-Expanded)

VI. **A rare ring chromosome 21 abnormality is associated with azoospermia in two different phenotypically normal cases.**
Berkay E. G., Karaman B., Başaran S.
Systems biology in reproductive medicine, pp.1-7, 2023 (SCI-Expanded)

VII. **Fetal skeletal dysplasia cohort of a single tertiary referral center in Istanbul, Turkey.**
Kalayci T., Altunoglu U., Çorbacioglu Esmer A., Avcı Ş., Sarac Sivrikoz T., Karaman B., Kalelioğlu İ., Has R., Uyguner Z. O., Yüksel A., et al.
American journal of medical genetics. Part A, vol.191, no.2, pp.498-509, 2023 (SCI-Expanded)

VIII. **A new enrichment approach for candidate gene detection in unexplained recurrent pregnancy loss and implantation failure**
Berkay E. G., Şoroğlu C. V., Kalaycı T., Uyguner Z. O., Akçapınar G. B., Başaran S.
MOLECULAR GENETICS AND GENOMICS, vol.298, no.1, pp.253-272, 2023 (SCI-Expanded)

IX. **Clinical and genetic spectrum from a prototype of ciliopathy: Joubert syndrome**
Aksu Uzunhan T., Ertürk B., Aydın K., Ayaz A., Altunoğlu U., Yarar M. H., Gezdirici A., İÇAĞASIOĞLU D. F., Gökpınar İli E., UYANIK B., et al.
Clinical Neurology and Neurosurgery, vol.224, 2023 (SCI-Expanded)

X. **Clinical and bi-genomic DNA findings of patients suspected to have mitochondrial diseases**
GEDİKBAŞI A., TOKSOY G., KARACA M., GÜLEÇ Ç., BALCI M. C., Gunes D., Gunes S., ASLANGER A. D., ÜNVERENGİL G., KARAMAN B., et al.
FRONTIERS IN GENETICS, vol.14, 2023 (SCI-Expanded)

XI. **PROKR2 Mutations in Patients with Short Stature Who Have Isolated Growth Hormone Deficiency and Multiple Pituitary Hormone Deficiency**
Kardelen A. D., Najafli A., Baş F., Karaman B., Toksoy G., Poyrazoğlu Ş., Avcı Ş., Altunoğlu U., Yavaş Abalı Z., Öztürk A. P., et al.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.15, no.4, pp.338-347, 2023 (SCI-Expanded)

XII. **Clinical, Cytogenetic and Molecular Cytogenetic Outcomes of Cell-Free DNA Testing for Rare Chromosomal Anomalies**
BAŞARAN S., HAS R., KALELİOĞLU İ. H., Sivrikoz T. S., KARAMAN B., Kirgiz M., Dehgan T., KALAYCI T., SELÇUK B. Ş., Miny P., et al.
GENES, vol.13, no.12, 2022 (SCI-Expanded)

XIII. **Mutations in AR or SRD5A2 Genes: Clinical Findings, Endocrine Pitfalls, and Genetic Features of Children with 46,XY DSD.**
Akcan N., Uyguner Z. O., Bas F., Altunoglu U., Toksoy G., Karaman B., Avci S., Abali Z. Y., Poyrazoglu Ş., Aghayev A., et al.
Journal of clinical research in pediatric endocrinology, vol.14, no.2, pp.153-171, 2022 (SCI-Expanded)

XIV. **Functional loss of ubiquitin-specific protease 14 may lead to a novel distal arthrogryposis phenotype.**
Turgut G. T., Altunoglu U., Sivrikoz T. S., Toksoy G., Kalayci T., Avci S., Karaman B., Gulec C., Basaran S., Sayin G., et al.
Clinical genetics, vol.101, no.4, pp.421-428, 2022 (SCI-Expanded)

XV. **Prenatal sonographic and cytogenetic/molecular findings of 22q11.2 microdeletion syndrome in 48**

confirmed cases in a single tertiary center.
Sarac Sivrikoz T., Basaran S., Has R., Karaman B., Kalelioglu I. H., Kirgiz M., Altunoglu U., Yuksel A.
Archives of gynecology and obstetrics, vol.305, no.2, pp.323-342, 2022 (SCI-Expanded)

XVI. **Clinical and molecular genetic findings of hereditary Parkinson's patients from Turkey.**
Emekli I., Tepgeç F., Samancı B., Toksoy G., Hasanoğulları Kına G., Tüfekçioğlu Z., Başaran S., Bilgiç B., Gürvit I. H., Emre M., et al.
Parkinsonism & related disorders, vol.93, pp.35-39, 2021 (SCI-Expanded)

XVII. **Growth and relationship of phenotypic characteristics with gonadal pathology and tumour risk in patients with 45, X/46, XY mosaicism**
Poyrazoglu Ş., Bas F., Karaman B., Yildiz M., Başaran S., Darendeliler F. F.
CLINICAL ENDOCRINOLOGY, vol.94, no.6, pp.973-979, 2021 (SCI-Expanded)

XVIII. **Array-comparative Genomic Hybridization Results in Clinically Affected Cases with Apparently Balanced Chromosomal Rearrangements.**
Satkin N. B., Karaman B., Ergin S., Kayserili H., Kalelioglu I. H., Has R., Yuksel A., Basaran S.
Balkan journal of medical genetics : BJMG, vol.23, no.2, pp.25-34, 2021 (SCI-Expanded)

XIX. **Two cases with central precocious puberty caused by paternally inherited novel variants in DLK1gene**
Karaman V., Ozturan E. K., Bas F., Başaran S., Uyguner Z. O.
EUROPEAN JOURNAL OF HUMAN GENETICS, no.SUPPL 1, pp.213, 2020 (SCI-Expanded)

XX. **A case report of a rare nonsense ZP1 variant in a patient with oocyte maturation defect**
Berkay E. G., Karaman B., Toksoy G., Selçuk B. Ş., Uyguner Z. O., Başaran S.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.28, no.SUPPL 1, pp.155-156, 2020 (SCI-Expanded)

XXI. **Clinical and Molecular Characterization of Fanconi Anemia Patients in Turkey**
Toksoy G., Uludağ Alkaya D., Bagirova G., Avcl Ş., Aghayev A., Günes N., Altunoğlu U., Alanay Y., Başaran S., Berkay E. G., et al.
MOLECULAR SYNDROMOLOGY, vol.11, no.4, pp.183-196, 2020 (SCI-Expanded)

XXII. **Follow-Up Studies of cf-DNA Testing from 101 Consecutive Fetuses and Related Ultrasound Findings.**
Basaran S., Has R., Kalelioglu I. H., Karaman B., Kirgiz M., Dehgan T., Satkin B. N., Sivrikoz T. S., Yuksel A.
Ultraschall in der Medizin (Stuttgart, Germany : 1980), vol.41, pp.175-185, 2020 (SCI-Expanded)

XXIII. **TWIST1 Gene Expression as a Biomarker for Predicting Primary Doxorubicin Resistance in Breast Cancer.**
Demir S., Muslumanoglu M. H., Muslumanoglu M., Basaran S., Calay Z. Z., Aydiner A., Vogt U., Cakir T., Kadioglu H., Artan S.
Balkan journal of medical genetics : BJMG, vol.22, no.2, pp.25-30, 2019 (SCI-Expanded)

XXIV. **Clinical and Genetic Investigation of Premature Ovarian Insufficiency Cases from Turkey**
Oral E., Toksoy G., Sofiyeva N., Celik H. G., Karaman B., Basaran S., Azami A., Uyguner Z. O.
JOURNAL OF GYNECOLOGY OBSTETRICS AND HUMAN REPRODUCTION, vol.48, pp.817-823, 2019 (SCI-Expanded)

XXV. **Array-CGH Analizlerinde Saptanan De Novo Değişimlere Klinik Genetik Yaklaşım**
Kumbasar G., TOKSOY G., BAŞARAN S., KARAMAN B.
Gazi Medical Journal, vol.30, no.4, pp.361-364, 2019 (SSCI)

XXVI. **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)

XXVII. **Genotype-Phenotype Correlation and Clinical Findings in 145 Patients with Congenital Adrenal Hyperplasia: Single Centre Experience**
Cilsaat G., Toksoy G., Bas F., Karaman B., Poyrazoglu Ş., Uyguner O., Basaran S., Altinoglu U., Darendeliler F. F.
HORMONE RESEARCH IN PAEDIATRICS, vol.91, pp.282, 2019 (SCI-Expanded)

XXVIII. **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)

XXIX. **Mutation spectrum of 260 dystrophinopathy patients from Turkey and important highlights for genetic counseling**
Toksoy G., Durmus H., Aghayev A., Bagirova G., Rustemoglu B. S., Basaran S., Avci S., Karaman B., Parman Y., Altunoglu U., et al.
NEUROMUSCULAR DISORDERS, no.8, pp.601-613, 2019 (SCI-Expanded)

XXX. **OriginalArticleClinicalandGeneticInvestigationofPrematureOvarianInsufficiencyCasesfromTurkey**
ORAL E., TOKSOY G., SOFIYEVA N., Göksever H., KARAMAN B., BAŞARAN S., AZAMI A., BAŞARAN S.
INTERNATIONAL JOURNAL OF GYNECOLOGY & OBSTETRICS, vol.1580, pp.1-7, 2019 (SCI-Expanded)

XXXI. **NORMAL KARYOTİPLİ PATOLOJİK ULTRASON BULGUSU OLAN FETUSLARDA MLPA (MULTİPLEX LİGATİON-DEPENDENT PROBE AMPLİFİCATİON) UYGULAMALARI**
TOKSOY G., KARAMAN B., UYGUNER Z. O., YILMAZ K., HAS R., KAYSERİLİ H., MINY P., BAŞARAN S.
İstanbul Tıp Dergisi, vol.82, no.1, pp.2-3, 2019 (SCI-Expanded)

XXXII. **Author Correction: RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6.**
Szenker-Ravi E., Altunoglu U., Leushacke M., Bosso-Lefèvre C., Khatoo M., Thi T., Naert T., Noelanders R., Hajamohideen A., Beneteau C., et al.
Nature, vol.561, 2018 (SCI-Expanded)

XXXIII. **Pallister-Killian syndrome: clinical, cytogenetic and molecular findings in 15 cases**
Karaman B., Kayserili H., Ghanbari A., Uyguner Z. O., Toksoy G., Altunoglu U., Basaran S.
MOLECULAR CYTOGENETICS, vol.11, 2018 (SCI-Expanded)

XXXIV. **A Rare Cause of Congenital Adrenal Hyperplasia: Clinical and Genetic Findings and Follow-up Characteristics of Six Patients with 17-Hydroxylase Deficiency Including Two Novel Mutations**
Kardelen A. D., Toksoy G., Bas F., Abali Z. Y., Gencay G., Poyrazoglu S., Bundak R., Altunoglu U., Avci S., Najafli A., et al.
Journal of clinical research in pediatric endocrinology, vol.10, no.3, pp.206-215, 2018 (SCI-Expanded)

XXXV. **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)

XXXVI. **RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6**
Szenker-Ravi E., Altunoglu U., Leushacke M., Boss-Lefevre C., Khatoo M., Hong Thi Tran H. T. T., Naert T., Noelanders R., Hajamohideen A., Beneteau C., et al.
NATURE, vol.557, pp.564-584, 2018 (SCI-Expanded)

XXXVII. **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)

XXXVIII. **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)

XXXIX. **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)

XL. **Copy-Number Variations of the Human Olfactory Receptor Gene Family in Patients with Macromastia and Prepubertal Gynecomastia**
Bas F., Karaman B., Kardelen A. A. D., Heidargholizadeh S., Najafli A., Toksoy G., Poyrazoglu S., Yildiz M., Uyguner O.,

Basaran S., et al.
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XLI. **Cell-free DNA testing: is it reliable? A case report**
Erzincan S. G., Saying N. C., Inan C., Yuce M. A., Varol F. G., Basaran S.
CLINICAL AND EXPERIMENTAL OBSTETRICS & GYNECOLOGY, vol.45, no.6, pp.939-941, 2018 (SCI-Expanded)

XLII. **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)

XLIII. **The Role of Mosaicism in Discordant cf DNA Testing Results**
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XLIV. **A new hereditary congenital facial palsy case supports arg5 in HOX-DNA binding domain as possible hot spot for mutations.**
Uyguner Z. O., Toksoy G., Altunoglu U., Ozgur H., Basaran S., Kayserili H.
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XLV. **False positive and false negative results of cell free DNA testing**
Basaran S., Yuksel A., Has R., Kirgiz M., Dehgan T., Karaman B.
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XLVI. **Novel indel Mutation in the GDF5 Gene Is Associated with Brachydactyly Type C in a Four-Generation Turkish Family.**
UYGUNER Z. O., KOCAOĞLU M., TOKSOY G., Basaran S., KAYSERILI H.
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XLVII. **Array-CGH Findings of de novo Apparently Balanced Chromosomal Rearrangements in Phenotypically Affected 20 Cases**
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XLVIII. **Familial Microdeletion of 3 Mb at 22q11.2 With Unusual Phenotype**
Toksoy G., Satkin B. N., Kayserili H., Karaman B., Basaran S.
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XLIX. **Cardiovascular abnormalities in Williams syndrome: 20 years' experience in Istanbul**
Ergul Y., Nisli K., Kayserili H., Karaman B., Basaran S., Koca B., Aydogan U., Omeroglu R. N., Dindar A.
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L. **Evaluation of coronary artery abnormalities in Williams syndrome patients using myocardial perfusion scintigraphy and CT angiography**
Ergul Y., Nisli K., Kayserili H., Karaman B., Basaran S., Dursun M., Yilmaz E., Ergul N., Unal S. N., Dindar A.
CARDIOLOGY JOURNAL, vol.19, no.3, pp.301-308, 2012 (SCI-Expanded)

LI. **Mild Nasal Malformations and Parietal Foramina Caused by Homozygous ALX4 Mutations**
Kayserili H., Altunoglu U., Ozgur H., Basaran S., Uyguner Z. O.
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LII. **Haploinsufficiency of SHH gene caused by deletion of 7q36 -> qter: holoprosencephaly sequence in 5 cases**
Satkin B., Kayserili H., Kalelioglu I., Karaman B., Uyguner O., Has R., Yukse A., Basaran S.
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LIII. **A de novo complex chromosomal rearrangement involving chromosomes 2, 8 and 13 in a dysmorphic case with polysyndactyly**
Karaman B., Rosti R. O., Yilmaz K., Ozturk H., Kayserili H., Basaran S.
TURKISH JOURNAL OF PEDIATRICS, vol.51, no.6, pp.613-616, 2009 (SCI-Expanded)

LIV. **Molecular genetic screening of MBS1 locus on chromosome 13 for microdeletions and exclusion of FGF9, GSH1 and CDX2 as causative genes in patients with Moebius syndrome**

Uzumcu A., Karaman B., Toksoy G., Uyguner Z. O., Candan S., Eris H., Tatli B., Geçkinli B., Yuksel A., Kayserili H., et al.
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LV. **Mutational screening of BASP1 and transcribed processed pseudogene TP Psi g-BASP1 in patients with Mobius syndrome**
Uzumcu A., Candan S., Toksoy G., Uyguner Z. O., Karaman B., Eris H., Tatli B., Kayserili H., Yuksel A., Geçkinli B., et al.
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Üzümcü A., Candan Ş., Toksoy G., UYGUNER Z. O., KARAMAN B., Eriş H., TATLI B., KAYSERİLİ H., YÜKSEL A., Geçkinli B., et al.
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Nisli K., Oner N., Candan S., Kayserili H., Tansel T., Tireli E., Karaman B., Omeroglu R. E., Dindari A., Aydogan U., et al.
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LVIII. **The molecular mechanism underlying Roberts syndrome involves loss of ESCO2 acetyltransferase activity**
Gordillo M., Vega H., Trainer A. H., Hou F., Sakai N., Luque R., Kayserili H., Basaran S., Skovby F., Hennekam R. C. M., et al.
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LIX. **Corpus callosum agenesis in trisomy 8p11.23 and monosomy 4q34 because of maternal translocation**
Işık U., BAŞARAN S., Dehgan T., Apak M.
PEDIATRIC NEUROLOGY, vol.39, no.1, pp.55-57, 2008 (SCI-Expanded)

LX. **Angelman syndrome: clinical findings and follow-up data of 14 patients**
Kara B., Karaman B., Ozmen M., Rosti R. O., Caliskan M., Kayserili H., Basaran S.
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LXI. **CYP21 gene mutations in congenital adrenal hyperplasia: Genotype-phenotype correlation in Turkish children**
Bas F., Darendeliler F. F., Kayserili H., Uyguner O., Wollnik B., Saka N., Yuksel-Apak M., Basaran S., Bundak R., Gunoz H.
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Memberships / Tasks in Scientific Organizations

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Congress and Symposium Activities

15. Ulusal Tıbbi genetik Kongresi, Invited Speaker, Muğla, Turkey, 2022
4. Jinekoloji ve Obstetrikte Tartışmalı Konular Kongresi, Panelists, Muğla, Turkey, 2022
Ümraniye Pediatri Nadir Hastalıklar Sempozyumu, Session Moderator, İstanbul, Turkey, 2022
Fetal Tıp ve Perinatoloji Kongresi, Invited Speaker, Ankara, Turkey, 2022

Türkiye Maternal Fetal Tıp ve Perinatoloji Derneği Ultrasonografi Kongresi, Invited Speaker, İstanbul, Turkey, 2021
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Fetal Tıp ve Perinatoloji Kongresi, Invited Speaker, Ankara, Turkey, 2021
56. Nörogenetik Kongresi, Invited Speaker, İstanbul, Turkey, 2020
56. Nörogenetik Kongresi, Invited Speaker, İstanbul, Turkey, 2020
14. Ulusal Tıbbi Genetik Kongresi, Session Moderator, Ankara, Turkey, 2020
9. İstanbul Üniversitesi Kadın Doğum Günleri, Invited Speaker, İstanbul, Turkey, 2019
Türkiye Maternal Fetal Tıp Derneği Ultrasonografi Kursu, Invited Speaker, İstanbul, Turkey, 2019
42. Dreiländertreffen, SGUM, DEGUM, ÖGUM, Invited Speaker, Basel, Switzerland, 2018
13. Tıbbi Genetik Kongresi, Invited Speaker, Antalya, Turkey, 2018
13. Tıbbi Genetik Kongresi, Invited Speaker, Antalya, Turkey, 2018
11. Türkiye Maternal ve Fetal Tıp Ulusal Kongresi, Panelists, İstanbul, Turkey, 2018
TMFP Derneği Fetal Tıp ve Prenatal Tanı Kongresi, Invited Speaker, İstanbul, Turkey, 2018
IV. Nadir Hastalıklar Sempozyumu ve Nörogenetik, Invited Speaker, İstanbul, Turkey, 2018
Erciyes Gevher Nesibe Tıp Genetik Günleri, Invited Speaker, Kayseri, Turkey, 2018
Erciyes Tıp Genetik Günleri 2017, Invited Speaker, Kayseri, Turkey, 2018
13. Ege Tıp Genetik Çalıştayı Sitogenetik Uygulamaları Sempozyumu, Invited Speaker, İzmir, Turkey, 2017
2. Ege Endokrin Hastalıklar ve Genetik Sempozyumu, Invited Speaker, İzmir, Turkey, 2017
Fetal Tıp ve Prenatal Tanı Subgrup Toplatısı, Panelists, Ankara, Turkey, 2016