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Research Areas

Medicine, Health Sciences, Fundamental Medical Sciences, Biochemistry, Medical Biology, Life Sciences, Molecular Biology and Genetics, Genetic Disorders, Genetic Engineering, Genomics, Natural Sciences

Academic Titles / Tasks

Professor, Hacettepe University, Çocuk Sağlığı Enstitüsü, Pediatrik Temel Bilimler A.B.D., 2013 - Continues
Associate Professor, Hacettepe University, Çocuk Sağlığı Enstitüsü, Pediatrik Temel Bilimler A.B.D., 2007 - 2013
Associate Professor, Hacettepe University, Çocuk Sağlığı Enstitüsü, Pediatrik Temel Bilimler A.B.D., 2006 - 2007
Lecturer PhD, Hacettepe University, Çocuk Sağlığı Enstitüsü, Pediatrik Temel Bilimler A.B.D., 2006 - 2007
Specialization Student, Hacettepe University, Çocuk Sağlığı Enstitüsü, Pediatrik Temel Bilimler A.B.D., 1997 - 2003

Academic and Administrative Experience

Head of Department, Hacettepe University, Çocuk Sağlığı Enstitüsü, Pediatrik Temel Bilimler A.B.D., 2023 - Continues
Assistant Manager of Research and Application Center, Hacettepe University, Çocuk Sağlığı Enstitüsü, 2017 - 2022
Ethics Committee Member, Hacettepe University, Çocuk Sağlığı Enstitüsü, 2011 - 2019

Courses

MET-703 KALITSAL HASTALIKLARDA PROTEOMİK VE METABOLOMİK, Doctorate, 2016 - 2017
KHB601-Kök Hücre ve Hücresel Tedaviler:Temel Kavramlar, Postgraduate, 2016 - 2017
KHB602-Kök Hücre Yapısının Moleküler Esasları, Postgraduate, 2016 - 2017
MET-705 KALITSAL METABOLİK HASTALIKLARIN MOLEKÜLER TEMELİ, Doctorate, 2016 - 2017
MET-706 SEMİNER, Doctorate, 2016 - 2017
KHB-702 KÖK HÜCRE LABORATUVAR YÖNTEMLERİ I, Doctorate, 2016 - 2017
MET-702 METABOLİK HASTALIKLARDA LABORATUVAR UYGULAMALARI I, Doctorate, 2016 - 2017

Advising Theses

Özgül R. K., Nörometabolik hastalık nedeni olan mutasyonların drosophila melanogasterda modellenmesi ve sinaps fonksiyonu üzerine etkilerinin araştırılması, Doctorate, R.KARATEPE(Student), 2022
Özgül R. K., Kalıtsal metabolik hastalıklarda antioksidan yanıt ve otofajinin P62/NRF2/KEAP1 yolağı aracılığıyla araştırılması, Doctorate, N.VARDAR(Student), 2020
Özgül R. K., İn vitro kültür ortamında kemik iliği nişi modellenmesine yönelik çalışmalar, Doctorate, Y.ANASIZ(Student), 2018
Özgül R. K., Duman M., Fenilketonüri tanısı için kağıt-tabanlı mikroakışkan analitik cihaz geliştirilmesi, Postgraduate, P.KAZANCI(Student), 2018
Özgül R. K., Metabolom ve transkriptom profillemesi ile insan kemik iliğinde hematopoetik niş karakterizasyonu, Doctorate, S.AYHAN(Student), 2015
ÖZGÜL R. K., Nörogenetik hastalıklarda ekzom dizileme analizi ile aday genlerin belirlenmesi ve zebrafish modellerinin oluşturulması, Doctorate, A.OZANTÜRK(Student), 2015
ÖZGÜL R. K., Mukopolisakkaridoz Tip I (MPS I) hastalarında IDUA gen mutasyonlarının analizi, Postgraduate, N.ATÇEKEN(Student), 2013
ÖZGÜL R. K., Kalıtsal retina dejenerasyonlarında DNA mikroarray yöntemiyle yüksek ölçekli genom taraması, Doctorate, D.YÜCEL(Student), 2010
ÖZGÜL R. K., Denatüre edici yüksek performanslı sıvı kromatografisi yöntemi ile biyotinidaz eksikliği ve lizinürik protein intoleransı görülen hastalarda mutasyon taraması, Postgraduate, A.GÜZEL(Student), 2010
ÖZGÜL R. K., Mikroarray tabanlı yeniden dizileme sistemi ile Wilson hastalarında ATP7B geninde moleküler genetik analizler, Doctorate, A.YILMAZ(Student), 2010

Published journal articles indexed by SCI, SSCI, and AHCI

I. **TRAPPC6B biallelic variants cause a neurodevelopmental disorder with TRAPP II and trafficking disruptions**
Almousa H., Lewis S. A., Bakhtiari S., Nordlie S. H., Pagnozzi A., Magee H., Efthymiou S., Heim J. A., Cornejo P., Zaki M. S., et al.
Brain : a journal of neurology, vol.147, no.1, pp.311-324, 2024 (SCI-Expanded)

II. **Predictors of eventual requirement of phenylalanine-restricted diet in young infants with phenylalanine hydroxylase deficiency initially managed with sapropterin monotherapy**
ÇIKI K., YILDIZ Y., KAHRAMAN A. B., ÖZGÜL R. K., COŞKUN T., DURSUN A., TOKATLI A., SİVRİ S.
Molecular Genetics and Metabolism, vol.140, no.3, 2023 (SCI-Expanded)

III. **A big picture of the mitochondria-mediated signals: From mitochondria to organism**
Vardar Acar N., ÖZGÜL R. K.
Biochemical and Biophysical Research Communications, vol.678, pp.45-61, 2023 (SCI-Expanded)

IV. **THE BRIDGE BETWEEN CELL SURVIVAL AND CELL DEATH: REACTIVE OXYGEN SPECIES-MEDIATED CELLULAR STRESS**
Acar N. V., ÖZGÜL R. K.
EXCLI Journal, vol.22, pp.520-555, 2023 (SCI-Expanded)

V. **Investigation of androgen receptor gene CAG repeat length polymorphism in pubertal gynecomastia**
Duzceker Y., Pehlivanturk-Kizilkan M., AKGÜL S., ÖZGÜL R. K., KANBUR N., DERMAN O.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.35, no.3, pp.349-354, 2022 (SCI-Expanded)

VI. **An investigation of different intracellular parameters for Inborn Errors of Metabolism: Cellular stress, antioxidant response and autophagy**
Vardar Acar N., DURSUN A., Aygün D., Gürses Cila H. E., LAY İ., GÜLBAKAN B., ÖZGÜL R. K.
Free Radical Biology and Medicine, vol.179, pp.190-199, 2022 (SCI-Expanded)

VII. **Biallelic mutations in ELFN1 gene associated with developmental and epileptic encephalopathy and joint laxity**
DURSUN A., YALNIZOĞLU D., Yilmaz D. Y., Oguz K. K., GÜLBAKAN B., KOŞUKCU C., AKAR H. T., Kahraman A. B., Acar N. V., GÜNBEY C., et al.

EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.64, no.11, 2021 (SCI-Expanded)

VIII. **Perplexing Etiology of Hyperphenylalaninemia in an Infant Referred via Newborn Screening.**
Çıkı K., Akar H. T., Özgül R. K., Gülbakan B., Yıldız Y.
Clinical chemistry, vol.67, no.10, pp.1428-1431, 2021 (SCI-Expanded)

IX. **Homozygous missense VPS16 variant is associated with a novel disease, resembling mucopolysaccharidosis-plus syndrome in two siblings**
Yıldız Y., Koşukcu C., Aygün D., Akçaboy M., Öztek Çelebi F. Z., Taşcı Yıldız Y., Şahin G., Aytekin C., Yüksel D., Lay İ., et al.
CLINICAL GENETICS, vol.100, no.3, pp.308-317, 2021 (SCI-Expanded)

X. **DNACJ12 deficiency in patients with unexplained hyperphenylalaninemia: two new patients and a novel variant**
Çıkı K., Yıldız Y., Yücel Yılmaz D., Pektaş E., Tokatlı A., Özgül R. K., Sivri H. S., Dursun A.
METABOLIC BRAIN DISEASE, vol.36, no.6, pp.1405-1410, 2021 (SCI-Expanded)

XI. **Clinical and molecular characteristics of carnitine-acylcarnitine translocase deficiency with c.270delC and a novel c.408C>A variant**
BİLGİNER GÜRBÜZ B., YÜCEL YILMAZ D., ÖZGÜL R. K., KOŞUKCU C., DURSUN A., SİVRİ H. S., COŞKUN T., TOKATLI A.
TURKISH JOURNAL OF PEDIATRICS, vol.63, no.4, pp.691-696, 2021 (SCI-Expanded)

XII. **Characterization of human bone marrow niches with metabolome and transcriptome profiling**
AYHAN S., NEMUTLU E., Cetinkaya D. U., KIR S., ÖZGÜL R. K.
JOURNAL OF CELL SCIENCE, vol.134, no.6, 2021 (SCI-Expanded)

XIII. **Autoinflammation in addition to combined immunodeficiency: SLC29A3 gene defect**
Cagdas D. N., Surucu N., TAN Ç., ÖZGÜL R. K., Akkaya-Ulum Y. Z., Aydinoglu A. T., Aytac Ş. S., GÜMRÜK F., Balci-Hayta B., Balci-Peynircioglu B., et al.
MOLECULAR IMMUNOLOGY, vol.121, pp.28-37, 2020 (SCI-Expanded)

XIV. **Complement Factor I Gene Polymorphism in a Turkish Age-Related Macular Degeneration Population**
Bezci Aygun F., KADAYIFÇILAR S., ÖZGÜL R. K., Eldem B.
OPHTHALMOLOGICA, vol.243, no.3, pp.187-194, 2020 (SCI-Expanded)

XV. **Presentation of 14 alkaptonuria patients from Turkey**
AKBABA A. İ., ÖZGÜL R. K., DURSUN A.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.33, no.2, pp.289-294, 2020 (SCI-Expanded)

XVI. **Detection of allele frequencies of common c. 511C > T and c.625G > A variants in the ACADS gene in the Turkish population**
Kilic M., Erguner B., KOŞUKCU C., ÖZGÜL R. K.
TURKISH JOURNAL OF PEDIATRICS, vol.62, no.1, pp.19-23, 2020 (SCI-Expanded)

XVII. **Exon 2 deletion represents a common mutation in Turkish patients with fructose-1,6-bisphosphatase deficiency**
Kilic M., Kasapkara C. S., Yilmaz D. Y., Ozguel R. K.
METABOLIC BRAIN DISEASE, vol.34, no.5, pp.1487-1491, 2019 (SCI-Expanded)

XVIII. **Clinical highlights of a very rare phospolipid remodeling disease due to MBOAT7 gene defect**
DURSUN A., YALNIZOĞLU D., ÖZGÜL R. K., Oguz K. K., Yucel-Yilmaz D.
AMERICAN JOURNAL OF MEDICAL GENETICS PART B-NEUROPSYCHIATRIC GENETICS, 2019 (SCI-Expanded)

XIX. **Expanding the phenotype of phospholipid remodelling disease due to MBOAT7 gene defect**
YALNIZOĞLU D., ÖZGÜL R. K., Oguz K. K., Ozer B., Yucel-Yilmaz D., Gurbuz B., Serdaroglu E., Erol I., Topcu M., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.42, no.2, pp.381-388, 2019 (SCI-Expanded)

XX. **Reply to 'contribution of the MRPS22 variant and a down mosaic to the phenotype'**
Kilic M., Kilic E., Yilmaz D. Y., ÖZGÜL R. K.
METABOLIC BRAIN DISEASE, vol.33, no.6, pp.1779-1780, 2018 (SCI-Expanded)

XXI. **Immunodeficiency in a Child with Alstrom Syndrome**
Ozdemir T. R., Karaca N. E., Marshall J. D., KÜTÜKÇÜLER N., AKSU G., ÖZGÜL R. K., Ozanturk A., IŞIK E., Akgun B., Ozdemir H. H., et al.

INDIAN JOURNAL OF PEDIATRICS, vol.85, no.10, pp.924-926, 2018 (SCI-Expanded)

XXII. **Five novel ALMS1 gene mutations in six patients with Alstrom syndrome**
Kilinc S., Yucel-Yilmaz D., Ardagil A., Apaydin S., Valverde D., ÖZGÜL R. K., Guven A.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.31, no.6, pp.681-687, 2018 (SCI-Expanded)

XXIII. **Clinical phenotype of hereditary spastic paraplegia due to KIF1C gene mutations across life span**
Yucel-Yilmaz D., Yucesan E., YALNIZOĞLU D., Oguz K. K., Sagiroglu M. S., Ozbek U., Serdaroglu E., Bilgic B., Erdem S., Iseri S. A. U., et al.
BRAIN & DEVELOPMENT, vol.40, no.6, pp.458-464, 2018 (SCI-Expanded)

XXIV. **The genotypic and phenotypic spectrum of MTO1 deficiency**
O'Byrne J. J., Tarailo-Graovac M., Ghani A., Champion M., Deshpande C., DURSUN A., ÖZGÜL R. K., Freisinger P., Garber I., Haack T. B., et al.
MOLECULAR GENETICS AND METABOLISM, vol.123, no.1, pp.28-42, 2018 (SCI-Expanded)

XXV. **Genotypic-phenotypic features and enzyme replacement therapy outcome in patients with mucopolysaccharidosis VI from Turkey**
Kilic M., DURSUN A., COŞKUN T., TOKATLI A., ÖZGÜL R. K., YUCEL-YILMAZ D., Karaca M., Dogru D., ALEHAN D., KADAYIFÇILAR S., et al.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.173, no.11, pp.2954-2967, 2017 (SCI-Expanded)

XXVI. **A patient with mitochondrial disorder due to a novel mutation in MRPS22**
Kilic M., Oguz K., Kilic E., YÜKSEL D., DEMİRCİ H., SAĞIROĞLU M. S., Yucel-Yilmaz D., ÖZGÜL R. K.
METABOLIC BRAIN DISEASE, vol.32, no.5, pp.1389-1393, 2017 (SCI-Expanded)

XXVII. **A New Chapter for Mesenchymal Stem Cells: Decellularized Extracellular Matrices**
Anasiz Y., ÖZGÜL R. K., Uckan-Cetinkaya D.
STEM CELL REVIEWS AND REPORTS, vol.13, no.5, pp.587-597, 2017 (SCI-Expanded)

XXVIII. **Novel and prevalent CYP11B1 gene mutations in Turkish patients with 11-beta hydroxylase deficiency**
KANDEMİR N., Yilmaz D. Y., GÖNÇ E. N., Ozon A., ALİKAŞİFOĞLU A., DURSUN A., ÖZGÜL R. K.
JOURNAL OF STEROID BIOCHEMISTRY AND MOLECULAR BIOLOGY, vol.165, pp.57-63, 2017 (SCI-Expanded)

XXIX. **A probable new syndrome with the storage disease phenotype caused by the VPS33A gene mutation**
DURSUN A., YALNIZOĞLU D., Gerdan O. F., Yucel-Yilmaz D., Sagiroglu M. S., YUKSEL B., GUCER S., Sivri S., ÖZGÜL R. K.
CLINICAL DYSMORPHOLOGY, vol.26, no.1, pp.1-12, 2017 (SCI-Expanded)

XXX. **Discovery of biomarkers in rare diseases: innovative approaches by predictive and personalized medicine**
GÜLBAKAN B., ÖZGÜL R. K., YÜZBAŞIOĞLU A., Kohl M., Deigner H., ÖZGÜÇ M.
EPMA JOURNAL, vol.7, 2016 (SCI-Expanded)

XXXI. **Allergy-specific Phenome-Wide Association Study for Immunogenes in Turkish Children**
Karaca S., Civelek E., Karaca M., ŞAHİNER Ü. M., ÖZGÜL R. K., Kocabas C. N., Polimanti R., ŞEKEREL B. E.
SCIENTIFIC REPORTS, vol.6, 2016 (SCI-Expanded)

XXXII. **Haplotype analysis of non-HLA immunogenetic loci in Turkish and worldwide populations**
Karaca S., Karaca M., Civelek E., ÖZGÜL R. K., ŞEKEREL B. E., Polimanti R.
GENE, vol.587, no.2, pp.132-136, 2016 (SCI-Expanded)

XXXIII. **Evaluation and identification of IDUA gene mutations in Turkish patients with mucopolysaccharidosis type I**
Atceken N., ÖZGÜL R. K., Yilmaz D. Y., TOKATLI A., COŞKUN T., SİVRİ H. S., DURSUN A., Karaca M.
TURKISH JOURNAL OF MEDICAL SCIENCES, vol.46, no.2, pp.404-408, 2016 (SCI-Expanded)

XXXIV. **Sapropterin dihydrochloride treatment in Turkish hyperphenylalaninemic patients under age four**
ÖZLEM U., GÖKMEN ÖZEL H., COŞKUN T., ÖZGÜL R. K., YÜCEL YILMAZ D., BURCU H., TOKATLI A., DURSUN A., SİVRİ H. S.
Turkish Journal Of Pediatrics, pp.213-218, 2015 (SCI-Expanded)

XXXV. **MCP1 2518 A/G polymorphism affects progression of childhood focal segmental glomerulosclerosis**
Besbas N., KALYONCU M., Cil O., ÖZGÜL R. K., Bakkaloglu A., ÖZALTIN F.

RENAL FAILURE, vol.37, no.9, pp.1435-1439, 2015 (SCI-Expanded)

XXXVI. **Detection of biotinidase gene mutations in Turkish patients ascertained by newborn and family screening**
KARACA M., ÖZGÜL R. K., ÜNAL O., Yucel-Yilmaz D., KILIÇ M., Hismi B., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S.
EUROPEAN JOURNAL OF PEDIATRICS, vol.174, no.8, pp.1077-1084, 2015 (SCI-Expanded)

XXXVII. **Ailevi Hiperkolesterolemili Hastaların Mutasyon Analiz Sonuçlarının Simone-Broome Kriterleriyle Değerlendirilmesi**
AYKAN H. H., ÖZGÜL R. K., Güzel A., COŞKUN T., DURSUN A.
Türkiye Çocuk Hastalıkları Dergisi, vol.9, no.3, pp.176-183, 2015 (SCI-Expanded)

XXXVIII. **Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction**
AKIZU N., CANTAGREL V., ZAKI M. S., Al-Gazali L., WANG X., ROSTI R. O., DIKOGLU E., GELOT A. B., ROSTI B., VAUX K. K., et al.
NATURE GENETICS, vol.47, no.5, pp.528-536, 2015 (SCI-Expanded)

XXXIX. **Polymorphisms in FAS and CASP8 genes may contribute to the development of ALPS phenotype: A study in 25 patients with probable ALPS**
TAN Ç., ÖZGÜL R. K., CAGDAS-AYVAZ D. N., TEZCAN I., SANAL O.
Turkish Journal of Pediatrics, vol.57, no.2, pp.141-145, 2015 (SCI-Expanded)

XL. **Retinitis pigmentosa caused by mutations in the ciliary MAK gene is relatively mild and is not associated with apparent extra-ocular features**
van Huet R. A. C., Siemiatkowska A. M., ÖZGÜL R. K., Yucel D., Hoyng C. B., Banin E., Blumenfeld A., ROTENSTREICH Y., RIEMSLAG F. C. C., DEN HOLLANDER A. I., et al.
ACTA OPHTHALMOLOGICA, vol.93, no.1, pp.83-94, 2015 (SCI-Expanded)

XLI. **The phenotypic and molecular genetic spectrum of Alstrom syndrome in 44 Turkish kindreds and a literature review of Alstrom syndrome in Turkey**
Ozanturk A., Marshall J. D., Collin G. B., Duzenli S., Marshall R. P., Candan S., Tos T., Esen I., Taskesen M., Cayir A., et al.
JOURNAL OF HUMAN GENETICS, vol.60, no.1, pp.1-9, 2015 (SCI-Expanded)

XLII. **Two Turkish siblings with MEGDEL syndrome due to novel SERAC1 genemutation.**
ÜNAL Ö., ÖZGÜL R. K., YÜCEL YILMAZ D., YALNIZOĞLU D., TOKATLI A., SİVRİ H. S., HİŞMİ B., COŞKUN T., DURSUN A.
Turkish Journal Of Pediatrics, vol.57, pp.388-393, 2015 (SCI-Expanded)

XLIII. **Genome-Wide Homozygosity Mapping in Families with Leber Congenital Amaurosis Identifies Mutations in AIPL1 and RDH12 Genes**
Yucel-Yilmaz D., TARLAN B., KIRATLI H., ÖZGÜL R. K.
DNA AND CELL BIOLOGY, vol.33, no.12, pp.876-883, 2014 (SCI-Expanded)

XLIV. **Phenotypic and genotypic spectrum of Turkish patients with isovaleric acidemia**
ÖZGÜL R. K., Karaca M., Kilic M., KUCUK O., YUCEL-YILMAZ D., UNAL O., HISMI B., Aliefendioglu D., SIVRI S., TOKATLI A., et al.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.57, no.10, pp.596-601, 2014 (SCI-Expanded)

XLV. **Dursun Syndrome Due to G6PC3 Gene Defect has a Fluctuating Pattern in All Blood Cell Lines**
ÖZGÜL R. K., YUCEL-YILMAZ D., DURSUN A.
JOURNAL OF CLINICAL IMMUNOLOGY, vol.34, no.3, pp.265-266, 2014 (SCI-Expanded)

XLVI. **High prevalence of cerebral venous sinus thrombosis (CVST) as presentation of cystathionine beta-synthase deficiency in childhood: Molecular and clinical findings of Turkish probands**
Karaca M., Hismi B., ÖZGÜL R. K., Karaca S., Yilmaz D. Y., COŞKUN T., SİVRİ H. S., TOKATLI A., DURSUN A.
GENE, vol.534, no.2, pp.197-203, 2014 (SCI-Expanded)

XLVII. **Isovaleric Acidemia Presenting as Diabetic Ketoacidosis: A Case Report**
Kilic M., Kaymaz N., ÖZGÜL R. K.
JOURNAL OF CLINICAL RESEARCH IN PEDIATRIC ENDOCRINOLOGY, vol.6, no.1, pp.59-61, 2014 (SCI-Expanded)

XLVIII. **Cobalamin C defect: a patient of late-onset type with homozygous p. R132*mutation**

KILIÇ M., ÖZGÜL R. K., DURSUN A., TOKATLI A., Kalkanoglu-Sivri H. S., Anlar B., Fowler B., COŞKUN T.
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XLIX. **Galactosemia in the Turkish population with a high frequency of Q188R mutation and distribution of Duarte-1 and Duarte-2 variations**
Oezgul R. K., Guezel-Ozantuerk A., Duendar H., Yuecel-Yilmaz D., COŞKUN T., Sivri S., Aydogdu S., Tokatli A., DURSUN A.
JOURNAL OF HUMAN GENETICS, vol.58, no.10, pp.675-678, 2013 (SCI-Expanded)

L. **Molecular and clinical evaluation of Turkish patients with lysinuric protein intolerance**
Guzel-Ozanturk A., ÖZGÜL R. K., Unal O., Hismi B., Aydin H. I., Sivri S., TOKATLI A., COŞKUN T., Aksoz E., DURSUN A.
GENE, vol.521, no.2, pp.293-295, 2013 (SCI-Expanded)

LI. **Dynamics of the Rhomboid-like Protein RHBDD2 Expression in Mouse Retina and Involvement of Its Human Ortholog in Retinitis Pigmentosa**
Ahmedli N. B., Gribanova Y., Njoku C. C., Naidu A., Young A., Mendoza E., Yamashita C. K., ÖZGÜL R. K., Johnson J. E., Fox D. A., et al.
JOURNAL OF BIOLOGICAL CHEMISTRY, vol.288, no.14, pp.9742-9754, 2013 (SCI-Expanded)

LII. **Atypical presentation and a novel mutation in ALMS1: implications for clinical and molecular diagnostic strategies for Alstrom syndrome**
Tasdemir S., Guzel-Ozanturk A., Marshall J. D., Collin G. B., ÖZGÜL R. K., Narin N., DÜNDAR M., Naggert J. K.
CLINICAL GENETICS, vol.83, no.1, pp.96-98, 2013 (SCI-Expanded)

LIII. **Association of CFH Y402H Polymorphism with Both Forms of Advanced Age-Related Macular Degeneration in Turkish Patients**
Yucel D., YILMAZ M., DURUKAN A. H., ÖZGÜL R. K.
OPHTHALMIC GENETICS, vol.33, no.3, pp.144-149, 2012 (SCI-Expanded)

LIV. **MOLECULAR CHARACTERISATION OF BIOTINIDASE GENE MUTATIONS IN TURKISH PATIENTS; AN UPDATE OF THE RESULTS**
Karaca M., Yucel D., Unal O., Guzel A., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S., ÖZGÜL R. K.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)

LV. **THE 48-HOUR TETRAHYDOBIOPTERIN (BH4) LOADING TEST AND LONG-TERM OUTCOME OF PATIENTS WITH BH4 TREATMENT IN TURKISH PHENYLKETONURIA (PKU) PATIENTS**
Unal O., Gokmen-Ozel H., Coskun T., Hismi B., Tokatli A., Dursun A., Ozgul R. K., Sivri H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)

LVI. **OCTN2 GENE MUTATIONS IN TURKISH PATIENTS WITH PRIMARY CARNITINE DEFICIENCY**
Yucel-Yilmaz D., Ersoy M., Candan S., Balci M., KILIÇ M., Gokcay G., DURSUN A., ÖZGÜL R. K.
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LVII. **Microarray based mutational analysis of patients with methylmalonic acidemia: Identification of 10 novel mutations**
Dundar H., ÖZGÜL R. K., Guzel-Ozanturk A., Dursun A., Sivri S., Aliefendioglu D., Coskun T., Tokatli A.
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