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Education Information

Postgraduate, Istanbul University, Spain 2022 - Continues
Doctorate, Istanbul University, Health Sciences Institute, İstanbul Tıp Fakültesi Bölümü, Turkey 2014 - 2022
Post Doctorate of Medicine, Istanbul University, Istanbul Medical Faculty, Division Of Medical Sciences , Turkey 2011 - 2014
Expertise In Medicine, Istanbul University, Istanbul Medical Faculty, Division Of Medical Sciences , Turkey 2006 - 2011

Foreign Languages

English, B2 Upper Intermediate

Certificates, Courses and Trainings

Education Management and Planning, ORPHEUS SÜRECİ VE DANIŞMANLIK EĞİTİMİ KURSU, İstanbul Üniversitesi, 2023
Health&Medicine, İyi klinik uygulamalar, Brookwood Global, 2021
Health&Medicine, Eğiticinin eğitimi, İstanbul Üniversitesi, 2020

Dissertations

Doctorate, NUTRITIONAL THERAPY APPLIED IN HEREDITARY METABOLISM DISEASES IN TERMS OF NON-COMMUNICATIVE CHRONIC DISEASES ASSESSMENT OF RISK FACTORS AND SOLUTION SUGGESTIONS, Istanbul University, Health Sciences Institute, İstanbul Tıp Fakültesi Bölümü, 2023
Expertise In Medicine, yağ asidi oksidasyon bozukluklarında kardiyolojik değerlendirme, Istanbul University, Istanbul Medical Faculty, Çocuk Sağlığı Ve Hastalıkları Anabilim Dalı, 2011

Research Areas

Health Sciences

Academic Titles / Tasks

Assistant Professor, Istanbul University, Istanbul Medical Faculty, Division Of Medical Sciences , 2020 - Continues
Lecturer PhD, Istanbul University, Istanbul Medical Faculty, Division Of Medical Sciences , 2017 - Continues

Courses

Case studies in inherited metabolic diseases, Undergraduate, 2023 - 2024
Healthy eating principles, Undergraduate, 2023 - 2024
Case examples in inherited metabolic diseases, Undergraduate, 2023 - 2024
Problems in feeding babies and young children, Undergraduate, 2023 - 2024
INTERACTIVE CASE DISCUSSION/ Vitamin B12 Deficiency, Undergraduate, 2023 - 2024
INTERACTIVE CASE DISCUSSION Feeding of Infant and Child, Undergraduate, 2023 - 2024
Complementary Feeding, Undergraduate, 2023 - 2024
The principles of healty nutrition, Undergraduate, 2023 - 2024
INTERACTIVE CASE DISCUSSION Common Congenital Metabolic Diseases, Undergraduate, 2023 - 2024
Definitions in congenital metabolic diseases, Undergraduate, 2023 - 2024
INTERACTIVE CASE DISCUSSION Principles of infant nutrition, Undergraduate, 2023 - 2024
Problems in feeding babies and young children, Undergraduate, 2023 - 2024
INTERACTIVE CASE DISCUSSION Frequent Inborn Errors of Metabolism, Undergraduate, 2023 - 2024
breastfeeding, Undergraduate, 2023 - 2024
definitions in inborn errors of metabolism, Undergraduate, 2023 - 2024
INTERACTIVE CASE DISCUSSION Approach to B12 Deficiency with Case Examples, Undergraduate, 2023 - 2024
Eating disorders in children, Undergraduate, 2023 - 2024
Breast Feeding, Undergraduate, 2023 - 2024
Functional Disorders of Organelles and Clinical Reflections, Associate Degree, 2023 - 2024

Jury Memberships

Doctoral Examination, Doctoral Examination, İstanbul Üniversitesi, December, 2023
Doctorate, Doctorate, İstanbul Üniversitesi, September, 2023
Doctorate, Doctorate, İstanbul Üniversitesi, September, 2023
Doctorate, Doctorate, İstanbul Üniversitesi, September, 2023
Doctoral Examination, Doctoral Examination, İstanbul Üniversitesi, September, 2023
Doctorate, Doctorate, İstanbul Üniversitesi, June, 2023

Published journal articles indexed by SCI, SSCI, and AHCI

I. **Clinical Presentation and Molecular Characterization of 3 Patients with Vici Syndrome: Two Novel Variants in the EPG5 Gene**
Selamioğlu A., Doğan B. Y., Balcl M. C., Kalaycl T., Karaca M., Ak B., Durmuş A., Körbeyli H. K., Gökçay G.
Molecular Syndromology, 2024 (SCI-Expanded)

II. **A different perspective into clinical symptoms in CPT I deficiency.**
Balci M. C., Karaca M., Selamioglu A., Korbeyli H. K., Durmus A., Ak B., Kozanoglu T., Gokcay G. F.
Molecular genetics and metabolism reports, vol.38, pp.101032, 2023 (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. **Evaluation of the risk factors for noncommunicable diseases in patients with inborn errors of amino acid metabolism receiving nutrition therapy.**
Balci M. C., Karaca M., Yesil A., Selamioglu A., Korbeyli H. K., Durmus A., Ak B., Kozanoglu T., Hacioglu I., Gokcay G. F.
Journal of pediatric endocrinology & metabolism : JPEM, 2023 (SCI-Expanded)

V. **Concurrent CobalaminC and plasminogen deficiencies in a patient with chronic thrombotic microangiopathy**
DİRİM A. B., Safak S., BALCI M. C., Ozyavuz P., Garayeva N., TİRYAKİ T. O., OTO Ö. A., ÖZLÜK M. Y., KILIÇASLAN I., SOLAKOĞLU S., et al.
NEPHRON, 2023 (SCI-Expanded)

VI. **Levodopa-refractory hyperprolactinemia and pituitary findings in inherited disorders of biogenic amine metabolism.**
YILDIZ Y., Kuseyri Hübschmann O., Akgöz Karaosmanoğlu A., Manti F., Karaca M., Schwartz I. V. D., Pons R., López-Laso E., Palacios N. A. J., Porta F., et al.
Journal of inherited metabolic disease, 2023 (SCI-Expanded)

VII. **Reanalysis of exome sequencing data reveals a treatable neurometabolic origin in two previously undiagnosed siblings with neurodevelopmental disorder**
Susgun S., Kesim Y., Khalilov D., Sirin N. G., Gezegen H., Salman B., Yucesan E., Gokcay G. F., Korbeyli H. K., Balci M. C., et al.
NEUROLOGICAL SCIENCES, vol.44, no.7, pp.2527-2540, 2023 (SCI-Expanded)

VIII. **Triosephosphate Isomerase Deficiency: E105D Mutation in Unrelated Patients and Review of the Literature**
Selamioğlu A., Karaca M., Balcl M. C., Körbeyli H. K., Durmuş A., Ylldlz E. P., Karaman S., Gökçay G. F.
MOLECULAR SYNDROMOLOGY, vol.14, no.3, pp.231-238, 2023 (SCI-Expanded)

IX. **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)

X. **Leucine tolerance in children with MSUD is not correlated with plasma leucine levels at diagnosis.**
Kozanoğlu T., Balcl M. C., Karaca M., Gökçay G. F.
Journal of pediatric endocrinology & metabolism : JPEM, vol.36, no.2, pp.167-173, 2022 (SCI-Expanded)

XI. **A multinational study of acute and long-term outcomes of Type 1 galactosemia patients who carry the S135L (c.404C > T) variant of GALT**
Katler Q. S., Stepien K. M., Paull N., Patel S., Adams M., BALCI M. C., Berry G. T., Bosch A. M., DeLaO A., Demirbas D., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.45, no.6, pp.1106-1117, 2022 (SCI-Expanded)

XII. **Cardiologic evaluation of Turkish mitochondrial fatty acid oxidation disorders**
Balci M. C., Karaca M., Ergul Y., Omeroglu R. E., Demirkol M., Gokcay G. F.
PEDIATRICS INTERNATIONAL, vol.64, no.1, 2022 (SCI-Expanded)

XIII. **Clinical presentation and outcome in a series of 32 patients with 2-methylacetoacetyl-coenzyme A thiolase (MAT) deficiency.**
Gruenert S. C., Schmitt R. N., Schlatter S. M., Gemperle-Britschgi C., Balci M. C., Berg V., ÇOKER M., Das A. M., Demirkol M., Derks T. G. J., et al.
Molecular genetics and metabolism, vol.122, pp.67-75, 2017 (SCI-Expanded)

XIV. **3-Hydroxy-3-methylglutaryl-coenzyme A lyase deficiency: Clinical presentation and outcome in a series of 37 patients.**
Gruenert S. C., Schlatter S. M., Schmitt R. N., Gemperle-Britschgi C., Mrazova L., Balci M. C., Bischof F., ÇOKER M., Das A. M., Demirkol M., et al.
Molecular genetics and metabolism, vol.121, no.3, pp.206-215, 2017 (SCI-Expanded)

XV. **Rapid Desensitization for Immediate Hypersensitivity to Galsulfase Therapy in Patients with MPS VI.**
Tamay Z. Ü., Gokcay G. F., Dilek F., Balci M. C., Ozceker D., Demirkol M., Guler N.

JIMD reports, vol.30, pp.53-57, 2016 (SCI-Expanded)

XVI. **Clinical manifestations of 17 patients affected with mucopolysaccharidosis type VI and eight novel ARSB mutations**
Kantaputra P. N., Kayserili H., Guven Y., Kantaputra W., Balci M. C., Tanpaiboon P., Tananuvat N., Uttarilli A., Dalal A.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.164, no.6, pp.1443-1453, 2014 (SCI-Expanded)

XVII. **Oral manifestations of 17 patients affected with mucopolysaccharidosis type VI**
KANTAPUTRA P. N., Kayserili H., Guven Y., KANTAPUTRA W., Balci M. C., Tanpaiboon P., Uttarilli A., DALAL A.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.37, no.2, pp.263-268, 2014 (SCI-Expanded)

XVIII. **Oral manifestations of 17 patients affected with mucopolysaccharidosis type VI.**
KANTAPUTRA P. N., KAYSERİLİ H., GÜVEN Y., KANTAPUTRA W., BALCI M. C., Tanpaiboon P., UTTARILI A., DALAL A.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.56, pp.10-20, 2013 (SCI-Expanded)

XIX. **OCTN2 GENE MUTATIONS IN TURKISH PATIENTS WITH PRIMARY CARNITINE DEFICIENCY**
Yucel-Yilmaz D., Ersoy M., Candan S., Balci M. C., Kilic M., Gokcay G. F., Dursun A., Ozgul R. K.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)

XX. **CONGENITAL DISORDERS OF GLYCOSYLATION TYPE IA: ARE VACUOLATED PERIPHERAL BLOOD CELLS A DIAGNOSTIC CLUE?**
Ersoy M., Balci M. C., Ozguven A. A., Unuvar A., Matthijs G., Jaeken J., Demirkol M., Gokcay G. F.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)

XXI. **alpha-METHYL-COA RACEMASE DEFICIENCY: REPORT OF A NEW MUTATION AND RESPONSE TO TREATMENT IN A PATIENT WITH NEONATAL CHOLESTATIC LIVER DISEASE AND ATTENTION DEFICIT HYPERACTIVITY DISORDER**
Ersoy M., Cakir N., Balci M. C., Demirkol M., Gokcay G. F.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)

XXII. **MAPLE SYRUP URINE DISEASE: USE OF LEUCINE FREE MEDICAL FOODS AND AMINO ACID SUPPLEMENTS IN DIETARY TREATMENT**
Cakir N., Tuncer S., Balci M. C., Ersoy M., Demirkol M., Gokcay G. F.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)

XXIII. **THE CARDIAC MANIFESTATION AND RESPONSE TO L-CARNITINE TREATMENT IN 14 CASES WITH PRIMARY SYSTEMIC CARNITINE DEFICIENCY: CORRELATION WITH GENOTYPE**
Balci M. C., Yucel D., Ergul Y., ÖZGÜL R. K., Baykal T., Aktuglu-Zeybek C., Ersoy M., Demirkol M., Eker-Omeroglu R. N., DURSUN A., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.34, 2011 (SCI-Expanded)

Articles Published in Other Journals

I. **The Use of Social Network in Daily Pediatric Practice and Education: Turkish Pediatric Atelier**
Gonullu E., Soysal A., Can I., Tutak E., Tunc T., Yildiz I., YEŞİLBAŞ O., Oner N., Anarat A., Soysal F. G., et al.
INTERNATIONAL JOURNAL OF PEDIATRICS, vol.2020, 2020 (ESCI)

II. **Öksürük ve öksürük şurupları.**
Balcı M. C., Somer A.
Klinik Tıp Pediatri Dergisi , vol.3, no.1, pp.1-6, 2011 (Non Peer-Reviewed Journal)

Books & Book Chapters

I. **"Çapa Çocuk" Sabah Vaka Arşivleri-I**
Balcı M. C., Karaca M., Pempegül Yıldız E., Demirkol D., Gökçay G. F.
in: "Çapa Çocuk" Sabah Vaka Arşivleri-I , Asuman Çoban,Demet Demirkol,Alev Yılmaz,Nuray Aktay Ayaz, Editor, Nobel Tıp Kitapevi, Ankara, pp.31-37, 2023

II. **"Çapa Çocuk" Sabah Vaka Arşivleri-I**

Kahraman H., Körbeyli H. K., Balcı M. C., Demirkol D., Gökçay G. F.
in: "Çapa Çocuk" Sabah Vaka Arşivleri-I, Asuman Çoban,Demet Demirkol,Alev Yılmaz,Nuray Aktay Ayaz, Editor, Nobel Tıp Kitapevi, Ankara, pp.21-30, 2023

III. **Tamamlayıcı beslenmeye güncel bakış**
BALCI M. C., GÖKÇAY G. F.
in: Genel Pediyatri- Pediyatrinin Temel Taşları, Fatma Oğuz, Editor, Ema Tıp Kitabevi, İstanbul, pp.73-100, 2022

IV. **Neurometabolic Strokes in Childhood**
Balcı M. C.
in: Stroke in Childhood, NUR AYDINLI,TUĞÇE AKSU UZUNHAN, Editor, Türkiye Klinikleri Yayınevi, Ankara, pp.14-20, 2021

V. **Mitokondriyal ve Diğer Enerji Metabolizması Bozuklukları**
Balcı M. C., Kozanoğlu T., Gökçay G. F.
in: Kalıtsal Metabolik Hastalıklarda Beslenme Tedavisi, Eminoğlu,Fatma Tuğba; Haspolat,Yusuf Kenan; Çeltik,Çoşkun; Çarman,Kürşat Bora; Akbulut,Ulaş Emre; Taş,Taşkın, Editor, Orient Yayınları, Ankara, pp.619-640, 2021

VI. **COVID-19 ve Doğumsal Metabolizma Hastalıkları**
Durmuş A., Karaca M., Balci M. C., Selamioğlu A., Körbeyli H. K., Gökçay G. F.
in: Çocuklarda Her Yönüyle COVID-19, Demirkol D,Karacabey BN,Karakaş Z., Editor, Selen Yayınevi, İstanbul, pp.259-268, 2021

VII. **MİTOKONDRİYAL ve DİĞER ENERJİ METABOLİZMASI BOZUKLUKLARI: UZUN ZİNCİRLİ YAĞ ASİDİ OKSİDASYON BOZUKLUKLARI ve BESLENME TEDAVİSİ**
BALCI M. C., Kozanoğlu T., GÖKÇAY G. F.
in: Kalıtsal Metabolik Hastalıklarda Beslenme Tedavisi, Eminoğlu Fatma Tuğba, Haspolat Yusuf Kenan, Çeltik Coşkun, Çarman Kürşat Bora, Akbulut Ulaş Emre, Taş Taşkın, Editor, Orient Yayınları, Ankara, pp.621-642, 2021

VIII. **Inborn errors of metabolic disease**
Demirkol M., Gökçay G. F., Balcı M. C.
in: Pediatric, Olcay Neyzi,Türkan Ertuğrul,Feyza Darendeliler, Editor, Nobel Tıp Kitapevi, İstanbul, pp.1003-1134, 2020

IX. **XI. Bölüm Metabolizma Hastalıkları: 11. Nörotransmitter Bozuklukları**
BALCI M. C., GÖKÇAY G. F.
in: PEDİYATRİ, Olcay Neyzi, Türkan Ertuğrul, Feyza Darendeliler, Editor, Nobel Tıp Kitapevleri, İstanbul, pp.1125-1128, 2020

X. **Bölüm 27: Çocukta Metabolizma Hastalıkları Semiyolojisi**
BALCI M. C., GÖKÇAY G. F.
in: PEDİYATRİK SEMİYOLOJİ, Fatma Oğuz, Editor, Ema Tıp Kitapevi, İstanbul, pp.343-355, 2020

XI. **XI. Bölüm Metabolizma Hastalıkları: 1. Doğumsal Metabolizma Hastalıklarına Yaklaşım**
DEMİRKOL M., BALCI M. C., GÖKÇAY G. F.
in: Pediyatri, Olcay Neyzi, Türkan Ertuğrul, Feyza Darendeliler, Editor, Nobel Tıp Kitapevleri, İstanbul, pp.1003-1014, 2020

XII. **Carbohydrate Metabolism Disorders**
Demirkol M., Balcı M. C.
in: Yurdakok Pediatrics, Murat Yurdakök,Turgay Coşkun,Serap Sivri, Editor, Güneş Kitabevi, Ankara, pp.146-181, 2018

XIII. **Çocukta metabolizma hastalıkları semiyolojisi**
BALCI M. C., GÖKÇAY G. F.
in: Pediyatrik Semiyoloji, Fatma Oğuz, Editor, İstanbul Tıp Kitabevleri, İstanbul, pp.323-334, 2018

XIV. **Rapid Desensitization for immediate hypersensitivity to Galsulfase therapy in patients with MPS VI**
TAMAY Z. Ü., GÖKÇAY G. F., DİLEK F., BALCI M. C., Ozceker D., DEMİRKOL M., Guler N.
in: JIMD Reports, Morava E, Baumgartner M, Patterson M, Rahman S, Zschocke J, Peters V, Editor, Springer, pp.53-57, 2016

XV. **Çocukta Metabolizma Hastalıkları Semiyolojisi**

Balcı M. C., GÖKÇAY G. F.
in: Pediyatrik Semiyoloji, Fatma Oğuz, Editor, İstanbul Tıp Kitabevi, İstanbul, pp.266-276, 2016

XVI. **Fatty Acid Oxidation Disorders and Lipid Storage Diseases**
Balcı M. C., Nişli K.
in: Cardiac Involvement in Pediatric Systemic Diseases, Nazmi Narin, Editor, Türkiye Klinikleri Yayınevi, Ankara, pp.40-45, 2015

Refereed Congress / Symposium Publications in Proceedings

I. **Cellular oxidative damage in congenital disorders of glycosylation**
Ak B., Karaca M., Gedikbaşı A., Aydın A. F., Balcı M. C., Bilgin A., Körbeyli H. K., Durmuş A., Kılıç Ş., Gökçay G. F.
SSIEM Annual Symposium 2023, Yerushalayim, Israel, 29 August - 01 September 2023, pp.745

II. **Pregnancy, Maternal and Child Health in Women with Inherited Metabolic Disorders**
Karaca M., Balcı M. C., Selamioğlu A., Körbeyli H. K., Durmuş A., Çakar E., Kozanoğlu T., Hacıoğlu İ., Gökçay G. F.
SSIEM Annual Symposıum 2023, Yerushalayim, Israel, 29 August - 01 September 2023, pp.270

III. **Rapid, accurate and comprehensive diagnostic method for the detection of Neuronal Ceroid Lipofuscinosis Type 2 (CLN2) Disease using long-read third-generation sequencing technology**
Teker B., Tatonyan S., Balcı M. C., Karaca M., Akan G., Özgen Ö., Kürekçi F., Güngör O., Deniz A., Gedikbaşı A., et al.
SSIEM Annual Symposium 2023, Tel-Aviv-Yafo, Israel, 29 August - 01 September 2023, vol.46, no.686, pp.361

IV. **Menstrual cycle characteristics, premenstrual syndrome and blood phenylalanine level relationship in women with PKU**
SELAMİOĞLU A., Tandoğan Z., BALCI M. C., KARACA M., KOZANOĞLU T., YEŞİL A., GÖKÇAY G. F.
SSIEM Annual Symposium 2023, Jerusalem, Israel, 29 August 2023, vol.46, pp.494

V. **Evaluation of the Risk Factors for Noncommunicable Diseases in Patients with Inborn Errors of Amino Acid Metabolism Receiving Nutrition Therapy**
BALCI M. C., KARACA M., Yeşil A., SELAMİOĞLU A., Körbeyli H. K., DURMUŞ A., AK B., Hacıoğlu İ., GÖKÇAY G. F.
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VI. **Retrospective Analysis Of Carbohydrate-Deficient Transferrin For CDG Screening: A Single Center Study**
Özgen Ö., Güdek Kılıç F., Gedikbaşi A., Balci M. C., Karaca M., Durmuş A., Ak B., Körbeyli H. K., Atalar F., Gökçay G. F.
SSIEM Annual Symposium 2023, Tel-Aviv-Yafo, Israel, 29 August 2023, vol.46, pp.257

VII. **Mitochondrial dysfunction in a disorder of transsulphuration: Cystathionine β-synthase deficiency**
Balcı M. C., Gedikbaşı A., Kahraman S., Tatonyan S., Tekin Neijmann Ş., Karaca M., Atalar F., Gökçay G. F.
SSIEM Annual Symposium 2023, Tel-Aviv-Yafo, Israel, 29 August 2023, vol.46, pp.148

VIII. **Fluorometric Analysis and Validation of Tripeptidyl Peptidase-1 in Dry Blood Samples and Leukocytes in the Diagnosis of Neuronal Ceroid Lipofuxinosis 2**
Teker B., Tatonyan S., Gedikbaşı A., Aydın A. F., Balcı M. C., Karaca M., Pempegül Yıldız E., Atalar F., Poda M., Gökçay G. F.
VIII. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi, Bursa, Turkey, 3 - 07 May 2023, pp.64

IX. **Immunomodulation and High-Dose Enzyme Replacement Therapy (ERT) Experience in Cross-Reactive Immunological Material (CRIM) Negative Infantile Pompe Patients**
Selamioğlu A., Durmuş A., Karaca M., Balcı M. C., Gökçay G. F.
VIII. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi, Bursa, Turkey, 3 - 07 May 2023, pp.60

X. **Neuraminidase Deficiency: 5 Case Reports**
Ak B., Balcı M. C., Karaca M., Gökçay G. F.
VIII. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi, Bursa, Turkey, 3 - 07 May 2023, pp.41

XI. **Farber Disease and New Treatment Options with Three Cases**
Durmuş A., Balcı M. C., Karaca M., Gökçay G. F.
VIII. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi, Bursa, Turkey, 3 May - 07 October 2023, pp.62

XII. **Pancreatic Involvement in Hereditary Metabolic Diseases**

Selamioğlu A., Karaca M., Balcı M. C., Gökçay G. F.
45. Pediatri Günleri / 24. Pediatri Hemşireliği Günleri, İstanbul, Turkey, 25 - 28 April 2023, pp.327-330

XIII. **Mitokondriyal Aminoasilt-RNA Sentetaz Eksikligi Olgularinda Serum FGF-21 Düzeylerinin Degerlendirilmesi**
Tekin Neijmann Ş., Gedikbaşı A., Güneş D., Balcı M. C., Karaca M., Atalar F., Gökçay G. F.
TÜRK KLİNİK BİYOKİMYA DERNEĞİ XXIII. Ulusal Klinik Biyokimya Kongresi, Antalya, Turkey, 27 - 30 April 2023, pp.234

XIV. **OTC Hastalarında Klinik Bulguların ve Laboratuvar Parametrelerinin Değerlendirilmesi**
Tırtır Yılmaz B., Ak B., Balcı M. C., Karaca M., Gökçay G. F.
45. Pediatri Günleri, İstanbul, Turkey, 25 - 28 April 2023, pp.1

XV. **Metabolik Hastalıklarda Endokrinolojik Sorunlar; Biz ne yapabiliriz ?**
İnan Balcı E., Balcı M. C., Karakılıç Özturan E., Yıldız M., Poyrazoğlu Ş., Baş F., Darendeliler F. F.
XXVI. Ulusal Pediatrik Endokrinoloji ve Diyabet Kongresi, Antalya, Turkey, 26 October - 30 November 2022, pp.91

XVI. **Demographic Characteristics and Phenotypic Findings of Patients with Dihydropterin Reductase Deficiency: Single Center Experience**
Eryılmaz C. C., Balcı M. C., Ak B., Durmuş A., Körbeyli H. K., Karaca M., Gökçay G. F.
XVI. Uluslararası Katılımlı Metabolik Hastalıklar ve Beslenme Kongresi, Hatay, Turkey, 28 May - 01 June 2022, pp.126-129

XVII. **Diagnostic Usefulness of Whole Exome Sequence Analysis in cases with suspected mitochondrial disease: Single center experience**
Gedikbaşı A., Balcı M. C., Karaca M., Toksoy G., Güleç Ç., Selamioğlu A., Durmuş A., Ak B., Körbeyli H. K., Uyguner Z. O., et al.
XVI. Uluslararası Katılımlı Metabolik Hastalıklar ve Beslenme Kongresi, Hatay, Turkey, 28 May - 01 June 2022, pp.144-146

XVIII. **CONGENITAL GLYCOSYLATION DISORDERS: CLINICAL IN 30 CASES EVALUATION**
Ak B., Balcı M. C., Karaca M., Gökçay G. F.
XVI. Uluslararası Katılımlı Metabolik Hastalıklar ve Beslenme Kongresi, Hatay, Turkey, 28 May - 01 June 2022, pp.117

XIX. **Retrospective Evaluation of Our Cases Diagnosed with Thiamine Biotin-Sensitive Basal Ganglion Disease**
Körbeyli H. K., Kılıç M. A., Balcı M. C., Pempegül Yıldız E., Gökçay G. F.
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XX. **Phenylketonuria Detected in Newborns Presenting with Elevated Phenylalanine in the National Newborn Screening of the Ministry of Health Other Congenital Metabolic Diseases**
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