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Education Information

Post Doctorate of Medicine, Hacettepe University, Tıp Fakültesi, Turkey 1977 - Continues

Expertise In Medicine, Hacettepe University, Tıp Fakültesi, Çocuk Sağlığı Ve Hastalıkları Anabilim Dalı, Turkey 1977 - 1981

Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Child Health and Diseases, Pediatric Endocrinology and Metabolism

Academic Titles / Tasks

Professor, Hacettepe University, Hacettepe Tıp Fakültesi, 1977 - Continues

Professor, Hacettepe University, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 1977 - 1981

Published journal articles indexed by SCI, SSCI, and AHCI

- I. **Electro-clinical features and long-term outcomes in guanidinoacetate methyltransferase (GAMT) deficiency**
YILDIZ Y., Ardiçlı D., GÖÇMEN R., YALNIZOĞLU D., Topçu M., Coşkun T., TOKATLI A., Haliloglu G. European Journal of Paediatric Neurology, vol.49, pp.66-72, 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. **Organic acidemias in the neonatal period: 30 years of experience in a referral center for inborn errors of metabolism**
ÜNSAL Y., YURDAKÖK M., YİĞİT Ş., ÇELİK H. T., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T. JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.35, no.11, pp.1345-1356, 2022 (SCI-Expanded)
- IV. **Recommendations on phenylketonuria in Turkey**
COŞKUN T., ÇOKER M., Mungan N. O., GÖKMEN ÖZEL H., SİVRİ H. S. TURKISH JOURNAL OF PEDIATRICS, vol.64, no.3, pp.413-434, 2022 (SCI-Expanded)
- V. **A new UHPLC-MS/MS method for the screening of urinary oligosaccharides expands the detection of storage disorders**
Semeraro M., Sacchetti E., Deodato F., COŞKUN T., LAY İ., Catesini G., Olivieri G., Rizzo C., Boenzi S., Dionisi-Vici C. Orphanet Journal of Rare Diseases, vol.16, no.1, 2021 (SCI-Expanded)
- VI. **Autism spectrum disorder in patients with inherited metabolic disorders-a large sample from a**

tertiary center

ÇELEN YOLDAŞ T., BİLGİNER GÜRBÜZ B., AKAR H. T., ÖZMERT E. N., COŞKUN T.

TURKISH JOURNAL OF PEDIATRICS, vol.63, no.5, pp.767-779, 2021 (SCI-Expanded)

- VII. **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)

- VIII. **Molecular etiology of isolated congenital cataract using next-generation sequencing: Single center exome sequencing data from Turkey**

Taylan Şekeroğlu H., Karaosmanoğlu B., Taşkıran E. Z., Şimşek Kiper P. Ö., Alikoşifoğlu M., Boduroğlu O. K., Coşkun T., Ütine G. E.

Molecular Syndromology, vol.11, pp.302-308, 2020 (SCI-Expanded)

- IX. **Glutaric aciduria type 1: Genetic and phenotypic spectrum in 53 patients**

BİLGİNER GÜRBÜZ B., YÜCEL YILMAZ D., COŞKUN T., TOKATLI A., DURSUN A., SİVRİ H. S.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.63, no.11, 2020 (SCI-Expanded)

- X. **Expanding the clinical spectrum of mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency with Turkish cases harboring novel HMGCS2 gene mutations and literature review**

Kılıç M., Dorum S., Topak A., Yazıcı M. U., EZGÜ F. S., COŞKUN T.

AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.182, no.7, pp.1608-1614, 2020 (SCI-Expanded)

- XI. **Predictors of acute metabolic decompensation in children with maple syrup urine disease at the emergency department**

YILDIZ Y., Akcan Yıldız L., DURSUN A., TOKATLI A., COŞKUN T., TEKŞAM Ö., SİVRİ H. S.

EUROPEAN JOURNAL OF PEDIATRICS, vol.179, no.7, pp.1107-1114, 2020 (SCI-Expanded)

- XII. **Two cases of Vici syndrome presenting with corpus callosum agenesis, albinism, and severe developmental delay**

Hızal M., Yeke B., YILDIZ Y., Öztürk A., Gürbüz B. B., COŞKUN T.

TURKISH JOURNAL OF PEDIATRICS, vol.62, no.3, pp.474-478, 2020 (SCI-Expanded)

- XIII. **Genotypes and estimated prevalence of phosphomannomutase 2 deficiency in Turkey differ significantly from those in Europe**

YILDIZ Y., Arslan M., Celik G., Kasapkara C. 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)

- XIV. **Caring for a Child with Phenylketonuria: Parental Experiences from a Eurasian Country**

ZENGİN AKKUŞ P., BİLGİNER GÜRBÜZ B., ÇIKI K., İLTER BAHADUR E., KARAHAN S., ÖZMERT E. N., COŞKUN T., Sıvri S.

JOURNAL OF DEVELOPMENTAL AND BEHAVIORAL PEDIATRICS, vol.41, no.3, pp.195-202, 2020 (SCI-Expanded)

- XV. **Oral health status of children with phenylketonuria**

BALLIKAYA E., YILDIZ Y., SİVRİ H. S., TOKATLI A., DURSUN A., Olmez S., COŞKUN T., Tekcicek M. U.

JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.33, no.3, pp.361-365, 2020 (SCI-Expanded)

- XVI. **Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency**

YILDIZ Y., TALİM B., Haliloglu G., Topaloglu H., AKÇÖREN Z., DURSUN A., SİVRİ H. S., COŞKUN T., TOKATLI A.

PEDIATRIC NEUROLOGY, vol.99, pp.69-75, 2019 (SCI-Expanded)

- XVII. **The effectiveness of enzyme replacement therapy on cardiac findings in patients with mucopolysaccharidosis**

BİLGİNER GÜRBÜZ B., AYPAR E., COŞKUN T., ALEHAN D., DURSUN A., TOKATLI A., SİVRİ H. S.

JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.32, no.10, pp.1049-1053, 2019 (SCI-Expanded)

- XVIII. **Imaging liver nodules in tyrosinemia type-1: A retrospective review of 16 cases in a tertiary pediatric hospital**

ÖZCAN H. N., KARÇAALTINCABA M., Pektaş E., SİVRİ H. S., OĞUZ B., DURSUN A., TOKATLI A., COŞKUN T., HALİLOĞLU M.

EUROPEAN JOURNAL OF RADIOLOGY, vol.116, pp.41-46, 2019 (SCI-Expanded)

- XIX. **International best practice for the evaluation of responsiveness to sapropterin dihydrochloride in**

- patients with phenylketonuria**
- Muntau A. C., Adams D. J., Belanger-Quintana A., Bushueva T. V., Cerone R., Chien Y., Chiesa A., COŞKUN T., de las Heras J., Feillet F., et al.
MOLECULAR GENETICS AND METABOLISM, vol.127, no.1, pp.1-11, 2019 (SCI-Expanded)
- XX. Do not Miss Rare and Treatable Cause of Early-Onset Hemolytic Uremic Syndrome: Cobalamin C Deficiency**
- TOPALOĞLU R., Inozu M., GÜLHAN B., BİLGİNER GÜRBÜZ B., TALİM B., COŞKUN T.
NEPHRON, vol.142, no.3, pp.258-263, 2019 (SCI-Expanded)
- XXI. TWO CASES WITH DIVERSE COURSE OF AHUS RELATED TO COBALAMIN C DEFECT**
- Inozu M., GÜLHAN B., TOKATLI A., COŞKUN T., TOPALOĞLU R.
PEDIATRIC NEPHROLOGY, vol.33, no.10, pp.1848, 2018 (SCI-Expanded)
- XXII. The potential role of gut microbiota and its modulators in the management of propionic and methylmalonic acidemia**
- Burlina A., Tims S., van Spronsen F., Sperl W., Burlina A. P., Kuhn M., Knol J., Rakhshandehroo M., COŞKUN T., Singh R. H., et al.
EXPERT OPINION ON ORPHAN DRUGS, vol.6, no.11, pp.683-692, 2018 (SCI-Expanded)
- XXIII. 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)
- XXIV. Deoxyguanosine kinase deficiency: a report of four patients**
- Unal O., ÖZTÜRK HİŞMİ B., KILIÇ M., HIZARCIOĞLU GÜLŞEN H., COŞKUN T., Sivri S. H., DURSUN A., YÜCE A., TOKATLI A.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.30, no.6, pp.697-702, 2017 (SCI-Expanded)
- XXV. Screening for mucopolysaccharidoses in the Turkish population: Analytical and clinical performance of an age-range specific, dye-based, urinary glycosaminoglycan assay**
- EL MOUSTAFA K., Sivri S., KARAHAN S., COŞKUN T., Akbilyik F., LAY İ.
CLINICA CHIMICA ACTA, vol.464, pp.72-78, 2017 (SCI-Expanded)
- XXVI. Partial hydatidiform mole in a phenylketonuria patient treated with sapropterin dihydrochloride**
- YILDIZ Y., DURSUN A., TOKATLI A., COŞKUN T., Sivri S.
GYNECOLOGICAL ENDOCRINOLOGY, vol.33, no.1, pp.19-20, 2017 (SCI-Expanded)
- XXVII. Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency**
- Olsen R. K. J., Konarikova E., Giancaspero T. A., Mosegaard S., Boczonadi V., Matakovic L., Veauville-Merllie A., Terrile C., Schwarzmayr T., Haack T. B., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.98, no.6, pp.1130-1145, 2016 (SCI-Expanded)
- XXVIII. Clinical phenotype, biochemical profile, and treatment in 19 patients with arginase 1 deficiency**
- Huemer M., Carvalho D. R., Brum J. M., Unal O., COŞKUN T., Weisfeld-Adams J. D., Schrager N. L., Scholl-Buergi S., Schlune A., Donner M. G., et al.
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- XXIX. Brown-Vialetto-Van Laere syndrome: two siblings with a new mutation and dramatic therapeutic effect of high-dose riboflavin**
- ÖZGÜR HOROZ Ö., Mungan N. O., Yildizdas D., Herguner O., Ceylaner S., KOR D., Waterham H., COŞKUN T.
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- XXX. Diagnostic and management practices for phenylketonuria in 19 countries of the South and Eastern European Region: survey results**
- Gizewska M., MacDonald A., Belanger-Quintana A., Burlina A., Cleary M., COŞKUN T., Feillet F., Muntau A. C., Trefz F. K., van Spronsen F. J., et al.
EUROPEAN JOURNAL OF PEDIATRICS, vol.175, no.2, pp.261-272, 2016 (SCI-Expanded)
- XXXI. A Nonvirilized form of Classic 3 beta-Hydroxysteroid Dehydrogenase Deficiency Due to a**

- Homozygous S218P Mutation in the HSD3B2 Gene in a Girl with Classic Phenylketonuria**
ALİKAŞIFOĞLU A., Buyukyilmaz G., GÖNC E. N., ÖZÖN Z. A., KANDEMİR N., DÜNDAR M., POLAT S., PEKTAŞ E., DURSUN A., Sivri S., et al.
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.281, 2016 (SCI-Expanded)
- XXXII. Late-diagnosed phenylketonuria in an eight-year-old boy with dyslexia and attention-deficit hyperactivity disorder**
YILDIZ Y., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.
TURKISH JOURNAL OF PEDIATRICS, vol.58, no.1, pp.94-96, 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. Detection of biotinidase gene mutations in Turkish patients ascertained by newborn and family screening**
KARACA M., ÖZGÜL R. K., ÜNAL O., Yuçel-Yılmaz 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)
- XXXVI. Ailevi Hipercolesterolemili 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)
- XXXVII. Conventional and advanced MR imaging in infantile Refsum disease**
KILIÇ M., Karli-Oguz K., Haliloglu G., TOPÇU M., Wanders R. J., COŞKUN T.
TURKISH JOURNAL OF PEDIATRICS, vol.57, no.3, pp.294-299, 2015 (SCI-Expanded)
- XXXVIII. Fundus autofluorescence and optical coherence tomography findings in glutathione synthetase deficiency**
TAYLAN ŞEKEROĞLU H., ÖZTÜRK HİŞMİ B., KADAYIFÇILAR S., COŞKUN T.
JOURNAL OF AAPOS, vol.19, no.1, pp.80-82, 2015 (SCI-Expanded)
- XXXIX. 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)
- XL. Phenotypic and genotypic spectrum of Turkish patients with isovaleric acidemia**
ÖZGÜL R. K., Karaca M., Kılıç 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)
- XLI. A case of fucosidosis type II: diagnosed with dysmorphological and radiological findings**
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TURKISH JOURNAL OF PEDIATRICS, vol.56, no.4, pp.430-433, 2014 (SCI-Expanded)
- XLII. Mucopolysaccharidosis: Otolaryngologic findings, obstructive sleep apnea and accumulation of glucosaminoglycans in lymphatic tissue of the upper airway**
GONULDAS B., YILMAZ T., SİVRİ H. S., GÜÇER K. Ş., KILINC K., GENÇ G. A., KILIÇ M., COŞKUN T.
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- XLIII. 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)

- XLIV. Guanidinoacetate methyltransferase (GAMT) deficiency: Outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring**
Stockler-Ipsiroglu S., van Karnebeek C., Longo N., Korenke G. C., Mercimek-Mahmutoglu S., Marquart I., Barshop B., Grolik C., Schlune A., Angle B., et al.
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- XLV. Pregnancy and Lactation Outcomes in a Turkish Patient with Lysinuric Protein Intolerance**
Unal O., COŞKUN T., ORHAN D., Tokatlı A., DURSUN A., ÖZTÜRK HİŞMİ B., ÖZYÜNCÜ Ö., Sivri S. H. K.
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- XLVI. A Patient With Pyruvate Carboxylase Deficiency and Nemaline Rods on Muscle Biopsy**
Unal O., ORHAN D., Ostergaard E., TOKATLI A., DURSUN A., ÖZTÜRK HİŞMİ B., COŞKUN T., Wibrand F., Kalkanoglu-Sivri H. S.
JOURNAL OF CHILD NEUROLOGY, vol.28, no.11, pp.1505-1508, 2013 (SCI-Expanded)
- XLVII. Vanishing White Matter With Hepatomegaly and Hypertriglyceridemia Attacks**
Unal O., ÖZGEN MOCAN B., ORHAN D., TOKATLI A., Hisimi B. O., DURSUN A., COŞKUN T., Kalkanoglu-Sivri H. S.
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- XLVIII. Cobalamin C defect: a patient of late-onset type with homozygous p. R132*mutation**
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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**
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- L. Molecular and clinical evaluation of Turkish patients with lysinuric protein intolerance**
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- LI. Genetic basis of hyperlysinemia**
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- LII. Serum alpha-fetoprotein levels in neonatal cholestasis**
DEMİR H., Hızal G., USLU KIZILKAN N., Gurakan F., TALİM B., COŞKUN T., KALE G., YÜCE A.
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- LIII. PROPIONIC ACIDEMIA PRESENTING WITH PERSISTENT PULMONARY HYPERTENSION IN TWO NEONATES**
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- 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.
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- LV. Detection of other inborn errors of metabolism in hyperphenylalaninemic babies picked up on narrow-spectrum screening programs**
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- LVI. When do we need to perform a diagnostic lumbar puncture for neurometabolic diseases? Positive yield and retrospective analysis from a tertiary center**
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- LVII. Genetic basis of cystinosis in Turkish patients: a single-center experience**
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- den Heuvel L., Kleta R., et al.
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- LVIII. **A Rare Galactosemia Complication: Vitreous Hemorrhage**
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- LIX. **Identification of Mutations and Evaluation of Cardiomyopathy in Turkish Patients with Primary Carnitine Deficiency**
KILIÇ M., ÖZGÜL R. K., COŞKUN T., Yucel D., KARACA M. A., SİVRİ H. S., TOKATLI A., ŞAHİN M., KARAGÖZ T., DURSUN A.
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- LX. **Effects of probiotic (Primalac 454) on nonalcoholic fatty liver disease in broilers**
YALÇIN S. S., Gucer S., Yalcin S., ONBAŞILAR İ., Kale G., COŞKUN T.
REVUE DE MEDECINE VETERINAIRE, vol.162, no.7, pp.371-376, 2011 (SCI-Expanded)
- LXI. **Annual symposium of the SSIEM 2010**
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- LXII. **Home visits in phenylketonuria: a 12-month longitudinal study**
GOKMEN-OZEL H., Buyuktuncer Z., Koksal G., KALKANOGLU-SIVRI H. S., COŞKUN T.
TURKISH JOURNAL OF PEDIATRICS, vol.53, no.2, pp.149-153, 2011 (SCI-Expanded)
- LXIII. **Gyrate atrophy of the choroid and retina: a case report**
Buyuktortop N., Alp M. N., Sivri S., COŞKUN T., Kural G.
TURKISH JOURNAL OF PEDIATRICS, vol.53, no.1, pp.94-96, 2011 (SCI-Expanded)
- LXIV. **MUTATION ANALYSIS IN ARSB GENE IN TURKISH PATIENTS WITH MPS TYPE VI: HIGH PREVALENCE OF L321P MUTATION**
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