# Prof. Hülya KAYSERİLİ KARABEY

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## Education Information

Doctorate, Istanbul University, Sağlık Bilimleri Enstitüsü/ Çocuk Sağlığı Enstitüsü , Genetik Anabilim Dalı , Turkey 1991 - 1998
Undergraduate, Istanbul University, Istanbul Medical Faculty, Tıp, Turkey 1978 - 1984

## Research Areas

Life Sciences, Molecular Biology and Genetics, Genetic Disorders, Genomics, Natural Sciences

## Academic Titles / Tasks

Professor, Istanbul University, Istanbul Medical Faculty, Tıbbi Genetik Ad , 2009 - Continues
Associate Professor, Istanbul University, Istanbul Medical Faculty, Dahili Bilimler , 2000 - Continues

## Published journal articles indexed by SCI, SSCI, and AHCI

I. **Clinicogenetic Study of Turkish Patients With Syndromic Craniosynostosis and Literature Review**
NUR B., Pehlivanoglu S., MIHÇI E., Caliskan M., Demir D., ALPER Ö., Kayserili H., Luleci G.
PEDIATRIC NEUROLOGY, vol.50, no.5, pp.482-490, 2014 (SCI-Expanded)

II. **Twins with hereditary sensory and autonomic neuropathy type IV with preserved periodontal sensation**
Guven Y., Altunoglu U., Aktoren O., Uyguner Z. O., Kayserili H., Kaewkahya M., Kantaputra P. N.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.57, no.5, pp.240-246, 2014 (SCI-Expanded)

III. **A patient with a de-novo deletion 3p25.3 and features overlapping with Rubinstein-Taybi syndrome**
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CLINICAL DYSMORPHOLOGY, vol.23, no.2, pp.67-70, 2014 (SCI-Expanded)

IV. **Newly Described Clinical Features in Two Siblings With MACS Syndrome and a Novel Mutation in RIN2**
Aslanger A. D., Altunoglu U., Aslanger E., Satkin B. N., Uyguner Z. O., Kayserili H.
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V. **Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders**
Novarino G., Fenstermaker A. G., Zaki M. S., Hofree M., Silhavy J. L., Heiberg A. D., Abdellateef M., Rosti B., Scott E.,

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VI. **The genetic basis of DOORS syndrome: an exome-sequencing study**
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VII. **Extreme Growth Failure is a Common Presentation of Ligase IV Deficiency**
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HUMAN MUTATION, vol.35, no.1, pp.76-85, 2014 (SCI-Expanded)

VIII. **A comprehensive molecular study on Coffin-Siris and Nicolaides-Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling**
Wieczorek D., Boegershausen N., Beleggia F., Steiner-Haldenstaett S., Pohl E., Li Y., Milz E., Martin M., Thiele H., Altmueller J., et al.
HUMAN MOLECULAR GENETICS, vol.22, no.25, pp.5121-5135, 2013 (SCI-Expanded)

IX. **Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans**
Halbritter J., Bizet A. A., Schmidts M., Porath J. D., Braun D. A., Gee H. Y., McInerney-Leo A. M., Krug P., Filhol E., Davis E. E., et al.
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X. **Genotype phenotype spectrum of PYCR1-related autosomal recessive cutis laxa**
Dimopoulou A., Fischer B., Gardeitchik T., Schroeter P., Kayserili H., Schlack C., Li Y., Brum J. M., Barisic I., Castori M., et al.
MOLECULAR GENETICS AND METABOLISM, vol.110, no.3, pp.352-361, 2013 (SCI-Expanded)

XI. **Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused by PAPSS2 Mutations**
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XIII. **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)

XIV. **Clinical and mutation data in 12 patients with the clinical diagnosis of Nager syndrome**
Czeschik J. C., Voigt C., ALANAY Y., Albrecht B., Avci S., FitzPatrick D., Goudie D. R., Hehr U., Hoogeboom A. J., Kayserili H., et al.
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XV. **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.
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XVI. **Developing a policy for paediatric biobanks: principles for good practice.**
Henz K., Van El C., Borry P., Cambom Thomsen A., Cornel M., Forzano F., Lucassen A., Patch C., Tranebjaerg L., Vermeulen E., et al.
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XVII. **A novel c.1255G>T (p.D419Y) mutation in SH3BP2 gene causes cherubism in a Turkish family.**
Dinçkan N., GÜVEN Y., KAYSERİLİ H., AKTÖREN O., UYGUNER Z. O.
ORAL SURGERY ORAL MEDICINE ORAL PATHOLOGY ORAL RADIOLOGY AND ENDODONTOLOGY, vol.11, no.4,

pp.42-46, 2012 (SCI-Expanded)

XVIII. **Mutations in BCKD-kinase Lead to a Potentially Treatable Form of Autism with Epilepsy**
Novarino G., El-Fishawy P., Kayserili H., Meguid N. A., Scott E. M., Schroth J., Silhavy J. L., Kara M., Khalil R. O., Ben-Omran T., et al.
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XIX. **Down Syndrome Diagnosis Based on Gabor Wavelet Transform**
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JOURNAL OF MEDICAL SYSTEMS, vol.36, no.5, pp.3205-3213, 2012 (SCI-Expanded)

XX. **Goldenhar syndrome : a new case expanding the phenotype by costal agenesis and pulmonary hypoplasia**
Çekmez F., İnce E., Çoban A., Yıldırım S., Bulut E., KAYSERİLİ H.
Eur Rev Med Pharmacol, vol.16, no.9, pp.1307-1308, 2012 (SCI-Expanded)

XXI. **Genotypic and phenotypic analysis of 396 individuals with mutations in Sonic Hedgehog.**
Solomon B., Bear K., Wyllie A., Keaton A., Dubourg C., David V., Mercier S., Odent S., Hehr U., Paulussen A., et al.
JOURNAL OF MEDICAL GENETICS, vol.49, no.7, pp.473-479, 2012 (SCI-Expanded)

XXII. **A large duplication involving the IHH locus mimics acrocallosal syndrome**
Yuksel-Apak M., Boegershausen N., Pawlik B., Li Y., Apak S., Uyguner O., Milz E., Nuernberg G., Karaman B., Gülgören A., et al.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.20, no.6, pp.639-644, 2012 (SCI-Expanded)

XXIII. **Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1.**
Bonnard C., Strobl A., Shboul M., Lee H., Merriman B., Nelson S., Ababneh O., Uz E., Güran T., KAYSERİLİ H., et al.
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XXIV. **Mutation analysis of 18 nephronophthisis associated ciliopathy disease genes using a DNA pooling and next generation sequencing strategy**
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XXV. **Novel TMEM67 mutations and genotype-phenotype correlates in meckelin-related ciliopathies.**
Iannicelli M., Brancati F., Mougou-Zerelli S., Mazzota A., Elkhartoufi N., Thomas S., Travaglini L., Gomes C., Ardissino G., E B., et al.
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XXVI. **Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival**
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HUMAN MOLECULAR GENETICS, vol.18, no.12, pp.2149-2165, 2009 (SCI-Expanded)

XXVII. **Mutational screening of BASP1 and transcribed processed pseudogene TPPsig-BASP1 in patients with Möbius syndrome.**
Üzümcü A., Candan Ş., Toksoy G., UYGUNER Z. O., KARAMAN B., Eriş H., TATLI B., KAYSERİLİ H., YÜKSEL A., Geçkinli B., et al.
journal genet BMC GENOMICS, vol.36, no.4, pp.251-6, 2009 (SCI-Expanded)

XXVIII. **Craniodentofacial manifestations in Hallerman-Streiff Syndrome.**
TUNA İNCE E. B., SÜLÜN T., KAYSERİLİ H., AKTÖREN O.
The Journal of Craniomandibular Practice, vol.27, no.1, pp.33-38, 2009 (SCI-Expanded)

XXIX. **Orodental findings of a family with lacrimo-auriculo-dento-digital (LADD) syndrome**
GÜVEN Y., Rosti R. Ö., TUNA İNCE E. B., KAYSERİLİ H., AKTÖREN O.
ORAL SURGERY ORAL MEDICINE ORAL PATHOLOGY ORAL RADIOLOGY, vol.106, pp.33-44, 2008 (SCI-Expanded)

XXX. **Cobblestone-like brain dysgenesis and altered glycosylation in congenital cutis laxa, Debre type.**
KAYSERİLİ H., Van Maldergem L.
Hum Genet, vol.11, no.71, pp.1602-1608, 2008 (SCI-Expanded)

XXXI. **Orodental findings of Lacrimo-Auriculo-Dento-Digital (LADD) Syndrome.**

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Oral Med Oral Pathol Oral Radio, vol.106, no.6, pp.33-40, 2008 (SCI-Expanded)

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Kara B., Karaman B., Ozmen M., Rosti R. O., Caliskan M., Kayserili H., Basaran S.
TURKISH JOURNAL OF PEDIATRICS, vol.50, no.2, pp.137-142, 2008 (SCI-Expanded)

XXXIII. **CYP21 gene mutations in congenital adrenal hyperplasia: Genotype-phenotype correlation in Turkish children**
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HORMONE RESEARCH, vol.70, pp.156-157, 2008 (SCI-Expanded)

XXXIV. **MYO15A (DFNB3) mutations in Turkish hearing loss families and functional modeling of a novel motor domain mutation.**
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AMERICAN JOURNAL OF MEDICAL GENETICS, vol.15, pp.2382-9, 2007 (SCI-Expanded)

XXXV. **Mutations in the lipoma HMGIC fusion partner-like 5 (LHFPL5) gene cause autosomal recessive nonsyndromic hearing loss.**
Kalay E., Li Y., Üzümcü A., UYGUNER Z. O., Karagüzel a., Becker C., Cremers C., KAYSERİLİ H., Hafız G., BAŞARAN S., et al.
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XXXVI. **The identification of small supernumerary marker chromosomes; the experiences of 15,792 fetal karyotyping from Turkey**
KARAMAN B., Aytan M., Yılmaz K., Toksoy G., Önal E. P., Ghanbari A., Engür A., KAYSERİLİ H., Yüksel Apak M., BAŞARAN S.
EUROPEAN JOURNAL OF HUMAN GENETICS, vol.49, no.3, pp.207-14, 2006 (SCI-Expanded)

XXXVII. **18q deletion syndrome associated with autoimmune thyroid disease presenting as hyperthyroidism**
Tütüncüler F., Darendeliler F. F., Günöz H., Karaman B., Kayserili H.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.18, no.4, pp.419-420, 2005 (SCI-Expanded)

XXXVIII. **Gender assignment in female congenital adrenal hyperplasia: a difficult experience**
Ozbey H., Darendeliler F. F., Kayserili H., Korkmazlar U., Salman T.
BJU INTERNATIONAL, vol.94, no.3, pp.388-391, 2004 (SCI-Expanded)

XXXIX. **Molecular characterisation of a new case of microphthalmia with linear skin defects (MLS).**
KAYSERİLİ H., Cox T. C., Cox L. L., BAŞARAN S., Kılıç G., Ballabio A., Yüksel Apak M.
Journal medical genetics, vol.37, pp.411-7, 2001 (SCI-Expanded)

XL. **Activation-induced cytidine deaminase (AID) deficiency causes the autosomal recessive form of the hyper-IgM syndrome (HIGM2)**
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XLI. **Mutation of the gene encoding the ROR2 tyrosine kinase causes autosomal recessive Robinow syndrome**
van Bokhoven H., Celli J., Kayserili H., van Beusekom E., Balci S., Brussel W., Skovby F., Kerr B., Percin E., Akarsu N., et al.
NATURE GENETICS, vol.25, no.4, pp.423-426, 2000 (SCI-Expanded)

XLII. **Glycine to tryptophan substitution in type I collagen in a patient with OI type III: a unique collagen mutation**
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JOURNAL OF MEDICAL GENETICS, vol.37, no.5, pp.371-375, 2000 (SCI-Expanded)

XLIII. **Screening of deletions in SMN, NAIP and BTF2p44 genes in Turkish spinal muscular atrophy patients**
Savas S., Gokgoz N., Kayserili H., Ozkinay F., Yuksel-Apak M., Kirdar B.
HUMAN HEREDITY, vol.50, no.3, pp.162-165, 2000 (SCI-Expanded)

XLIV. **A rare mutation [IVS-I-130 (G-A)] in a Turkish beta-thalassemia major patient**
Tadmouri G., Bilenoglu O., Kantarci S., Kayserili H., Perrin P., Basak A.
AMERICAN JOURNAL OF HEMATOLOGY, vol.63, no.4, pp.223-225, 2000 (SCI-Expanded)

XLV. **Beckwith Wiedemann sendromlu 8 olguda klinik yaklaşım ve izlem süreci**
BAŞ F., KAYSERİLİ H., DARENDELİLER F. F., TÜKEL T., BUNDAK R., KABATAŞ ERYILMAZ S., APAK-YÜKSEL M., SAKA H. N., GÜNÖZ H.
İstanbul Tıp Dergisi, vol.63, no.2, pp.181-187, 2000 (SCI-Expanded)

XLVI. **17 beta-hydroxysteroid dehydrogenase-3 deficiency: Diagnosis, phenotypic variability, population genetics, and worldwide distribution of ancient and de novo mutations**
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XLVII. **Identification of point mutations in Turkish DMD/BMD families using multiplex-single stranded conformation analysis (SSCA)**
ERASLAN S., Kayserili H., Apak M., KIRDAR B.
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XLVIII. **Genotype and phenotype in patients with dihydropyrimidine dehydrogenase deficiency**
Van Kuilenburg A., Vreken P., Abeling N., Bakker H., Meinsma R., Van Lenthe H., De Abreu R., Smeitink J., Kayserili H., Apak M., et al.
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XLIX. **Analysis of infertile brothers with congenital bilateral absence of vas deferens for mutations in the CFTR gene**
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L. **Seven cases of Wiedemann-Beckwith syndrome, including the first reported case of mosaic paternal isodisomy along the whole chromosome 11**
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LI. **Analysis of the CFTR gene in Turkish cystic fibrosis patients: identification of three novel mutations (3172delAC, P1013L and M1028I)**
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HUMAN GENETICS, vol.102, no.2, pp.224-230, 1998 (SCI-Expanded)

## Articles Published in Other Journals

I. **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)

II. **Türk Noonan Sendromlu Hastalarda Genotip Fenotip İlişkisi**
Altunoğlu U., Denmeyer E., Rosti R. Ö., KARAMAN B., KAYSERİLİ H.
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III. **Mandibuloakral displazi: Vaka sunumu ve laminopatilere genel bakış**
Pehlivan D., BAŞ F., Rosti R. Ö., DARENDELİLER F. F., KAYSERİLİ H.
ÇOCUK DERGİSİ, vol.8, no.4, pp.251-256, 2008 (Peer-Reviewed Journal)

IV. **Down Sendromlu 1416 Postnatal Olgunun Kromozom Analiz Sonuçları**
KARAMAN B., Öztürk H., Yılmaz K., BAŞARAN S., KAYSERİLİ H., Yüksel Apak M.
Çocuk Kliniği Dergisi, vol.7, pp.15-17, 2007 (Peer-Reviewed Journal)

V. **Frajil-X Sendromu Tanısında 20 Yıllık Süreçteki Gelişmeler ve Deneyimlerimiz**

KAYSERİLİ H., Tükel T., KARAMAN B., UYGUNER Z. O., Rosti R. Ö., Açarsöz D., Eriş H., BAŞARAN S., Yüksel Apak M.
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VI. **Gebelikte trizomi 21 ve 18 için biokimyasal tarama testleri**
BAŞARAN S., KARAMAN B., Öztürk H., Yılmaz K., Tükel T., KAYSERİLİ H., Yüksel Apak M.
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VII. **Kuşkulu genitalyalı 103 hastanın etyoloji, tanı ve cinsel kimlik açısından değerlendirilmesi**
DARENDELİLER F. F., BAŞ F., Saka H. N., KAYSERİLİ H., Apak M., Karaaslan N., BUNDAK R., SALMAN F. T., Günöz H.
KLİNİK GELİŞİM, vol.13, pp.109-114, 2000 (Peer-Reviewed Journal)

VIII. **Genetik Hastalıkların Prenatal Tanısı, 1995–1997 yıllarındaki uygulama ve araştırmalarımızın sonuçları**
Yüksel Apak M., BAŞARAN S., Aydınlı K., KAYSERİLİ H., KARAMAN B., Açarsöz D., Eriş H., YÜKSEL A., İbrahimoğlu L., Ermiş H., et al.
İst. Tıp Fakültesi Mecmuası, vol.62, no.4, pp.354-362, 1999 (Peer-Reviewed Journal)

IX. **Duchenne Kas Distrofisi İçin Riskli Ailelerde Taşıyıcılığın Belirlenmesi ve Prenatal Tanı Uygulamalarında Karşılaşılan Sorunlar**
KAYSERİLİ H., Açarsöz D., Gökgöz N., BAŞARAN S., KARAMAN B., Kırdar B., Tolun A., Aydınlı K., YÜKSEL A., Yüksel Apak M.
İstanbul Çocuk Kliniği Dergisi, vol.30, pp.200-206, 1995 (Peer-Reviewed Journal)

## Books & Book Chapters

I. **Tıbbi Genetik**
KAYSERİLİ H., BAŞARAN S., KARAMAN B., UYGUNER Z. O.
in: Pediatride Rutinler, ömer devecioğlu, Editor, İstanbul Tıp Kitabevi, İstanbul, pp.300-305, 2014

II. **Konjenital Adrenal Hiperplazi; Moleküler Tanı, Fenotip/Genotip Korelasyonu ve Antenatal İzlem Deneyimlerimiz (1990-2009).**
Kayserili H., Uyguner Z. O., Karaman B., Başaran S.
in: Endokrinoloji Diyabet Yıllığı, Yarman S,Alagöl F., Editor, İstanbul Tıp Fakültesi, İstanbul, pp.103-113, 2012

III. **Ekstremite Anomalileri**
KALELİOĞLU İ. H., YAŞA C., KAYSERİLİ KARABEY H.
in: Obstetrik ve Jinekolojide Ultrason, Özden S, Editor, Doğan Tıb Kitabevi, İstanbul, pp.336-358, 2009

## Refereed Congress / Symposium Publications in Proceedings

I. **The Application of array CGH for Monogenic Disorders; Clinical and Molecular Cytogenetic Characterization of Twenty Patients.**
KARAMAN B., NAJAFLı A., Toksoy G., KAYSERİLİ KARABEY H., BAŞARAN S.
European Cytogenetic Conference, Floransa, Italy, 29 June - 02 July 2017, pp.8

II. **Familial Microdeletion of 3 Mb at 22q11.2 With Unusual Phenotype**
Toksoy G., Satkın B. N., KAYSERİLİ H., KARAMAN B., BAŞARAN S.
European Cytogenetic Conference, Ireland, 1 - 04 June 2013, pp.69

III. **Molecular Test Results of Syndromic Craniosynostosis Patients:genotype-phenotype correlations**
Karaman V., Altunoğlu U., Toksoy G., KARAMAN B., KAYSERİLİ H., UYGUNER Z. O.
European Human Genetic Congress, France, 1 - 04 June 2013, pp.99

IV. **Array-CGH Findings of de novo Apparently Balanced Chromosomal Rearrangements in Phenotypically Affected 20 Cases**
KARAMAN B., Satkın B. N., KAYSERİLİ H., BAŞARAN S.
European Cytogenetic Conference, Ireland, pp.122

V. **A novel molecular and functional mechanism predisposing to ototoxicity**
KAYSERİLİ H.
American Society of Human Genetics 62nd Annual Meeting, United States Of America, 1 - 04 October 2012, pp.10-15

VI. **Konjenital Eritropoietik Porfiride Eritrodonti:Bir Olgu Raporu**
AKTÖREN O., KAYSERİLİ H., GÜVEN Y.
19.Türk Pedodonti Derneği Kongresi, Antalya, Turkey, 1 - 04 October 2012, pp.163

VII. **Next generation sequencing detects mutations in ISPD as a common cause of Walker-Warburg syndrome with defective glycosylation of adystroglycan**
KAYSERİLİ H.
American Society of Human Genetics 62nd Annual Meeting, United States Of America, 1 - 04 October 2012, pp.48-9

VIII. **DYNC2H1 mutations are commonly found in Juene Asphyxating Thoracic Dyspilasia (JATD) without extraskeletal features while IFT140 mutations cause JATD with renal involvement**
KAYSERİLİ H.
American Society of Human Genetics 62nd Annual Meeting, United States Of America, 1 - 04 October 2012, pp.23-24

IX. **Continuun from monogenic to polygenic and multifactorial disease**
KAYSERİLİ H.
P4 Predictive Preventive Personalized Participatory Mediicine Meeting, Turkey, 1 - 04 September 2012, pp.3-6

X. **Further molevular characterization of PYCR1-related cutis laxa**
KAYSERİLİ H.
European Human Genetics Conferance, Germany, 1 - 04 June 2012, pp.60-61

XI. **Increased sensitivity to DNA damage in a recessive form of Weawer syndrome caused by functional loss of an E3 ubiquitin ligase**
KAYSERİLİ H.
European Human genetics Conference 2012, Germany, 1 - 04 June 2012, pp.3-5

XII. **Mutation in RIPK4 that encodes Receptor-Interacting Serine/Threonine Kinase Protein 4 couse the autosomal recessive form of popliteal ptertgium syndrome**
KAYSERİLİ H.
European Human Genetics Conferance, Germany, 1 - 04 June 2012, pp.45-46

XIII. **Novel c.1731delC mutation in RIN2 gene in two Turkish siblings with MACS/RIN2 syndrome**
KAYSERİLİ H.
European Human Genetics Conference, Germany, 1 - 04 June 2012, pp.57-58

XIV. **Indications for Fetal Karyotyping and Ultrasonographic Findings in Common Trisomies; Alterations in over 2 Decades**
Başaran S., Karaman B., Aytan M., Toksoy G., Kalelioğlu İ. H., Has R., Aydınlı K., Ermiş H., Kayserili H., Dehgan T., et al.
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XV. **Chromosomal Imbalances in Holoprosencephaly Sequence; Results of 87 Cases Diagnosed Prenatally**
KARAMAN B., Satkın B. N., Kalelioğlu İ. H., HAS R., KAYSERİLİ H., Ermiş H., YÜKSEL A., BAŞARAN S.
European Human Genetic Congress, Germany, pp.148-149

XVI. **The Efficiency Of Multiplex Ligation-Dependent Probe AmplificationTechnique In The Diagnosis Of Fetal Chromosomal Abnormalities**
Toksoy G., Karaman B., Uyguner Z. O., Yılmaz K., Has R., Kayserili H., Başaran S.
European Human Genetics Conference 2012, Nuremberg, Germany, 23 - 26 June 2012, pp.150

XVII. **Array-CGH results in fetuses with central nervous system**
Ergin S., KARAMAN B., Satkın B. N., Kalelioğlu İ. H., KAYSERİLİ H., YÜKSEL A., BAŞARAN S.
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XVIII. **How toapproach lissencephaly/subcortical band heterotopia spectrum.**
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XIX. **CLINICAL AND ULTRASONOGRAPHIC FINDINGS OF FETUSES WITH TRISOMY 21, 18 AND 13 CASES DIAGNOSED PRENATALLY IN AMNIOTIC FLUID SAMPLES. FROM JANUARY 1989 TO JULY 2010, 142 TRISOMY 21, 39 TRISOMY 18, 12 TRISOMY 13 CASES WERE DIAGNOSED IN AMNIOTIC FLUID SAMPLES**
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XX. **Novel P.D419Y Mutation in SH3P2 Gene Cause Cherubism in a Turkish Family.**
AKTÖREN O., KAYSERİLİ H., GÜVEN Y.
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XXI. **A novel molecular and functional mechanism predisposing to ototoxicity**
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XXII. **Girl with left hemiatrophy reveals confined mosaicisms for r(13)in fibroblasts**
Altunoğlu U., KARAMAN B., BAŞARAN S., KAYSERİLİ H.
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XXIII. **Investigation of CYFIP1 and CYFIP2 genes in patients with autosomal recessive non-syndromic mental retardation**
Güven G., KAYSERİLİ H., Üzümcü A., Eriş H., KARAMAN B., BAŞARAN S., UYGUNER Z. O.
European Human Genetics, Spain, pp.271

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Semerci C. N., Tufan Şatıroğlu L., Bereket A., TÜYSÜZ B., Yılmaz E., KAYSERİLİ H., KARAMAN B., Düzcan F., Bağcı H.
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XXVI. **A case of monosomy 8p23.3Yter and trisomy 16p13.3Yter; cytogenetic, molecular cytogenetic and clinical findings**
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GÜVEN Y., Rosti R. Ö., TUNA İNCE E. B., KAYSERİLİ H., AKTÖREN O.
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## Supported Projects

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UYGUNER Z. O., KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, NON-SENDROMİK DİŞ AGENEZİSİ İLE İLİŞKİLİ YENİ GENLERİN VE SİNYAL YOLAKLARININ YENİ NESİL DİZİLEME TEKNOLOJİLERİ İLE ARAŞTIRILMASI, 2014 - 2016
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KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, Non-Spesifik Mental Retardasyonlu Olgularda submikroskobik Mikrodelesyon/Mikroduplikasyonların Array Teknolojisi ile Araştırılması, 2011 - 2014
KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, Endoplazmik Retikulum (ER) stres transdüksiyonu OASIS'i kodlayan CREB3L1'in yokluğu otozomal resesif kalıtılan ağır osteogenezis imperfektaya neden olmaktdır., 2013 - 2013
KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, Frontonazal dizostozlu olgularda klinik sınıflandırma ve nörokristopati ile ilişkilendirilmiş genlerin araştırılması, 2010 - 2013
KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, MACS /RIN2 sendromlu iki Türk kardeşte yeni mutasyon tanımlanması, 2012 - 2012
KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, DOĞUMSAL AĞIZ-ÇENE MALFORMASYONLARININ SINIFLANDIRILMASI, GENETİK SENDROMLARLA İLİŞKİLENDİRİLMESİ VE ETYOPATOGENEZİ, 2011 - 2012
KAYSERİLİ KARABEY H., EU Framework Program Project, Kraniyofasiyel Malformasyonlara Klinik ve Genetik Yaklaşım CRANIRARE, 2009 - 2012
KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, Gelişmekte olan ülkelerde 1. basamak hizmetlerinde genetiğin rolü, 2011 - 2011
KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, nöronal migrasyon anomalilerinde genotip-fenotip ilişkisi, 2009 - 2011
KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, Otozomal resesif mikrosefali'de klinik ve moleküler çalışmalar, 2008 - 2011
KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, Nadir Hastalıkların Moleküler Temellerinin Araştırılması, 2008 - 2011
KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, JUBERT SENDROMUNUN OKÜLO-RENAL FORMUNDA SAPTANAN CEP290 MUTASYON ARAŞTIRMALARI, 2007 - 2010
KAYSERİLİ KARABEY H., Project Supported by Higher Education Institutions, 25 ve 31 Haftalarda Kısa Femur Saptanması Nedeniyle Yapılan Testlerde Leroy I-Cell Tanısı alan Fetusların Postnatal Takibi, 2007 - 2008

## Activities in Scientific Journals

American Journal of Medical Genetics Part A , Publication Committee Member, 2013 - Continues
European Journal of Medical Genetics , Publication Committee Member, 2005 - Continues

## Memberships / Tasks in Scientific Organizations

European Society of Human Genetics, Member, 1994 - Continues
Tıbbi Genetik Derneği, Board Member, 2007 - 2011

## Scientific Refereeing

European Journal of Medical Genetics , SCI Journal, July 2013
American Journal of Medical Genetics Part A , SCI Journal, May 2013
American Journal of Medical Genetics Part A, SCI Journal, April 2013
Cocuk Dergisi İstanbul Tıp Fakültesi / İstanbul Üniversitesi , National Scientific Refreed Journal, October 2012

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## Congress and Symposium Activities

6. Dismorfoloji Günleri, Attendee, Salt Galata Karaköy / İstanbul, Turkey, 2013
10.Ulusal Tıbbi Genetik Kongresi, Attendee, Uludağ Üniversitesi / Bursa, Turkey, 2012
Tıbbi Genetikle Tanışma, Attendee, İÜ Baltalimanı Sosyal Tesisleri / İstanbul, Turkey, 2012
24.Endokrinoloji ve Metabolizma Hastalıkları Mezuniyet Sonrası Eğitim Kursu, Attendee, Askeri Müze Kültür Sitesi / İstanbul, Turkey, 2012
P4 Predictive Preventive Personalized Participatory, Attendee, Anadolu Üniversitesi / Eskişehir, Turkey, 2012
European Human Genetics Conference 2012, Attendee, Nürnberg / Almanya, Germany, 2012
3.Pediatri Günleri ve 13. Pediatri Hemşireliği Günleri, Attendee, Ceylan Intercontinental, Turkey, 2012
Acıbadem Üniv Pediatri Anabili Dalı Toplantıları OTİZM PANELİ, Attendee, Acıbadem Üniversitesi Tıp Fakültesi, Turkey, 2012
Mezuniyet Sonrası Eğitim Programı 2011-2012, Attendee, İstanbul Tıp Fakültesi İSTEM Salonu / İstanbul, Turkey, 2012
Fourth European Course in Clinical Dysmorphology, Attendee, Roma / İtalya, Italy, 2011
International Genetics Education Network, Attendee, Montreal , United Kingdom, 2011
European Biotechnology Congress, Attendee, İstanbul, Turkey, 2011
European of Human Genetics Conference, Attendee, Amsterdam , Netherlands, 2011
5. Dismorofoloji Günleri, Attendee, Nippon Otel / Taksim / İstanbul, Turkey, 2011
33.Pediatri Günleri ve 12.Pediatri Hemşireliği Günleri, Attendee, Ceylan İntercontinental / İstanbul, Turkey, 2011
7.Uludağ Pediatri Kış Kongresi, Attendee, Uludağ Üniv. Tıp Fak. / Bursa, Turkey, 2011
1. Nörometabolik Dismorfoloji Sempozyumu, Attendee, İstanbul , Turkey, 2011
Mezuniyet Sonrası Eğitim Programı 2010-2011, Attendee, İstanbul Tıp Fakültesi Çocuk Sağ. ve Hastalıkları AD, Turkey, 2011
4. Dismorfoloji Günleri, Attendee, Nippon Otel / Taksim / İstanbul, Turkey, 2009