

CURRICULUM VITAE

MARIOS KAMBOURIS, PhD, FACMG

Medical & Molecular Genetics

Assistant Professor Adjunct, Yale University School of Medicine, USA

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PERSONAL DATA

DoB: 3 March 1961
Citizenship: USA / Hellenic (Dual)

EDUCATION

- Board Certifications:** American Board of Medical Genetics: **Clinical Molecular Genetics** 1996
Certification #: 96091 **Ph.D. Medical Genetics** 1996
- Post-Doctoral Fellowships:** **Clinical Molecular Genetics** 1993-1995
Medical Genetics 1993-1995
Medical Genetics & Birth Defects Center, **Henry Ford Hospital**, Detroit, Michigan, USA
- Ph.D.:** Major: **Medical and Molecular Genetics** 1993
Minor: **Life Sciences**
Department of Medical & Molecular Genetics, **Indiana University School of Medicine**, Indianapolis, Indiana, USA
- B.S.:** Major: **Biochemistry** 1984
Minor: **Molecular Biology**
Departments of Biochemistry and Biology, **State University of New York**, Buffalo, New York, USA

MEMBERSHIPS

- Member,** American Society of Human Genetics 1993 - Present
- Diplomate,** American Board of Medical Genetics 1996 - Present
- Fellow,** American College of Medical Genetics 1996 - Present

EMPLOYMENT HISTORY

- Assistant Professor Adjunct** Department of Genetics 1995 - Present
Yale University School of Medicine, New Haven, Connecticut, USA
- Biotechnology Consultant** **Zenican Consulting Limited** 2009 - Present
Limassol, Cyprus
- Chief Scientific Officer** **Geno-Type Biotechnology** 2005 – 2009
Athens - Greece
- Chief Scientific Officer** **Synergene Biotechnology Group** 2003 – 2005
Qormi, Malta
- Scientist** Head, Laboratory for Molecular Genetics & DNA Diagnostics 1995 - 2003
Department of Genetics, **King Faisal Specialist Hospital & Research Center**, Riyadh, Saudi Arabia

EXPERIENCE*** SCIENTIFIC & BUSINESS DEVELOPMENT****Scientific & strategic planning**

- Research & Development programs
- Molecular diagnostic services
- “Know-how” marketing in Medical & Molecular Genetics and Genomics
- Formulation of Global Alliances
- Venture capital recruiting
- Private investment fund raising

Genomics Business Development

Jointly responsible for the formulation of the scientific plan and for negotiating the formation of a Genomics company (AraGene) on behalf of the Research Center of King Faisal Hospital (Riyadh, Saudi Arabia) with British Aerospace Systems (part of an offset program) and Merlin Ventures (a UK based Venture Capitalist Company).

*** MOLECULAR GENETICS**

- **Head: Laboratory for Molecular Genetics & DNA Diagnostics** **1995 - 2003**
Department of Genetics, **King Faisal Specialist Hospital & Research Center**, Riyadh, Saudi Arabia

Established and directed at King Faisal Specialist Hospital & Research Center a Molecular Genetics laboratory with dual function. DNA Diagnostic and Molecular Genetics Research. DNA diagnostic testing was performed for inherited diseases and predisposition factors such as **Factor V Leiden, Cystic Fibrosis, Fragile-X syndrome, Huntington disease** (& disorders caused by trinucleotide repeat expansion), **Multiple Endocrine Neoplasia 2A & 2B, Gaucher disease, Sickle cell disease** and many more. Research activity was focused on mapping human disease genes. Directly involved with the localization and identification or characterization of more than ten novel human genes. In addition to mapping Mendelian disorders directly involved in utilization of populations with unique genetic characteristics for target gene discovery in polygenic multifactorial disorders (Type 2 Diabetes Mellitus, Cardiovascular diseases etc).

- **Post-Doctoral Fellow: DNA Diagnostics Laboratory** **1993-1995**
Medical Genetics and Birth Defects Center, **Henry Ford Hospital**, Detroit, Michigan USA

Established, Performed, Interpreted & Signed DNA Diagnostic testing (clinical & prenatal) for: **Cystic Fibrosis, Fragile-X Syndrome, 2A & 2B, Myotonic Dystrophy & Sickle Cell Disease, etch**

- **Post Doctoral Fellow: Molecular Cytogenetics Laboratory** **1995**
Medical Genetics and Birth Defects Center, **Henry Ford Hospital**, Detroit, Michigan, USA

Performed and interpreted Fluorescent *In Situ* Hybridization (FISH) for Critical Deletion Syndromes (Prader-Willie, Angelman, Cri-du-Chat), Cosmid, α -Satellite and Centromere Probes

- **Doctoral Training: Molecular Genetics Laboratory** **1985-1992**
Medical and Molecular Genetics Department, **Indiana University School of Medicine**, Indianapolis, Indiana USA

* **CLINICAL GENETICS**

- **Post Doctoral Fellow: Medical Genetics Clinic** **1993-1995**
Medical Genetics and Birth Defects Center, **Henry Ford Hospital**, Detroit, Michigan, USA

Diagnosis, Risk assessment, Counseling and Management of patients with a multitude of genetic disorders, congenital malformations, dysmorphic features and mental retardation. Patients were seen in daily **Genetics clinics**, **In-Patient Consultations** and monthly **Field Clinics**

- **Doctoral Training: Medical Genetics Clinic** **1985-1992**
Department of Medical and Molecular Genetics, **Riley Hospital for Children**, Indianapolis, Indiana, USA

Diagnosis, Risk assessment and Counseling for patients seen in weekly **Genetics Clinics**, **In-Patient Consultations** (congenital malformations, dysmorphic features, mental retardation), **Growth Clinic** (skeletal dysplasias and growth abnormalities) and **Neurology Clinic** (neurogenetic disorders)

- **Genetic Associate: Prenatal Diagnosis Clinic** **1986-1989**
Department of Obstetrics and Gynecology, **Indiana University Hospital**, Indianapolis, Indiana, USA

Risk assessment and Genetic counseling for high risk pregnancy patients (maternal age, family history of genetic disorders, teratogenic exposures, abnormal MSAFP).

PUBLICATIONS* **ORIGINAL ARTICLES (refereed):**

1. **Kambouris M, Dlouhy SR, Trofatter JA, Connealy PM, Hodes ME: Localization of the Gene for X-linked Nephrogenic Diabetes Insipidus to Xq28. *American Journal of Medical Genetics* 29: 239-247, 1988.**
2. **Kambouris M, Sangameswaran L, Dlouhy SR, Ghetti B, Hodes ME, Triarhou LC: Cellular Distribution of the RNA Transcripts of a Newly Discovered Gene in the Brain of Normal, Weaver, Purkinje Cell Degeneration and Reeler Mutant Mice as Evidenced by *In Situ* Hybridization Histochemistry. *Molecular Brain Research* 18: 321-328, 1993.**
3. **Kambouris M, Triarhou LC, Dlouhy SR, Sangameswaran L, Luo F, Ghetti B, Hodes ME: Novel cDNA Clones Obtained by Antibody Screening of a Mouse Cerebellar cDNA Expression Library. *Molecular Brain Research* 25: 183-191, 1994.**
4. **Kambouris M, Sangameswaran L, Triarhou LC, Kozak CA, Dlouhy SR, Ghetti B, Hodes ME: Molecular Characterization of a Novel cDNA from Murine Cerebellum: Developmental Expression and Distribution in Brain. *Molecular Brain Research* 25: 192-199, 1994.**
5. Goldstein DJ, **Kambouris M**, Ward, RE: **Familial Crossed Polysyndactyly. *American Journal of Medical Genetics* 50: 215-223, 1994.**
6. Feldman GL, **Kambouris M**, Talpos GB, Mulligan LM, Ponder BAJ, Jackson CE: **Clinical Value of Direct DNA Analysis of the *RET* Proto-oncogene in Families with Multiple Endocrine Neoplasia Type 2A. *Surgery* 116: 1042-1047, 1994.**
7. Dennehy PJ, Feldman GL, **Kambouris M**, O'Malley ER, Sanders CY, Jackson CE: **Relationship of Familial Prominent Corneal Nerves and Lesions of the Tongue Resembling Neuromas to Multiple Endocrine Neoplasia Type 2B. *American Journal of Ophthalmology* 120: 456-461, 1995.**
8. **Kambouris M, Jackson CE, Feldman GL: Diagnosis of Multiple Endocrine Neoplasia (MEN) 2A, 2B and Familial Medullary Thyroid Carcinoma (FMTC) by Multiplex PCR and Heteroduplex Analyses of *RET* Proto-oncogene Mutations. *Human Mutation* 8: 64-70, 1996.**

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9. **Kambouris M, Snow K, Thibodeau S, Green M, Bluhm D, Feldman GL: Unusual Somatic Instability in the FMR-1 Locus in a Mildly Affected Male.** *American Journal of Medical Genetics* 64: 404-407, 1996.
 10. Stephens JC, ...**Kambouris M, ... Dean M: Dating of the Origin of the CCR5Δ32 AIDS-Resistance Allele by the Coalescence of Haplotypes.** *American Journal of Human Genetics* 62: 1507-1515, 1998.
 11. Quari MH, Khalil SH, **Kambouris M, Meyer BF: Mutation of p16, p21 or Cyclin Dependent Kinase 4 is Rare in Acute Lymphoblastic Leukemia.** *British Journal of Hematology* 103: 467-472, 1998.
 12. Al-Jishi E, Meyer BF, Al-Essa M, Rashed M, Al-Hamed MH, Sakati N, Sanjad S, Ozand PT, **Kambouris M: Clinical, Biochemical and Molecular Characterization of Patients with Glutathione Synthetase Deficiency.** *Clinical Genetics* 55: 444-449, 1999.
 13. Diaz GA, Gelb BD, Ali F, Sakati N, Sanjad S, Meyer BF, **Kambouris M: The Sanjad-Sakati and Autosomal Recessive Kenny-Caffey Syndromes are Allelic: Evidence for an Ancestral Founder Mutation and Locus Refinement.** *American Journal of Medical Genetics* 85: 48-52, 1999.
 14. Bohlega S, **Kambouris M, Shahid M, Homsy M, Al-Sous W: Gaucher Disease with Oculomotor Apraxia and Cardiovascular Calcification. (Gaucher Type IIIC).** *Neurology*, 54: 261-3, 2000.
 15. **Kambouris M, Bohlega S, Al-Tahan A, Meyer BF: Localization of the Gene for a Novel Autosomal Recessive Neurodegenerative Huntington-like Disorder to Hsa 4p15.3** *American Journal of Human Genetics*, 66: 445-452, 2000.
 16. **Kambouris M, Meyer BF Interpretation of linkage data for a Huntington-like disorder mapping to 4p15.3.** *American Journal of Human Genetics* 67: 263, 2000.
 17. **Kambouris M, Banjar H, Moggari I, Nazer H, Al-Hamed M, Meyer BF: Identification of Novel Mutations in Arabs with Cystic Fibrosis and their Impact on the Cystic Fibrosis Transmembrane Regulator Mutation Detection Rate in Arab Populations.** *European Journal of Pediatrics*, 159: 303-309, 2000.
 18. Bohlega S, Al-Tahan A, **Kambouris M, Divakaran M: Neurodegenerative Huntington-Like Disorder.** *Movement Disorders*, 16: 533-534, 2001.
 19. Martignetti JA, Al Aqeel A, Al Sewairi W, Boumah CE, **Kambouris M, Al Mayouf S, Sheth KV, Al Eid W, Dowling O, Harris J, Glucksman MJ, Bahabri S, Meyer BF, Desnick RJ. Inherited Mutations in the Matrix Metalloproteinase 2 Gene (mmp-2) Result in a Syndrome of Multicentric Osteolysis and Arthritis.** *Nature Genetics*, 28: 261-265, 2001.
 20. Abu-Amero K, Wyngaard C, **Kambouris M, Dzimir N: Prevalence of the 20210 G→A Prothrombin Variant and its Association with Coronary Artery Disease in Middle-Eastern Arab Population.** *Archives of Pathology and Laboratory Medicine*, 126: 1087-1090, 2002.
 21. The HRD/Sanjad-Sakati/Autosomal Recessive Kenny-Caffey Syndrome Consortium: Group 1: Parvari R, Hershkovitz E, Grossman N, Gorodischer R, Loeys B, Zecic A, Mortier G, Gregory S, Sharony R, Group 2: **Kambouris M, Sakati N, Meyer BF, Group 3: Al Aqeel AI, Al Humaidan A, Al Zahrani F, Al Swaid A, Al Othman J, Group 4: Diaz GA, Weiner R, K. Khan TS, Gordon R, Gelb BD: Mutation of a Tubulin-Specific Chaperone Gene, TBCE, Causes the HRD/Sanjad-Sakati/Autosomal Recessive Kenny-Caffey Syndrome.** *Nature Genetics*: 32 448-452, 2002
 22. Hodgkinson CA, Bohlega SA, Abu-Amero SN, Cupler EJ, **Kambouris M, Meyer BF, Bharucha VA: A Novel Autosomal Recessive Pure Hereditary Spastic Paraplegia in a Saudi Family Showing Linkage to Chromosome 13q14.** *Neurology*, 59: 1905-1909, 2002.
 23. Abu-Amero, K, Wyngaard CA, Al-Boudari OM, **Kambouris M, Dzimir N: Lack of Association of Lipoprotein Lipase Gene Polymorphisms with Coronary Artery Disease in Arab Populations.** *Archives of Pathology and Laboratory Medicine*. 127: 597-600, 2003.
 24. Bohlega S. Lach B. Meyer BF. Al Said Y. **Kambouris M. Al Homsy M. Cupler EJ: Autosomal dominant hyaline body myopathy: clinical variability and pathologic findings.** *Neurology* 61: 1519-23, 2003.

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25. Khan A. Al-Saif A. **Kambouris M: A Novel KERA Mutation Associated With Autosomal Recessive Cornea Plana.** *Ophthalmic Genetics* 25: 147-52, 2004.
 26. Khan AO. Shamsi FA. Al-Saif A. **Kambouris M: A Novel Missense Norrie Disease Mutation Associated With A Severe Ocular Phenotype.** *Journal of Pediatric Ophthalmology & Strabismus.* 41: 361-3, 2004.
 27. **Kambouris M.: Target Gene Discovery In Extended Families With Type 2 Diabetes Mellitus.** *Atherosclerosis Supplement* 6: 31-36, 2005.
 28. Coucke Paul J; Willaert Andy; Wessels Marja W; Callewaert Bert; Zoppi Nicoletta; De Backer Julie; Fox Joyce E; Mancini Grazia M S; **Kambouris Marios**; Gardella Rita; Facchetti Fabio; Willems Patrick J; Forsyth Ramses; Dietz Harry C; Barlati Sergio; Colombi Marina; Loeys Bart; De Paepe Anne: **Mutations in the facilitative glucose transporter GLUT10 alter angiogenesis and cause arterial tortuosity syndrome.** *Nature genetics*;38:452-457, 2006

* **ORIGINAL ARTICLES OF REGIONAL INTEREST (refereed):**

1. Banjar H, **Kambouris M**, Meyer BF, al-Mehaidib A, Mogarri I **Geographic distribution of cystic fibrosis transmembrane regulator gene mutations in Saudi Arabia.** *Annals of Tropical Paediatrics* 19: 69-73, 1999.
2. Abdul Wahab A, Al Thani G, Dawod ST, **Kambouris M**, Al Hamed M **Heterogeneity of the cystic fibrosis phenotype in a large kindred family in Qatar with cystic fibrosis mutation (I1234V).** *Journal of Tropical Paediatrics* 47: 110-112, 2001.
3. Wahab AA, Janahi IA, Hebi S, al-Hamed M, **Kambouris M** **Cystic Fibrosis in a Child from Syria.** *Annals of Tropical Paediatrics* 22: 53-55, 2002.
4. Wahab A, Al Thani G, Dawod S, **Kambouris M**, Al Hamed M **Rare CFTR mutation 1525-1G→A in a Pakistani patient.** *Journal of Tropical Pediatrics.* 50:120-122, 2004.

• **ABSTRACTS:**

1. **Kambouris M**, Dlouhy SR, Trofatter JA, Hodes ME: **Preliminary Localization of X-linked Nephrogenic Diabetes Insipidus in the XqTerminal Region.** *American Journal of Human Genetics [Supplement]* 41: A171, 1987.
2. Goldstein DJ, **Kambouris M**: **Familial Mixed Polysyndactyly.** *American Journal of Human Genetics [Supplement]* 41: A63, 1987.
3. **Kambouris M**, Hodes ME, Connealy PM, Trofatter JA, Dlouhy SR: **Nephrogenic Diabetes Insipidus Linked to Xq28.** *Abstracts of the Ninth International Workshop on Human Gene Mapping*, 1987.
4. **Kambouris M**, Hodes ME, Connealy PM, Trofatter JA, Dlouhy SR: **Nephrogenic Diabetes Insipidus Linked to Xq28.** *Cytogenetics and Cell Genetics* 46: 636, 1987.
5. **Kambouris M**, Sangameswaran L, Dlouhy SR, Triarhou LC, Ghetti B, Hodes ME: **Granule Cell Antiserum Identifies a Brain Specific cDNA in an Expression Library Derived from Neonatal Heterozygous Weaver Cerebella.** *Society for Neuroscience Abstracts* 17: 556, 1991.
6. **Kambouris M**, Sangameswaran L, Triarhou LC, Dlouhy SR, Ghetti B, Hodes ME: **Granule Cell Antiserum Identifies GC cDNAs in an Expression Library Derived from Neonatal Heterozygous Weaver Cerebella.** *Journal of Neuropathology and Experimental Neurology* 51: 350, 1992.
7. **Kambouris M**, Triarhou LC, Sangameswaran L, Dlouhy SR, Ghetti B, Hodes ME.: **Molecular Characterization of a Novel Cerebellar cDNA, Prenatal Ontogeny, and Cellular Distribution in the Brains of Normal, Weaver, PCD and Reeler Mutant Mice.** *Society for Neuroscience Abstracts* 18: 53, 1992.

8. **Kambouris M, Triarhou LC, Sangameswaran L, Dlouhy SR, Ghetti B, Hodes ME: Identification of 15 Novel Cerebellar cDNAs by Screening a Neonatal Heterozygous Weaver Cerebellar Expression Library with an Anti-Granule Cell Antiserum; Molecular Characterization, Prenatal Ontogeny, and Cellular Distribution of One Novel cDNA in Neurological Mutant Mice and in Human Cerebellum.** *American Journal of Human Genetics [Supplement] 51: A121, 1992.*
9. **Kambouris M, Hodes ME, Ghetti B, Triarhou LC: Cellular Distribution of a Novel Cerebellar cDNA in the Brain of Normal, Weaver, Purkinje Cell Degeneration and Reeler Mutant Mice as Revealed by *In Situ* Hybridization Histochemistry.** *Abstracts of the International Brain Research Organization Workshop on Mechanism of Neuronal Plasticity*, p. 86, 1992.
10. **Kambouris M, Sangameswaran L, Triarhou LC, Kozak CA, Dlouhy SR, Ghetti B, Hodes ME: Molecular Characterization of Two Novel cDNAs Obtained by Antibody Screening of a Mouse Cerebellar cDNA Expression Library.** *Journal of Neuropathology and Experimental Neurology 52: 287, 1993.*
11. **Kambouris M, Sangameswaran L, Triarhou LC, Kozak CA, Dlouhy SR, Ghetti B, Hodes ME: Mouse Cerebellar cDNA Expression Library Screening: Novel clones.** *Clinical Neuropathology [Supplement 1]: 12: S49, 1993.*
12. **Feldman GL, Li P, Kambouris M, Ponder BAJ, Mulligan LM, Jackson CE: Presymptomatic Diagnosis of Multiple Endocrine Neoplasia 2A (MEN 2A) by Direct Mutation Analysis of the *RET*-Proto Oncogene.** *American Journal of Medical Genetics, 52: 366, 1994.*
13. **Kambouris M, Snow K, Thibodeau S, Green M, Bluhm D, Feldman GL: Segregation of the Fragile-X Mutation from an Affected Male: Evidence of Unusual Somatic Instability in the *FMR-1* Locus.** *American Journal of Human Genetics [Supplement] 55: A225, 1994.*
14. **Kambouris, M, Jackson CE, Feldman GL: Presymptomatic Diagnosis of MEN 2A, MEN 2B and FMTC Using Multiplex PCR and Mutation Detection Enhancement (MDE) for Simultaneous Heteroduplex Identification of Multiple *RET* Proto-oncogene mutations.** *26th March of Dimes Clinical Genetics Conference; 2nd Annual Meeting of the American College of Medical Genetics.* 1994.
15. **Kambouris M, Jackson CE, Feldman GL: Diagnosis of Multiple Endocrine Neoplasia [MEN] 2A, 2B & Familial Medullary Thyroid Cancer [FMTC] by multiplex PCR and heteroduplex analyses of *RET* proto-oncogene mutations.** *American Journal of Human Genetics [Supplement] 57: A68, 1995.*
16. **Kambouris M, Meyer BF, Banjar H, Al-Hamed MH, Moggari I, Ozand P. Identification of Two Novel CFTR Exonic Deletions [425del142 & 1549delG] in Cystic Fibrosis (CF) Patients by Mutation Detection Enhancement (MDE) Heteroduplex Analyses. Possible Founder Effect Associated with the 1540A→G Polymorphism.** *American Journal of Human Genetics [Supplement] 59: A397 1996.*
17. **Meyer BF and Kambouris M. Resolution of Homozygous Sequence Alterations in the CFTR Gene by Mutation Detection Enhancement (MDE) Analysis Independent of Heteroduplex Formation Reveals a Novel Mutation [548A→T] that Causes Cystic Fibrosis in Homozygous Patients.** *American Journal of Human Genetics [Supplement] 59: A399 1996.*
18. **Bohlega S, Shahid M, Kambouris M, Al-Sous W. Gaucher's Disease Variant with Oculomotor Apraxia & Cardiovascular Calcification: Unique Genotype with 1342C (D409H) Mutation.** *European Neurological Society Abstracts*, 1997
19. **Kambouris M, Rahbeeni Z, Meyer BF, Al-Yamani EA, Ozand PT, Rashed M: Mutation screening of the MCAD gene in Patients with Biochemical Medium-Chain Fatty Acid Oxidation Defects (MCFAOD).** *American Journal of Human Genetics [Supplement] 61: A254 1997.*
20. **Al-Jishi E, Meyer BF, Rashed M, Al-Hamed MH, Sakati N, Sanjad S, Ozand PT, Kambouris M: Molecular Analyses of the Glutathione Synthetase (GSS) Gene in Patients with Pyroglutamic Aciduria.** *American Journal of Human Genetics [Supplement] 61: A248 1997.*
21. **Meyer BF, Qari M, Kambouris M, Khalil S: Screening of Primary Acute Lymphoblastic Leukemia Samples for the Presence of Mutations in the p16, p21 & CDK4 Genes.** *Blood 90: 3626 1997.*

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22. **Kambouris M, Bohlega S, Tahan A, Meyer BF: Localization of the Gene for a Novel Basal Ganglia Progressive Neurodegenerative Disorder to Hsa 4p16.1** *American Journal of Human Genetics [Supplement] 63: A294, 1998.*
 23. **Ali F, Diaz G, Sakati N, Sanjad S, Meyer B, Gelb B, Kambouris M: Localization of the Gene for the Sanjad-Sakati syndrome to 1q43.** *American Journal of Human Genetics [Supplement] 6: A279, 1998.*
 24. **Meyer BF, Kambouris M: Mutation Detection Enhancement (MDE) Heteroduplex Analysis Reveals Single Nucleotide Polymorphisms Associated with Uninformative Microsatellites.** *American Journal of Human Genetics [Supplement] 63: A301, 1998.*
 25. **Diaz GA; Khan KT; Sakati N; Sanjad S; Meyer BF; Kambouris M; Gelb BD Linkage mapping of the Kenny-Caffey syndrome and evidence for allelism with the Sanjad-Sakati syndrome.** *Pediatric Research 45: 798 1999*
 26. **Diaz GA; Khan KT; Sakati N; Sanjad S; Meyer BF; Kambouris M; Gelb BD Linkage mapping of the Kenny-Caffey syndrome and evidence for allelism with the Sanjad-Sakati syndrome.** *Journal of Investigative Medicine 47: 164A, 1999*
 27. **Kambouris M, Meyer BF, Bahabri S: Localization of the Gene for NAO Syndrome (Nodulosis, Arthropaty & Osteolysis) to 16p11.2-21.** *American Journal of Human Genetics [Supplement] 65: A59, 1999.*
 28. **Kambouris M, Shabib S, Nazer H, Al-Mehaidib A, Abu-Amero S, Meyer BF: Localization of the gene for a novel disease characterized by Intestinal Lymphangiectasia to 6p21-22. Evidence for genetic heterogeneity.** *American Journal of Human Genetics [Supplement] 67: 324, 2000.*
 29. **Abu-Amero S, Meyer BF, Boumah CE, Hodgkinson C, Barucha V, Butt A, Cupler E, Kambouris M, Bohlega S: A Novel Congenital Autosomal Dominant Hyaline Body Myopathy in a Saudi Family Showing Linkage to Chromosome 6q.** *American Journal of Human Genetics [Supplement] 67: 274, 2000.*
 30. **Meyer BF, Kambouris M, El-Samadi S, Boumah C, Abu-Amero S, Butt A, Al-Fadley A, Crossner C, Pedersen K, Ullbro C: Phenotype/Genotype Correlation of a Cohort of Patients with Papillon-Lefevre Syndrome.** *American Journal of Human Genetics [Supplement] 67: 2329, 2000.*
 31. **Cupler E, Hodgkinson C, Abu-Amero S, Meyer BF, Kambouris M, Boumah C, Bohlega S, Bharucha VA: Nonaka Myopathy In A Large Saudi Kindred Not Linked To Chromosome 9** *Neurology 56: A440, 2001.*
 32. **Hejazi NS; Chaves EC; Boumah C; Abu-Amero SSN; Dabbagh O; Ozand P; Essa M; Kambouris M; Meyer B Linkage of hyperekplexia (HEK) in three Saudi families to chromosome 4q31.3 associated with the glycine receptor beta subunit (GLR beta)** *Neurology 56: A132, 2001.*
 33. **Martignetti JA, Al Aqeel A, Al Sewairi W, Boumah CE, Kambouris M, Al Mayouf S, Sheth KV, Al Eid W, Dowling O, Harris J, Glucksman MJ, Bahabri S, Meyer BF, Desnick RJ: The First Matrix Metalloproteinase Disease: MMP-2 Deficiency Results in A Multicentric Osteolysis Syndrome.** *American Journal of Human Genetics [Supplement] 69: 189, 2001.*
 34. **Hodgkinson CA, Bohlega SA, Cupler EJ, Abu-Amero SN, Boumah CE, Kambouris M, Meyer BF, Bharucha VA: A Novel Autosomal Recessive Pure Hereditary Spastic Paraplegia in a Saudi Family Showing Linkage to Chromosome 13q.** *American Journal of Human Genetics [Supplement] 69: 499, 2001.*
 35. **Bharucha VA, Cupler EJ, Bohlega SA, Boumah CE, Abu-Amero SN, Kambouris M, Meyer BF, Hodgkinson CA: A Novel Distal Myopathy with Rimmed Vacuoles in a Large Saudi Kindred is Linked to Chromosome 12.** *American Journal of Human Genetics [Supplement] 69: 538, 2001.*
 36. **Abu-Amero K, Bohlega S, Al-Shubili A, Kambouris M: Clinical and Genetic Spectrum of CADASIL in Three Arab Families.** *British Human Genetics Conference, 2002.*
 37. **Abu-Amero S, Bohlega S, Barucha V, Hodgkinson C, Lach B, Boumah CE, Cupler EJ, Kambouris M, Meyer BF: Autosomal Dominant Hyaline Body Myopathy in a Saudi Family is Linked to Chromosome 14q.** *British Human Genetics Conference, 2002.*

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38. **Kambouris M, Bohlega S, Trabzuni D, Meyer BF: Localization of the Gene for a Novel Autosomal Recessive Neuromuscular Disease Featuring Tremulous and Myoclonic Dystonia with MRI White Matter Alterations.** *American Journal of Human Genetics [Supplement] 71*: 443, 2002
 39. Hodgkinson CA, Bohlega S, Abu-Amero SN, **Kambouris M**, Cupler E, Meyer BF, Bharucha VA: **A Refined Interval for the Autosomal Recessive Nonsyndromic Deafness Locus DFNB6.** *American Journal of Human Genetics [Supplement] 71*: 431, 2002.
 40. The HRD/Sanjad-Sakati/Autosomal Recessive Kenny-Caffey Syndrome Consortium. Diaz GA, Al Aqeel A, Gelb BD, Gordon R, Gorodischer R, Gregory S, Grossman N, Hershkovitz E, **Kambouris M**, Khan KTS, Loeys B, Meyer BF, Mortier G, Parvari R, Sakati N, Sharony R, Weiner R, Zecic A **Mutation of a Tubulin-Specific Chaperone, TBCE, Causes the HRD/Sanjad-Sakati/Autosomal Recessive Kenny-Caffey Syndrome.** *American Journal of Human Genetics [Supplement] 71*: 209, 2002.
 41. Coucke1 PJ, Willaert A, Callewaert B, Wessels MW, Mancinni GM, De Backer J, Fox JE, Kambouris M, Gardella R, Barlati S, Colombi M, Dietz HC, Loeys B, Willems PJ, De Paepe A **Mutations in GLUT10/SLC2A10 a Facilitative Glucose Transporter Cause Arterial Tortuosity Syndrome.** *American Journal of Human Genetics [Supplement] XX*: 39, 2005.