Form 500.1.3

# Academic Personnel Short Profile / Short CV

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| **University:** | University of Nicosia |
| **Surname:** | Toumba |
| **Name:** | Meropi |
| **Rank/Position:** | Clinical Associate Professor |
| **Faculty:** | Medical School |
| **Department:** | Basic & Clinical Sciences |
| **Scientific Domain:** | Paediatric Endocrinology |

## Academic qualifications

| Qualification | Year | Awarding Institution | Department | Thesis title (Optional Entry) |
|---|---|---|---|---|
| Doctor of Medicine, MD | 1997 | Aristotelion University of Thessaloniki | Medical School | |
| Specialist in Pediatrics | 2003 | Aristotelion University of Thessaloniki | Hippokration Hospital | |
| Specialist in Pediatric Endocrinology | 2007 | The University College of London hospitals | Pediatric Endocrinology at The Great Ormond st Hospital for Sick Children | |

Academic Staff Short Profile

| Employment history in Academic Institutions/Research Centers | | | | |
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| **Period of employment** | | **Employer** | **Location** | **Position** |
| **From** | **To** | | | |
| 2023 | Present | The University of Nicosia | Nicosia, CY | Clinical Associate Professor |
| 2007 | Present | Private practice-self employed and visiting consultant at the Aretaeion and AMC hospitals | Nicosia, Limassol, CY | Consultant in Pediatric Endocrinology |
| 2005 | 2007 | The Great Ormond Street Hospital and the Institute of Child Health | London, UK | Registrar<br>Clinical and Research Fellow in Pediatric Endocrinology |

| Key ***refereed*** journal papers, monographs, books, conference publications etc. | | | | | | |
|---|---|---|---|---|---|---|
| **Ref. Number** | **Year** | **Title** | **Other authors** | **Journal and Publisher / Conference** | **Vol.** | **Pages** |
| 1 | 2023 | The pathogenic p.Gln319Ter variant is not causing congenital adrenal hyperplasia when inherited in one of the duplicated CYP21A2 genes | Fanis P, Skordis N, **Toumba M,** Picolos M, Tanteles G. A., Neocleous V, Phylactou, L. A. | Front Endocrinol (Lausanne) | 14 | doi.org/10.3389/fendo.2023.1156616 |
| 2 | 2023 | Reduced serum concentrations of biomarkers reflecting Leydig and Sertoli cell function in male patients with congenital adrenal hyperplasia. | Johannsen TH, Albrethsen J, Neocleous V, Baronio F, Cools M, Aksglaede L, Jørgensen N, Christiansen P, **Toumba M**, Fanis P, Ljubicic ML, Juul A. | Endocr Connect | 23 | doi: 10.1530/EC-230073. |

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|---|---|---|---|---|---|---|
| 3 | 2023 | Methylation status of Fanis P, hypothalamic *Mkrn3* promoter across puberty. **Toumba M**, | Morrou M, Tomazou Front M, Michailidou K, Spyrou Skordis N, .1075341 Neocleous V, Phylactou LA. | Endocrinol 13 doi: GM, (Lausanne) | | 10.3389/fendo.2022 |
| 4 | 2023 | Stress, Thyroid Dysregulation, and Thyroid Cancer in Children and Adolescents: Proposed Impending Mechanisms. | Kyriacou A, Tziaferi V, **Toumba M**. | Horm Res Paediatr | 96 | 2023;96(1):44-53. doi: 10.1159/000524477 |
| 5 | 2022 | Current clinical practice for thromboprophylaxis management | van Haalen FM, Kaya M, Pelsma ICM, Dekkers OM, | Orphanet J Rare Dis | 17 | 2022 May 3;17(1):178. doi: |
| | | in patients with Cushing's syndrome across reference centers of the European Reference Network on Rare Endocrine Conditions (EndoERN). | Biermasz NR, Cannegieter SC, Huisman MV, van Vlijmen BJM, Feelders RA, Klok FA, Pereira AM; Endo-ERN Cushing and Thrombosis **study group** | | | 10.1186/s13023-022-02320-x |
| 6 | 2022 | Molecular modelling of novel ADCY3 variant predicts a molecular target for tackling obesity. | **Toumba M**, Fanis P, Vlachakis D, Neocleous V, Phylactou LA, Skordis N, Mantzoros CS, Pantelidou M | Int J Mol Med. | 49 | 49(1):10. doi: 10.3892/ijmm.2021.5065. |

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| 7 | 2017 | Diagnosis and management of Silver-Russell syndrome: first international consensus statement. | Wakeling EL, Brioude F, Lokulo-Sodipe O, O'Connell SM, Salem J, Bliek J, Canton AP, Chrzanowska KH, Davies JH, Dias RP, Dubern B, Elbracht M, Giabicani E, Grimberg A, Grønskov K, Hokken-Koelega AC, Jorge AA, Kagami M, Linglart A, Maghnie M, Mohnike K, Monk D, Moore GE, Murray PG, Ogata T, Petit IO, Russo S, Said E, **Toumba M**, Tümer Z, Binder G, Eggermann T, Harbison MD, Temple IK, Mackay DJ, Netchine | Nature Rev Endocrinol. | 13 | 13(2):105-124. doi: 10.1038/nrendo.2016.138. |
| 8 | 2010 | Effect of long-term growth hormone treatment on final height of children with RussellSilver syndrome. | **Toumba M**, Albanese A, Azcona C, Stanhope R | Horm Res Paediatr. | 74 | 2010;74(3):212-7. doi: 10.1159/000295924. |
| 9 | 2009 | Typical Growth Patterns: Infancy, Childhood and Adolescence. In: A comprehensive Guide. Understanding aspects of | **M Toumba**, J B Salem | In: A comprehensive Guide. Understanding aspects of children diagnosed with | | |
| | | children diagnosed with Russell Silver Syndrome or born Small for Gestational Age. Copyright by Quadco Printing Inc, CA 95928. | | Russell Silver Syndrome or born Small for Gestational Age. Copyright by Quadco Printing Inc, CA 95928, USA | | |

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| 10. | 2005<br>2012 | Thalassaemia and its impact on growth. | **M Toumba**, N Skordis | The Handbook of Growth and Growth Monitoring in Health and Disease. (***Ed:*** ***Springer:*** Victor R. Preedy | | 978-1-4419-1795-9 |

**Exhibitions**

| Ref. Number | Date | Topic | International / Local | Location | Role in Exhibition |
|---|---|---|---|---|---|
| 1 | September 2023 | The chimeric CYP21A1P/CYP21A2 and TNX A/TNXB gene deficiencies in patients with Congenital Adrenal Hyperplasia | The 61st ESPE Meeting - European Society for Paediatric Endocrinology, | The Hague, The Netherlands | Oral Presentation |
| 2 | September 2022 | DNA methylation status of MKRN3 in puberty: studies in Central Precocious Puberty girls and in pubertal mouse | The 60th ESPE Meeting - European Society for Paediatric Endocrinology | Rome, Italy | Oral Presentation |
| 3 | September 2022 Precocious | Pathogenic and Low-Frequency genetic determinants in children with Central Puberty. Paediatric Endocrinology | The 60th ESPE Meeting - European Society for | Rome, Italy | Oral Presentation |
| 4 | September 2022 | Homology modelling analysis provides insight into ADCY3, a candidate obesity gene | The 60th ESPE Meeting - European Society for Paediatric Endocrinology | Rome, Italy | Poster Presentation |
| 5 | December, 2020 | GnRH Deficient Patients with Congenital Hypogonadotropic Hypogonadism: Novel Genetic Findings in *ANOS1, RNF216, WDR11, FGFR1, CHD7, and POLR3A* Genes | 8th International conference of The Cyprus Society of Human Genetics. | Nicosia, Cyprus | Oral Presentation |

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| 6 | September 2019 | Central Precocious Puberty Caused by Novel Mutations in the Promoter and UTR Region of the Imprinted MKRN3 Gene. | The 58th ESPE Meeting - European Society for Paediatric Endocrinology | Vienna, Austria | Oral Presentation |
| 7 | September 2017 | MC4R variants are related with severe early onset obesity and extreme growth in children younger than 3 years of age | 10th International Meeting of Pediatric Endocrinology | Washington DC, USA | Poster Presentation |
| 8 | November 2016 | Clinical diagnosis of Silver Russell, Syndrome: First international consensus guidelines. | The 17th Manchester Dysmorphology Conference, November | Manchester, UK | Oral Presentation |
| 9 | October 2015 | A novel melanocortin 4 receptor (MC4R) gene mutation associated with early onset severe obesity. | The 54th ESPE- Annual Meeting of the European Society of Paediatric Endocrinology | Barcelona, Spain. | Poster Presentation |
| 10 | June 2011 | Hyperandrogenism in heterozygous Congenital Adrenal Hyperplasia females with 21-hydroxylase deficiency. | ENDO 2011: The Endocrine Society 93rd Annual Meeting | Boston, MA, USA | Oral Presentation |

| Research Projects | | | | |
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| **Ref. Number** | **Date** | **Title** | **Funded by** | **Project Role** |
| 1 | 2016 | The genetic basis of Endocrine Disorders: Identification of molecular defects in patients with Delayed Puberty, Congenital | RPF “RESTART 2016 – 2020” Excellence Projects call (€ 250.0*00). | Co-Investigator |
| | | Hypogonadotropic Hypogonadism (CHH) and Central Precocious Puberty (CPP) | | |

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| 2 | 2015 | Early onset childhood obesity-genetic variations | AG Leventis Foundation (grant no. 3317312) and the RCB Bank Ltd. (grant no. 33173151). | Co-Investigator |
| 3 | 2015 | Congenital adrenal hyperplasia- molecular defects | AG Leventis Foundation | Co-Investigator |
| 4 | 2014 | Hyperandrogenemia / PCOS genetic variants | AG Leventis Foundation | Co-Investigator |
| 5 | 2014-2017 | Congenital Imprinting disorders- RSS | COST-European network | Member of the Research Team |
| 6 | 2004-2007 | Children born Small for gestational age | NA | Project coordinator |

| Academic Consulting Services and/or Participation in Councils / Boards/ Editorial Committees | | | | |
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| **Ref. Number** | **Period** | **Organization** | **Title of Position or Service** | **Key Activities** |
| 1 | 2004-present | European Society for Paediatric Endocrinology | Member | |
| 2 | 2005-present | Society for Endocrinology, UK | Member | |
| 3 | 2008-present | Hellenic Society of Adolescent and Paediatric Endocrinology | Member | |
| 4 | 2008-present | Cyprus Endocrine Society | Member of council | |
| 5 | 2003-present | Cyprus Pediatric Society | Member of council | |