ROSER PONS MD

Child Neurologist

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CURRENT POSITION

Associate professor at First Department of Pediatrics of the National and Kapodistrian University of Athens , "Agia Sofia" Children's hospital, Athens, Greece

ACADEMIC TRAINING

MD from Medical School of the Autonomous University of Barcelona, Spain (1988)
PhD from Medical School of the Autonomous University of Barcelona, Spain (2007)

TRAINEESHIP

- 1989-1992: Residency in Pediatrics at Children's Hospital Vall d'Hebron, Autonomous University of Barcelona, Spain
- 1993-1995: Postdoctoral research fellowship in the Division of Pediatric Neurology at Columbia Presbyterian Medical Center, Columbia University, New York, USA.
- 1995-1996: Postdoctoral research fellowship in the Department of Biochemistry and Genetics at the Neurological Institute Carlo Besta, Milano, Italy
- 1997-2000: Residency in Pediatric Neurology at Columbia University, New York, USA.
- 2000-2001: Movement Disorders Fellowship in the Department of Neurology at Columbia University, New York, USA

PUBLICATIONS (last 5 years)

1. Ortez C, Duarte ST, Ormazábal A, Serrano M, Pérez A, Pons R, Pineda M, Yapici Z, Fernández-Álvarez E, Domingo-Jiménez R, De Castro P, Artuch R, García-Cazorla A.Cerebrospinal fluid synaptic proteins as useful biomarkers in tyrosine hydroxylase deficiency. Mol Genet Metab. 2015;114(1):34-40.
2. Pons R, De Vivo DC Movement Disorders in Glucose Transporter Type 1 Deficiency. J Pediatr Neurol 2015; 13(4): 168-173
3. Syrengelas D, Kalampoki V, Kleisiouni P, Manta V, Mellos S, Pons R, Chrousos GP, Siahanidou T . Alberta Infant Motor Scale (AIMS) Performance of Greek Preterm Infants: Comparisons with Full-Term Infants of the Same Nationality and Impact of Prematurity-Related Morbidity Factors. Phys Ther. 2016; 96(7):1102-8.
4. Polymeris AA, Tessa A, Anagnostopoulou K, Rubegni A, Galatolo D, Dinopoulos A, Gika AD, Youroukos S, Skouteli E, Santorelli FM, Pons R. A series of Greek children with pure hereditary spastic paraplegia: clinical features and genetic findings.J Neurol. 2016; 263(8):1604-11
5. Pons R, Kekou K, Antonellou R, Svingou M, Kanavakis E, Stefanis L. Analysis of a founder mutation in the TH gene in a cohort of greek patients with Parkinson's disease. Mov Disord. 2016;31(11):1753-1754
6. Bacopoulou F, Apostolaki D, Pons R. External genitalia hypertrophy in an adolescent girl with Sanfilippo syndrome. Eur J Contracept Reprod Health Care. 2016;21(5):412-3
7. Kekou K, Sofocleous C, Papadimas G, Petichakis D, Svingou M, Pons RM, Vorgia P, Gika A, Kitsiou-Tzeli S, Kanavakis E. A dynamic trinucleotide repeat (TNR) expansion in the DMD gene. Mol Cell Probes. 2016;30(4):254-60.
8. Opladen T, Cortès-Saladelafont E, Mastrangelo M, Horvath G, Pons R, Lopez-Laso E, Fernández-Ramos JA, Honzik T, Pearson T, Friedman J, Scholl-Bürgi S, Wassenberg T, Jung-Klawitter S, Kuseyri O, Jeltsch K, Kurian MA, Garcia-Cazorla A. The International Working Group on Neurotransmitter related Disorders (iNTD): A worldwide research project focused on primary and secondary neurotransmitter disorders. Molecular Genetics and Metabolism Reports. 2016; 9:61–66
9. Yubero D, Brandi N, Ormazabal A, Garcia-Cazorla À, Pérez-Dueñas B, Campistol J, Ribes A, Palau F, Artuch R, Armstrong J; Targeted Next Generation Sequencing in Patients with Inborn Errors of Metabolism. Working Group. PLoS One. 2016 31;11(5): 1-10
10. Pons R, Kekou K, Gkika A, Papadimas G, PhD N, Svingou M, Papadopooulos C, Nikas I, Dinopoulos A, Youroukos S, Kanavakis E. A single amino acid loss in the dystrophin protein associated with a mild clinical phenotype. Muscle Nerve. 2017; 55(1):46-50
11. Wassenberg T, Molero Luis M, Jeltsch K, Hoffmann G, Assmann B, Blau N, Garcia Cazorla A, Artuch R, Pons R, Pearson T, Leuzzi V, Mastrangelo M, Pearl P, Lee WT, Kurian M, Heales S, Flint L, Verbeek M, Willemsen M, Opladen T. Consensus Guideline for the Diagnosis and Treatment of Aromatic l-Amino acid Decarboxylase (AADC) Deficiency. Orphanet J Rare Dis. 2017; 12 (1):12.
12. Makrygianni EA, Papadimas GK, Georgala M, Tzetis M, Poulou M, Kitsiou-Tzeli S, Pons R. CCFDN syndrome: new clinical features. Ped Neurol 2017; 67:e5-e6
13. Pearson TS*, Pons R*, Engelstad K, Kane SA, Goldberg ME, De Vivo DC. Paroxysmal eye-head movements in Glut1 deficiency syndrome. Neurology. 2017, 25;88(17):1666-1673. *joint first authorship
14. Carecchio M, Mencacci NE, Iodice A, Pons R, Panteghini C, Zorzi G, Zibordi F, Bonakis A, Dinopoulos A, Jankovic J, Stefanis L, Bhatia KP, Monti V, R'Bibo L, Veneziano L, Garavaglia B, Fusco C, Wood N, Stamelou

M, Nardocci N. ADCY5-related movement disorders: Frequency, disease course and phenotypic variability in a cohort of paediatric patients. Parkinsonism Relat Disord. 2017; 41:37-43.
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37. Leuzzi V, Nardecchia F, Pons R, Galosi S. Parkinsonism in children: clinical classification and etiological spectrum Parkinsonism Relat Disord 2020; S1353-8020(20)30777-X. doi: 10.1016/j.parkreldis.2020.10.002. Online ahead of print.
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