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Σημερινή θέση:

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Παρόν ερευνητικό έργο

- "Web-based patient registry for inherited defects of biogenic amines, pterin, folate, serine, glycine and GABA metabolism" International Working Group on Neurotransmitter Related Disorders (iNTD)" (K.E. T.Opladen)
- "Biomarcadores y genes en la necrosis estriatal bilateral de la infancia" Fondo de investigacion Sanitaria, Ministerio de Economia y competitividad, Espana (K.E. B.Perez Duenas)
- "Ανάπτυξη κλιμάκων αξιολόγησης της κινητικής λειτουργίας, του παρκινσονισμού, της δυστονίας και της φάρμακο-προκαλούμενης δυσκινησίας σε ασθενείς με διαταραχές του μεταβολισμού των βιογενών αμινών" (K.E. R. Pons) Επιχορήγηση από το AADC Research Trust (UK)
- "A retrospective and prospective observational registry to explore the epidemiology, clinical profile and therapeutic management of SMA patients in Greece» Εθνική μελέτη με Επιχορήγηση από Genesis Pharma

Συγγραφικό έργο (τελευταία 5 χρόνια)

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.
15. Mavrogeni S, Giannakopoulou A, Papavasiliou A, Markousis-Mavrogenis G, Pons R, Karanasios E, Noutsias M, Kolovou G, Papadopoulos G. Cardiac profile of asymptomatic children with Becker and Duchenne muscular dystrophy under treatment with steroids and with/without perindopril. BMC Cardiovasc Disord. 2017 24;17(1):197.
16. Pons R, Vanezis A, Skouteli H, Papavasiliou A, Tziomaki M, Syrengelas D, Darras N. Upper Limb Function, Kinematic Analysis, and Dystonia Assessment in Children With Spastic Diplegic Cerebral Palsy and Periventricular Leukomalacia. J Child Neurol. 2017;32(11):936-941.
17. Darling A, Tello C, Martí MJ, Garrido C, Aguilera-Albesa S, Tomás Vila M, Gastón I, Madruga M, González Gutiérrez L, Ramos Lizana J, Pujol M, Gavilán Iglesias T, Tustin K, Lin JP, Zorzi G, Nardocci N, Martorell L, Lorenzo Sanz G, Gutiérrez F, García PJ, Vela L, Hernández Lahoz C, Ortigoza Escobar JD, Martí Sánchez L, Moreira F, Coelho M, Correia Guedes L, Castro Caldas A, Ferreira J, Pires P, Costa C, Rego P, Magalhães M, Stamelou M, Cuadras Pallejà D, Rodríguez-Blazquez C, Martínez-Martín P, Lupo V, Stefanis L, Pons R, Espinós C, Temudo T, Pérez Dueñas B. Clinical rating scale for pantothenate kinase-associated neurodegeneration: A pilot study. Mov Disord. 2017;32(11):1620-1630
18. Pons R, Nikas I, Papadopoulos G, Verganelakis DA, Kolovou G, Chrousos GP. Brain and heart magnetic resonance imaging/spectroscopy in duchenne muscular dystrophy. Mavrogeni S. Eur J Clin Invest. 2017, 47(12).
19. Mavrogeni S, Papavasiliou A, Giannakopoulou K, Markousis-Mavrogenis G, Pons MR, Karanasios E, Nikas I, Papadopoulos G, Kolovou G, Chrousos GP. Oedema-fibrosis in Duchenne Muscular Dystrophy: Role of cardiovascular magnetic resonance imaging. Eur J Clin Invest. 2017, 47(12).
20. Batllori M, Molero-Luis M, Arrabal L, Heras JL, Fernandez-Ramos JA, Gutiérrez-Solana LG, Ibáñez-Micó S, Domingo R, Campistol J, Ormazabal A, Sedel F, Opladen T, Zouvelou B, Pons R, Garcia-Cazorla A, Lopez-Laso E, Artuch R. Urinary sulphatoxymelatonin as a biomarker of serotonin status in biogenic amine-deficient patients. Sci Rep. 2017 7;7(1):14675.
21. Dinopoulos A, Tsirouda MA, Bonakis A, Pons R, Pavlopoulou ID, Tsoumakas K. Sleep architecture and epileptic characteristics of drug naïve patients in childhood absence epilepsy spectrum. A prospective study. Seizure. 2018 Apr 30;59:99-107
22. Stamelou M, Petrucci S, Ginevrino M, Pons R, Papagiannakis N, Stefanis L, Valente EM. Intrafamilial variability in a polish family harbouring a frameshift THAP1 mutation. J Neurol Sci. 2018;388:158.
23. Papadopoulou A, Dinopoulos A, Koutsodontis G, Pons R, Vorgia P, Koute V, Vratimos A, Zafeiriou D. Screening for TSC1 and TSC2 mutations using NGS in Greek children with tuberous sclerosis syndrome. Eur J Paediatr Neurol. 2018;22(3):419-426
24. Tsipi M, Fylaktou E, Kosma K, Tsoutsou E, Poulou M, Pons MR, Kokkinou E, Kitsiou- Tzeli S, Fryssira H, Tzetis M. Phenotypic expression of a spectrum of Neurofibromatosis Type 1 (NF1) mutations identified through NGS and MLPA. J Neurol Sci 2018; 15;395:95-105.
25. D'Amore A, Tessa A, Casali C, Dotti MT, Filla A, Silvestri G, Antenora A, Astrea G, Barghigiani M, Battini R, Battisti C, Bruno I, Cereda C, Dato C, Di Iorio G, Donadio V, Felicori M, Fini N, Fiorillo C, Gallone S, Gemignani F, Gigli GL, Graziano C, Guerrini R, Gurrieri F, Kariminejad A, Lieto M, Marques LourenÇo C, Malandrini A, Mandich P, Marcotulli C, Mari F, Massacesi L, Melone MAB, Mignarri A, Milone R, Musumeci O, Pegoraro E, Perna A, Petrucci A, Pini A, Pochiero F, Pons MR, Ricca I, Rossi S, Seri M, Stanzial F, Tinelli F[1], Toscano A, Valente M, Federico A, Rubegni A, Santorelli FM
Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Front Neurol 2018; 9:981.
26. Zouvelou V, Yubero D, Apostolakopoulou L, Kokkinou E, Bilanakis M, Dalivigka Z, Nikas I, Kollia E, Perez-Dueñas B, Macaya A, Marcé-Grau A, Voutetakis A, Anagnostopoulou K, Kekou K, Sofocleus C, Veltra D, Kokkinis X, Fryssira H, Torres RJ, Amstrong J, Santorelli FM, Artuch R, Pons R. The genetic etiology in cerebral palsy mimics: the results from a Greek tertiary care center. Eur J Paediatr Neurol. 2019, 23(3):427-437.
27. Pearson TS, Pons R, Ghaoui R, Sue CM. Genetic Mimics of Cerebral Palsy. Mov Disord. 2019;34(5):625-636
28. Pearson TS, Pons R Movement disorders in Children. Continuum (Minneap Minn). 2019; 25(4):1099-1120
29. Kokkinou E, Roka K, Alexopoulos A, Tsina E, Nikas I, Krallis P, Thanopoulou I, Nasi L, Makrygianni E, Tsoutsou E, Kosma K, Tsipi M, Tzetis M, Frysira H, Kattamis A, Pons R. Development of a multidisciplinary clinic of neurofibromatosis type 1 and other neurocutanous disorders in Greece. A three year experience. Postgrad Med. 2019 doi: 10.1080/00325481.2019.1659708.

30. Valari M, Theodoraki M, Loukas I, Gkantseva-Patsoura S, Karavana G, Falaina V, Lykopoulou L, Pons R, Athanasiou I, Wertheim-Tysarowska K, Kanaka-Gantenbein C, Kiritsi D. Novel PLEC Variant Causes Only Mild Skin Fragility, but Pyloric Atresia, Muscular Dystrophy and Urological Manifestations. Acta Derm Venereol. 2019 Sep 12. doi: 10.2340/00015555-3317.
31. Mavrogeni S, Giannakopoulou A, Katsalouli M, Pons RM, Papavasiliou A, Kolovou G, Noutsias M, Papadopoulos G, Karanasios E, Chrousos GP. Friedreich's Ataxia: Case series and the Additive Value of Cardiovascular Magnetic Resonance. J NeuromusculDis. 2019 Nov 27. doi: 10.3233/JND-180373.
32. Pearson T, Gilbert L, Opladen T, Garcia-Cazorla A, Mastrangelo M, Leuzzi V, Tay SKH, Sykut-CegielskaJ, Pons R, Mercimek-Andrews S; KatoM, Lücke T, Oppebøen M, Kurian M, Steel D, Manti F, Black KD, Jeltsch K, Flint L. AADC Deficiency From Infancy to Adulthood Symptoms and Developmental Outcome in an International Cohort of 63 Patients. J Inherit Metab Dis 2020 May 5.
33. Opladen T,López-Laso E, Cortès-Saladelafont E, Pearson T, Sivri HS, Yildiz Y, Assmann B, Kurian MA; Leuzzi V; Heales S; Pope S, Porta F, Garcia-Cazorla A, Honzik T, Pons R, Regal L, Goez H, Artuch R, Hoffmann GF, Horvath G, Thöny B, Scholl-Bürgi S, Burlina A, MVerbeek M, Mastrangelo M, Friedman J, Wassenberg T, Jeltsch K, Kulhanek J, Kuseyri Hübschmann O, International Working Group on Neurotransmitter related Disorders Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies Orphanet J Rare Dis. 2020 May 26;15(1):126
34. Kekou K, Svingou M, SofocleousC, Mourtzi N, Giouroukos S, Skiadas K, Katsalouli M, Pons R, Papavasileiou A, Kotsalis C, Pavlou E, Evangeliou A, Katsarou E, Boudris K, Ntinopoulos A, Vorgia P, Niotakis G, Diamantopoulos N, Nakou I, Kote B, Papadimas GK, Papadopoulos K, Tsivgoulis G, Fryssira H. 5q- Linked Spinal Muscular Atrophy (SMA) in Greece: Twenty-two years of experienceJ Neuromuscul Dis 2020;7(3):247-256
35. Vanegas MI, Marcé-Grau A, Martí-Sánchez L, Mellid S, Baide-Mairena H, Correa-Vela M, Rodríguez C, Toledo L, Macaya A, Fernández-Ramos J, Pons R, Aguilera S, Martí MJ, Eiris J, Iglesias G, De Fábregas O, Olivé-Cirera G, Garriz M, Madruga M, Cabrera CJ, Pérez-Dueñas B. Delineating the motor phenotype of SGCE-myoclonus dystonia syndrome. Parkinsonism Relat Disord 2020; 80:165-174
36. Balestrini S, Mikati MA, Alvarez Garcia-Roves R, Carboni M, Hunanyan Arsen S, Kherallah B, McLean M, Prange L, de Grandis E, Gagliardi A, Pisciotta L, Stagnaro M, Veneselli E, Campistol J, Carmen Fons C, Leticia Pias L, Allison Brashear A, Charlotte Miller C, Raquel Samoes R, Vesna Brankovic V, Quasar S Padiath QS, Ana Potic A, Jacek Pilch J, Vezyroglou K, Hollingsworth G, Scheffer IE, Granata T, Nardocci N, Ragona FG, Arzimanoglou A, Panagiotakaki E, Carrilho I, Zucca C, Novy J, Dzieżyc K, Parowicz M, Mazurkiewicz-Bełdzińska M, Weckhuysen S, Pons R, Groppa S, Daniel S Sinden DS, Pitt GS, Tinker A, Ashworth M, Michalak Z, Thom M, Cross H, Vavassori R, Kaski JP, Sisodiya SM Cardiac phenotype in *ATP1A3* related-syndromes: treatment implications from a multicentre cohort study. Neurology 2020; 95(21):e2866-e2879
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.
38. Klepper J, Akman C, Armeno M, Auvin S, Cervenka M, Cross HJ, De Giorgis V, Della Marina A, Engelstad K, Heussinger N, Kossoff EH, Leen WG, Leiendecker B, Monani UR, Oguni H, Neal E, Pascual JM, Pearson TS, Pons R, Scheffer IE, Veggiotti P, Willemsen M, Zuberi SM, De Vivo DC Glut1 Deficiency Syndrome (Glut1DS): State of the art in 2020 and recommendations of the international Glut1DS study group.Epilepsia Open 2020; 5(3):354-365.
39. Tristán-Noguero A, Borràs E, Molero-Luis M, Wassenberg T, Peters T, Verbeek MM, Willemsen M, Opladen T, Jeltsch K, Pons R, Thony B, Horvath G, Yapici Z, Friedman J, Hyland K, Agosta GE, López-Laso E, Artuch R, Sabidó E, García-Cazorla À. Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. Mov Disord. 2020 Nov 5. doi: 10.1002/mds.28362. Online ahead of print.PMID: 33152132
40. Marti-Sanchez L, Baide-Mairena H, Marcé-Grau A, Pons R, López-Lazo E, Sigatullina M, Rizzo C, Semeraro M, Martinelli D, Carrozzo R, Dionisi-Vici C, González-Gutiérrez-Solana L, Correa-Vela M, Ortigoza-Escobar JD, Sánchez-Montañez A, Vazquez E, Delgado I, Aguilera-Albesa S, Yoldi ME, Ribes A, Pollini L, Galosi S, Leuzzi V, Pérez-Gay L, Aldamiz-Echavarría L, Del Toro M, Arranz A, Roelens F, Urreizti R, Artuch R, Macaya A, Pérez-Dueñas B. Delineating the neurological phenotype in children with ECHS1 and HIBCH genetic defects. J Inherit Metab Dis. 2020,doi: 10.1002/jimd.12288. Online ahead of print.
41. Kuseyri Hübschmann O, Friedman J, Manti F, Horvath G, Cortès-Saladelafont E, Andrews S, Yildiz Y, Pons R, Kulhánek J, Oppenboen M, Aimee Koht J, Zafeiriou DI, López-Laso E, Guder P, Santer R, Honzík T, Hoffmann GF, Garbade SF, Sivri HS, Leuzzi V, Jeltsch K, García-Cazorla A, Opladen T, Harting I. Spectrum and patterns of MR imaging changes in inherited disorders of monoamine neurotransmitters: A retrospective, systematic analysis of 70 patients (in press)
42. Papadimitriou I, Dalivigka Z, Outsika C, Scarmeas N, Pons R. Dystonia Assessment in Children with Cerebral Palsy and Periventricular Leukomalacia (submitted)