Dott. Chiara Pantaleoni

Pubblicazioni scientifiche recenti

1) D'Arrigo S, Tessarollo V, Taroni F, Baratta S, **Pantaleoni C**, Schiaffi E, Ciano C. A Case of Severe Early-Onset Neuropathy Caused by a Compound Heterozygous Deletion of the PMP22 Gene: Clinical and Neurographic Aspects. Neuropediatrics. 2019 Nov 29.

2) Barresi S, Dentici ML, Manzoni F, Bellacchio E, Agolini E, Pizzi S, Ciolfi A, Tarnopolsky M, Brady L, Garone G, Novelli A, Mei D, Guerrini R, Capuano A, **Pantaleoni C**, Tartaglia M. Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. Pediatr Neurol. 2019 Oct 19.

3) Melloni G, Eoli M, Cesaretti C, Bianchessi D, Ibba MC, Esposito S, Scuvera G, Morcaldi G, Micheli R, Piozzi E, Avignone S, Chiapparini L, **Pantaleoni C**, Natacci F, Finocchiaro G, Saletti V. Risk of Optic Pathway Glioma in Neurofibromatosis Type 1: No Evidence of Genotype-Phenotype Correlations in A Large Independent Cohort. Cancers (Basel). 2019;11(12).

4) Ferlazzo E, Franceschetti S, Gasparini S, Elia M, Canafoglia L, **Pantaleoni C**, Ascoli M, D'Agostino T, Sueri C, Ferrigno G, Panzica F, Cianci V, Aguglia U. Connectivity measures suggest a sub-cortical generator of myoclonus in Angelman syndrome. Clin Neurophysiol. 2019;130(12):2231-2237.

5) Gatti M, Magri S, Nanetti L, Sarto E, Di Bella D, Salsano E, **Pantaleoni C**, Mariotti C, Taroni F. From congenital microcephaly to adult onset cerebellar ataxia: Distinct and overlapping phenotypes in patients with PNKP gene mutations. Am J Med Genet A. 2019;179(11):2277-2283.

6) Ciaccio C, Castello R, Esposito S, Pinelli M, Nigro V, Casari G, Chiapparini L, **Pantaleoni C**; TUDP Study Group, D'Arrigo S. Consolidating the Role of TDP2 Mutations in Recessive Spinocerebellar Ataxia Associated with Pediatric Onset Drug Resistant Epilepsy and Intellectual Disability (SCAR23).Cerebellum. 2019;18(5):972-975.

7) Tomson T, Battino D, Bonizzoni E, Craig J, Lindhout D, Perucca E, Sabers A, Thomas SV, Vajda F; EURAP Study Group (including **Pantaleoni C**.). Declining malformation rates with changed antiepileptic drug prescribing: An observational study. Neurology. 2019;93(9):e831-e840.

8) Saletti V, Viganò I, Melloni G, **Pantaleoni C**, Vetrano IG, Valentini LG. Chiari I malformation in defined genetic syndromes in children: are there common pathways? Childs Nerv Syst. 2019; 35(10):1727-1739.

9) D'Arrigo S, Tessarollo V, Maselli E, **Pantaleoni C**, Canafoglia L. Flunarizine and Aspirin for Transient Hemiparesis in Sturge-Weber Syndrome. Neuropediatrics. 2019 Dec;50(6):406-407.

10) Esposito S, Moscatelli M, Schiariti MP, Viganò I, **Pantaleoni C**, Marucci G. Pott's Disease: An Emerging Source of Potentially Inappropriate Treatment. Neuropediatrics. 2019; 50(5):334-335.

11) Scheuerle AE, Holmes LB, Albano JD, Badalamenti V, Battino D, Covington D, Harden C, Miller D, Montouris GD, **Pantaleoni C**, Thorp J, Tofighy A, Tomson T, Golembesky AK. Levetiracetam Pregnancy Registry: Final results and a review of the impact of registry methodology and definitions on the prevalence of major congenital malformations. Birth Defects Res. 2019;111(13):872-887.

12) Griffiths S, Loveday C, Zachariou A, Behan LA, Chandler K, Cole T, D'Arrigo S, Dieckmann A, Foster A, Gibney J, Hunter M, Milani D, **Pantaleoni C**, Roche E, Sherlock M, Springer A, White SM; Childhood Overgrowth Collaboration, Tatton-Brown K. EED and EZH2 constitutive variants: A study to expand the Cohen-Gibson syndrome phenotype and contrast it with Weaver syndrome. Am J Med Genet A. 2019;179(4):588-594.

13) Bersano A, Bedini G, Nava S, Acerbi F, Sebastiano DR, Binelli S, Franceschetti S, Faragò G, Grisoli M, Gioppo A, Ferroli P, Bruzzone MG, Riva D, Ciceri E, **Pantaleoni C**, Saletti V, Esposito S, Nardocci N, Zibordi F, Caputi L, Marzoli SB, Zedde ML, Pavanello M, Raso A, Capra V, Pantoni L, Sarti C, Pezzini A, Caria F, Dell' Acqua ML, Zini A, Baracchini C, Farina F, Sanguigni S, De Lodovici ML, Bono G, Capone F, Di Lazzaro V, Lanfranconi S, Toscano M, Di Piero V, Sacco S, Carolei A, Toni D, Paciaroni M, Caso V, Perrone P, Calloni MV, Romani A, Cenzato M, Fratianni A, Ciusani E, Prontera P, Lasserve ET, Blecharz K, Vajkoczy P, Parati EA; GEN-O-MA study group. GEN-O-MA project: an Italian network studying clinical course and pathogenic pathways of moyamoya disease-study protocol and preliminary results. Neurol Sci. 2019;40(3):561-570.

14) Ciaccio C, Saletti V, D'Arrigo S, Esposito S, Alfei E, Moroni I, Tonduti D, Chiapparini L, **Pantaleoni C**, Milani D. Clinical spectrum of PTEN mutation in pediatric patients. A bicenter experience. Eur J Med Genet. 2019;62(12):103596.

15) 19) Luisa SF, Rizzo A, Bedini G, Capone F, Di Lazzaro V, Nava S, Acerbi F, Rossi DS, Binelli S, Faragò G, Gioppo A, Grisoli M, Bruzzone MG, Ferroli P, **Pantaleoni C**, Caputi L, Gomez JV, Parati EA, Bersano A.Microduplication of 15q13.3 and Microdeletion of 18q21.32 in a Patient with Moyamoya Syndrome. Int J Mol Sci. 2018; 19(11). pii: E3675.

16) Magri S, Fracasso V, Plumari M, Alfei E, Ghezzi D, Gellera C, Rusmini P, Poletti A, Di Bella D, Elia AE, **Pantaleoni C**, Taroni F.; Concurrent AFG3L2 and SPG7 mutations associated with syndromic parkinsonism and optic atrophy with aberrant OPA1 processing and mitochondrial network fragmentation. Hum Mutat. 2018;39(12):2060-2071.

17) Tonduti D, Panteghini C, Pichiecchio A, Decio A, Carecchio M, Reale C, Moroni I, Nardocci N, Campistol J, Garcia-Cazorla A, Perez Duenas B; Cerebral Calcification International Study Group (… **Pantaleoni C**., …), Chiapparini L, Garavaglia B, Orcesi S. Encephalopathies with intracranial calcification in children: clinical and genetic characterization. Orphanet J Rare Dis. 2018; 13(1):135

18) Rizzo A, Alfei E, Zibordi F, Saletti V, Zorzi G, Freri E, Estienne M, Girgenti V, D'Arrigo S, Esposito S, Buldrini B, Moroni I, Milani D, Granata T, Ardissone A, Eoli M, Molteni B, Bigoni S, **Pantaleoni C**, Nardocci N, Sciacca FL. The noncoding RNA AK127244 in 2p16.3 locus: A new susceptibility region for neuropsychiatric disorders. Am J Med Genet B Neuropsychiatr Genet. 2018;177(6):557-562.

19) Tomson T, Battino D, Bonizzoni E, Craig J, Lindhout D, Perucca E, Sabers A, Thomas SV, Vajda F; EURAP Study Group (including **Pantaleoni C**.) Comparative risk of major congenital malformations with eight different antiepileptic drugs: a prospective cohort study of the EURAP registry. Lancet Neurol. 2018;17(6):530-538.

20) Pagliano E, Baranello G, Masson R, Foscan M, Arnoldi MT, Marchi A, Aprile G, **Pantaleoni C**. Outcome measures for children with movement disorders. Eur J Paediatr Neurol. 2018; 22(3):346-353. Review.

21) Ivanovski I, Djuric O, Caraffi SG, Santodirocco D, Pollazzon M, Rosato S, Cordelli DM, Abdalla E, Accorsi P, Adam MP, Ajmone PF, Badura-Stronka M, Baldo C, Baldi M, Bayat A, Bigoni S, Bonvicini F, Breckpot J, Callewaert B, Cocchi G, Cuturilo G, De Brasi D, Devriendt K, Dinulos MB, Hjortshøj TD, Epifanio R, Faravelli F, Fiumara A, Formisano D, Giordano L, Grasso M, Grønborg S, Iodice A, Iughetti L, Kuburovic V, Kutkowska-Kazmierczak A, Lacombe D, Lo Rizzo C, Luchetti A, Malbora B, Mammi I, Mari F, Montorsi G, Moutton S, Møller RS, Muschke P, Nielsen JEK, Obersztyn E, Pantaleoni C, Pellicciari A, Pisanti MA, Prpic I, Poch-Olive ML, Raviglione F, Renieri A, Ricci E, Rivieri F, Santen GW, Savasta S, Scarano G, Schanze I, Selicorni A, Silengo M, Smigiel R, Spaccini L, Sorge G, Szczaluba K, Tarani L, Tone LG, Toutain A, Trimouille A, Valera ET, Vergano SS, Zanotta N, Zenker M, Conidi A, Zollino M, Rauch A, Zweier C, Garavelli L. Phenotype and genotype of 87 patients with Mowat-Wilson syndrome and recommendations for care. Genet Med. 2018 Sep;20(9):965-975.

22) Esposito S, Carecchio M, Tonduti D, Saletti V, Panteghini C, Chiapparini L, Zorzi G, **Pantaleoni C**, Garavaglia B, Krainc D, Lubbe SJ, Nardocci N, Mencacci NE. A PDE10A de novo mutation causes childhood-onset chorea with diurnal fluctuations. Mov Disord 2017; 32(11):1646-1647.

23) Alcantara D, Timms AE, Gripp K, Baker L, Park K, Collins S, Cheng C, Stewart F, Mehta SG, Saggar A, Sztriha L, Zombor M, Caluseriu O, Mesterman R, Van Allen MI, Jacquinet A, Ygberg S, Bernstein JA, Wenger AM, Guturu H, Bejerano G, Gomez-Ospina N, Lehman A, Alfei E, **Pantaleoni C**, Conti V, Guerrini R, Moog U, Graham JM Jr, Hevner R, Dobyns WB, O'Driscoll M, Mirzaa GM. Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain 2017; 140(10):2610-2622.

24) Zanzottera C, Milani D, Alfei E, Rizzo A, D'Arrigo S, Esposito S, **Pantaleoni C**. ZC4H2 deletions can cause severe phenotype in female carriers. Am J Med Genet A 2017;173(5):1358-1363.

25) Garavelli L, Ivanovski I, Caraffi SG, Santodirocco D, Pollazzon M, Cordelli DM, Abdalla E, Accorsi P, Adam MP, Baldo C, Bayat A, Belligni E, Bonvicini F, Breckpot J, Callewaert B, Cocchi G, Cuturilo G, Devriendt K, Dinulos MB, Djuric O, Epifanio R, Faravelli F, Formisano D, Giordano L, Grasso M, Grønborg S, Iodice A, Iughetti L, Lacombe D, Maggi M, Malbora B, Mammi I, Moutton S, Møller R, Muschke P, Napoli M, **Pantaleoni C,** Pascarella R, Pellicciari A, Poch-Olive ML, Raviglione F, Rivieri F, Russo C, Savasta S, Scarano G, Selicorni A, Silengo M, Sorge G, Tarani L, Tone LG, Toutain A, Trimouille A, Valera ET, Vergano SS, Zanotta N, Zollino M, Dobyns WB, Paciorkowski AR. Neuroimaging findings in Mowat-Wilson syndrome: a study of 54 patients. Genet Med 2017, 19(6):691-700

26) Kasher PR, Schertz KE, Thomas M, Jackson A, Annunziata S, Ballesta-Martinez MJ, Campeau PM, Clayton PE, Eaton JL, Granata T, Guillén-Navarro E, Hernando C, Laverriere CE, Liedén A, Villa-Marcos O, McEntagart M, Nordgren A, **Pantaleoni C**, Pebrel-Richard C, Sarret C, Sciacca FL, Wright R, Kerr B, Glasgow E, Banka S. Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. Am J Hum Genet 2016; 98(2):363-372.

27) Natacci F, Alfei E, Tararà L, D'Arrigo S, Zuffardi O, Gentilin B, **Pantaleoni C**. Chromosome 17q21.31 duplication syndrome: Description of a new familiar case and further delineation of the clinical spectrum. Eur J Paediatr Neurol 2016; 20(1):183-187.

28) D'Arrigo S, Gavazzi F, Alfei E, Zuffardi O, Montomoli C, Corso B, Buzzi E, Sciacca FL, Bulgheroni S, Riva D, **Pantaleoni C**. The Diagnostic Yield of Array Comparative Genomic Hybridization Is High Regardless of Severity of Intellectual Disability/Developmental Delay in Children. J Child Neurol 2016; 31(6):691-699.

29) Nicita F, Garone G, Spalice A, Savasta S, Striano P, **Pantaleoni C**, Spartà MV, Kluger G, Capovilla G, Pruna D, Freri E, D'Arrigo S, Verrotti A. Epilepsy is a possible feature in Williams-Beuren syndrome patients harboring typical deletions of the 7q11.23 critical region. Am J Med Genet A 2016; 170A(1):148-155.

30) Bedini G, Blecharz KG, Nava S, Vajkoczy P, Alessandri G, Ranieri M, Acerbi F, Ferroli P, Riva D, Esposito S, **Pantaleoni C**, Nardocci N, Zibordi F, Ciceri E, Parati EA, Bersano A. Vasculogenic and Angiogenic Pathways in Moyamoya Disease. Curr Med Chem 2016; 23(4):315-345.

31) Grozeva D, Carss K, Spasic-Boskovic O, Tejada MI, Gecz J, Shaw M, Corbett M, Haan E, Thompson E, Friend K, Hussain Z, Hackett A, Field M, Renieri A, Stevenson R, Schwartz C, Floyd JA, Bentham J, Cosgrove C, Keavney B, Bhattacharya S; Italian X-linked Mental Retardation Project (including **Pantaleoni C**.); UK10K Consortium.; GOLD Consortium., Hurles M, Raymond FL. Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. Hum Mutat 2015; 36(12):1197-1204.

32) Tomson T, Battino D, Bonizzoni E, Craig J, Lindhout D, Perucca E, Sabers A, Thomas SV, Vajda F; Dose-dependent teratogenicity of valproate in mono- and polytherapy: an observational study. EURAP Study Group (including **Pantaleoni C**.). Neurology 2015; 85(10):866-872.

33) Baranello G, Saredi S, Sansanelli S, Savadori P, Canioni E, Chiapparini L, Balestri P, Malandrini A, Arnoldi MT, **Pantaleoni C**, Morandi L, Mora M. A novel homozygous ISPD gene mutation causing phenotype variability in a consanguineous family. Neuromuscul Disord 2015; 25(1):55-59.