

1: 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 Nov 21;11(12). pii: E1838. doi: 10.3390/cancers11121838. PubMed PMID: 31766501.

2: Taddei M, Erbetta A, **Esposito S**, Saletti V, Bulgheroni S, Riva D. Brain Tumors in NF1 Children: Influence on Neurocognitive and Behavioral Outcome. *Cancers (Basel)*. 2019 Nov 11;11(11). pii: E1772. doi: 10.3390/cancers11111772. PubMed PMID: 31717965; PubMed Central PMCID: PMC6896178.

3: Tritto V, Ferrari L, **Esposito S**, Zuccotti P, Bianchessi D, Natacci F, Saletti V, Eoli M, Riva P. Non-Coding RNA and Tumor Development in Neurofibromatosis Type 1: ANRIL Rs2151280 Is Associated with Optic Glioma Development and a Mild Phenotype in Neurofibromatosis Type 1 Patients. *Genes (Basel)*. 2019 Nov 5;10(11). pii: E892. doi: 10.3390/genes10110892. PubMed PMID: 31694342; PubMed Central PMCID: PMC6895873.

4: Koczkowska M, Callens T, Chen Y, Gomes A, Hicks AD, Sharp A, Johns E, Uhas KA, Armstrong L, Bosanko KA, Babovic-Vuksanovic D, Baker L, Basel DG, Bengala M, Bennett JT, Chambers C, Clarkson LK, Clementi M, Cortés FM, Cunningham M, D'Agostino MD, Delatycki MB, Digilio MC, Dosa L, **Esposito S**, Fox S, Freckmann ML, Fauth C, Giugliano T, Giustini S, Goetsch A, Goldberg Y, Greenwood RS, Griffis C, Gripp KW, Gupta P, Haan E, Hachen RK, Haygarth TL, Hernández-Chico C, Hodge K, Hopkin RJ, Hudgins L, Janssens S, Keller K, Kelly-Mancuso G, Kochhar A, Korf BR, Lewis AM, Liebelt J, Lichty A, Listernick RH, Lyons MJ, Maystadt I, Martinez Ojeda M, McDougall C, McGregor LK, Melis D, Mendelsohn N, Nowaczyk MJM, Ortenberg J, Panzer K, Pappas JG, Pierpont ME, Piluso G, Pinna V, Pivnick EK, Pond DA, Powell CM, Rogers C, Ruhrman Shahar N, Rutledge SL, Saletti V, Sandaradura SA, Santoro C, Schatz UA, Schreiber A, Scott DA, Sellars EA, Sheffer R, Siqveland E, Slopis JM, Smith R, Spalice A, Stockton DW, Streff H, Theos A, Tomlinson GE, Tran G, Trapane PL, Trevisson E, Ullrich NJ, Van den Ende J, Schrier Vergano SA, Wallace SE, Wangler MF, Weaver DD, Yohay KH, Zackai E, Zonana J, Zurcher V, Claes KBM, Eoli M, Martin Y, Wimmer K, De Luca A, Legius E, Messiaen LM. Clinical spectrum of individuals with pathogenic NF1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotype-phenotype study in neurofibromatosis type 1. *Hum Mutat*. 2020 Jan;41(1):299-315. doi: 10.1002/humu.23929. Epub 2019 Oct 26. PubMed PMID: 31595648.

5: 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 Oct;18(5):972-975. doi: 10.1007/s12311-019-01069-7. PubMed PMID: 31410782.

6: **Esposito S**, Moscatelli M, Schiariti MP, Viganò I, Pantaleoni C, Marucci G. Pott's Disease: An Emerging Source of Potentially Inappropriate Treatment. *Neuropediatrics*. 2019 Oct;50(5):334-335. doi: 10.1055/s-0039-1691833. Epub 2019 May 29. PubMed PMID: 31141827.

7: Bulgheroni S, Taddei M, Saletti V, **Esposito S**, Micheli R, Riva D. Visuoperceptual Impairment in Children with NF1: From Early Visual Processing to Procedural Strategies. *Behav Neurol*. 2019 Jan 13;2019:7146168. doi: 10.1155/2019/7146168. eCollection 2019. PubMed PMID: 30733835; PubMed Central PMCID: PMC6348799.

8: 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, Lasserre 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 Mar;40(3):561-570. doi: 10.1007/s10072-018-3664-z. Epub 2019 Jan 3. PubMed PMID: 30604336.

9: 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 Dec;62(12):103596. doi: 10.1016/j.ejmg.2018.12.001. Epub 2018 Dec 4. PubMed PMID: 30528446.

10: 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 including **Esposito S**, Chiapparini L, Garavaglia B, Orcesi S. Encephalopathies with intracranial calcification in children: clinical and genetic characterization. *Orphanet J Rare Dis*. 2018 Aug 16;13(1):135. doi: 10.1186/s13023-018-0854-y. PubMed PMID: 30111349; PubMed Central PMCID: PMC6094574.

11: 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, Ardisson 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 Sep;177(6):557-562. doi: 10.1002/ajmg.b.32649. Epub 2018 Aug 14. PubMed PMID: 30105822.

12: Ramos EM, Carecchio M, Lemos R, Ferreira J, Legati A, Sears RL, Hsu SC, Panteghini C, Magistrelli L, Salsano E, **Esposito S**, Taroni F, Richard AC, Tranchant C, Anheim M, Ayrignac X, Goizet C, Vidailhet M, Maltete D, Wallon D, Frebourg T, Pimentel L, Geschwind DH, Vanakker O, Galasko D, Fogel BL, Innes AM, Ross A, Dobyns WB, Alcantara D, O'Driscoll M, Hannequin D, Champion D; French PFBC study group, Oliveira JR, Garavaglia B, Coppola G, Nicolas G. Primary brain calcification: an international study reporting novel variants and associated phenotypes. *Eur J Hum Genet*. 2018 Oct;26(10):1462-1477. doi: 10.1038/s41431-018-0185-4. Epub 2018 Jun 28. PubMed PMID: 29955172; PubMed Central PMCID: PMC6138755.

13: **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 Nov;32(11):1646-1647. doi: 10.1002/mds.27175. Epub 2017 Sep 26. PubMed PMID: 28949041.

14: Tucci A, Saletti V, Menni F, Cesaretti C, Scuvera G, Esposito S, Melloni G, **Esposito S**, Milani D, Cereda C, Cigada M, Tresoldi L, Viola F, Natacci F. The absence that makes the difference: choroidal abnormalities in Legius syndrome. *J Hum Genet*. 2017 Nov;62(11):1001-1004. doi: 10.1038/jhg.2017.78. Epub 2017 Jul 27. Erratum in: *J Hum Genet*. 2018 Mar;63(3):391. PubMed PMID: 28747691.

15: Saletti V, **Esposito S**, Maccaro A, Giglio S, Valentini LG, Chiapparini L. Chiari I malformation in a child with PTEN hamartoma tumor syndrome: Association or coincidence? *Eur J Med Genet*. 2017 May;60(5):261-264. doi: 10.1016/j.ejmg.2017.03.002. Epub 2017 Mar 7. PubMed PMID: 28286253.

- 16: Riva D, Vago C, Erbetta A, Saletti V, **Esposito S**, Micheli R, Bulgheroni S. The Key Search Subtest of the Behavioural Assessment of the Dysexecutive Syndrome in Children (BADS-C) Instrument Reveals Impaired Planning Without External Constraints in Children With Neurofibromatosis Type 1. *J Child Neurol*. 2017 Mar;32(4):387-396. doi: 10.1177/0883073816683322. Epub 2016 Dec 20. PubMed PMID: 28193119.
- 17: 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):315-45. Review. PubMed PMID: 26861126.
- 18: Bianchessi D, Morosini S, Saletti V, Ibba MC, Natacci F, **Esposito S**, Cesaretti C, Riva D, Finocchiaro G, Eoli M. 126 novel mutations in Italian patients with neurofibromatosis type 1. *Mol Genet Genomic Med*. 2015 Jul 7;3(6):513-25. doi: 10.1002/mgg3.161. eCollection 2015 Nov. PubMed PMID: 26740943; PubMed Central PMCID: PMC4694136.
- 19: Erbetta A, Bulgheroni S, Contarino VE, Chiapparini L, **Esposito S**, Annunziata S, Riva D. Low-Functioning Autism and Nonsyndromic Intellectual Disability: Magnetic Resonance Imaging (MRI) Findings. *J Child Neurol*. 2015 Oct;30(12):1658-63. doi: 10.1177/0883073815578523. Epub 2015 Apr 20. PubMed PMID: 25895913.
- 20: Bianchi M, Saletti V, Micheli R, **Esposito S**, Molinaro A, Gagliardi S, Orcesi S, Cereda C. Legius Syndrome: two novel mutations in the SPRED1 gene. *Hum Genome Var*. 2015 Dec 3;2:15051. doi: 10.1038/hgv.2015.51. eCollection 2015. PubMed PMID: 27081556; PubMed Central PMCID: PMC4785569.
- 21: Erbetta A, Bulgheroni S, Contarino V, Chiapparini L, **Esposito S**, Vago C, Riva D. Neuroimaging findings in 41 low-functioning children with autism spectrum disorder: a single-center experience. *J Child Neurol*. 2014 Dec;29(12):1626-31. doi: 10.1177/0883073813511856. Epub 2013 Dec 16. PubMed PMID: 24346312.
- 22: Riva D, Cazzaniga F, **Esposito S**, Bulgheroni S. Executive functions and cerebellar development in children. *Appl Neuropsychol Child*. 2013;2(2):97-103. doi: 10.1080/21622965.2013.791092. Epub 2013 Jun 9. Review. PubMed PMID: 23745837.
- 23: Baranello G, Cesaretti C, Zambonin F, Casalone R, Granata P, **Esposito S**, Alfei E, Natacci F. Partial Trisomy 13 and Partial Monosomy 8 Mosaicism Secondary to an Unbalanced De Novo Translocation: Highlighting an Uncommon Chromosomal Abnormality. *J Child Neurol*. 2013 Nov;28(11):1463-1466. Epub 2013 Apr 22. PubMed PMID: 23611886.
- 24: Novara F, Rizzo A, Bedini G, Girgenti V, **Esposito S**, Pantaleoni C, Ciccone R, Sciacca FL, Achille V, Della Mina E, Gana S, Zuffardi O, Estienne M. MEF2C deletions and mutations versus duplications: a clinical comparison. *Eur J Med Genet*. 2013 May;56(5):260-5. doi: 10.1016/j.ejmg.2013.01.011. Epub 2013 Feb 10. PubMed PMID: 23402836.
- 25: Riva D, Franceschetti S, Erbetta A, Baranello G, **Esposito S**, Bulgheroni S. Congenital brain damage: cognitive development correlates with lesion and electroencephalographic features. *J Child Neurol*. 2013 Apr;28(4):446-54. doi: 10.1177/0883073812447684. Epub 2012 Jun 29. PubMed PMID: 22752481.
- 26: Vidmer S, Sergio C, Veronica S, Flavia T, **Silvia E**, Sara B, Valentini LG, Daria R, Solero CL. The neurophysiological balance in Chiari type 1 malformation (CM1), tethered cord and related syndromes. *Neurol Sci*. 2011 Dec;32 Suppl 3:S311-6. doi: 10.1007/s10072-011-0692-3. PubMed PMID: 22012627.

27: Saletti V, **Esposito S**, Frittoli M, Valentini LG, Chiapparini L, Bulgheroni S, Riva D. Neurological pictures in paediatric Chiari I malformation. *Neurol Sci.* 2011 Dec;32 Suppl 3:S295-8. doi: 10.1007/s10072-011-0744-8. PubMed PMID: 21983864.

28: Riva D, Usilla A, Saletti V, **Esposito S**, Bulgheroni S. Can Chiari malformation negatively affect higher mental functioning in developmental age? *Neurol Sci.* 2011 Dec;32 Suppl 3:S307-9. doi: 10.1007/s10072-011-0779-x.