

**Dr.ssa Veronica Saletti – Pubblicazione degli ultimi 5 anni (2015-2019)**

**Risk of Optic Pathway Glioma in Neurofibromatosis Type 1: No Evidence of Genotype-Phenotype Correlations in A Large Independent Cohort.**

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.  
Cancers (Basel). 2019 Nov 21;11(12).

**Brain Tumors in NF1 Children: Influence on Neurocognitive and Behavioral Outcome.**

Taddei M, Erbetta A, Esposito S, Saletti V, Bulgheroni S, Riva D.  
Cancers (Basel). 2019 Nov 11;11(11).

**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.**

Tritto V, Ferrari L, Esposito S, Zuccotti P, Bianchessi D, Natacci F, Saletti V, Eoli M, Riva P.  
Genes (Basel). 2019 Nov 5;10(11).

**Clinical spectrum of individuals with pathogenic NF1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotype-phenotype study in neurofibromatosis type 1.**

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.  
Hum Mutat. 2020 Jan;41(1):299-315.

**Neurological malignancies in neurofibromatosis type 1.**

Eoli M, Saletti V, Finocchiaro G.  
Curr Opin Oncol. 2019 Nov;31(6):554-561.

**Fluorescein-guided resection of plexiform neurofibromas: how I do it.**

Vetrano IG, Saletti V, Nazzi V.  
Acta Neurochir (Wien). 2019 Oct;161(10):2141-2145.

**Expanding the phenotypic spectrum of Allan-Herndon-Dudley syndrome in patients with SLC16A2 mutations.**

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Dev Med Child Neurol. 2019 Dec;61(12):1439-1447.

**Chiari I malformation in defined genetic syndromes in children: are there common pathways?**

Saletti V, Viganò I, Melloni G, Pantaleoni C, Vetrano IG, Valentini LG.  
Childs Nerv Syst. 2019 Oct;35(10):1727-1739.

**Novel mutations in SLC16A2 associated with a less severe phenotype of MCT8 deficiency.**

Masnada S, Groenweg S, Saletti V, Chiapparini L, Castellotti B, Salsano E, Visser WE, Tonduti D. *Metab Brain Dis.* 2019 Dec;34(6):1565-1575.

**Chiari 1 malformation and untreated sagittal synostosis: a new subset of complex Chiari?**

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**Molecular Genetics and Interferon Signature in the Italian Aicardi Goutières Syndrome Cohort: Report of 12 New Cases and Literature Review.**

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**Visuoperceptual Impairment in Children with NF1: From Early Visual Processing to Procedural Strategies.**

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**GEN-O-MA project: an Italian network studying clinical course and pathogenic pathways of moyamoya disease-study protocol and preliminary results.**

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**The molecular landscape of glioma in patients with Neurofibromatosis 1.**

D'Angelo F, Ceccarelli M, Tala, Garofano L, Zhang J, Frattini V, Caruso FP, Lewis G, Alfaro KD, Bauchet L, Berzero G, Cachia D, Cangiano M, Capelle L, de Groot J, DiMeco F, Ducray F, Farah W, Finocchiaro G, Goutagny S, Kamiya-Matsuoka C, Lavarino C, Loiseau H, Lorgis V, Marras CE, McCutcheon I, Nam DH, Ronchi S, Saletti V, Seizeur R, Slopis J, Suñol M, Vandenbos F, Varlet P, Vidaud D, Watts C, Tabar V, Reuss DE, Kim SK, Meyronet D, Mokhtari K, Salvador H, Bhat KP, Eoli M, Sanson M, Lasorella A, Iavarone A. *Nat Med.* 2019 Jan;25(1):176-187.

**Clinical spectrum of PTEN mutation in pediatric patients. A bicenter experience.**

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**Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970\_2972del): an update of genotype-phenotype correlation.**

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Genet Med. 2019 Apr;21(4):867-876. Erratum in: Genet Med. 2019 Mar;21(3):764-765.

**Progressive bone impairment with age and pubertal development in neurofibromatosis type I.**

Rodari G, Scuvera G, Olivieri FM, Profka E, Menni F, Saletti V, Esposito S, Bergamaschi S, Ferrante E, Eller-Vainicher C, Esposito S, Arosio M, Giavoli C.  
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**Encephalopathies with intracranial calcification in children: clinical and genetic characterization.**

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**The noncoding RNA AK127244 in 2p16.3 locus: A new susceptibility region for neuropsychiatric disorders.**

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.  
Am J Med Genet B Neuropsychiatr Genet. 2018 Sep;177(6):557-562.

**Correction: The absence that makes the difference: choroidal abnormalities in Legius syndrome.**

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.  
J Hum Genet. 2018 Mar;63(3):391.

**Unique combination of myxopapillary ependymoma and conus lipoma with subcutaneous extension in an 11-month-old child.**

Vetrano IG, Erbetta A, Pollo B, Saletti V, Valentini LG.  
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**Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844-848.**

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**A PDE10A de novo mutation causes childhood-onset chorea with diurnal fluctuations.**

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**The absence that makes the difference: choroidal abnormalities in Legius syndrome.**

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.  
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**Chiari I malformation in a child with PTEN hamartoma tumor syndrome: Association or coincidence?**

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**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.**

Riva D, Vago C, Erbetta A, Saletti V, Esposito S, Micheli R, Bulgheroni S.  
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**126 novel mutations in Italian patients with neurofibromatosis type 1.**

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Mol Genet Genomic Med. 2015 Jul 7;3(6):513-25.

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**Mutations in the PP2A regulatory subunit B family genes PPP2R5B, PPP2R5C and PPP2R5D cause human overgrowth.**

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**Electroencephalographic (EEG) Photoparoxysmal Responses Under 5 Years of Age: Diagnostic Implications and Peculiarities.**

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