Pubblicazioni su riviste scientifiche peer review (aggiornate al 1/2020)

Numero totale pubblicazioni: 46, di cui 14 come primo autore
H index: 13 (fonte Scopus ID: 26646312900 e Research gate https://www.researchgate.net/profile/Anna_Ardissone)

1) "Fukutin gene mutations in an Italian patient with early onset muscular dystrophy but no central nervous system involvement"
Saredi S, Ruggieri A, Mottarelli E, **Ardissone A,** Zanotti S, Farina L, Morandi L, Mora M, Moroni I. Muscle Nerve. 2009 Jun;39(6):845-8 IF 2.3

2) "*Novel POMGNT1 point mutations and intragenic rearrangements associated with muscle-eye-brain disease"*
Saredi S, **Ardissone A,** Ruggieri A, Mottarelli E, Farina L, Rinaldi R, Silvestri E, Gandioli C, D'Arrigo S, Salerno F, Morandi L, Grammatico P, Pantaleoni C, Moroni I, Mora M. J Neurol Sci. 2012 Jul 15;318(1-2):45-50 IF 2.2

3) "*Novel PTRF-cavin mutation associated with myopathy and mild congenital lipodystrophy"*
Anna Ardissone, Cinzia Bragato, Lorella Caffi, Renato Mantegazza, Lucia Morandi, Isabella Moroni, Marina Mora; BMC Medical Genetics 14(1),89 IF 2.5

4) *A fourth case of POMT2-related limb girdle muscle dystrophy with mild reduction of alfa-dystroglycan glycosylation*
S. Saredi, S. Gibertini, **A. Ardissone**, I. Fusco, S. Zanotti, F. Blasevich, L.Morandi. M.Mora. I. Moroni European Journal of Paediatric Neurology 18(3), 404-408 IF 2.3

5) *Double-trouble in pediatric neurology: Myotonia Congenita combined with Charcot-Marie-Tooth disease type 1A*
Anna Ardissone, Raffaella Brugnoni , Claudia Gandioli, Micaela Milani, Claudia Ciano, Graziella Uziel, Isabella Moroni Muscle and Nerve, 2014 Jul;50(1):145-7 IF 2.3

6) Mitochondrial dysfunction in CNS white matter disorders
Laia Morató L, Enrico Bertini, Daniela Verrigni, Graziella Uziel , **Anna Ardissone** , Isidre Ferrer, Aurora Pujol Glia, 2014 Nov;62(11):1878-94 IF 6

7) *Mitochondrial diseases in childhood- Review*
Anna Ardissone , Eleonora Lamantea, Federica Invernizzi , Silvia Genitrini, Isabella Moroni Graziella Uziel Current Molecular Medicine 14(8), pp. 1069-1078 IF 3.7

8) *Mutations in APOPT1, encoding a mitochondrial protein, cause cavitating leukoencephalopathy with cytochrome c oxidase deficiency*
Melchionda L, Haack TB, Hardy S, Abbink TE, Fernandez-Vizarra E, Lamantea E, Marchet S, Morandi L, Moggio M, Carrozzo R, Torraco A, Diodato D, Strom TM, Meitinger T, Tekturk P, Yapici Z, Al-Murshedi F, Stevens R, Rodenburg RJ, Lamperti C, **Ardissone A**, Moroni I, Uziel G, Prokisch H, Taylor RW, Bertini E, van der Knaap MS, Ghezzi D, Zeviani M.

American Journal of Human Genetics, 2014 Sep 4;95(3):315-25 IF 11

9) *Cavitating leukoencephalopathy with multiple mitochondrial dysfunction syndrome and NFU1 mutations*
Invernizzi F, **Ardissone A** (co-first autor), Lamantea E, Garavaglia B, Zeviani M, Farina L, Ghezzi D and Moroni I, Frontiers genetics, 2014 Nov 20;5:412

10) Childhood onset of acquired neuromyotonia: association with a ganglioneuroma
Anna Ardissone, Giovanna Zorzi, Claudia Ciano, Isabella Moroni
Muscle and nerve, 2015 Apr;51(4):620-1 IF 2.7

11) *A novel homozygous YARS2 mutation in two italian siblings and a review of literature* **Anna Ardissone**, Eleonora Lamantea, Jade Quartararo, Cristina Dallabona, Franco Carrara, Isabella Moroni,Claudia Donnini, Barbara Garavaglia, Massimo Zeviani, Graziella Uziel
Journal of Inherited Metabolic Disease, JIMD Rep. 2015;20:95-101

12) *A slowly progressive mitochondrial encephalomyopathy widens the clinical spectrum of AIFM1-related disorders*
Anna Ardissone, Giuseppe Piscosquito, Andrea Legati, Tiziana Langella, Eleonora Lamantea, Barbara Garavaglia, Ettore Salsano, Laura Farina, Isabella Moroni, Davide Pareyson, Daniele Ghezzi
Neurology 84(21), pp. 2193-2195 IF 8.2

13) *Long term follow-up to evaluate the efficacy of miglustat treatment in Italian patients with Niemann-Pick disease type C.*
Simona Fecarotta, Diana Bruschini, Roberto Della Casa, Alfonso Romano, Giuseppina Mansi, Ennio Del Giudice, Bruno Bembi, Agata Fiumara, Maja Di Rocco, Graziella Uziel, **Anna Ardissone**, Dario Roccatello, Mirella Alpa, Enrico Bertini, Adele D'Amico, Carlo Dionisi Vici, Federica Deodato, Antonio Federico, Silvia Palmeri, Orazio Gabrielli, Lucia Santoro, Antonio Filla, Cinzia Russo, Giancarlo Parenti, Generoso Andria Orphanet Journal of Rare Diseases 2015 Feb 27;10:22 IF 3.6

14) *Mitochondrial complex III deficiency caused by TTC19 defects: report of a novel mutation and review of literature*
Anna Ardissone, Tiziana Granata, Andrea Legati, Daria Diodato, Laura Melchionda, Eleonora Lamantea, Barbara Garavaglia, Daniele Ghezzi, Isabella Moroni Journal of Inherited Metabolic Disease 2015;22:115-20

15)*Mitochondrial leukoencephalopathy and complex II deficiency associated with a recessive SDHB mutation with reduced penetrance*
A.Ardissone, F.Invernizzi, A.Nasca, I.Moroni, L.Farina, D.Ghezzi
Molecular Genetics and Metabolism Reports 5:51-54, December 2015 IF 3.1

16) *MRI spectrum of Succinate Dehydrogenase-related infantile leukoencephalopathy.*
G Helman, BS, L Caldovic, PhD, MT. Whitehead, MD, Cas Simons, PhD, K Brockmann, MD, S Edvardson, MD, R Bai, MD, PhD, I Moroni, MD, JM Taylor, MD, SDH Work Group (…**A.Ardissone** …), RJ. Taft, PhD, A Vanderver, MD, MS. van der Knaap, MD, PhD.
Ann Neurol. 2016 Mar;79(3):379-86 IF 9.9

17) *LYRM7 mutations cause a multifocal cavitating leukoencephalopathy with a distinct magnetic resonance imaging*
C.Dallabona, E.M. Abbink, R.Carrozzo, A.Torraco, A.Legati,C.G.M. van Berkel, M.Niceta, T.Langella, D.Verrigni, T.Rizza, D.Diodato, F.Piemonte, E.Lamantea, M.Fang, J.Zhang, D.Martinelli, E.Bevivino, C.Dionisi-Vici, A.Vanderver, S.G. Philip, M.Kurian, C. Verma, S.Bijarnia-Mahay, S.Jacinto, F.Furtado, P.Accorsi, **A.Ardissone**, I.Moroni, I.Ferrero, M.Tartaglia, P.Goffrini, D.Ghezzi, M.S. van der Knaap, E. Bertini
Brain. 2016 Mar;139(Pt 3):782-94 IF 10

18) *SEPN1-Related Myopathy In Three Patients: Novel Mutations And Diagnostic Clues*
A. Ardissone, C. Bragato , F.Blasevich , E.Maccagnano, F.Salerno, C.Gandioli, L.Morandi, M.Mora , I.Moroni
European Journal of Pediatrics 175(8), pp. 1113-1118 IF 1.9

19) *Unusual presentations and intrafamilial phenotypic variability in infantile onset Alexander disease*.
Tonduti D, **Ardissone A,** Ceccherini I, Giaccone G, Farina L, Moroni I.
Neurological Sciences 37(6), pp. 973-977 IF 1.7

20) *New genes and pathomechanisms in mitochondrial disorders unraveled by NGS technologies*.
Legati A, Reyes A, Nasca A, Invernizzi F, Lamantea E, Tiranti V, Garavaglia B, Lamperti C, **Ardissone A**, Moroni I, Robinson A, Ghezzi D, Zeviani M.
Biochimica et Biophysica Acta – Bioenergetics 1857(8), pp. 1326-1335 IF 4.9

21) *Elevated aspartate aminotransferase and lactate dehydrogenase levels are a constant finding in PLA2G6-associated neurodegeneration*.
Kraoua I, Romani M, Tonduti D, BenRhouma H, Zorzi G, Zibordi F, **Ardissone A**, Gouider-Khouja N, Ben Youssef-Turki I, Nardocci N, Valente EM. European Journal of Neurology
23(4), pp. e24-e25 IF 3.9

22) "*Mitochondrial neuropathies": A survey from the large cohort of the Italian Network*.
Mancuso M, Orsucci D, Angelini C, Bertini E, Carelli V, Comi GP, Federico A, Minetti C, Moggio M, Mongini T, Tonin P, Toscano A, Bruno C, Ienco EC, Filosto M, Lamperti C, Diodato D, Moroni I, Musumeci O, Pegoraro E, Spinazzi M, Ahmed N, Sciacco M, Vercelli L, **Ardissone A**, Zeviani M, Siciliano G.
Neuromuscular Disorders 26(4-5), pp. 272-276 IF 2.9

23) *Neurological Disorders Associated with Striatal Lesions: Classification and Diagnostic Approach.* Tonduti D, Chiapparini L, Moroni I, **Ardissone A**, Zorzi G, Zibordi F, Raspante S, Panteghini C, Garavaglia B, Nardocci N
Curr Neurol Neurosci Rep. 2016 Jun;16(6):54. IF 3.3

24) *Biallelic Mutations in DNM1L are Associated with a Slowly Progressive Infantile Encephalopathy.*
Nasca A, Legati A, Baruffini E, Nolli C, Moroni I, **Ardissone A**, Goffrini P, Ghezzi D.
Human Mutation 37(9), pp. 898-903 IF 4.6

25) *COA7 (C1orf163/RESA1) mutations associated with mitochondrial leukoencephalopathy and cytochrome c oxidase deficiency*.

Martinez Lyons A, **Ardissone A**, Reyes A, Robinson AJ, Moroni I, Ghezzi D, Fernandez-Vizarra E, Zeviani M.
Journal of Medical Genetics 53(12), pp. 846-849 IF 5.4

26) *Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes*
Torraco A, **Ardissone A**, Invernizzi F, Rizza T, Fiermonte G, Niceta M, Zanetti N, Martinelli D, Vozza A, Verrigni D, Di Nottia M, Lamantea E, Diodato D, Tartaglia M, Dionisi-Vici C, Moroni I, Farina L, Bertini E, Ghezzi D, Carrozzo R.
Journal of Neurology 264(1), pp. 102-111 IF 3.8

27) *Intrafamilial phenotypic variability in Andersen-Tawil syndrome: A diagnostic challenge in a potentially treatable condition.*
Ardissone A, Sansone V, Colleoni L, Bernasconi P, Moroni I.
Neuromuscul Disord. 2017 Mar;27(3):294-296 IF 2.5

28) *Diagnosis of Duchenne Muscular Dystrophy in Italy in the last decade: Critical issues and areas for improvements.*
D'Amico A, Catteruccia M, Baranello G, Politano L, Govoni A, Previtali SC, Pane M, D'Angelo MG, Bruno C, Messina S, Ricci F, Pegoraro E, Pini A, Berardinelli A, Gorni K, Battini R, Vita G, Trucco F, Scutifero M, Petillo R, D'Ambrosio P, **Ardissone A**, Pasanisi B, Vita G, Mongini T, Moggio M, Comi GP, Mercuri E, Bertini E Neuromuscul Disord. 2017 May;27(5):447-451 IF 2.5

29) *Mutations in Epigenetic Regulation Genes Are a Major Cause of Overgrowth with Intellectual Disability.*
Tatton-Brown K, Loveday C, Yost S, Clarke M, Ramsay E, Zachariou A, Elliott A, Wylie H, **Ardissone A**, Rittinger O, Stewart F, Temple IK, Cole T; Childhood Overgrowth Collaboration., Mahamdallie S, Seal S, Ruark E, Rahman N. Am J Hum Genet. 2017 May 4;100(5):725-736. IF 8.9

30) *Not only dominant, not only optic atrophy: expanding the clinical spectrum associated with OPA1 mutations*.
Nasca A, Rizza T, Doimo M, Legati A, Ciolfi A, Diodato D, Calderan C, Carrara G, Lamantea E, Aiello C, Di Nottia M, Niceta M, Lamperti C, **Ardissone A**, Bianchi-Marzoli S, Iarossi G, Bertini E, Moroni I, Tartaglia M, Salviati L, Carrozzo R, Ghezzi D.
Orphanet J Rare Dis. 2017 May 12;12(1):8 IF 3.6

31) *Congenital myasthenic syndrome: phenotypic variability in patients harbouring p.T159P mutation in CHRNE gene*.
Ardissone A, Moroni I, Bernasconi P, Brugnoni R. Acta Myol. 2017 Mar;36(1):28-32.

32) *Revisiting mitochondrial ocular myopathies: a study from the Italian Network.*
Orsucci D, Angelini C, Bertini E, Carelli V, Comi GP, Federico A, Minetti C, Moggio M, Mongini T, Santorelli FM, Servidei S, Tonin P, **Ardissone A**, Bello L, Bruno C, Ienco EC, Diodato D, Filosto M, Lamperti C, Moroni I, Musumeci O, Pegoraro E, Primiano G, Ronchi D, Rubegni A, Salvatore S, Sciacco M, Valentino ML, Vercelli L, Toscano A, Zeviani M, Siciliano G, Mancuso M. Journal of Neurology 264(8), pp. 1777-1784 IF 3.8

33) *Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults.*
Charif M, Nasca A, Thompson K, Gerber S, Makowski C, Mazaheri N, Bris C, Goudenège D, Legati A, Marrofian R, Shariati G, Lamantea E, Hopton S, **Ardissone A**, Moroni I, Giannotta M, Siegel C, Strom TM, Prokisch H, Vignal-Clermont C, Derrien S, Zanlonghi X, Kaplan J, Hamel CP, Leruez S, Procaccio V, Bonneau D, Reynier P, White FE, Hardy SA, Barbosa IA, Simpson MA, Vara R, Perdomo Trujillo Y, Galehdari H, Deshpande C, Haack TB, Rozet JM, Taylor RW, Ghezzi D, Amati-Bonneau P, Lenaers G.
JAMA Neurology 75(1), pp. 105-113 IF 12

34) *Clinical, biochemical and genetic features associated with VARS2-related mitochondrial disease.*
Bruni F, Meo ID, Bellacchio E, Webb BD, McFarland R, Chrzanowska-Lightowlers ZMA, He L, Skorupa E, Moroni I, **Ardissone A**, Walczak A, Tyynismaa H, Isohanni P, Mandel H, Prokisch H, Haack T, Bonnen PE, Enrico B, Pronicka E, Ghezzi D, Taylor RW, Diodato D.
Human Mutation 39(4), pp. 563-578 IF 5.4

35) Compound heterozygous missense and deep intronic variants in NDUFAF6 unraveled by exome sequencing and mRNA analysis.
Catania A, **Ardissone A**, Verrigni D, Legati A, Reyes A, Lamantea E, Diodato D, Tonduti D, Imperatore V, Pinto AM, Moroni I, Bertini E, Robinson A, Carrozzo R, Zeviani M, Ghezzi D.
Journal of Human Genetics 63(5), pp. 563-568 IF 2.9

36) *KARS-related diseases: progressive leukoencephalopathy with brainstem and spinal cord calcifications as new phenotype and a review of literature.*
Ardissone A, Tonduti D, Legati A, Lamantea E, Barone R, Dorboz I, Boespflug-Tanguy O, Nebbia G, Maggioni M, Garavaglia B, Moroni I, Farina L, Pichiecchio A, Orcesi S, Chiapparini L, Ghezzi D
Orphanet J Rare Dis. 2018 Apr 4;13(1):45. IF 4.1

37) *Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?*
Repp BM, Mastantuono E, Alston CL, Schiff M, Haack TB, Rötig A, **Ardissone A**, Lombès A, Catarino CB, Diodato D, Schottmann G, Poulton J, Burlina A, Jonckheere A, Munnich A, Rolinski B, Ghezzi D, Rokicki D, Wellesley D, Martinelli D, Wenhong D, Lamantea E, Ostergaard E, Pronicka E, Pierre G, Smeets HJM, Wittig I, Scurr I, de Coo IFM, Moroni I, Smet J, Mayr JA, Dai L, de Meirleir L, Schuelke M, Zeviani M, Morscher RJ, McFarland R, Seneca S, Klopstock T, Meitinger T, Wieland T, Strom TM, Herberg U, Ahting U, Sperl W, Nassogne MC, Ling H, Fang F, Freisinger P, Van Coster R, Strecker V, Taylor RW, Häberle J, Vockley J, Prokisch H, Wortmann S.
Orphanet Journal of Rare Diseases 13(1),120 IF 4.1

38) *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, **Ardissone 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 IF 3

39) *Encephalopathies with intracranial calcification in children: clinical and genetic characterization*.

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, Chiapparini L, Garavaglia B, Orcesi S. Orphanet J Rare Dis. 2018 Aug 16;13(1):135 IF 4.1

40) *Substantia nigra swelling and dentate nucleus T2-hyperintensity may be early MRI signs of BPAN: Early MRI features in four cases of BPAN*
Russo C., **Ardissone A.,** Freri E., Gasperini S., Moscatelli M., Zorzi G., Panteghini C, Castellotti B. , Garavaglia B., Nardocci N., Chiapparini L., Movement Disorders Clinical Practice October 2018 DOI: 10.1002/mdc3.12693

41) Clinical-genetic features and peculiar muscle histopathology in infantile DNM1L-related mitochondrial epileptic encephalopathy.
Verrigni D, Di Nottia M, **Ardissone A** (co-first author), Baruffini E, Nasca A, Legati A, Bellacchio E, Fagiolari G, Martinelli D, Fusco L, Battaglia D, Trani G, Versienti G, Marchet S, Torraco A, Rizza T, Verardo M, D'Amico A, Diodato D, Moroni I, Lamperti C, Petrini S, Moggio M, Goffrini P, Ghezzi D, Carrozzo R, Bertini E.
Hum Mutat. 2019 May;40(5):601-618

42) *Exome sequencing detects compound heterozygous nonsense LAMA2 mutations in two siblings with atypical phenotype and nearly normal brain MRI.*
Saredi S, Gibertini S, Matalonga L, Farina L, **Ardissone A**, Moroni I, Mora M.
Neuromuscul Disord. 2019 May;29(5):376-38

43) *Epileptic phenotypes in children with early-onset mitochondrial diseases.*
Matricardi S, Canafoglia L, **Ardissone A**, Moroni I, Ragona F, Ghezzi D, Lamantea E, Nardocci N, Franceschetti S, Granata T.
Acta Neurol Scand. 2019 May 18

44) *HIST1H1E heterozygous protein-truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals.*
Burkardt DD, Zachariou A, Loveday C, Allen CL, Amor DJ, **Ardissone A**, Banka S, Bourgois A, Coubes C, Cytrynbaum C, Faivre L, Marion G, Horton R, Kotzot D, Lay-Son G, Lees M, Low K, Luk HM, Mark P, McConkie-Rosell A, McDonald M, Pappas J, Phillipe C, Shears D, Skotko B, Stewart F, Stewart H, Temple IK, Mau-Them FT, Verdugo RA, Weksberg R, Zarate YA, Graham JM, Tatton-Brown K.

Am J Med Genet A. 2019 Oct;179(10):2049-2055

45) *Pre-diagnosing and managing patients with GM1 gangliosidosis and related disorders by the evaluation of GM1 ganglioside content*.

Tonin R, Caciotti A, Procopio E, Fischetto R, Deodato F, Mancardi MM, Di Rocco M, **Ardissone A,** Salviati A, Marangi A, Strisciuglio P, Mangone G, Casini A, Ricci S, Fiumara A, Parini R, Pavone FS, Guerrini R, Calamai M, Morrone A Sci Rep. 2019 Nov 27;9(1):17684. doi: 10.1038/s41598-019-53995-5.

46) *Mitochondrial epilepsy: a cross-sectional nationwide Italian survey.*

Ticci C, Sicca F, **Ardissone A,** Bertini E, Carelli V, Diodato D, Di Vito L, Filosto M, La Morgia C, Lamperti C, Martinelli D, Moroni I, Musumeci O, Orsucci D, Pancheri E, Peverelli L, Primiano G, Rubegni A, Servidei S, Siciliano G, Simoncini C, Tonin P, Toscano A, Mancuso M, Santorelli FM.

Neurogenetics. 2020 Jan 3