

1. Mol Genet Metab Rep. 2016 Sep 1;9:12-4. "Mitochondrial m.3243A > G mutation and carotid artery dissection." Mancuso M, Montano V, Orsucci D, Peverelli L, Caputi L, Gambaro P, Siciliano G, Lamperti C.
2. Neuromuscul Disord. 2013 Nov;23(11):907-10. "An "inflammatory" mitochondrial myopathy. A case report." Mancuso M, Orsucci D, Ienco EC, Ricci G, Ali G, Servadio A, Fontanini G, Filosto M, Vielmi V, Rocchi A, Petrozzi L, Logerfo A, Siciliano G.
3. Neurology. 2013 May 28;80(22):2049-54. "Phenotypic heterogeneity of the 8344A>G mtDNA "MERRF" mutation." Mancuso M, Orsucci D, Angelini C, Bertini E, Carelli V, Comi GP, Minetti C, Moggio M, Mongini T, Servidei S, Tonin P, Toscano A, Uziel G, Bruno C, Caldarazzo Ienco E, Filosto M, Lamperti C, Martinelli D, Moroni I, Musumeci O, Pegoraro E, Ronchi D, Santorelli FM, Sauchelli D, Scarpelli M, Sciacco M, Spinazzi M, Valentino ML, Vercelli L, Zeviani M, Siciliano G.
4. Am J Hum Genet. 2013 Feb 7;92(2):293-300. "Mutations in DNA2 link progressive myopathy to mitochondrial DNA instability." Ronchi D, Di Fonzo A, Lin W, Bordoni A, Liu C, Fassone E, Pagliarani S, Rizzuti M, Zheng L, Filosto M, Ferrò MT, Ranieri M, Magri F, Peverelli L, Li H, Yuan YC, Corti S, Sciacco M, Moggio M, Bresolin N, Shen B, Comi GP.
5. Neuromuscul Disord. 2012 Dec;22 Suppl 3:S226-9. "Fatigue and exercise intolerance in mitochondrial diseases. Literature revision and experience of the Italian Network of mitochondrial diseases." Mancuso M, Angelini C, Bertini E, Carelli V, Comi GP, Minetti C, Moggio M, Mongini T, Servidei S, Tonin P, Toscano A, Uziel G, Zeviani M, Siciliano G; Nation-wide Italian Collaborative Network of Mitochondrial Diseases.
6. Biochim Biophys Acta. 2013 Mar;1832(3):445-52. "Cybrid studies establish the causal link between the mtDNA m.3890G>A/MT-ND1 mutation and optic atrophy with bilateral brainstem lesions." Caporali L, Ghelli AM, Iommari L, Maresca A, Valentino ML, La Morgia C, Liguori R, Zanna C, Barboni P, De Nardo V, Martinuzzi A, Rizzo G, Tonon C, Lodi R, Calvaruso MA, Cappelletti M, Porcelli AM, Achilli A, Pala M, Torroni A, Carelli V.
7. Neuromuscul Disord. 2012 Dec;22 Suppl 3:S172-7. "Effects of aerobic training on exercise-related oxidative stress in mitochondrial myopathies." Siciliano G, Simoncini C, Lo Gerfo A, Orsucci D, Ricci G, Mancuso M.
8. Curr Neurovasc Res. 2013 Feb;10(1):76-80. "Vascular factors and mitochondrial dysfunction: a central role in the pathogenesis of Alzheimer's disease." Orsucci D, Mancuso M, Ienco EC, Simoncini C, Siciliano G, Bonuccelli U.
9. Brain. 2012 Nov;135(Pt 11):3404-15. "Next-generation sequencing reveals DGUOK mutations in adult patients with mitochondrial DNA multiple deletions." Ronchi D, Garone C, Bordoni A, Gutierrez Rios P, Calvo SE, Ripolone M, Ranieri M, Rizzuti M, Villa L, Magri F, Corti S, Bresolin N, Mootha VK, Moggio M, DiMauro S, Comi GP, Sciacco M.
10. Mol Genet Metab. 2012 Nov;107(3):383-8. "EPI-743 reverses the progression of the pediatric mitochondrial disease--genetically defined Leigh Syndrome." Martinelli D, Catteruccia M, Piemonte F, Pastore A, Tozzi G, Dionisi-Vici C, Pontrelli G, Corsetti T, Livadiotti S, Kheifets V, Hinman A, Shrader WD, Thoolen M, Klein MB, Bertini E, Miller G.
11. Neurogenetics. 2012 Nov;13(4):375-86. "TMEM70: a mutational hot spot in nuclear ATP synthase deficiency with a pivotal role in complex V biogenesis." Torraco A1, Verrigni D, Rizza T, Meschini MC, Vazquez-Memije ME, Martinelli D, Bianchi M, Piemonte F, Dionisi-Vici C, Santorelli FM, Bertini E, Carrozzo R.
12. Nat Protoc. 2012 May 31;7(6):1235-46. "Assessment of mitochondrial respiratory chain enzymatic activities on tissues and cultured cells." Spinazzi M, Casarin A, Pertegato V, Salviati L, Angelini C.

13. J Inherit Metab Dis. 2013 Jan;36(1):43-53 "Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients." Cassandrini D, Cilio MR, Bianchi M, Doimo M, Balestri M, Tessa A, Rizza T, Sartori G, Meschini MC, Nesti C, Tozzi G, Petruzzella V, Piemonte F, Bisceglia L, Bruno C, Dionisi-Vici C, D'Amico A, Fattori F, Carrozzo R, Salviati L, Santorelli FM, Bertini E.
14. Expert Opin Pharmacother. 2012 Mar;13(4):527-43 "Drugs and mitochondrial diseases: 40 queries and answers." Mancuso M, Orsucci D, Filosto M, Simoncini C, Siciliano G.
15. J Neurol Sci. 2012 Apr 15;315(1-2):146-9. "Optic atrophy plus phenotype due to mutations in the OPA1 gene: two more Italian families." Ranieri M, Del Bo R, Bordoni A, Ronchi D, Colombo I, Riboldi G, Cosi A, Servida M, Magri F, Moggio M, Bresolin N, Comi GP, Corti S.
16. Neurol Sci. 2013 Jan;34(1):71-4. "Psychiatric involvement in adult patients with mitochondrial disease." Mancuso M, Orsucci D, Ienco EC, Pini E, Choub A, Siciliano G.
17. Biochem Biophys Res Commun. 2011 Nov 18;415(2):300-4. "Novel large-range mitochondrial DNA deletions and fatal multisystemic disorder with prominent hepatopathy." Bianchi M, Rizza T, Verrigni D, Martinelli D, Tozzi G, Torraco A, Piemonte F, Dionisi-Vici C, Nobili V, Francalanci P, Boldrini R, Callea F, Santorelli FM, Bertini E, Carrozzo R.
18. Mitochondrion. 2011 Nov;11(6):893-904. "Optimization of respiratory chain enzymatic assays in muscle for the diagnosis of mitochondrial disorders." Spinazzi M1, Casarin A, Pertegato V, Ermani M, Salviati L, Angelini C.
19. Curr Med Chem. 2011;18(26):4053-64. "Targeting mitochondrial dysfunction and neurodegeneration by means of coenzyme Q10 and its analogues." Orsucci D1, Mancuso M, Ienco EC, LoGerfo A, Siciliano G.
20. Biochem Biophys Res Commun. 2011 Aug 26;412(2):245-8. "Unusual adult-onset Leigh syndrome presentation due to the mitochondrial m.9176T>C mutation." Ronchi D1, Bordoni A, Cosi A, Rizzuti M, Fassone E, Di Fonzo A, Servida M, Sciacco M, Collotta M, Ronzoni M, Lucchini V, Mattioli M, Moggio M, Bresolin N, Corti S, Comi GP.
21. Neurol Sci. 2012 Apr;33(2):449-52. "Nerve and muscle involvement in mitochondrial disorders: an electrophysiological study." Mancuso M1, Piazza S, Volpi L, Orsucci D, Calsolaro V, Caldarazzo Ienco E, Carlesi C, Rocchi A, Petrozzi L, Calabrese R, Siciliano G
22. BMC Neurol. 2011 Jul 12;11:85. "Clinical and molecular features of an infant patient affected by Leigh Disease associated to m.14459G>A mitochondrial DNA mutation: a case report." Ronchi D, Cosi A, Tonduti D, Orcesi S, Bordoni A, Fortunato F, Rizzuti M, Sciacco M, Collotta M, Cagdas S, Capovilla G, Moggio M, Berardinelli A, Veggiani P, Comi GP.
23. J Neurol Sci. 2011 Sep 15;308(1-2):173-6. "Two novel mutations in PEO1 (twinkle) gene associated with chronic external ophthalmoplegia." Ronchi D, Fassone E, Bordoni A, Sciacco M, Lucchini V, Di Fonzo A, Rizzuti M, Colombo I, Napoli L, Ciscato P, Moggio M, Cosi A, Collotta M, Corti S, Bresolin N, Comi GP.
24. J Alzheimers Dis. 2011;24 Suppl 2:111-26. "Oxidative stress treatment for clinical trials in neurodegenerative diseases." Ienco EC1, LoGerfo A, Carlesi C, Orsucci D, Ricci G, Mancuso M, Siciliano G.
25. J Mol Neurosci. 2011 May;44(1):17-24. "POLG1-related and other "mitochondrial Parkinsonisms": an overview." Orsucci D, Caldarazzo Ienco E, Mancuso M, Siciliano G.
26. Neurogenetics. 2011 Feb;12(1):9-17. "Progressive cavitating leukoencephalopathy associated with respiratory chain complex I deficiency and a novel mutation in NDUFS1." Ferreira M, Torraco A, Rizza T, Fattori F, Meschini MC, Castana C, Go NE, Nargang FE, Duarte M, Piemonte F, Dionisi-Vici C, Videira A, Vilarinho L, Santorelli FM, Carrozzo R, Bertini E.
27. Neuropsychiatr Dis Treat. 2010 Sep 7;6:491-9. "Current and emerging treatment options in the management of Friedreich ataxia." Mancuso M, Orsucci D, Choub A, Siciliano G.

28. J Neurol Sci. 2012 Apr 15;315(1-2):146-9. "Optic atrophy plus phenotype due to mutations in the OPA1 gene: two more Italian families." Ranieri M, Del Bo R, Bordoni A, Ronchi D, Colombo I, Riboldi G, Cosi A, Servida M, Magri F, Moggio M, Bresolin N, Comi GP, Corti S.
29. Eur J Hum Genet. 2012 Mar;20(3):357-60. "The novel mitochondrial tRNAAsn gene mutation m.5709T>C produces ophthalmoparesis and respiratory impairment." Ronchi D, Sciacco M, Bordoni A, Raimondi M, Ripolone M, Fassone E, Di Fonzo A, Rizzuti M, Ciscato P, Cosi A, Servida M, Moggio M, Corti S, Bresolin N, Comi GP.
30. Mov Disord. 2014 May;29(6):722-8. "Myoclonus in mitochondrial disorders." Mancuso M, Orsucci D, Angelini C, Bertini E, Catteruccia M, Pegoraro E, Carelli V, Valentino ML, Comi GP, Minetti C, Bruno C, Moggio M, Ienco EC, Mongini T, Vercelli L, Primiano G, Servidei S, Tonin P, Scarpelli M, Toscano A, Musumeci O, Moroni I, Uziel G, Santorelli FM, Nesti C, Filosto M, Lamperti C, Zeviani M, Siciliano G.
31. Neuroepidemiology. 2012;38(3):194-5. "Quality of life in adult patients with mitochondrial myopathy." Orsucci D, Calsolaro V, Siciliano G, Mancuso M.
32. Neurol Res Int. 2013;2013:293893 "Mitochondrial fusion proteins and human diseases." Ranieri M, Brajkovic S, Riboldi G, Ronchi D, Rizzo F, Bresolin N, Corti S, Comi GP.
33. J Neurol. 2014 Sep;261 Suppl 2:S528-41. "The genetics of ataxia: through the labyrinth of the Minotaur, looking for Ariadne's thread." Mancuso M, Orsucci D, Siciliano G, Bonuccelli U.
34. JIMD Rep. 2015;23:85-9. "Expanding the Clinical and Magnetic Resonance Spectrum of Leukoencephalopathy with Thalamus and Brainstem Involvement and High Lactate (LTBL) in a Patient Harboring a Novel EARS2 Mutation." Biancheri R, Lamantea E, Severino M, Diodato D, Pedemonte M, Cassandrini D, Ploederl A, Trucco F, Fiorillo C, Minetti C, Santorelli FM, Zeviani M, Bruno
35. Int J Alzheimers Dis. 2011 Feb 22;2011:709061 "May "mitochondrial eve" and mitochondrial haplogroups play a role in neurodegeneration and Alzheimer's disease?" Ienco EC, Simoncini C, Orsucci D, Petrucci L, Filosto M, Mancuso M, Siciliano G.
36. Acta Myol. 2011 Jun;30(1):9-15. "Cardiological manifestations of mitochondrial respiratory chain disorders." Berardo A, Musumeci O, Toscano A.
37. Acta Myol. 2011 Oct; 30(2): 164. "P-9 Construction of a database for a nation-wide Italian collaborative network of mitochondrial diseases" M. Mancuso, C. Angelini, E. Bertini, E. Caldarazzo Ienco, V. Carelli, G.P. Comi, C. Minetti, T. Mongini, D. Orsucci, S. Servidei, P. Tonin, A. Toscano, G. Uziel, P. Santantonio, and G. Siciliano
38. Neuromuscul Disord. 2016 Apr-May;26(4-5):272-6. doi: 10.1016/j.nmd.2016.02.008. Epub 2016 Feb 23. "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, Ardissoni A, Zeviani M, Siciliano G.
39. J Neurol. 2015 May;262(5):1301-9. "Redefining phenotypes associated with mitochondrial DNA single deletion." Mancuso M, Orsucci D, Angelini C, Bertini E, Carelli V, Comi GP, Donati MA, Federico A, Minetti C, Moggio M, Mongini T, Santorelli FM, Servidei S, Tonin P, Toscano A, Bruno C, Bello L, Caldarazzo Ienco E, Cardaioli E, Catteruccia M, Da Pozzo P, Filosto M, Lamperti C, Moroni I, Musumeci O, Pegoraro E, Ronchi D, Sauchelli D, Scarpelli M, Sciacco M, Valentino ML, Vercelli L, Zeviani M, Siciliano G.
40. J Neurol. 2014 Mar;261(3):504-10."The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?" Mancuso M, Orsucci D, Angelini C, Bertini E, Carelli V, Comi GP, Donati A, Minetti C, Moggio M, Mongini T, Servidei S, Tonin P, Toscano A, Uziel G, Bruno C, Ienco EC, Filosto M, Lamperti C, Catteruccia M, Moroni I,

Musumeci O, Pegoraro E, Ronchi D, Santorelli FM, Sauchelli D, Scarpelli M, Sciacco M, Valentino ML, Vercelli L, Zeviani M, Siciliano G.