

# MARCELLO VILLANOVA

## ISTRUZIONE

1984 Liceo Classico “Quinto Ennio”, Taranto

1990 Laurea in Medicina, Università di Siena (110 cum laude) Titolo della tesi: atrofia cerebellare e ipogonadismo: studio di due casi (Pubblicata nel 1993 su J Submicrosc Cytol Pathol 25 (3):371-375)

1995 Dottorato di Ricerca in Neuroscienze

1998 Specializzazione in Neurologia, 70 cum laude, Università di Siena

2002 Riconoscimento abilitazione Terapia Fisica e Riabilitazione, ordine dei Medici di Bologna

## PREMI, BORSE DI STUDIO E CONTRATTI

1990 “Premio di Laurea”, Università di Siena

1991 Borsa di studio-Progetto Europeo “Erasmus”, 7 mesi

1993-1996 Borsa di studio Telethon

1995 Premio Glaxo per la Ricerca 1997 Contratto di Ricerca - Università di Siena (durata 1/1 - 31/12 1997)

1997-98 Contratto di Consulenza presso il Servizio di Neurofisiopatologia (dr. L. Merlini) degli Istituti Oropedici Rizzoli

1998-2001 Contratto di Ricerca presso il Servizio di Neurofisiopatologia (dr. L. Merlini) degli Istituti Oropedici Rizzoli

2001-2004 Contratto di ricerca in Neuroscienze presso l’Università di Siena

2004-2008 Contratto di consulenza Dipartimento di Neurologia, Università di Bologna

## POSIZIONE

1990 Abilitazione all’esercizio della professione Medica

1991-1995 Dottorato di Ricerca in Neuroscienze indirizzo Neuropatologia (4 anni, terminato in Ottobre 1995), svolto all'estero.

Titolo della tesi : Miosite ad inclusioni (IBM): ipotesi sul ruolo della amiloide, della proteina precursore della -amiloide e della proteina tau )

1998 Specializzazione in Neurologia

1990-91; 1995-97 Medico interno Istituto Scienze Neurologiche, Università di Siena

1998-2001 Contrattista, Servizio di Neurofisiopatologia, Istituto Ortopedico Rizzoli

1999-2000 Libero Professionista, Unità Funzionale Neuromotoria, ospedale privato accreditato "Villa Salus", Bologna

2001-2021 Responsabile, Unità di Riabilitazione e Recupero Funzionale Malattie Neuromuscolari, Ospedale PA "Nigrisoli", Bologna

2022 - ... Responsabile Unità di Riabilitazione e Recupero Funzionale Malattie Neuromuscolari, Presidio Ospedaliero Accreditato Villa Bellombra, Bologna

## **DOCENZA**

1999-2000 Insegnamento di "Terapia Neurologica" presso la Scuola di Specializzazione in Neurologia dell'Università di Siena

2001 - 2005 Insegnamento di "Riabilitazione Neurologica" presso la Scuola di Specializzazione in Neurologia dell'Università di Siena

## **ESPERIENZA ALL'ESTERO**

1991- 04/1994 Post-graduate fellow in Neuropatologia, Born-Bunge Foundation, Università di Anversa, Belgio (Prof. Dr. J.J. MARTIN).

05/1994 -2/95 Post-graduate fellow in Biologia e Patologia delle malattie neuromuscolari, Hospital Pitié-Salpêtrière, Parigi, Francia (Prof. Dr. M. FARDEAU, e Prof. Dr. F.M.S Tomè).

2001 Observer in the Center of ventilatory management of the University Hospital- New Jersey Medical school, New York (Prof. JR Bach)

2009 Observer in the Center of ventilatory management of the University Hospital- New Jersey Medical school, New York (Prof. JR Bach)

## ARTICOLI SELEZIONATI (FULL PAPER)

1. Respiratory muscle involvement in HNRNPDL LGMD D3 muscular dystrophy: an extensive clinical description of the first Italian patient. Malfatti E, Cassandrini D, Rubegni A, Sartorelli FM, Villanova M. *Acta Myol.* 2020 Jun 1;39(2):98-100. doi: 10.36185/2532-1900-013. eCollection 2020 Jun.
2. A new congenital multicore titinopathy associated with fast myosin heavy chain deficiency. Perrin A, Metay C, Villanova M, Carlier RY, Pegoraro E, Juntas Morales R, Stojkovic T, Richard I, Richard P, Romero NB, Granzier H, Koenig M, Malfatti E, Cossée M. *Ann Clin Transl Neurol.* 2020 May;7(5):846-854. doi: 10.1002/acn3.51031. Epub 2020 Apr 19.
3. Airway clearance techniques in neuromuscular disorders: A state of the art review. Chatwin M, Toussaint M, Gonçalves MR, Sheers N, Mellies U, Gonzales-Bermejo J, Sancho J, Fauroux B, Andersen T, Hov B, Nygren-Bonnier M, Lacombe M, Pernet K, Kampelmacher M, Devaux C, Kinnett K, Sheehan D, Rao F, Villanova M, Berlowitz D, Morrow BM. *Respir Med.* 2018 Mar;136:98-110. doi: 10.1016/j.rmed.2018.01.012. Epub 2018 Feb
4. 228th ENMC International Workshop:: Airway clearance techniques in neuromuscular disorders Naarden, The Netherlands, 3-5 March, 2017.
5. Expanded access program with Nusinersen in SMA type I in Italy: Strengths and pitfalls of a successful experience. Messina S, Pane M, Sansone V, Bruno C, Catteruccia M, Vita G, Palermo C, Albamonte E, Pedemonte M, Bertini E, Binetti L, Mercuri E; Italian EAP working Group. *Neuromuscul Disord.* 2017 Dec;27(12):1084-1086. doi: 10.1016/j.nmd.2017.09.006. Epub 2017 Sep 21. No abstract available.
6. New Survival Target for Duchenne Muscular Dystrophy. Villanova M, Kazibwe S. *Am J Phys Med Rehabil.* 2017 Feb;96(2):e28-e30. doi: 10.1097/PHM.0000000000000569.
7. Allogeneic mesenchymal stem cell therapy outcomes for three patients with spinal muscular atrophy type 1. Villanova M, Bach JR. *Am J Phys Med Rehabil.* 2015 May;94(5):410-5. doi: 10.1097/PHM.
8. 1st Italian SMA Family Association Consensus Meeting: Management and recommendations for respiratory involvement in spinal muscular atrophy (SMA) types I-III, Rome, Italy, 30-31 January 2015. Toussaint M, Chatwin M, Gonzales J, Berlowitz DJ; ENMC Respiratory Sansone VA, Racca F, Ottonello G, Vianello A, Berardinelli A, Crescimanno G, Casiraghi JL; Italian SMA Family Association. *Neuromuscul Disord.* 2015 Dec;25(12):979-89. doi: 10.1016/j.nmd.2015.09.009. Epub 2015 Sep 18.

9. Prevalence of congenital muscular dystrophy in Italy: a population study.

Graziano A, Bianco F, D'Amico A, Moroni I, Messina S, Bruno C, Pegoraro E, Mora M, Astrea G, Magri F, Comi GP, Berardinelli A, Moggio M, Morandi L, Pini A, Petillo R, Tasca G, Monforte M, Minetti C, Mongini T, Ricci E, Gorni K, Battini R, Villanova M, Politano L, Gualandi F, Ferlini A, Muntoni F, Santorelli FM, Bertini E, Pane M, Mercuri E.

Neurology. 2015 Mar 3;84(9):904-11. doi: 10.1212/WNL.0000000000001303. Epub 2015 Feb 4.

10. Duchenne muscular dystrophy: life prolongation by noninvasive ventilatory support. Villanova M, Brancalione B, Mehta AD.

Am J Phys Med Rehabil. 2014 Jul;93(7):595-9. doi: 10.1097/PHM.0000000000000074 Therapy Consortium.

11. Duchenne muscular dystrophy and epilepsy.

Pane M, Messina S, Bruno C, D'Amico A, Villanova M, Brancalione B, Sivo S, Bianco F, Striano P, Battaglia D, Lettori D, Vita GL, Bertini E, Gualandi F, Ricotti V, Ferlini A, Mercuri E.

Neuromuscul Disord. 2013 Apr;23(4):313-5. doi: 10.1016/j.nmd.2013.01.011. Epub

12. Mutations in the N-terminal actin-binding domain of filamin C cause a distal myopathy.

Duff RM, Tay V, Hackman P, Ravenscroft G, McLean C, Kennedy P, Steinbach A, Schöffler W, van der Ven PFM, Fürst DO, Song J, Djinić-Carugo K, Penttilä S, Raheem O, Reardon K, Malandrini A, Gambelli S,

Villanova M, Nowak KJ, Williams DR, Landers JE, Brown RH Jr, Udd B, Laing NG.

Am J Hum Genet. 2011 Jun 10;88(6):729-740. doi: 10.1016/j.ajhg.2011.04.021. Epub 2011 May 27.

13. Reliability of the North Star Ambulatory Assessment in a multicentric setting.

Mazzone ES, Messina S, Vasco G, Main M, Eagle M, D'Amico A, Doglio L, Politano L, Cavallaro F, Frosini S, Bello L, Magri F, Corlatti A, Zucchini E, Brancalione B, Rossi F, Ferretti M, Motta MG, Cecio MR, Berardinelli A, Alfieri P, Mongini T, Pini A, Astrea G, Battini R, Comi G, Pegoraro E, Morandi L, Pane M, Angelini C, Bruno C,

Villanova M, Vita G, Donati MA, Bertini E, Mercuri E.

Neuromuscul Disord. 2009 Jul;19(7):458-61. doi: 10.1016/j.nmd.2009.06.368. Epub

14. Hepatitis C among former athletes: association with the use of injectable

stimulants in the past. Passos AD, Figueiredo JF, Martinelli Ade L, Villanova M, Nascimento MM, Secaf M.

Mem Inst Oswaldo Cruz. 2008 Dec;103(8):809-12.

15. Spastic paraplegia with thinning of the corpus callosum and white matter abnormalities: further mutations and relative frequency in ZFYVE26/SPG15 in the Italian population.  
Denora PS, Muglia M, Casali C, Truchetto J, Silvestri G, Messina D, Boukrhis A, Magariello A, Modoni A, Masciullo M, Malandrini A, Morelli M, de Leva MF, Villanova M, Giugni E, Citrigno L, Rizza T, Federico A, Pierallini A, Quattrone A, Filla A, Brice A, Stevanin G, Santorelli FM.  
J Neurol Sci. 2009 Feb 15;277(1-2):22-5. doi: 10.1016/j.jns.2008.09.039. Epub 2008 Dec 13.
16. Fine-mapping the gene for X-linked myopathy with excessive autophagy.  
Munteanu I, Ramachandran N, Mnatzakanian GN, Villanova M, Fardeau M, Levy N, Kissel JT, Minassian BA. Neurology. 2008 Sep 16;71(12):951-3. doi: 10.1212/01.wnl.0000325991.01899.35.
15. Daily salbutamol in young patients with SMA type II.  
Pane M, Staccioli S, Messina S, D'Amico A, Pelliccioni M, Mazzone ES, Cuttini M, Alfieri P, Battini R, Main M, Muntoni F, Bertini E, Villanova M, Mercuri E.  
Neuromuscul Disord. 2008 Jul;18(7):536-40. doi: 10.1016/j.nmd.2008.05.004. Epub
17. Tracheostomy tubes are not needed for Duchenne muscular dystrophy.  
Bach JR, Bianchi C, FINDER J, Fragasso T, Goncalves MR, Ishikawa Y, Ramlall AK, McKim D, Servera E, Vianello A, Villanova M, Winck JC.  
Eur Respir J. 2007 Jul;30(1):179-80; author reply 180-1. No
18. The Hammersmith functional score correlates with the SMN2 copy number: a multicentric study.  
Tiziano FD, Bertini E, Messina S, Angelozzi C, Pane M, D'Amico A, Alfieri P, Fiori S, Battini R, Berardinelli A, Boffi P, Bruno C, Cini C, Minetti C, Mongini T, Morandi L, Orcesi S, Pelliccioni M, Pini A, Villanova M, Vita G, Locatelli M, Mercuri E, Brahe C.  
Neuromuscul Disord. 2007 May;17(5):400-3.  
Neurology. 2007 Jan 2;68(1):51-5.
19. Randomized, double-blind, placebo-controlled trial of phenylbutyrate in spinal muscular atrophy.  
Mercuri E, Bertini E, Messina S, Solari A, D'Amico A, Angelozzi C, Battini R, Berardinelli A, Boffi P, Bruno C, Cini C, Colitto F, Kinali M, Minetti C, Mongini T, Morandi L, Neri G, Orcesi S, Pane M, Pelliccioni M, Pini A, Tiziano FD, Villanova M, Vita G, Brahe C.  
Neurology. 2007 Jan 2;68(1):51-5.
20. Reliability of the Hammersmith functional motor scale for spinal muscular atrophy in a multicentric study.  
Mercuri E, Messina S, Battini R, Berardinelli A, Boffi P, Bono R, Bruno C, Carboni N, Cini C, Colitto F, D'Amico A, Minetti C, Mirabella M, Mongini T, Morandi L, Dlamini N, Orcesi S, Pelliccioni M, Pane M, Pini A, Swan AV, Villanova M, Vita G, Main M, Muntoni F, Bertini E.  
Neuromuscul Disord. 2006 Feb;16(2):93-8. Epub

21. Clinical and genetic studies in hereditary spastic paraplegia with thin corpus callosum.  
Casali C, Valente EM, Bertini E, Montagna G, Criscuolo C, De Michele G, Villanova M, Damiano M, Pierallini A, Brancati F, Scarano V, Tessa A, Cricchi F, Grieco GS, Muglia M, Carella M, Martini B, Rossi A, Amabile GA, Nappi G, Filla A, Dallapiccola B, Santorelli FM.  
*Neurology*. 2004 Jan 27;62(2):262-8.
22. Phenylbutyrate increases SMN expression in vitro: relevance for treatment of spinal muscular atrophy.  
Andreassi C, Angelozzi C, Tiziano FD, Vitali T, De Vincenzi E, Boninsegna A, Villanova M, Bertini E, Pini A, Neri G, Brahe C.  
*Eur J Hum Genet*. 2004 Jan;12(1):59-65.
23. Neurological presentation of Ehlers-Danlos syndrome type IV in a family with parental mosaicism.  
Palmeri S, Mari F, Meloni I, Malandrini A, Ariani F, Villanova M, Pompilio A, Schwarze U, Byers PH, Renieri A.  
*Clin Genet*. 2003 Jun;63(6):510-5.
24. Nerve growth factor expression in human dystrophic muscles.  
Toti P, Villanova M, Vatti R, Schuerfeld K, Stumpo M, Barbagli L, Malandrini A, Costantini M.  
*Muscle Nerve*. 2003 Mar;27(3):370-3.
25. Asymptomatic cores and paracrystalline mitochondrial inclusions in CADASIL.  
Malandrini A, Albani F, Palmeri S, Fattapposta F, Gambelli S, Berti G, Bracco A, Tammaro A, Calzavara S, Villanova M, Ferrari M, Rossi A, Carrera P.  
*Neurology*. 2002 Aug 27;59(4):617-20.
26. Narrowing in on the causative defect of an intriguing X-linked myopathy with excessive autophagy.  
Minassian BA, Aiyar R, Alic S, Banwell B, Villanova M, Fardeau M, Mandell JW, Juel VC, Rafii M, Auranen M, Kalimo H.  
*Neurology*. 2002 Aug 27;59(4):596-601.
27. Mutations of the selenoprotein N gene, which is implicated in rigid spine muscular dystrophy, cause the classical phenotype of multiminicore disease: reassessing the nosology of early-onset myopathies.  
Ferreiro A, Quijano-Roy S, Pichereau C, Moghadaszadeh B, Goemans N, Bönnemann C, Jungbluth H, Straub V, Villanova M, Leroy JP, Romero NB, Martin JJ, Muntoni F, Voit T, Estournet B, Richard P, Fardeau M, Guicheney P.  
*Am J Hum Genet*. 2002 Oct;71(4):739-49. Epub 2002 Aug 21.
28. Electrophysiological findings in X-linked myopathy with excessive autophagy.  
Jääskeläinen SK, Juel VC, Udd B, Villanova M, Liguori R, Minassian BA, Falck B, Niemi P, Kalimo H.  
*Ann Neurol*. 2002 May;51(5):648-52.
29. Ultrastructural findings in the peripheral nerve in a family with the intermediate form of Charcot-Marie-Tooth disease.  
Malandrini A, Ceuterick C, Villanova M, Gambelli S, Berti G, Rossi A, Guazzi GC.  
*J Submicrosc Cytol Pathol*. 2001 Jan-Apr;33(1-2):59-63. PMID: 11686409

30. Localization of the gene for the intermediate form of Charcot-Marie-Tooth to chromosome 10q24.1-q25.1.

Verhoeven K, Villanova M, Rossi A, Malandrini A, De Jonghe P, Timmerman V.  
Am J Hum Genet. 2001 Oct;69(4):889-94. Epub 2001 Aug 30.

31. Neuropathological findings associated with retained lead shot pellets in a man surviving two months after a suicide attempt.

Malandrini A, Villanova M, Salvadori C, Gambelli S, Berti G,  
Di Paolo M. J Forensic Sci. 2001 May;46(3):717-21.

32. The T9176G mtDNA mutation severely affects ATP production and results in Leigh syndrome.

Carozzo R, Tessa A, Vázquez-Memije ME, Piemonte F, Patrono C, Malandrini A, Dionisi-Vici C,  
Vilarinho L, Villanova M, Schägger H, Federico A, Bertini E, Santorelli FM.  
Neurology. 2001 Mar 13;56(5):687-90.

33. X-linked vacuolated myopathy: TNF- $\alpha$  and IFN- $\gamma$  expression in muscle fibers with MHC class I on sarcolemma.

Rouger K, Louboutin JP, Villanova M, Cherel  
Y, Fardeau M. Am J Pathol. 2001 Feb;158(2):355-9.

34. Image analysis quantification of substance P immunoreactivity in the trapezius muscle of patients with

fibromyalgia and myofascial pain syndrome.

De Stefano R, Selvi E, Villanova M, Frati E, Manganelli S, Franceschini E, Biasi G,  
Marcolongo R. J Rheumatol. 2000 Dec;27(12):2906-10.

35. Unusual laminin  $\alpha$ 2 processing in myoblasts from a patient with a novel variant of congenital muscular dystrophy.

Lattanzi G, Muntoni F, Sabatelli P, Squarzone S, Maraldi NM, Cenni V, Villanova M, Columbaro M, Merlini  
L, Marmiroli S.

Biochem Biophys Res Commun. 2000 Nov

36. Neurological involvement in Werner's syndrome: clinical and biopsy study of a familial case.

Malandrini A, Dotti MT, Villanova M, Battisti C, Federico A.  
Eur Neurol. 2000;44(3):187-9. No

37. Congenital muscular dystrophy associated with calf hypertrophy, microcephaly and severe mental retardation in three Italian families: evidence for a novel CMD syndrome.

Villanova M, Mercuri E, Bertini E, Sabatelli P, Morandi L, Mora M, Sewry C, Brockington M, Brown SC,  
Ferreiro A, Maraldi NM, Toda T, Guicheney P, Merlini L, Muntoni F.

Neuromuscul Disord. 2000

Dec;10(8):541-7. PMID: 11053679

38. A novel SURF1 mutation results in Leigh syndrome with peripheral neuropathy caused by cytochrome c oxidase deficiency.

Santoro L, Carrozzo R, Malandrini A, Piemonte F, Patrono C, Villanova M, Tessa A, Palmeri S, Bertini E, Santorelli FM.

Neuromuscul Disord. 2000 Aug;10(6):450-3. PMID: 10899453

39. Autonomic nervous system and smooth muscle cell involvement in systemic sclerosis: ultra-structural study of 3 cases.

Malandrini A, Selvi E, Villanova M, Berti G, Sabadini L, Salvadori C, Gambelli S, De Stefano R, ernillo R, Marcolongo R, Guazzi G. J Rheumatol. 2000 May;27(5):1203-6.

40. X-linked vacuolar myopathies: two separate loci and refined genetic mapping. Auranen M, Villanova M, Muntoni F, Fardeau M, Scherer SW,

Kalino H, Minassian BA. Ann Neurol. 2000 May;47(5):666-9.

41. Type I sialidosis: a clinical, biochemical and neuroradiological study.

Palmeri S, Villanova M, Malandrini A, van Diggelen OP, Huijmans JG, Ceuterick C, Rufa A, DeFalco D, Ciacci G, Martin JJ, Guazzi G.

Eur Neurol. 2000;43(2):88-94.

42. Hepatitis C virus infection and myositis: a polymerase chain reaction study.

Villanova M, Caudai C, Sabatelli P, Toti P, Malandrini A, Luzi P, Maraldi NM, Valensin PE, Merlini L. Acta Neuropathol. 2000 Mar;99(3):271-6.

43. Chronic diarrhea associated with the A3243G mtDNA mutation.

Santorelli FM, Villanova M, Malandrini A, Grieco GS, Palmeri S, Merlini L, Casali C.

Neurology. 2000 Jan 11;54(1):266-7. No abstract available.

44. Inheritance of a 38-kb fragment in apparently sporadic facioscapulohumeral muscular dystrophy.

Vitelli F, Villanova M, Malandrini A, Bruttini M, Piccini M, Merlini L, Guazzi G, Renieri A.

Muscle Nerve. 1999 Oct;22(10):1437-41.

45. Detection of beta-A4 amyloid and its precursor protein in the muscle of a patient with juvenile neuronal ceroid lipofuscinosis (Spielmeyer-Vogt-Sjögren).

Villanova M, Ceuterick C, Dotti MT, Santorelli FM, Casali C, Malandrini A, De Stefano N, Lübke U, Martin JJ, Guazzi GC, Federico A.

Acta Neuropathol. 1999 Jul;98(1):78-84.

46. Decreased expression of laminin beta 1 in chromosome 21-linked

Bethlem myopathy. Merlini L, Villanova M, Sabatelli P, Malandrini A, Maraldi NM.

Neuromuscul Disord. 1999 Jul;9(5):326-9.

47. Nuclear changes in a case of X-linked Emery-Dreifuss muscular dystrophy.

Ognibene A, Sabatelli P, Petrini S, Squarzone S, Riccio M, Santi S, Villanova M, Palmeri S, Merlini L,

Maraldi NM. Muscle Nerve. 1999 Jul;22(7):864-9.



48. Mitochondrial G8363A mutation presenting as cerebellar ataxia and lipomas in an Italian family. Casali C, Fabrizi GM, Santorelli FM, Colazza G, Villanova M, Dotti MT, Cavallaro T, Cardaioli E, Battisti C, Manneschi L, DiGennaro GC, Fortini D, Spadaro M, Morocutti C, Federico A. *Neurology*. 1999 Mar 23;52(5):1103-4. No abstract available.
49. Acute inflammatory neuropathy in Charcot-Marie-Tooth disease. Malandrini A, Villanova M, Dotti MT, Federico A. *Neurology*. 1999 Mar 10;52(4):859-61.
50. Mitochondrial myopathy mimicking fibromyalgia syndrome. Villanova M, Selvi E, Malandrini A, Casali C, Santorelli FM, De Stefano R, Marcolongo R. *Muscle Nerve*. 1999 Feb;22(2):289-91. No abstract available.
51. Ultrastructural study of enteric ganglia in three patients with Rett syndrome. Malandrini A, Hayek G, Villanova M, Aucone AM, Berti G, Vernillo R, Zappella M, Guazzi GC. *Brain Dev*. 1998 Dec;20(8):586-8.
52. Immunofluorescence study of a muscle biopsy from a 1-year-old patient with Walker-Warburg syndrome. Villanova M, Sabatelli P, He Y, Malandrini A, Petrini S, Maraldi NM, Merlini L. *Acta Neuropathol*. 1998 Dec;96(6):651-4.
53. Altered expression of the alpha2 laminin chain in psoriatic skin: the effect of treatment with cyclosporin. Toti P, Pellegrino M, Villanova M, Flori ML, Miracco C, Bartolommei S, Andreassi L. *Br J Dermatol*. 1998 Sep;139(3):375-9.
54. Neuronal intranuclear inclusion disease: neuropathologic study of a case. Malandrini A, Villanova M, Tripodi S, Palmeri S, Sicurelli F, Parrotta E, Berti G, Salvadori C, Cintorino M, Guazzi GC. *Brain Dev*. 1998 Aug;20(5):290-4.
55. CAG repeat expansion in an Italian family with spinocerebellar ataxia type 2 (SCA2): a clinical and genetic study. Malandrini A, Galli L, Villanova M, Palmeri S, Parrotta E, DeFalco D, Cappelli M, Grieco GS, Renieri A, Guazzi G. *Eur Neurol*. 1998 Oct;40(3):164-8.
56. Charcot-Marie-Tooth disease: an intermediate form. Villanova M, Timmerman V, De Jonghe P, Malandrini A, Rizzuto N, Van Broeckhoven C, Guazzi G, Rossi A. *Neuromuscul Disord*. 1998 Aug;8(6):392-3. No abstract available. PMID: 97138
57. Giant axonal neuropathy with subclinical involvement of the central nervous system: case report. Malandrini A, Dotti MT, Battisti C, Villanova M, Capocchi G, Federico A. *J Neurol Sci*. 1998 Jun 30;158(2):232-5.
58. Expression of laminin 1 and 2 in brain tumor vessels. An immunohistochemical study. Toti P, Villanova M, De Felice C, Megha T, Bartolommei S, Tosi P. *J Submicrosc Cytol Pathol*. 1998 Apr;30(2):227-30. PMID: 9648286

59. Hereditary motor and sensory neuropathy Lom type in an Italian Gypsy family. Merlini L, Villanova M, Sabatelli P, Trogu A, Malandrini A, Yanakiev P, Maraldi NM, Kalaydjieva L. *Neuromuscul Disord*. 1998 May;8(3-4):182-5.
60. X-linked vacuolated myopathy: membrane attack complex deposition on the surface membrane of injured muscle fibers is not accompanied by S-protein. Louboutin JP, Navenot JM, Villanova M, Rouger K, Merlini L, Fardeau M. *Muscle Nerve*. 1998 Jul;21(7):932-5.
61. Increase of neuronal nitric oxide synthase in rat skeletal muscle during ageing. Capanni C, Squarzoni S, Petrini S, Villanova M, Muscari C, Maraldi NM, Guarnieri C, Caldarera CM. *Biochem Biophys Res Commun*. 1998 Apr 7;245(1):216-9.
62. Juvenile Leigh syndrome with protracted course presenting as chronic sensory motor neuropathy, ataxia, deafness and retinitis pigmentosa: a clinicopathological report. Malandrini A, Palmeri S, Fabrizi GM, Villanova M, Berti G, Salvadori C, Gardini G, Motti L, Solimé F, Guazzi GC. *J Neurol Sci*. 1998 Mar 5;155(2):218-21.
63. FAFL4, a new gene encoding long-chain acyl-CoA synthetase 4, is deleted in a family with Alport syndrome, elliptocytosis, and mental retardation. Piccini M, Vitelli F, Bruttini M, Pober BR, Jonsson JJ, Villanova M, Zollo M, Borsani G, Ballabio A, Renieri A. *Genomics*. 1998 Feb 1;47(3):350-8.
64. Localization of laminin alpha 2 chain in normal human central nervous system: an immunofluorescence and ultrastructural study. Villanova M, Malandrini A, Sabatelli P, Sewry CA, Toti P, Torelli S, Six J, Scarfó G, Palma L, Muntoni F, Squarzoni S, Tosi P, Maraldi NM, Guazzi GC. *Acta Neuropathol*. 1997 Dec;94(6):567-71.
65. Selective bilateral amyotrophy of the anterior tibial muscle: a case report. Villanova M, Malandrini A, Louboutin JP, Palmeri S, Ginanneschi F, Six J, Volterrani L, Guazzi G. *Muscle Nerve*. 1997 Oct;20(10):1335-6. No abstract available.
66. Detection of *Borrelia burgdorferi* DNA and complement membrane attack complex deposits in the sural nerve of a patient with chronic polyneuropathy and tertiary Lyme disease. Maimone D, Villanova M, Stanta G, Bonin S, Malandrini A, Guazzi GC, Annunziata P. *Muscle Nerve*. 1997 Aug;20(8):969-75.
67. Immunolocalization of several laminin chains in the normal human central and peripheral nervous system. Villanova M, Sewry C, Malandrini A, Toti P, Muntoni F, Merlini L, Torelli S, Tosi P, Maraldi NM, Guazzi GC. *J Submicrosc Cytol Pathol*. 1997 Jul;29(3):409-13.
68. Polymyositis associated with simvastatin. Giordano N, Senesi M, Mattii G, Battisti E, Villanova M, Gennari C. *Lancet*. 1997 May 31;349(9065):1600-1. No

69. Unusual clinical features and early brain MRI lesions in a family with cerebral autosomal dominant arteriopathy.

Malandrini A, Carrera P, Ciacci G, Gonnelli S, Villanova M, Palmeri S, Vismara L, Brancolini V, Signorini E, Ferrari M, Guazzi GC.

Neurology. 1997 May;48(5):1200-3.

70. A syndrome of autosomal recessive pontocerebellar hypoplasia with white matter abnormalities and protracted course in two brothers.

Malandrini A, Palmeri S, Villanova M, Parrotta E, Sicurelli F, Amato D, DeFalco D, Guazzi GC. Brain Dev. 1997 Apr;19(3):209-11.

71. Intracellular detection of laminin alpha 2 chain in skin by electron microscopy immunocytochemistry:

comparison between normal and laminin alpha 2 chain deficient subjects.

Squarzoni S, Villanova M, Sabatelli P, Malandrini A, Toti P, Pini A, Merlini L, Guazzi GC, Maraldi NM.

Neuromuscul Disord. 1997 Mar;7(2):91-8.

72. Localization of laminin chains in the human retina: possible implications for congenital muscular dystrophy associated with alpha 2-chain of laminin deficiency.

Toti P, De Felice C, Malandrini A, Megha T, Cardone C, Villanova M. Neuromuscul Disord. 1997 Jan;7(1):21-5.

73. Focal myositis: a polymerase chain reaction analysis for a viral etiology.

Toti P, Romano L, Villanova M, Zazzi M, Luzi P.

Hum Pathol. 1997 Jan;28(1):111-3.

74. X-linked vacuolated myopathy: membrane attack complex deposition on muscle fiber membranes with calcium accumulation on sarcolemma.

Louboutin JP, Villanova M, Lucas-Héron B, Fardeau M. Ann Neurol. 1997

Jan;41(1):117-20.

75. Expression of CD59, a regulator of the membrane attack complex of complement, on human skeletal muscle fibers

Navenot JM, Villanova M, Lucas-Héron B, Malandrini A, Blanchard D, Louboutin JP.

Muscle Nerve. 1997 Jan;20(1):92-6.

76. An unusual type of primary cerebral hemihypotrophy with signs of dysfunctional neuronal migration.

Malandrini A, Lo Russo F, Villanova M, Salvestroni R, Sicurelli F, Salvadori C, Paolozzi C, Guazzi GC.

Acta Neuropathol. 1996 Dec;92(6):631-4.

77. Acanthocytosis, retinitis pigmentosa, pallidal degeneration. Report of two cases without serum lipid abnormalities.

Malandrini A, Cesaretti S, Mulinari M, Palmeri S, Fabrizi GM, Villanova M, Parrotta E, Montagnani A, Montagnani M, Anichini M, Guazzi GC.

J Neurol Sci. 1996 Sep 1;140(1-2):129-31.

78. Elevated levels of complement components C5 and C9 and decreased antitrypsin activity in the serum of patients with X-linked vacuolated myopathy.

Louboutin JP, Villanova M, Ulrich G, De Clerck LS, Fardeau M, Sagniez M. *Muscle Nerve*. 1996 Sep;19(9):1144-7.

79. Clinicopathological and genetic studies of two further Italian families with cerebral autosomal dominant arteriopathy.

Malandrini A, Carrera P, Palmeri S, Cavallaro T, Fabrizi GM, Villanova M, Fattapposta M, Vismara L, Brancolini V, Tanganelli P, Calì A, Morocutti C, Zeviani M, Ferrari M, Guazzi GC. *Acta Neuropathol*. 1996 Aug;92(2):115-22.

80. Merosin positive congenital muscular dystrophy with severe involvement of the central nervous system.

De Stefano N, Dotti MT, Villanova M, Scarano G, Federico A. *Brain Dev*. 1996 Jul-Aug;18(4):323-6.

81. Autosomal recessive paraparesis with amyotrophy of hands and feet and white matter lesions.

Malandrini A, Scarpini C, Villanova M, Sicurelli F, Parrotta E, DeFalco D, Guazzi GC. *Acta Neurol Scand*. 1996 Jul;94(1):60-2.

82. Muscular dystrophy, mental retardation and cardiomyopathy not associated with dystrophin deficiency.

Villanova M, Malandrini A, Biancotti R, Löfgren A, Mongini T, Six J, Salvestroni R, Parrotta E, Van Broeckhoven C, Paolozzi C, Guazzi G. *Neuromuscul Disord*. 1996

83. Localization of merosin in the normal human brain: implications for congenital muscular dystrophy with merosin deficiency.

Villanova M, Malandrini A, Toti P, Salvestroni R, Six J, Martin JJ, Guazzi GC. *J Submicrosc Cytol Pathol*. 1996 Jan;28(1):1-4.

84. Palatal myoclonus and unusual MRI findings in a patient with membranous lipodystrophy.

Malandrini A, Scarpini C, Palmeri S, Villanova M, Parrotta E, Tripodi S, Giani S, DeFalco D, Guazzi GC. *Brain Dev*. 1996 Jan-Feb;18(1):59-63.

85. Neuronal intranuclear inclusion disease: polymerase chain reaction and ultrastructural study of rectal biopsy specimen in a new case.

Malandrini A, Fabrizi GM, Cavallaro T, Zazzi M, Parrotta E, Romano L, Berti G, Villanova M, Guazzi GC. *Acta Neuropathol*. 1996;91(2):215-8.

86. X-linked vacuolated myopathy: complement membrane attack complex on surface membrane of injured muscle fibers.

Villanova M, Louboutin JP, Chateau D, Eymard B, Sagniez M, Tomé FM, Fardeau M. *Ann Neurol*. 1995 May;37(5):637-45.

87. Apolipoprotein E expression at neuromuscular junctions in mouse, rat and human skeletal muscle. A kaaboune M, Villanova M, Festoff BW, Verdière-Sahuqué M, Hantai D. FEBS Lett. 1994 Sep 5;351(2):246-8.
88. Eosinophilia myalgia syndrome: absence of immunoglobulin reactivity suggests a cellular rather than humoral mechanism. Villanova M, De Clerck LS, Cras P, Guazzi GC, Martin JJ. Acta Neurol Belg. 1994;94(3):200-4.
90. Eosinophilia-myalgia syndrome: a clinicopathological study of four patients. Villanova M, De Clerck LS, Cras P, Ceuterick C, Van Marck E, Guazzi GC, Martin JJ. Clin Neuropathol. 1993 Jul-Aug;12(4):201-3.
91. Ultrastructural sperm abnormalities and cerebellar atrophy: does a correlation exist? Report of two cases without endocrine hypogonadism. Malandrini A, Villanova M, Piomboni P, Collodel G, Spadaro M, Giunti P, Salvadori C, Morocutti C, Guazzi GC. Submicrosc Cytol Pathol. 1993 Jul;25(3):371-5. PMID: 8402537
92. Rimmed vacuoles of inclusion body myositis and oculopharyngeal muscular dystrophy contain amyloid precursor protein and lysosomal markers. Villanova M, Kawai M, Lübke U, Oh SJ, Perry G, Six J, Ceuterick C, Martin JJ, Cras P. Brain Res. 1993 Feb 19;603(2):343-7.
93. Inflammatory pathogenesis of cortical polymicrogyria: an autopsy study. Toti P, De Felice C, Palmeri ML, Villanova M, Martín JJ, Buonocore G. Pediatr Res. 1998 Sep;44(3):291-6. PMID: 9727703 [PubMed - indexed or MEDLINE] Similar articles