Curriculum Vitae

Dr. Davide Tonduti, MD, PhD

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Lingue straniere
Francese ottimo, Inglese ottimo

Istruzione e Formazione

- Settembre 2006 conseguita Laurea in Medicina e Chirurgia a pieni voti con tesi dal titolo "Implicazioni diagnostiche e terapeutiche dello studio dei metaboliti dei neurotrasmettitori e delle pterine liquorali nella patologia neurologica progressiva ad esordio infantile."

- Luglio 2012 conseguito Diploma di Specializzazione in Neuropsichiatria Infantile a pieni voti con tesi dal titolo "Stroke perinatale: caratteristiche cliniche e neuroradiologiche di un campione selezionato e risultati preliminari del progetto SVE-la sui fattori genetici predisponenti"

- Settembre 2013 conseguito Diploma InterUniversitario (DIU) in Malattie Ereditarie del Metabolismo, Université Paris Descartes (Paris 5)

- Novembre 2017 conseguito a pieni voti dottorato di ricerca (PhD) in Scienze Biomediche presso L'Università degli Studi di Pavia in cotutela con la Scuola di Dottorato "C2iD in Genetica, Cellule, Immunologia, Infettivologia, Sviluppo" dell'Università Paris Diderot (Paris 8). Tesi dal titolo "Dal fenotipo al genotipo: classificazione e identificazione di nuovi geni nell'ambito delle leucodistrofie e delle leucoencefalopatie genetiche"

Esperienze professionali

Posizione	Organizzazione/dipartimento	Paese	Data di inizio-fine (mese/anno)
Neuropsichiatra Infantile (Dirigente Medico)	U.O.C Neurologia Pediatrica, Ospedale dei Bambini V. Buzzi	Milano (Italia)	06/2017-in corso
Neuropsichiatra Infantile (Collaboratore Coordinato Continuativo)	U.O. Neuropsichiatria Infantile, Fondazione I.R.C.C.S. Istituto Neurologico Carlo Besta	Milano (Italia)	03/2014-06/2017
Frequentante Neuropsichiatra Infantile	U.O. Neuropsichiatria Infantile, Fondazione I.R.C.C.S. Istituto Neurologico Carlo Besta	Milano (Italia)	01/2014-02/2014

Consulente Neuropsichiatra Infantile	U.O. Neuropsichiatria Infantile, Fondazione I.R.C.C.S. Istituto Neurologico Carlo Besta	Milano (Italia)	09/2013-12/2013
Ricercatore, Studente PhD	Centro di referenza per le leucodistrofie, Servizio di Neurologia e Metabolismo, INSERM U676 Hôpital Robert Debré, Paris.	Francia (Parigi)	08/2012-08/2013
Specializzando in neuropsichiatria infantile	Children's National Medical Center, Center for Genetic Medicine Research, White Matter Disorders Clinic	USA (Washington, DC)	05/2011-12/2011
Specializzando in neuropsichiatria infantile	Service de Médecine Néonatale de Port Royale – Paris	Francia (Parigi)	06/2009
Specializzando in neuropsichiatria infantile)	Struttura complessa di Neuropsichiatria Infantile I. Neurologico Nazionale IRCCS Fondazione"C. Mondino"	Italia (Pavia)	07/2007-7/2012
Studente universitario	U.O. Neuropsichiatria Infantile, Fondazione I.R.C.C.S. Istituto Neurologico Carlo Besta	Italia (Milano)	03/2005 – 06/2007
Studente universitario	Departmenti di Reumatologia; Endocrinologia; Cardiochirurgia. Hopital Pitié-Salpetrière	Francia (Parigi)	10/2003-07/2004

Publicazioni

1. Mirchi A, Pelletier F, Tran LT, Keller S, Braverman N, **Tonduti D**, Vanderver A, Pizzino A, Dilenge ME, Poulin C, Shevell M, Majnemer A, Sébire G, Srour M, Osterman B, Boucher RM, Vanasse M, Rossignol E, Mitchell J, Venkateswaran S, Pohl D, Kauffman M, Schiffmann R, Goizet C, Moutton S, Roncarolo F, Bernard G Health-Related Quality of Life for Patients With Genetically Determined Leukoencephalopathy. Pediatr Neurol. 2018 Apr 9

2. 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. *KARS-related diseases: progressive leukoencephalopathy with brainstem and spinal cord calcifications as new phenotype and a review of literature*. Orphanet J Rare Dis. 2018 Apr 4;13(1):45. doi: 10.1186/s13023-018-0788-4.

3. 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. *Compound heterozygous missense and deep intronic variants in NDUFAF6 unraveled by exome sequencing and mRNA analysis*.J Hum Genet. 2018 Mar 12. doi: 10.1038/s10038-018-0423-1

4. **Tonduti D**, Invernizzi F, Panteghini C, Pinelli L, Battaglia S, Fazzi E, Zorzi G, Moroni I, Garavaglia B, Chiapparini L, Nardocci N. *SLC19A3 related disorder: Treatment implication and clinical outcome of 2 new patients*. Eur J Paediatr Neurol. 2017 Dec 16. pii: S1090-3798(17)31698-7. doi: 10.1016/j.ejpn.2017.11.012

5. 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.

6. Adang LA, Sherbini O, Ball L, Bloom M, Darbari A, Amartino H, DiVito D, Eichler F, Escolar M, Evans SH, Fatemi A, Fraser J, Hollowell L, Jaffe N, Joseph C, Karpinski M, Keller S, Maddock R, Mancilla E, McClary B, Mertz J, Morgart K, Langan T, Leventer R, Parikh S, Pizzino A, Prange E, Renaud DL, Rizzo W, Shapiro J, Suhr D, Suhr T, **Tonduti D**, Waggoner J, Waldman A, Wolf NI, Zerem A, Bonkowsky JL, Bernard G, van Haren K, Vanderver A; Global Leukodystrophy Initiative (GLIA) Consortium. *Revised consensus statement on the preventive and symptomatic care of patients with leukodystrophies.* Mol Genet Metab. 2017 Sep;122(1-2):18-32. doi: 10.1016/j.ymgme.2017.08.006.

7. Invernizzi F, Panteghini C, Chiapparini L, Moroni I, Nardocci N, Garavaglia B, **Tonduti D.** *Thiamine-responsive disease due to mutation of tpk1: Importance of avoiding misdiagnosis.* Neurology. 2017 Aug 22;89(8):870-871. doi: 10.1212/WNL.0000000000004270.

8. Armangue T, Orsini JJ, Takanohashi A, Gavazzi F, Conant A, Ulrick N, Morrissey MA, Nahhas N, Helman G, Gordish-Dressman H, Orcesi S, **Tonduti D**, Stutterd C, van Haren K, Toro C, Iglesias AD, van der Knaap MS, Goldbach Mansky R, Moser AB, Jones RO, Vanderver A. *Neonatal detection of Aicardi Goutières Syndrome by increased C26:0 lysophosphatidylcholine and interferon signature on newborn screening blood spots.* Mol Genet Metab. 2017 Jul 20. pii: S1096-7192(17)30369-4. doi: 0.1016/j.ymgme.2017.07.006.

9. Heimer G, Kerätär JM, Riley LG, Balasubramaniam S, Eyal E, Pietikäinen LP, Hiltunen JK, Marek-Yagel D, Hamada J, Gregory A, Rogers, Penelope Hogarth, Martha Nance, Nechama Shalva, Alvit Veber, Michal Tzadok, Andreea Nissenkorn, **Davide Tonduti**, Florence Renaldo, University of Washington Center for Mendelian Genomics, Ichraf Kraoua, Celeste Panteghini, Lorella Valletta, Barbara Garavaglia, Mark J. Cowley, Velimir Gayevskiy, Tony Roscioli, Jonathon M Silberstein, Orly Elpeleg, Annick Raas-Rothschild, Valeria Tiranti, Yair Anikster, John Christodoulou, Alexander J. Kastaniotis, Bruria Ben-Zeev and Susan J. Hayflick, C. MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. The American Journal of Human Genetics. 2016 Nov 3.

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11. **Tonduti D**, Chiapparini L, Moroni I, Ardissone A, Zorzi G, Zibordi F, Raspante S, Panteghini C, Garavaglia B, Nardocci N. Neurological Disorders Associated with Striatal Lesions: Classification and Diagnostic Approach. Curr Neurol Neurosci Rep. 2016 Jun;16(6):54. doi: 10.1007/s11910-016-0656-3. Review.

12. 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. Elevated aspartate aminotransferase and lactate dehydrogenase levels are a constant finding in PLA2G6-associated neurodegeneration. Eur J Neurol. 2016 Apr;23(4):e24-5. doi: 10.1111/ene.12927.

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14. Sarret C, Lemaire JJ, **Tonduti D**, Sontheimer A, Coste J, Pereira B, Feschet F, Roche B, Boespflug-Tanguy O. Time-course of myelination and atrophy on cerebral imaging in 35 patients with PLP1-related disorders. Dev Med Child Neurol. 2016 Jan 19. doi: 10.1111/dmcn.13025. [Epub ahead of print]

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17. La Piana R, Uggetti C, Roncarolo F, Vanderver A, Olivieri I, **Tonduti D**, Helman G, Balottin U, Fazzi E, Crow YJ, Livingston J, Orcesi S Neuroradiologic patterns and novel imaging findings in Aicardi-Goutières syndrome Neurology. 2016 Jan 5;86(1):28-35. doi: 10.1212/WNL.0000000000002228. Epub 2015 Nov 18.

18. Kevelam SH, Taube JR, van Spaendonk RM, Bertini E, Sperle K, Tarnopolsky M, **Tonduti D**, Valente EM, Travaglini L, Sistermans EA, Bernard G, Catsman-Berrevoets CE, van Karnebeek CD, Østergaard JR, Friederich RL, Fawzi Elsaid M, Schieving JH, Tarailo-Graovac M, Orcesi S, Steenweg ME, van Berkel CG, Waisfisz Q, Abbink TE, van der Knaap MS, Hobson GM, Wolf NI. Altered PLP1 splicing causes hypomyelination of early myelinating structures. Ann Clin Transl Neurol. 2015 Jun;2(6):648-61. doi: 10.1002/acn3.203

19. **Tonduti D**, Dorboz I, Renaldo F, Masliah-Planchon J, Elmaleh-Bergès M, Dalens H, Rodriguez D, Boespflug-Tanguy O. Cystic leukoencephalopathy with cortical dysplasia related to LAMB1 mutations. Neurology. 2015 May 26;84(21):2195-7. doi: 10.1212/WNL.0000000000001607

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21. **Tonduti D**, Dorboz I, Imbard A, Slama A, Boutron A, Pichard S, Elmaleh M, Vallée L, Benoist J, Ogier H, Boespflug-Tanguy O. *New spastic paraplegia phenotype associated to mutation of NFU1*. Orphanet J Rare Dis. 2015 Feb 8;10(1):13.

22. Decio A, **Tonduti D**, Pichiecchio A, Vetro A, Ciccone R, Limongelli I, Giorda R, Caffi L, Balottin U, Zuffardi O, Orcesi S. A novel mutation in COL4A1 gene: A possible cause of early postnatal cerebrovascular events. Am J Med Genet A. 2015 Apr;167(4):810-5. doi: 10.1002/ajmg.a.36907.

23. Vanderver A, Prust M, **Tonduti D**, Mochel F, Hussey HM, Helman G, Garbern J, Eichler F, Labauge P, Aubourg P, Rodriguez D, Patterson MC, Van Hove JL, Schmidt J, Wolf NI, Boespflug-Tanguy O, Schiffmann R, van der Knaap MS; on behalf of the GLIA Consortium. *Case definition and classification of leukodystrophies and Zeukoencephalopathies*. Mol Genet Metab. 2015 Jan 29. pii: S1096-7192(15)00028-1. doi: 10.1016/j.ymgme.2015.01.006.

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Battini R, Beresford MW, Bernard G, Bianchi M, Billette de Villemeur T, Blair EM, Bloom M, Burlina AB, Luisa Carpanelli M, Carvalho DR, Castro-Gago M, Cavallini A, Cereda C, Chandler KE, Chitayat DA, Collins AE, Sierra Corcoles C, Cordeiro NJ, Crichiutti G, Dabydeen L, Dale RC, D Arrigo S, De Goede CG, De Laet C, De Waele LM, Denzler I, Desguerre I, Devriendt K, Di Rocco M, Fahey MC, Fazzi E, Ferrie CD, Figueiredo A, Gener B, Goizet C, Gowrinathan NR, Gowrishankar K, Hanrahan D, Isidor B, Kara B, Khan N, King MD, Kirk EP, Kumar R, Lagae L, Landrieu P, Lauffer H, Laugel V, Piana RL, Lim MJ, Lin JP, Linnankivi T, Mackay MT, Marom DR, Marques Lourenço C, McKee SA, Moroni I, Morton JE, Moutard ML, Murray K, Nabbout R, Nampoothiri S, Nunez-Enamorado N, Oades PJ, Olivieri I, Ostergaard JR, Pérez-Dueñas B, Prendiville JS, Ramesh V, Rasmussen M, Régal L, Ricci F, Rio M, Rodriguez D, Roubertie A, Salvatici E, Segers KA, Sinha GP, Soler D, Spiegel R, Stödberg TI, Straussberg R, Swoboda KJ, Suri M, Tacke U, Tan TY, Te Water Naude J, Wee Teik K, Mary Thomas M, Till M, **Tonduti D,** Maria Valente E, Noel Van Coster R, van der Knaap MS, Vassallo G, Vijzelaar R, Vogt J, Wallace GB, Wassmer E, Webb HJ, Whitehouse WP, Whitney RN, Zaki MS, Zuberi SM, Livingston JH, Rozenberg F, Lebon P, Vanderver A, Orcesi S, Rice GI. *Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1.* Am J Med Genet A. 2015 Feb;167(2):296-312. doi: 10.1002/ajmg.a.36887.

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26. Vanderver A, **Tonduti D**, Schiffmann R, Schmidt J, Van der Knaap MS. *Leukodystrophy Overview* In Pagon RA, Adam MP, Bird TD, Dolan CR, Fong CT, Smith RJH, Stephens K, editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 2014 Feb 06.

27. La Piana R, Uggetti C, Olivieri I, **Tonduti D**, Balottin U, Fazzi E, Orcesi S *Bilateral striatal necrosis in two subjects with Aicardi-Goutières syndrome due to mutations in ADAR1 (AGS6)*. Am J Med Genet A. 2013 Dec 20. doi: 10.1002/ajmg.a.36360

28. La Piana, Roberta, **Davide Tonduti**, Heather Gordish Dressman, Johanna L. Schmidt, Jonathan Murnick, Bernard Brais, Genevieve Bernard, and Adeline Vanderver. "Brain magnetic resonance imaging (MRI) pattern recognition in Pol III-related leukodystrophies." Journal of child neurology (2013): 0883073813503902.

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leukoencephalopathy hypomyelination with atrophy of the basal ganglia and cerebellum. Am J Hum Genet. 2013 May 2;92(5):767-73.

32. **Tonduti D**, Pichiecchio A, Wolf NI, Ariaudo G, van der Knaap MS, Bastianello S, Balottin U, Orcesi S. "*Novel Hypomyelinating Leukoencephalopathy Affecting Early Myelinating Structures: Clinical Course in Two Brothers*" Neuropediatrics. 2013 Jan 24. [Epub ahead of print]

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"*Characteristic Brain Magnetic Resonance Imaging Pattern in PTEN associated disorders*" Ann Neurol 2012 December 28, 72(S16):S205

35. **Tonduti D**, Pichiecchio A, La Piana R, Livingston JH, Doherty DA, Majumdar A, Tomkins S, Mine M, Ceroni M, Ricca I, Balottin U, Orcesi S. *COL4A1-Related Disease : Raised Creatine Kinase and Cerebral Calcification as Useful Pointers* Neuropediatrics. 2012 Aug 29

36. Adeline Vanderver, **Davide Tonduti**; Sarah Auerbach; Johanna L Schmidt; Sumit Parikh; Gordon C Gowans; Kelly E Jackson; Pamela L Brock; Marc Patterson; Michelle Nehrebecky; Rena Godfrey; Wadih M Zein; William Gahl; Camilo Toro "*Neurotransmitter abnormalities and response to supplementation in SPG11"* Mol Genet Metab. 2012 Sep;107(1-2):229-33

37. **Davide Tonduti**, Adeline Vanderver, Angela Berardinelli, Johanna L Schmidt, Christin D Collins, Francesca Novara, Antonia Di Genni, Alda Mita, Fabio Triulzi, Janice E Brunstrom-Hernandez, Orsetta Zuffardi, Umberto Balottin, Simona Orcesi *"MCT8 deficiency: extrapyramidal symptoms and delayed myelination as prominent features"* J Child Neurol, J Child Neurol. 2012 Jul 17. J Child Neurol 28(6): 795-800

38. **Davide Tonduti**, Adeline Vanderver, Ivana Olivieri, Johanna Loewenstein, Elisa Fazzi, Roberta La Piana, Simona Orcesi "*Neopterin and Tetrahydrobiopterin Cerebrospinal Fluid Elevations in Aicardi Goutieres Syndrome: Confirmation of Findings in Mutation Confirmed Subjects*" Neurology 2012 Aprile, 78(S1)

39. Orcesi S., **Tonduti D**, La Piana R *Calcifying leukoencephalopathies: new overlapping phenotypes* Am J Med Genet A. Am J Med Genet A. 2012 Mar 14.

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42. Tétreault M, Choquet K, Orcesi S, **Tonduti D**, Balottin U, Teichmann M, Fribourg S, Schiffmann R, Brais B, Vanderver A, Bernard G. *Recessive Mutations in POLR3B, Encoding the Second Largest Subunit of Pol III, Cause a Rare Hypomyelinating Leukodystrophy*. Am J Hum Genet. 2011 Oct 26.

43. 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, Veggiotti P, Comi GP. *Clinical and molecular features of an infant patient affected by Leigh Disease*

associated to m.14459G>A mitochondrial DNA mutation: a case report. BMC Neurol. 2011 Jul 12;11:85.

44. Orcesi S, La Piana R, Uggetti C, **Tonduti D**, Pichiecchio A, Pasin M, Viselner G, Comi G, Del Bo R, Ronchi D, Bastianello S, Balottin U. *Spinal cord calcification in an early-progressive leukoencephalopathy*. J Child Neurol. 2011 Jul;26(7):876-80. Epub 2011 Mar 22.

45. Brun L, Ngu LH, Keng WT, Ch'ng GS, Choy YS, Hwu WL, Lee WT, Willemsen MA, Verbeek MM, Wassenberg T, Régal L, Orcesi S, **Tonduti D**, Accorsi P, Testard H, Abdenur JE, Tay S, Allen GF, Heales S, Kern I, Kato M, Burlina A, Manegold C, Hoffmann GF, Blau N. *Clinical and biochemical features of aromatic L-amino acid decarboxylase deficiency*. Neurology. 2010 Jul 6;75(1):64-71. Epub 2010 May 26. Erratum in: Neurology. 2010 Aug 10;75(6):576.

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Milano, 12.06.2018

- Si autorizza il trattamento dei propri dati personali ai sensi del D. Lgs. n. 196 del 30/06/2003.
- Il presente CV ha funzione di autocertificazione ai sensi del D.P.R. n. 445 del 28/12/2000.