EUROPEAN CURRICULUM VITAE FORMAT

INFORMAZIONI PERSONALI

Nome	**LORENZO NANETTI**
Telefono	(+39) 0223942519
Fax	(+39) 0223942140
E-mail	**lorenzo.nanetti@istituto-besta.it**

ESPERIENZA LAVORATIVA

• Data

Dal 01 maggio 2020 a oggi

• Datore di lavoro

Fondazione IRCCS Istituto Neurologico "Carlo Besta" – Milano,

UO Genetica Medica e Neurogenetica

• Posizione lavorativa

Dirigente Medico, Neurologia

• attività e responsabilità principali

Attività clinica:

Presa in carico, diagnosi e cura di pazienti affetti da Malattia di Huntington, atassia spinocerebellare, paraparesi spastica e altre malattie neurologiche genetiche rare (circa 700 visite ambulatoriali/anno).

Attività di ricerca:

- "Principal Investigator" progetto Ricerca Finalizzata GR-2013-02357821: "An Italian Study of intermediate-lenght polyq-tract expansions: frequency, clinical variability and brain morphometry of subjects at risk for late-onset neurodegenerative diseases" (budget 285.550 euro)
- "Co-Principal Investigator" progetto Ricerca Finalizzata GR-2016-02363337: "Epileptogenic encephalopathies and complex generalized epilepsies of infancy: NGS analysis and functional characterization of novel causative variants for an efficient diagnosis and a personalized treatment" (budget 411.750 euro)

Partecipazione a sperimentazioni multicentriche osservazionali e farmacologiche per la malattia di Huntington e le atassie spinocerebellari in accordo con i criteri di "Good Cinical Practice ":

- dal 2019 a oggi Sperimentatore per la sperimentazione farmacologica in doppio cieco per la malattia di Huntington GENERATIONHD1, Roche
- dal 2018 a oggi Sperimentatore per la sperimentazione farmacologica randomizzata in doppio cieco per la malattia di Friedreich MOXIe, REATA
- 2015-2016, Sperimentatore per la sperimentazione farmacologica randomizzata in doppio cieco per la Malattia di Huntington LEGATO-HD, TEVA
- dal 2014 a oggi, Coordinatore Clinico e Sperimentatore per il protocollo di studio osservazionale per la Malattia di Huntington ENROLL-HD
- dal 2010 a oggi, Sperimentatore per il protocollo di studio osservazionale per la Malattia di Friedreich EFACTS
- 2011-2012, Coordinatore Clinico e Sperimentatore per la sperimentazione farmacologica randomizzata in doppio cieco per la Malattia di Huntington SEN0014196 (SIENA-BIOTECH)
- 2011-2012 Sperimentatore per la sperimentazione farmacologica randomizzata in doppio cieco per la Malattia di Friedreich CEPO (Lu AA24493 –*LUNDBECK*)
- 2008-2013 Sperimentatore per il protocollo di studio osservazionale per la Malattia di Huntington Registry-3
- 2008-2010 Sperimentatore per la sperimentazione farmacologica randomizzata in doppio cieco per la malattia di Charcot Marie Tooth tipo 1A CMT-TRIAAL

(TELETHON)

- 2008-2009 Sperimentatore per la sperimentazione farmacologica randomizzata in doppio cieco per la Malattia di Friedreich EPOFA (*AIFA*)
- 2008-2013 Sperimentatore per il protocollo di studio osservazionale per i pazienti e i soggetti a rischio di trasmissione per le atassie spinocerebellari (EUROSCA, RISCA)

ESPERIENZA LAVORATIVA

• Data	**Dal 01 maggio 2013 al 30/04/2020**
• Datore di lavoro	Fondazione IRCCS Istituto Neurologico "Carlo Besta" – Milano, UO Genetica Medica e Neurogenetica
• Posizione lavorativa	Medico specialista in Neurologia, contratto di Collaborazione Coordinata e Continuativa dal titolo "Fisiopatologia, storia naturale e trattamento delle malattie neurodegenerative da espansione di triplette" (40 ore/settimana)

ESPERIENZE FORMATIVE

Data	2008 -2013
Titolo di studio	Specializzazione in Neurologia (votazione 70/70)
	Università degli Studi di Milano – Specializzazione in Neurologia
Nome dell'Ateneo o dell'Istituto di formazione	Fondazione IRCCS Istituto Neurologico "Carlo Besta" – Milano
	SOSD Genetica delle Malattie Neurodegenerative e Metaboliche
Data	Settembre 2006
Titolo di studio	Diploma di "Clinical attachment"
Nome dell'Ateneo o dell'Istituto di formazione	National Hospital for Neurology Queen Square - London, UK
	Frequenza volontaria per "Clinical Attachment"
Data	2000 -2006
Titolo di studio	Laurea in Medicina (votazione 110/110)
Nome dell'Ateneo o dell'Istituto di formazione	Università degli Studi di Milano, corso di laurea in Medicina

ALTRE COMPETENZE

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LINGUA MADRE	**ITALIANO**
ALTRE LINGUE	
	INGLESE
• abilità nella lettura	BUONO
• abilità nella scrittura	BUONO
• abilità nel parlare	BUONO

PUBBLICAZIONI AUTORE DI 61 LAVORI SCIENTIFICI PUBBLICATI IN RIVISTE PEER-REVIEWED (13 IN QUALITÀ DI PRIMO/CORRESPONDING AUTHOR* E 48 COAUTORE). H-INDEX= 18 (03/03/2021)

PUBBLICAZIONI

1. Magri S, Nanetti*, et al. Missing the pathological expansion in Huntington disease: de novo c.51C>G variant on the expanded allele causing intrafamilial allele dropout.. Am J Med Genet A. 2021
2. Nigri A, Sarro L, Mongelli A, Pinardi C, Porcu L, Castaldo A, Ferraro S, Grisoli M, Bruzzone MG, Gellera C, Taroni F, Mariotti C, Nanetti L. Progression of Cerebellar Atrophy in Spinocerebellar Ataxia Type 2 Gene Carriers: A Longitudinal MRI Study in Preclinical and Early Disease Stages. Front Neurol. 2020

3. Mongelli A, Magri S, Salvatore E, Rizzo E, De Rosa A, Fico T, Gatti M, Gellera C, Taroni F, Mariotti C, Nanetti L. Frequency and distribution of polyQ disease intermediate-length repeat alleles in healthy Italian population. Neurol Sci. 2020
4. Nanetti L, et al. ANO10 mutational screening in recessive ataxia: genetic findings and refinement of the clinical phenotype. J Neurol. 2019
5. Nanetti.l et al. Cortical thickness, stance control, and arithmetic skill: An exploratory study in premanifest Huntington disease. Parkinsonism Relat Disord. 2018
6. Nanetti L, et al. Stance instability in preclinical SCA1 mutation carriers: A 4-year prospective posturography study. Gait Posture 2017
7. Ferraro S, Nanetti L*, et al. Frontal cortex BOLD signal changes in premanifest Huntington disease: A possible fMRI biomarker. Neurology 2014
8. Nanetti L, et al. SETX mutations are a frequent genetic cause of adult onset cerebellar ataxia with neuropathy and elevated serum alpha-fetoprotein. Orphanet J Rare Dis 2013
9. Nanetti L, et al. Novel and recurrent spastin mutations in a large series of SPG4 Italian families. Neurosci Lett 2012
10. Nanetti L, et al. PEX7 Mutations Cause Congenital Cataract Retinopathy and Late-Onset Ataxia and Cognitive Impairment: Report of Two Siblings and Review of the Literature. J Clin Neurol. 2015
11. Nanetti L, et al. Rare association of motor neuron disease and spinocerebellar ataxia type 2 (SCA2): a new case and review of the literature. J Neurol 2009
12. Nanetti L, et al. Slowly progressive sensory hemisyndrome: unusual presentation of paraneoplastic sensory neuronopathy. J Peripher Nerv Syst 2010
13. Vanotti A, Nanetti L*, et al. Somatosensory conduction pathway in spastic paraplegia type 5. J Clin Neurol 2014
14. Sarro L, Nanetti L, et al. Monitoring disease progression in spinocerebellar ataxias: implications for treatment and clinical research. Expert Rev Neurother. 2017
15. Politi LS, Bianchi Marzoli S, Godi C, Panzeri M, Ciasca P, Brugnara G, Castaldo A, Di Bella D, Taroni F, Nanetti L, Mariotti C. MRI Evidence of Cerebellar and Extraocular Muscle Atrophy Differently Contributing to Eye Movement Abnormalities in SCA2 and SCA28 Diseases. Invest Ophthalmol Vis Sci. 2016
16. Jacobi H, du Montcel ST, Romanzetti S, Harmuth F, Mariotti C, Nanetti L, et al. Conversion of individuals at risk for spinocerebellar ataxia types 1, 2, 3, and 6 to manifest ataxia (RISCA): a longitudinal cohort study. Lancet Neurol. 2020
17. Diallo A, Jacobi H, Cook A, Labrum R, Durr A, Brice A, Charles P, Marelli C, Mariotti C, Nanetti L, et al. Survival in patients with spinocerebellar ataxia types 1, 2, 3, and 6 (EUROSCA): a longitudinal cohort study.Lancet Neurol 2018
18. Jacobi H, du Montcel ST, Bauer P, Giunti P, Cook A, Labrum R, Parkinson MH, Durr A, Brice A, Charles P, Marelli C, Mariotti C, Nanetti L, et al. Long-term disease progression in spinocerebellar ataxia types 1, 2, 3, and 6: a longitudinal study. Lancet Neurol 2015
19. Reetz K, Dogan I, Costa AS, Dafotakis M, Fedosov K, Giunti P, Parkinson MH, Sweeney MG, Mariotti C, Panzeri M, Nanetti L, et al. Biological and clinical characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS) cohort: a cross-sectional analysis of baseline data. Lancet Neurol 2015
20. Jacobi H, Reetz K, du Montcel ST, Bauer P, Mariotti C, Nanetti L, et al. Biological and clinical characteristics of individuals at risk for spinocerebellar ataxia types 1, 2, 3, and 6 in the longitudinal RISCA study: analysis of baseline data. Lancet Neurol 2013
21. Synofzik M, Smets K, Mallaret M, Di Bella D, Gallenmüller C, Baets J, Schulze M, Magri S, Sarto E, Mustafa M, Deconinck T, Haack T, Züchner S, Gonzalez M, Timmann D, Stendel C, Klopstock T, Durr A, Tranchant C, Sturm M, Hamza W, Nanetti L, et al. SYNE1 ataxia is a common recessive ataxia with major non-cerebellar features: a large multi-centre study. Brain 2016
22. Pensato V, Castellotti B, Gellera C, Pareyson D, Ciano C, Nanetti L, et al. Overlapping phenotypes in complex spastic paraplegias SPG11, SPG15, SPG35 and SPG48. Brain 2014
23. Canafoglia L, Robbiano A, Pareyson D, Panzica F, Nanetti L, et al. Expanding sialidosis spectrum by genome-wide screening: NEU1 mutations in adult-onset myoclonus. Neurology 2014
24. Tezenas du Montcel S, Durr A, Rakowicz M, Nanetti L, et al. Prediction of the age at onset in spinocerebellar ataxia type 1, 2, 3 and 6. J Med Genet 2014
25. Gellera C, Ticozzi N, Nanetti L, et al. ATAXIN2 CAG-repeat length in Italian patients with amyotrophic lateral sclerosis: risk factor or variant phenotype? Implication for genetic testing and counseling. Neurobiol Aging 2012
26. Nigri A, Visani E, Bertolino N, Nanetti L, et al. Cerebellar Involvement in Patients with Mild to Moderate Myoclonus Due to EPM1: Structural and Functional MRI Findings in Comparison with Healthy Controls and Ataxic Patients. Brain Topogr. 2016
27. Diallo A, Jacobi H, Cook A, Giunti P, Parkinson MH, Labrum R, Durr A, Brice A, Charles P, Marelli C, Mariotti C, Nanetti L, et al. Prediction of Survival With Long-Term Disease Progression in Most Common Spinocerebellar Ataxia. Mov Disord. 2019
28. Mariotti C, Fancellu R, Caldarazzo S, Nanetti L, et al. Erythropoietin in Friedreich ataxia: no effect on frataxin in a randomized controlled trial. Mov Disord 2012
29. Boesch S, Nachbauer W, Mariotti C, Sacca F, Filla A, Klockgether T, Klopstock T, Schöls L, Jacobi H, Büchner B, Vom Hagen JM, Nanetti L, Manicom K. Safety and tolerability of carbamylated erythropoietin in Friedreich's ataxia. Mov Disord 2014
30. Criscuolo C, Guacci A, Carbone R, Lieto M, Salsano E, Nanetti L, et al. Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa and cataracts (PHARC) screening in an Italian population. Eur J Neurol 2013

31. Jacobi H, du Montcel ST, Bauer P, Giunti P, Cook A, Labrum R, Parkinson MH, Durr A, Brice A, Charles P, Marelli C,Mariotti C, Nanetti L, et al. Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. J Neurol 2018
32. Bassani R, Mariotti C, Nanetti L, et al. Pendular nystagmus in progressive ataxia and palatal tremor. J Neurol 2011
33. Gatti M, Magri S, Nanetti L, et al. From congenital microcephaly to adult onset cerebellar ataxia: Distinct and overlapping phenotypes in patients with PNKP gene mutations. Am J Med Genet A. 2019
34. Catania A, Legati A, Peverelli L, Nanetti L, et al. Homozygous variant in OTX2 and possible genetic modifiers identified in a patient with combined pituitary hormone deficiency, ocular involvement, myopathy, ataxia, and mitochondrial impairment. Am J Med Genet A. 2019
35. Piras IS, Picinelli C, Iennaco R, Baccarin M, Castronovo P, Tomaiuolo P, Cucinotta F, Ricciardello A, Turriziani L, Nanetti L, et al. Huntingtin gene CAG repeat size affects autism risk: Family-based and case-control association study. Am J Med Genet B Neuropsychiatr Genet. 2020
36. Schindler A, Pizzorni N, Sassone J, Nanetti L, et al. Fiberoptic endoscopic evaluation of swallowing in early-to-advanced stage Huntington's disease. Sci Rep. 2020
37. Pozzi E, Giorgio E, Mancini C, Lo Buono N, Augeri S, Ferrero M, Di Gregorio E, Riberi E, Vinciguerra M, Nanetti L, et al. In vitro dexamethasone treatment does not induce alternative ATM transcripts in cells from Ataxia Telangiectasia patients. Sci Rep. 2020
38. Visani E, Mariotti C, Nanetti L, et al. Cortical network dysfunction revealed by magnetoencephalography in carriers of spinocerebellar ataxia 1 or 2 mutation. Clin Neurophysiol. 2020
39. Visani E, Mariotti C, Nanetti L, et al. Different patterns of movement-related cortical oscillations in patients with myoclonus and in patients with spinocerebellar ataxia. Clin Neurophysiol 2019
40. Leoni V, Mariotti C, Nanetti L, et al. Whole body cholesterol metabolism is impaired in Huntington's disease. Neurosci Lett 2011
41. Mongelli A, Sarro L, Rizzo E, Nanetti L, et al. Multiple system atrophy and CAG repeat length: A genetic screening of polyglutamine disease genes in Italian patients. Neurosci Lett 2018
42. Salsano E, Fancellu R, Di Fede G, Ciano C, Scaioli V, Nanetti L, et al. Lower limb areflexia without central and peripheral conduction abnormalities is highly suggestive of Gerstmann-Sträussler-Scheinker disease Pro102Leu. J Neurol Sci 2011
43. Capiluppi E, Romano L, Rebora P, Nanetti L, et al. Late-onset Huntington's disease with 40-42 CAG expansion. Neurol Sci. 2020
44. Mariotti C, Ferruta A, Gellera C, Nespolo C, Fancellu R, Genitrini S, Di Bella D, Panzeri M, Nanetti L, et al. Predictive Genetic Tests in Neurodegenerative Disorders: A Methodological Approach Integrating Psychological Counseling for At-Risk Individuals and Referring Clinicians. Eur Neurol. 2010
45. Diallo A, Jacobi H, Schmitz-Hubsch T, Cook A, Durr A, Brice A, Charles P, Marelli C, Mariotti C, Nanetti L, et al. Body Mass Index Decline Is Related to Spinocerebellar Ataxia Disease Progression. Mov Disord Clin Pract 2017
46. Marchesi C, Ciano C, Salsano E, Nanetti L, et al. Co-occurrence of amyotrophic lateral sclerosis and Charcot-Marie-Tooth disease type 2A in a patient with a novel mutation in the mitofusin-2 gene. Neuromuscul Disord 2011
47. Rota S, Marchina E, Todeschini A, Nanetti L, et al. Very late-onset friedreich ataxia with laryngeal dystonia. Case Rep Neurol 2014
48. Minati L, Piacentini S, Ferrè F, Nanetti L, et al. Choice-option evaluation is preserved in early Huntington and Parkinson's disease. Neuroreport 2011
49. Moss DJH, Pardiñas AF, Langbehn D, Lo K, Leavitt BR, Roos R, Durr A, Mead S; TRACK-HD investigators. Identification of genetic variants associated with Huntington's disease progression: a genome-wide association study. Lancet Neurol. 2017
50. Reetz K, Dogan I, Hilgers RD, Giunti P, Mariotti C, Durr A, Klopstock T, de Rivera FJ, Schöls L, Klockgether T, Bürk K, Rai M, Pandolfo M, Schulz JB; EFACTS Study Group. Progression characteristics of the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS): a 2 year cohort study. Lancet Neurol 2015
51. Pareyson D, Reilly MM,; CMT-TRIAAL group. Ascorbic acid in Charcot-Marie-Tooth disease type 1A (CMT-TRIAAL and CMT-TRAUK): a double-blind randomised trial. Lancet Neurol 2011
52. Renaud M, Tranchant C, Martin JVT, Mochel F, Synofzik M, van de Warrenburg B, Pandolfo M, Koenig M, Kolb SA, Anheim M; RADIAL Working Group. A recessive ataxia diagnosis algorithm for the next generation sequencing era. Ann Neurol 2017
53. Tanguy Melac A, Mariotti C, Filipovic Pierucci A, Giunti P, Arpa J, Boesch S, Klopstock T, Müller Vom Hagen J, Klockgether T, Bürk K, Schulz JB, Reetz K, Pandolfo M, Durr A, Tezenas du Montcel S; EFACTS group. Friedreich and dominant ataxias: quantitative differences in cerebellar dysfunction measurements. J Neurol Neurosurg Psychiatry 2017
54. Nobbio L, Visigalli D, Radice D, Fiorina E, Solari A, Lauria G, Reilly MM, Santoro L, Schenone A, Pareyson D; CMT-TRIAAL Group. PMP22 messenger RNA levels in skin biopsies: testing the effectiveness of a Charcot-Marie-Tooth 1A biomarker. Brain 2014
55. Piscosquito G, Reilly MM, Padua L, Gemignani F, Visioli F, Laurà M, Solari A, Pareyson D; for the CMT-TRIAAL group. Is overwork weakness relevant in Charcot-Marie-Tooth disease? J Neurol Neurosurg Psychiatry 2014
56. Baake V, Reijntjes RHAM, Dumas EM, Thompson JC; REGISTRY Investigators of the European Huntington's Disease Network, Roos RAC. Cognitive decline in Huntington's disease expansion gene carriers. Cortex 2017
57. Vittori A, Breda C, Repici M, Orth M,; REGISTRY investigators of the EHDN. Copy-number variation of the neuronal glucose transporter gene SLC2A3 and age of onset in Huntington's disease. Hum Mol Genet 2014

58. Hubers AA, van Duijn E, Roos RA, Craufurd D, Rickards H, Bernhard Landwehrmeyer G, van der Mast RC, Giltay EJ; REGISTRY investigators of the EHDN. Suicidal ideation in a European Huntington's disease population. J Affect Disord 2013
59. Metzger S, Walter C, Riess O, Roos RA, Nielsen JE, Craufurd D; REGISTRY Investigators of the EHDN. The V471A polymorphism in autophagy-related gene ATG7 modifies age at onset specifically in Italian Huntington disease patients. PLoS One 2013
60. Quarrell OW, Handley O, O'Donovan K, European Huntington's Disease Network. Discrepancies in reporting the CAG repeat lengths for Huntington's disease. Eur J Hum Genet 2012
61. Orth M, Handley OJ, Schwenke C, European Huntington's Disease Network. Observing Huntington's disease: the European Huntington's Disease Network's REGISTRY. J Neurol Neurosurg Psychiatry 2011

Consapevole delle sanzioni penali nel caso di dichiarazioni non veritiere, di formazione o uso di atti falsi, richiamate dall'articolo 76 del DPR 445/2000 e della decadenza dai benefici eventualmente conseguenti al provvedimento emanato sulla base di dichiarazioni non veritiere, attesto che le dichiarazioni contenute nel presente documento sono sostitutive di certificazione ai sensi dell'articolo 46 del DPR 445/2000
Manifesto il mio consenso affinchè i dati forniti possano essere trattati nel rispetto del GDPR 679/2016 (regolamento europeo in materia di protezione dei dati personali) per gli adempimenti connessi alla presente procedura, nonché all'eventuale procedura di assunzione.

Milano, 03/03/2021

Lorenzo Nanetti

Fondazione I.R.C.C.S.
Istituto Neurologico " C. Besta"
Dr. LORENZO NANETTI
C.F.: NNTLNZ81B08F205J
Specialista in Neurologia