Nome E Cognome Matilde Valeria Ursini

FORMAZIONE

Gennaio 1980: Laurea in Scienze Biologiche presso l'Università degli Studi di Napoli con votazione "summa cum laude"

ATTIVITA' LAVORATIVE

1981- 1982: Borsista del CNR e poi Assistente Associato presso l'Universite' Claude Bernard, Lyon I, Centre de Genetique Moleculaire et Cellulaire, (CGMC)

1982-1988: Borsista del CNR, ex Legge 285, presso il Dipartimento di Patologia e Biologia Cellulare e Molecolare, Universita' degli Studi di Napoli

1985: Guest scientist presso National Institutes of Health (NIH), Bethesda, USA Aprile 1987-Giugno

1987: Guest scientist presso National Institutes of Health (NIH), Bethesda, MD, USA

1988: Ricercatore del Consiglio Nazionale delle Ricerche nell'Organico di Ruolo dell'Istituto Internazionale di Genetica e Biofisica (IIGB) del CNR di Napoli

2001- Primo-Ricercatore II Livello del CNR

2019- Dirigente di Ricerca I Livello del CNR

2014- Abilitazione scientifica nazionale come Professore di Prima Fascia nel settore 05/F1 Biologia Applicata

ALTRO

Finanziamenti

1993-1995 VI-VIII Progetto di Ricerche sull'AIDS. *Tat mediated regulation of GP6D*

2000-2002 VII Progetto Telethon *Human Nude/SCID phenotype* (co-PI n.E0934)

2007 Progetto finanziato da Banco Napoli-Fondazione*: Identification of gene variants associated with NEMO Incontinentia Pigmenti*.

2008-2011 Progetto Telethon: *Unraveling the molecular mechanisms of impaired NEMO function in Incontinentia Pigmenti (IP) pathogenesis* (GGP08125)

2009-2012 Progetto finanziato da France Incontinentia Pigmenti Association

2011-2012 Progetto finanziato da Fondazione Roma - Terzo Settore Sportello della Solidarietà (co-PI)

2013-2015 Progetto finanziato da France Incontinentia Pigmenti Association

2014-2019 Progetto finanziato da IPASSI Onlus (Incontinentia Pigmenti ASSociation Italy), *Biobanca genetica dell'Incontinentia Pigmenti*

2015. Progetto finanziato da IPIF (Incontinentia Pigmenti International Foundation) USA *Incontinentia Pigmenti Genetic Biobank*

2015 Progetto finanziato da Regione Campania Legge 5/2007 (Detection Of Mutation In NEMO, DOMINO)

2016-2019 Progetto finanziato da France Incontinentia Pigmenti Association

2017. Progetto finanziato da CNR-DSB Progetto Bandiera InterOmics *(*co-PI*, OMIC-IP)*

2015-2016 Progetto Operativo Regionale:Terapie Innovative di Malattie Infiammatorie croniche, metaboliche, Neoplastiche e Geriatriche (POR-TIMING) (co-PI)

2019 Research grant from the Incontinentia Pigmenti International Foundation, USA: *The built of the global Incontinentia Pigmenti genetic Biobank*

Società scientifiche

2004- a tutt'oggi Membro dell'American Society of Human Genetics (ASHG)

2015- a tutt'oggi Membro dell'European Society of Human Genetics (ESHG)

2020–2022 Membro dell' ASHG Online Programs & Professional Education (OPPE) Working Group

Brevetti

Brevetto n.0001423541 August 2, 2016 "*Diagnostic Kit and a method for the Incontinentia pigmenti genetic diagnosis*" Inventors: **MV Ursini** (CNR), M Paciolla, F Fusco (CNR), MB Lioi (University of Basilicata)

Sito IGB: http://www.igb.cnr.it/index.php?id=90&staff_detail=ursini

Sito Orphanet site

http://www.orpha.net/consor/cgibin/Directory_Professionals.php?lng=IT&data_id=12995&MISSING%20CONTENT=URSINI&search=Directory_Professionals_Simple&title=Dr-Matilde-Valeria-URSINI

Pubblicazioni

Libri

NF-κB-Related Genetic Diseases, 2016 SpringerBriefs in Biochemistry and Molecular Biology Authors**:** Courtois**,** G., Pescatore**,** A., Gautheron**,** J., Fusco**,** F., **Ursini**, **M.V**., Senegas**,** A.

UNIVERSITA' DEGLI STUDI DELLA BASILICATA
DIPARTIMENTO DI SCIENZE

Pubblicazioni

1. Fusco F, Pescatore A, Steffann J, Bonnefont JP, De Oliveira J, Lioi MB, **Ursini MV**. Clinical utility gene card: for incontinentia pigmenti. Eur J Hum Genet. 2019 Dec;27(12):1894-1900.
2. Fusco F, Valente V, Fergola D, Pescatore A, Lioi MB, **Ursini MV**. The Incontinentia Pigmenti Genetic Biobank: study design and cohort profile to facilitate research into a rare disease worldwide. Eur J Hum Genet. 2019 Oct;27(10):1509-1518.
3. Cammarata-Scalisi F, Fusco F, **Ursini MV**. Incontinentia Pigmenti. Actas Dermosifiliogr. 2019 May;110(4):273-278.
4. Romano R, Grasso F, Gallo V, Cirillo E, Prencipe R, Mamone G, Mollica C, **Ursini MV**, De Ville De Goyet J, Pignata C, Giardino G. A case of incontinentia pigmenti associated with congenital absence of portal vein system and nodular regenerative hyperplasia. Br J Dermatol. 2019 Mar;180(3):674-675.
5. Cuomo F, Coppola A, Botti C, Maione C, Forte A, Scisciola L, Liguori G, Caiafa I, **Ursini MV**, Galderisi U, Cipollaro M, Altucci L, Cobellis G. Pro-inflammatory cytokines activate hypoxia-inducible factor 3α via epigenetic changes in mesenchymal stromal/stem cells. Sci Rep. 2018 Apr 11;8(1):5842.
6. Scheuerle AE, **Ursini MV**. Incontinentia Pigmenti. 1999 Jun 8 [updated 2017 Dec 21]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2018. Available from http://www.ncbi.nlm.nih.gov/books/NBK1472/
7. Pizzamiglio MR, Piccardi L, Bianchini F, Canzano L, Palermo L, Fusco F, D'Antuono G, Gelmini C, Garavelli L, **Ursini MV**. Cognitive-behavioural phenotype in a group of girls from 1.2 to 12 years old with the Incontinentia Pigmenti syndrome: Recommendations for clinical management. Appl Neuropsychol Child. 2017 Oct-Dec;6(4):327-334.
8. Fusco F, Conte MI, Diociauti A, Bigoni S, Branda MF, Ferlini A, El Hachem M and **Ursini MV** Unusual Father-To-Daughter Transmission of Incontinentia Pigmenti due to Mosaicism in IP Males Pediatrics 2017 pii: e20162950.
9. Bal E, Laplantine E, Hamel Y, Dubosclard V, Boisson B, Pescatore A, Picard C, Hadj-Rabia S, Royer G, Steffann J, Bonnefont JP, **Ursini MV**, Vabres P, Munnich A, Casanova JL, Bodemer C, Weil R, Agou F, Smahi A. Lack of interaction between NEMO and SHARPIN impairs linear ubiquitination and NF-kB activation and leads to incontinentia pigmenti. J Allergy Clin Immunol. 2017 Feb 27. pii: S0091-6749(17)30321-4.
10. Müller K, Courtois G, **Ursini MV** and Schwaninger M New Insight Into the Pathogenesis of Cerebral Small-Vessel Diseases Stroke. 2017 Feb;48(2):520-527.Pescatore A, Esposito E, Draber P, Walczak H, **Ursini MV**. NEMO regulates a cell death switch in TNF signaling by inhibiting recruitment of RIPK3 to the cell death-inducing complex II. Cell Death Dis. 2016 Aug 25;7(8):e2346.
11. Pescatore A, Esposito E, Draber P, Walczak H, **Ursini MV**. NEMO regulates a cell death switch in TNF signaling by inhibiting recruitment of RIPK3 to the cell death-inducing complex II. Cell Death Dis. 2016 Aug 25;7(8):e2346.
12. Esposito E, Napolitano G, Pescatore A, Calculli G, Incoronato MR, Leonardi A, **Ursini MV**. COMMD7 as a novel NEMO interacting protein involved in the termination of NF-kB signaling. J Cell Physiol. 2016, 231(1):152-61
13. Giardino G, Somma D, Cirillo E, Ruggiero G, Terrazzano G, Rubino V, **Ursini MV**, Vairo D, Badolato R, Carsetti R, Leonardi A, Puel A, Pignata C. Novel STAT1 gain-of-function mutation and suppurative infections. Pediatr Allergy Immunol. 2016, 27(2):220-3.
14. Giardino G, Gallo V, Somma D, Farrow EG, Thiffault I, D'Assante R, Donofrio V, Paciolla M, **Ursini MV**, Leonardi A, Saunders CJ, Pignata C. Targeted next-generation sequencing revealed MYD88 deficiency in a child with chronic yersiniosis and granulomatous lymphadenitis. J Allergy Clin Immunol. 2016, 137(5):1591-1595
15. Giardino G, Cirillo E, Gallo V, Esposito T, Fusco F, Conte MI, Quinti I, **Ursini MV**, Carsetti R, Pignata C. B cells from nuclear factor kB essential modulator deficient patients fail to differentiate to antibody secreting cells in response to TLR9 ligand. Clin Immunol. 2015, 161(2):131
16. Fusco F Pescatore A Conte MI Mirabelli P Paciolla M Esposito E Lioi MB **Ursini MV** EDA-ID and IP, two faces of the same coin: how the same *IKBKG*/*NEMO* mutation affecting the NF-kB pathway can cause immunodeficiency and/or inflammation International Review in Immunology 2015;34(6):445-59
17. Paciolla M Pescatore A Conte MI, Esposito E, Incoronato M, Lioi MB, Fusco F, **Ursini MV** Rare Mendelian Primary Immunodeficiency diseases associated to impaired NF-kB signaling Genes and Immunity 2015, 16(4):239-46
18. Fusco F, Paciolla M, Conte MI, Pescatore A, Esposito E, Mirabelli P, Lioi MB, **Ursini MV**. Incontinentia pigmenti: report on data from 2000 to 2013. Orphanet J Rare Dis. 2014 Jun 24;9:93.

19. Pizzamiglio MR, Piccardi R, Bianchini F, Canzano L, Palermo L, Fusco F, D'Antuono G, Gelmini C, Garavelli L, **Ursini MV**.Incontinentia pigmenti: identification of learning disabilities as a fundamental hallmark of the disease. PLOSone 2014; 9:Article number e87771

20. Conte MI, Pescatore A, Paciolla M, Esposito E, Miano MG, Lioi MB, McAleer MA, Giardino G, Pignata C, Irvine AD, Scheuerle AE, Royer G, Hadj-Rabia S, Bodemer C, Bonnefont JP, Munnich A, Smahi A, Steffann J, Fusco F, **Ursini MV**. Insight into IKBKG/NEMO Locus: Report of New Mutations and Complex Genomic Rearrangements Leading to Incontinentia Pigmenti Disease. Hum Mutat. 2013 Nov 12. 35: 165-177

21. d'Alessio PA, Ostan R, Bisson JF, Schulzke JD, **Ursini MV**, Bene MC. Oral administration of d-limonene controls inflammation in rat colitis and displays anti-inflammatory properties as diet supplementation in humans. Life Sci. 2013 Jul 10;92(24-26):1151-6.

22. Cecchini S, Paciolla M, Biffali E, Borra M, **Ursini MV**, Lioi MB. Ontogenetic profile of innate immune related genes and their tissue-specific expression in brown trout, Salmo trutta (Linnaeus, 1758). Fish Shellfish Immunol. 2013Sep;35(3):988-92.

23. van de Vosse E, van Dissel JT, Palamaro L, Giardino G, Santamaria F, Romano R, Fusco A, Montella S, Salerno M, **Ursini MV**, Pignata C. The R156H variation in IL-12Rbeta1 is not a mutation. Ital J Pediatr. 2013 Feb 14;39:12.

24. Palamaro L, Giardino G, Santamaria F, Romano R, Fusco A, Montella S, Salerno M, **Ursini MV**, Pignata C. Interleukin 12 receptor deficiency in a child with recurrent bronchopneumonia and very high IgE levels. Ital J Pediatr. 2012 Sep 19;38:46.

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27. Fusco F, Pescatore A, Steffann J, Royer G, Bonnefont JP, **Ursini MV**. Clinical Utility Gene Card for: incontinentia pigmenti. Eur J Hum Genet. 2013 Jul;21(7):792.

28. **Ursini MV**, Conte MI, Pescatore A, Miano MG and Fusco F. *Molecular Genetics of Incontinentia Pigmenti.* In**: eLS.** John Wiley & Sons, Ltd: Chichester**. 2012**doi:10.1002/9780470015902.a0024332.

29. Fusco F, Paciolla M, Napolitano F, Pescatore A, D'Addario I, Bal E, Lioi MB, Smahi A, Miano MG, **Ursini MV**. Genomic architecture at the Incontinentia Pigmenti locus favours de novo pathological alleles through different mechanisms. Hum Mol Genet. 2012 Mar 15;21(6):1260-71.

30. Fusco F, Paciolla M, Chen E, Li X, Genesio R, Conti A, Jones J, Poeta L, Lioi MB, **Ursini MV**, Miano MG. Genetic and molecular analysis of a new unbalanced X;18 rearrangement: localization of the diminished ovarian reserve disease locus in the distal Xq POF1 region. Hum Reprod. 2011 Nov;26(11):3186-96.

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32. Paciolla M, Boni R, Fusco F, Pescatore A, Poeta L, **Ursini MV**, Lioi MB, Miano MG. Nuclear factor-kappa-B-inhibitor alpha (NFKBIA) is a developmental marker of NF-ŒJB/p65 activation during in vitro oocyte maturation and early embryogenesis. Hum Reprod. 2011 May;26(5):1191-201.

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34. Gautheron J#, Pescatore A#, Fusco F, Esposito E, Yamaoka S, Agou F, **Ursini MV**#, Courtois G#. Identification of a new NEMO/TRAF6 interface affected in incontinentia pigmenti pathology. Hum Mol Genet. 2010 Aug 15;19(16):3138-49. #joint first author. #joint last author

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36. Monfregola J, Napolitano G, D'Urso M, Lappalainen P, **Ursini MV**. Functional characterization of Wiskott-Aldrich syndrome protein and scar homolog (WASH), a bi-modular nucleation-promoting factor able to interact with biogenesis of lysosome-related organelle subunit 2 (BLOS2) and gamma-tubulin. J Biol Chem. 2010 May 28;285(22):16951-7.

37. Fusco F, Paciolla M, Pescatore A, Lioi MB, Ayuso C, Faravelli F, Gentile M, Zollino M, D'Urso M, Miano MG, **Ursini MV**. Microdeletion/duplication at the Xq28 IP locus causes a de novo IKBKG/NEMO/IKKgamma exon4_10 deletion in families with Incontinentia Pigmenti. Hum Mutat. 2009 Sep;30(9):1284-91.

38. Napolitano G, Mirra S, Monfregola J, Lavorgna A, Leonardi A, **Ursini MV**. NESCA: a new NEMO/IKKgamma and TRAF6 interacting protein. J Cell Physiol. 2009 Aug;220(2):410-7.

39. Fusco F, Pescatore A, Bal E, Ghoul A, Paciolla M, Lioi MB, D'Urso M, Rabia SH, Bodemer C, Bonnefont JP, Munnich A, Miano MG, Smahi A, **Ursini MV**. Alterations of the IKBKG locus and diseases: an update and a report of 13 novel mutations. Hum Mutat. 2008 May;29(5):595-604.

40. Sebban-Benin H#, Pescatore A#, Fusco F, Pascuale V, Gautheron J, Yamaoka S, Moncla A, **Ursini MV**#, Courtois G#. Identification of TRAF6-dependent NEMO polyubiquitination sites through analysis of a new NEMO mutation causing incontinentia pigmenti. Hum Mol Genet. 2007 Dec 1;16(23):2805-15. #joint first author. #joint last author

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42. Miano MG, Laperuta C, Chiurazzi P, D'Urso M, **Ursini MV**. Ovarian dysfunction and FMR1 alleles in a large Italian family with POF and FRAXA disorders: case report. BMC Med Genet. 2007 Apr 11;8:18.

43. Monfregola J, Napolitano G, Conte I, Cevenini A, Migliaccio C, D'Urso M, **Ursini MV**. Functional characterization of the TMLH gene: promoter analysis, in situ hybridization, identification and mapping of alternative splicing variants. Gene. 2007 Jun 15;395(1-2):86-97.

44. Fusco F, Fimiani G, Tadini G, Michele D, **Ursini MV**. Clinical diagnosis of incontinentia pigmenti in a cohort of male patients. J Am Acad Dermatol. 2007 Feb;56(2):264-7.

45. Falabella P, Varricchio P, Provost B, Espagne E, Ferrarese R, Grimaldi A, de Eguileor M, Fimiani G, **Ursini MV**, Malva C, Drezen JM, Pennacchio F. Characterization of the IkappaB-like gene family in polydnaviruses associated with wasps belonging to different Braconid subfamilies. J Gen Virol. 2007 Jan;88(Pt 1):92-104.

46. Fusco F, Mercadante V, Miano MG, **Ursini MV**. Multiple regulatory regions and tissue-specific transcription initiation mediate the expression of NEMO/IKKgamma gene. Gene. 2006 Nov 15;383:99-107.

47. Busiello R, Fimiani G, Miano MG, Arico M, Santoro A, **Ursini MV**, Pignata C. A91V perforin variation in healthy subjects and FHLH patients. Int J Immunogenet. 2006 Apr;33(2):123-5.

48. Fimiani G, Laperuta C, Falco G, Ventruto V, D'Urso M, **Ursini MV**, Miano MG. Heterozygosity mapping by quantitative fluorescent PCR reveals an interstitial deletion in Xq26.2-q28 associated with ovarian dysfunction. Hum Reprod. 2006 Feb;21(2):529-35.

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51. Fusco F, Bardaro T, Fimiani G, Mercadante V, Miano MG, Falco G, Israel A, Courtois G, D'Urso M, **Ursini MV**. Molecular analysis of the genetic defect in a large cohort of IP patients and identification of novel NEMO mutations interfering with NF-kappaB activation. Hum Mol Genet. 2004 Aug 15;13(16):1763-73.

52. Adriani M, Martinez-Mir A, Fusco F, Busiello R, Frank J, Telese S, Matrecano E, **Ursini MV**, Christiano AM, Pignata C. Ancestral founder mutation of the nude (FOXN1) gene in congenital severe combined immunodeficiency associated with alopecia in southern Italy population. Ann Hum Genet. 2004 May;68(Pt 3):265-8.

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56. Bardaro T, Falco G, Sparago A, Mercadante V, Gean Molins E, Tarantino E, **Ursini MV**, D'Urso M. Two cases of misinterpretation of molecular results in incontinentia pigmenti, and a PCR-based method to discriminate NEMO/IKKgamma gene deletion. Hum Mutat. 2003 Jan;21(1):8-11.

57. **Ursini MV**, Gaetaniello L, Ambrosio R, Matrecano E, Apicella AJ, Salerno MC, Pignata C. Atypical X-linked SCID phenotype associated with growth hormone hyporesponsiveness. Clin Exp Immunol. 2002 Sep;129(3):502-9.

58. Ambrosio R, Fimiani G, Monfregola J, Sanzari E, De Felice N, Salerno MC, Pignata C, D'Urso M, **Ursini MV**. The structure of human STAT5A and B genes reveals two regions of nearly identical sequence and an alternative tissue specific STAT5B promoter. Gene. 2002 Feb 20;285(1-2):311-8.

59. Salerno M, Balestrieri B, Matrecano E, Officioso A, Rosenfeld RG, Di Maio S, Fimiani G, **Ursini MV**, Pignata C. Abnormal GH receptor signaling in children with idiopathic short stature. J Clin Endocrinol Metab. 2001 Aug;86(8):3882-8.

60. Del Giudice E, Gaetaniello L, Matrecano E, Cosentini E, **Ursini MV**, Racioppi L, Arrigo G, Pignata C. Brain migration disorder and T-cell activation deficiency associated with abnormal signaling through TCR/CD3 complex and hyperactivity of Fyn tyrosine kinase. Neuropediatrics. 2000 Oct;31(5):265-8.

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