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"

Attività professionali

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

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

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

2016-2024 Coordinatore della Incontinentia Pigmenti Genetic Biobank (IPGB, http://www.igb.cnr.it/ipgb)

2017-2024 Coordinatore del Centro di risorse Biologiche dell'IGB (http://www.igb.cnr.it/centro-di-risorse-biologiche) consorziato con BBMRI-ERIC

2015 Esperto nominato del G-ERN (European Reference Network of Centres of Expertise for Genodermatosis) per l' Ectodermal Dysplasia & Incontinentia Pigmenti

2017-2024, Coordinatore del Centro di Risorse Biologiche dell' IGB associato al consorzio BBMRI-ERIC (Biobanking an BioMolecular resources Research Infrastructure European Research Infrastructure Consortium)

2019- Dirigente di Ricerca I Livello del CNR

2023, Associato senior all'IGB, Napoli

Elenco titoli

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: *Molecular characterization and pathophysiology of IP and associated neurologic abnormalities*

2011-2012 Progetto finanziato da Fondazione Roma - Terzo Settore Sportello della Solidarietà: Neuropsychological characteristics of the syndrome IP and correlation with genetic data (co-PI)

2013-2015 Progetto finanziato da France Incontinentia Pigmenti Association: *Molecular characterization and pathophysiology of Incontinetia Pigmenti*

2014-2020 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: *Molecular characterization and pathophysiology of Incontinetia Pigmenti*

2017. Progetto finanziato da CNR-DSB Progetto Bandiera InterOmics: *TranscriptOMIC strategy to identify the altered signal pathways in patients with severe forms of Incontinentia pigmenti* (*OMIC-IP)* (co-PI)

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*

2019-2022 Progetto finanziato dal MISE (Prog n. F/050011/01/X32): *Genomica Funzionale di malattie genetiche rare: Realizzazione di strumenti innovativi ad alto potere diagnostico* (co-PI) (1/08/2019-31/07/2022)

Altri incarichi professionali

2019-2020-2021 Coordinatore del Rare Disease Working Group (WG RD) del BBMRI-ERIC (Biobanking an BioMolecular resources Research Infrastructure European Research Infrastructure Consortium), https://www.bbmri.it/nodo-nazionale/gruppi-di-lavoro/biobanche-e-malattie-rare/

https://www.ashg.org/about/committees/digital-learning-committee/

2022 Rappresentante CNR nel Comitato di Controllo e nel Comitato Scientifico del Progetto: The Italian Network of Biobanks and Biomolecular Resourches (BBMRI-it) (FOE 2020) Decreto nomina della Presidente CNR del 9-6-22

2023-2026 Membro del Comitato di Controllo per il progetto PNRR Infrastrutture di Ricerca: BBMRI.it - Strengthening of the Biobanking and Biomolecular Resources Research Infrastructure of Italy (in qualità di Referente Scientifico) Decreto nomina del DG del 24-11-2023

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

2023 a tutt'oggi Membro dell'ASHG Professional Practice & Social Implications Committee

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)

Attività didattica a livello universitario

2020-2024 Professore a contratto di Genetica Umana e di Popolazioni (8 CFU, 64 ore/anno, SSD: Bio/18), Università degli Studi della Basilicata, Potenza

Attività di revisore esterno

1995-1999 Comitato di revisione (Peer Review): Programma Nazionale AIDS

1999 Comitato di revisione dell'Institut Universitaire de France (IUF)

2009 Comitato di revisione dell'ANR (Agence National pour la Recherche): progetto JCJC SVSE 2

2011 Comitato di revisione ANR Reviewer Committee: progetto CESA

2011-2013 Comitato di revisione ANR Reviewer Committee: progetto Labex

2016 Comitato di valutazione per la promozione alla qualifica di Professore (Qualification Promotion to Professor for faculty members) per la Case Western Reserve University School of Medicine

2016, Revisore esperto per la Agencia per la Qualitat del sistema Universitari de Catalunya

2020 Revisore esperto per la Agency for Health Quality and Assessment of Catalonia (AQuAS), Fundaciò la Maratò de TV3 call for rare disease

2024, Expert of the European Commission for the EIC Pathfinder programme, Horizon call for research proposal.

WWWW sites

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

CRB site: (http://www.igb.cnr.it/centro-di-risorse-biologiche)

Sito Orphanet

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

Brevetti

Brevetto n.0001423541, rilasciato il 2 agosto 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)

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.

Pubblicazioni

1. Scheuerle AE, Ursini MV. Incontinentia Pigmenti. 1999 Jun 8 [updated 2025 Apr 3]. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2025.

2. Rosain J, Le Voyer T, Liu X, Gervais A, Polivka L, Cederholm A, Berteloot L, Parent AV, Pescatore A, Spinosa E, Minic S, Kiszewski AE, Tsumura M, Thibault C, Esnaola Azcoiti M, Martinovic J, Philippot Q, Khan T, Marchal A, Charmeteau-De Muylder B, Bizien L, Deswarte C, Hadjem L, Fauvarque MO, Dorgham K, Eriksson D, Falcone EL, Puel M, Ünal S, Geraldo A, Le Floc'h C, Li H, Rheault S, Muti C, Bobrie-Moyrand C, Welfringer-Morin A, Fuleihan RL, Lévy R, Roelens M, Gao L,Materna M, Pellegrini S, Piemonti L, Catherinot E, Goffard JC, Fekkar A, Sacko-Sow A, Soudée C, Boucherit S, Neehus AL, Has C, Hübner S, Blanchard-Rohner G, Amador-Borrero B, Utsumi T, Taniguchi M, Tani H, Izawa K, Yasumi T, Kanai S, Migaud M, Aubart M, Lambert N, Gorochov G, Picard C, Soudais C, L'Honneur AS, Rozenberg F, Milner JD, Zhang SY, Vabres P, Trpinac D, Marr N, Boddaert N, Desguerre I, Pasparakis M, Miller CN, Poziomczyk CS, Abel L, Okada S, Jouanguy E, Cheynier R, Zhang Q, Cobat A, Béziat V, Boisson B, Steffann J, Fusco F, Ursini MV, Hadj-Rabia S, Bodemer C, Bustamante J, Luche H, Puel A, Courtois G, Bastard P, Landegren N, Anderson MS, Casanova JL. Incontinentia pigmenti underlies thymic dysplasia, autoantibodies to type I IFNs, and viral diseases. J Exp Med. 2024 Nov 4;221(11):e20231152.

3. Marchal A, Cirulli ET, Neveux I, Bellos E, Thwaites RS, Schiabor Barrett KM, Zhang Y, Nemes-Bokun I, Kalinova M, Catchpole A, Tangye SG, Spaan AN, Lack JB, Ghosn J, Burdet C, Gorochov G, Tubach F, Hausfater P; COVID Human Genetic Effort; COVIDeF Study Group; French COVID Cohort Study Group; CoV-Contact Cohort; COVID-STORM Clinicians; COVID Clinicians; Orchestra Working Group; Amsterdam UMC COVID-19 Biobank; NIAID-USUHS COVID Study Group; Dalgard CL, Zhang SY, Zhang Q, Chiu C, Fellay J, Grzymski JJ, Sancho-Shimizu V, Abel L, Casanova, JL, Cobat A, Bolze A. Lack of association between classical HLA genes and asymptomatic SARS-CoV-2 infection. HGG Adv. 2024 Jul 18;5(3):100300.

4. Matuozzo D, Talouarn E, Marchal A, Zhang P, Manry J, Seeleuthner Y, Zhang Y, Bolze A, Chaldebas M, Milisavljevic B, Gervais A, Bastard P, Asano T, Bizien L, Barzaghi F, Abolhassani H, Tayoun AA, Aiuti A, Darazam IA, Allende LM, Alonso-Arias R, Arias AA, Aytekin G, Bergman P, Bondesan S, Bryceson YT, Bustos IG, Cabrera-Marante O, Carcel S, Carrera P, Casari G, Chaïbi K, Colobran R, Condino-Neto A, Covill LE, Delmonte OM, Zein LE, Flores C, Gregersen PK, Gut M, Haerynck F, Halwani R, Hancerli S, Hammarström L, Hatipoğlu N, Karbuz A, Keles S, Kyheng C, Leon-Lopez R, Franco JL, Mansouri D, Martinez-Picado J, Akcan OM, Migeotte I, Morange PE, Morelle G, Martin-Nalda A, Novelli G, Novelli A, Ozcelik T, Palabiyik F, Pan-Hammarström Q, de Diego RP, Planas-Serra L, Pleguezuelo DE, Prando C, Pujol A, Reyes LF, Rivière JG, Rodriguez-Gallego C, Rojas J, Rovere- Querini P, Schlüter A, Shahrooei M, Sobh A, Soler-Palacin P, Tandjaoui-Lambiotte Y, Tipu I, Tresoldi C, Troya J, van de Beek D, Zatz M, Zawadzki P, Al-Muhsen SZ, Alosaimi MF, Alsohime FM, Baris-Feldman H, Butte MJ, Constantinescu SN, Cooper MA, Dalgard CL, Fellay J, Heath JR, Lau YL, Lifton RP, Maniatis T, Mogensen TH, von Bernuth H, Lermine A, Vidaud M, Boland A, Deleuze JF, Nussbaum R, Kahn-Kirby A, Mentre F, Tubiana S, Gorochov G, Tubach F, Hausfater P; COVID Human Genetic Effort; COVIDeF Study Group; French COVID Cohort Study Group; CoV-Contact Cohort; COVID Clinicians; Orchestra Working Group; Amsterdam UMC Covid-19 Biobank; NIAID-USUHS COVID Study Group; Meyts I, Zhang SY, Puel A, Notarangelo LD, Boisson-Dupuis S, Su HC, Boisson B, Jouanguy E, Casanova JL, Zhang Q, Abel L, Cobat A. Correction: Rare predicted loss-of-function variants of type I IFN immunity genes are associated with life-threatening COVID-19. Genome Med. 2024 Jan 6;16(1):6.

5. Le Voyer T, Parent AV, Liu X, Cederholm A, Gervais A, Rosain J, Nguyen T, Perez Lorenzo M, Rackaityte E, Rinchai D, Zhang P, Bizien L, Hancioglu G, Ghillani-Dalbin P, Charuel JL, Philippot Q, Gueye MS, Maglorius Renkilaraj MRL, Ogishi M, Soudée C, Migaud M, Rozenberg F, Momenilandi M, Riller

Q, Imberti L, Delmonte OM, Müller G, Keller B, Orrego J, Franco Gallego WA, Rubin T, Emiroglu M, Parvaneh N, Eriksson D, Aranda-Guillen M, Berrios DI, Vong L, Katelaris CH, Mustillo P, Raedler J, Bohlen J, Bengi Celik J, Astudillo C, Winter S; NF-κB Consortium; COVID Human Genetic Effort; McLean C, Guffroy A, DeRisi JL, Yu D, Miller C, Feng Y, Guichard A, Béziat V, Bustamante J, Pan-Hammarström Q, Zhang Y, Rosen LB, Holland SM, Bosticardo M, Kenney H, Castagnoli R, Slade CA, Boztuğ K, Mahlaoui N, Latour S, Abraham RS, Lougaris V, Hauck F, Sediva A, Atschekzei F, Sogkas G, Poli MC, Slatter MA, Palterer B, Keller MD, Pinzon-Charry A, Sullivan A, Droney L, Suan D, Wong M, Kane A, Hu H, Ma C, Grombiříková H, Ciznar P, Dalal I, Aladjidi N, Hie M, Lazaro E, Franco J, Keles S, Malphettes M, Pasquet M, Maccari ME, Meinhardt A, Ikinciogullari A, Shahrooei M, Celmeli F, Frosk P, Goodnow CC, Gray PE, Belot A, Kuehn HS, Rosenzweig SD, Miyara M, Licciardi F, Servettaz A, Barlogis V, Le Guenno G, Herrmann VM, Kuijpers T, Ducoux G, Sarrot-Reynauld F, Schuetz C, Cunningham-Rundles C, Rieux-Laucat F, Tangye SG, Sobacchi C, Doffinger R, Warnatz K, Grimbacher B, Fieschi C, Berteloot L, Bryant VL, Trouillet Assant S, Su H, Neven B, Abel L, Zhang Q, Boisson B, Cobat A, Jouanguy E, Kampe O, Bastard P, Roifman CM, Landegren N, Notarangelo LD, Anderson MS,Casanova JL, Puel A. Autoantibodies against type I IFNs in humans with alternative NF-κB pathway deficiency. Nature. 2023 Nov;623(7988):803- 813.

6. Matuozzo D, Talouarn E, Marchal A, Zhang P, Manry J, Seeleuthner Y, Zhang Y,Bolze A, Chaldebas M, Milisavljevic B, Gervais A, Bastard P, Asano T, Bizien L,Barzaghi F, Abolhassani H, Abou Tayoun A, Aiuti A, Alavi Darazam I, Allende LM, Alonso-Arias R, Arias AA, Aytekin G, Bergman P, Bondesan S, Bryceson YT, Bustos IG, Cabrera-Marante O, Carcel S, Carrera P, Casari G, Chaïbi K, Colobran R,Condino-Neto A, Covill LE, Delmonte OM, El Zein L, Flores C, Gregersen PK, Gut M, Haerynck F, Halwani R, Hancerli S, Hammarström L, Hatipoğlu N, Karbuz A, Keles S, Kyheng C, Leon-Lopez R, Franco JL, Mansouri D, Martinez-Picado J, Metin Akcan O, Migeotte I, Morange PE, Morelle G, Martin-Nalda A, Novelli G, Novelli A, Ozcelik T, Palabiyik F, Pan-Hammarström Q, de Diego RP, Planas-Serra L, Pleguezuelo DE, Prando C, Pujol A, Reyes LF, Rivière JG, Rodriguez-Gallego C, Rojas J, Rovere-Querini P, Schlüter A, Shahrooei M, Sobh A, Soler-Palacin P, Tandjaoui-Lambiotte Y, Tipu I, Tresoldi C, Troya J, van de Beek D, Zatz M Zawadzki P, Al-Muhsen SZ, Alosaimi MF, Alsohime FM, Baris-Feldman H, Butte MJ,Constantinescu SN, Cooper MA, Dalgard CL, Fellay J, Heath JR, Lau YL, Lifton RP, Maniatis T, Mogensen TH, von Bernuth H, Lermine A, Vidaud M, Boland A, Deleuze JF, Nussbaum R, Kahn-Kirby A, Mentre F, Tubiana S, Gorochov G, Tubach F, Hausfater P; COVID Human Genetic Effort; COVIDeF Study Group; French COVID Cohort Study Group; CoV-Contact Cohort; COVID-STORM Clinicians; COVID Clinicians; Orchestra Working Group; Amsterdam UMC Covid-19 Biobank; NIAID-USUHS COVID Study Group; Meyts I, Zhang SY, Puel A, Notarangelo LD, Boisson-Dupuis S, Su HC, Boisson B, Jouanguy E, Casanova JL, Zhang Q, Abel L, Cobat A. Rare predicted loss-of-function variants of type I IFN immunity genes are associated with life-threatening COVID-19. Genome Med. 2023 Apr 5;15(1):22.

7. Johnson JL, Meneses-Salas E, Ramadass M, Monfregola J, Rahman F, Carvalho Gontijo R, Kiosses WB, Pestonjamasp K, Allen D, Zhang J, Osborne DG, Zhu YP,Wineinger N, Askari K, Chen D, Yu J, Henderson SC, Hedrick CC, Ursini MV,Grinstein S, Billadeau DD, Catz SD. Differential dysregulation of granule subsets in WASH-deficient neutrophil leukocytes resulting in inflammation. NatCommun. 2022 Sep 21;13(1):5529.

8. Pescatore A, Spinosa E, Casale C, Lioi MB, Ursini MV and Fusco F Human genetic diseases linked to the absence of NEMO: an obligatory Somatic Mosaic Disorder in male Int. J. Mol. Sci. 2022 Feb.1, 23 (3):1179 doi.org/10.3390/ijms23031179

9. Zhang Q, Bastard P, Liu Z, Le Pen J, Moncada-Velez M, Chen J, Ogishi M, Sabli IKD, Hodeib S, Korol C, Rosain J, Bilguvar K, Ye J, Bolze A, Bigio B, Yang R, Arias AA, Zhou Q, Zhang Y, Onodi F, Korniotis S, Karpf L, Philippot Q, Chbihi M, Bonnet-Madin L, Dorgham K, Smith N, Schneider WM, Razooky BS, Hoffmann HH, Michailidis E, Moens L, Han JE, Lorenzo L, Bizien L, Meade P, Neehus AL, Ugurbil AC, Corneau A, Kerner G, Zhang P, Rapaport F, Seeleuthner Y, Manry J, Masson C, Schmitt Y, Schlüter A, Le Voyer T, Khan T, Li J, Fellay J, Roussel L, Shahrooei M, Alosaimi MF, Mansouri D, Al-Saud H, Al-Mulla F, Almourfi F, Al-Muhsen SZ, Alsohime F, Al Turki S, Hasanato R, van de Beek D, Biondi A, Bettini LR, D'Angio' M, Bonfanti P, Imberti L, Sottini A, Paghera S, Quiros-Roldan E, Rossi C, Oler AJ, Tompkins MF, Alba C, Vandernoot I, Goffard JC, Smits G, Migeotte I, Haerynck F, Soler-Palacin P, Martin-Nalda A, Colobran R, Morange PE, Keles S, Çölkesen F, Ozcelik T, Yasar KK, Senoglu S, Karabela ŞN, Rodríguez-Gallego C, Novelli G, Hraiech S, Tandjaoui-Lambiotte Y, Duval X, Laouénan C; COVID-STOR Clinicians; COVID Clinicians; Imagine COVID Group; French COVID Cohort Study Group; CoV-Contact Cohort; Amsterdam UMC Covid-19 Biobank; COVID Human Genetic Effort; NIAID-USUHS/TAGC COVID Immunity Group, Snow AL, Dalgard CL, Milner JD, Vinh DC, Mogensen TH, Marr N, Spaan AN, Boisson B, Boisson-Dupuis S, Bustamante J, Puel A, Ciancanelli MJ, Meyts I, Maniatis T, Soumelis V, Amara A, Nussenzweig M, García-Sastre A, Krammer F, Pujol A, Duffy D, Lifton RP, Zhang SY, Gorochov G, Béziat V, Jouanguy E, Sancho-Shimizu V, Rice CM, Abel L, Notarangelo LD, Cobat A, Su HC, Casanova JL. Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science. 2020 Oct 23;370(6515):eabd4570. doi: 10.1126/science.abd4570

10. Bastard P, Rosen LB, Zhang Q, Michailidis E, Hoffmann HH, Zhang Y, Dorgham K, Philippot Q, Rosain J, Béziat V, Manry J, Shaw E, Haljasmägi L, Peterson P, Lorenzo L, Bizien L, Trouillet-Assant S, Dobbs K, de Jesus AA, Belot A, Kallaste A, Catherinot E, Tandjaoui-Lambiotte Y, Le Pen J, Kerner G, Bigio B, Seeleuthner Y, Yang R, Bolze A, Spaan AN, Delmonte OM, Abers MS, Aiuti A, Casari G, Lampasona V, Piemonti L, Ciceri F, Bilguvar K, Lifton RP, Vasse M, Smadja DM, Migaud M, Hadjadj J, Terrier B, Duffy D, Quintana-Murci L, van de Beek D, Roussel L, Vinh DC, Tangye SG, Haerynck F, Dalmau D, Martinez-Picado J, Brodin P, Nussenzweig MC, Boisson-Dupuis S, Rodríguez-Gallego C, Vogt G, Mogensen TH, Oler AJ, Gu J, Burbelo PD, Cohen JI, Biondi A, Bettini LR, D'Angio M, Bonfanti P, Rossignol P, Mayaux J, Rieux-Laucat F, Husebye ES, Fusco F, **Ursini MV**, Imberti L, Sottini A, Paghera S, Quiros-Roldan E, Rossi C, Castagnoli R, Montagna D, Licari A, Marseglia GL, Duval X, Ghosn J; HGID Lab; NIAID-USUHS Immune Response to COVID Group; COVID Clinicians; COVID-STORM Clinicians; Imagine COVID Group; French COVID Cohort Study Group; Milieu Intérieur

Consortium; CoV-Contact Cohort; Amsterdam UMC Covid-19 Biobank; COVID Human Genetic Effort, Tsang JS, Goldbach-Mansky R, Kisand K, Lionakis MS, Puel A, Zhang SY, Holland SM, Gorochov G, Jouanguy E, Rice CM, Cobat A, Notarangelo LD, Abel L, Su HC, Casanova JL. Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science. 2020 Oct 23;370(6515):eabd4585. doi:10.1126/science.abd4585..

11. Zazzeroni F, Verzella D, Pescatore A, Capece D, Vecchiotti D, **Ursini MV**, Franzoso G, and Alesse E. Life, death and autophagy in cancer: NF-κB turn up everywhere Cell Death & Disease 2020 Mar 30;11(3):210. doi: 10.1038/s41419-020-2399-y.

12. 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.

13. 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.

14. Cammarata-Scalisi F, Fusco F, **Ursini MV**. Incontinentia Pigmenti. Actas Dermosifiliogr. 2019 May;110(4):273-278.

15. 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.

16. 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.

17. 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/

18. 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.

19. 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.

20. 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.

21. 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.

22. 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.

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