Curriculum vitae Dorianna Sandonà

Personal information

- Born: April 28, 1964, Horgen (CH)
- Citizenship: Italian
- Residence: Via Riviera 41 Polverara, Padova (Italy)
- Married with Francesco Di Virgilio

Education

- 1988 Degree in Biological Sciences "summa cum laude", University of Padova.
- 1994 PhD in Molecular and Cellular Biology and Pathology, University of Padova.
- 2014 National academic qualification as Associate professor of Molecular Biology (BIO/11)

Employment

- 01.04.1989/31.12.1989: University of Padova, Institute of Pathology, fellowship by FIDIA S.p.A.
- 01.11.1993/31.10.1994: Telethon fellowship in the project “Molecular analysis of mitochondrial cytopathology with deficit of cytochrome c oxidase.”
- 01.11.1994/31.05.98 technical collaborator at the “Istituto Policattedra della Facoltà di Biotecnologie Agro Industriali”, University of Verona.
- 01/06/1998/30.09.2001 technical collaborator at the “Department of Biomedical and Experimental Sciences”, University of Padova.
- 01.10.2001/30/11/2015 Researcher, grouping discipline Molecular Biology (BIO/11), Department of Biomedical Sciences, School of Medicine, University of Padova.
- 01/12/2015 to date Associate Professor of Molecular Biology, Department of Biomedical Sciences, School of Medicine, University of Padova.

Teaching

- 2002-2012: course of Molecular Biology Laurea Triennale Interfacoltà in Biotecnologie Sanitarie University of Padova
- 2010-2011, 2011-2012: course of Molecular Biology Laurea Magistrale in Farmacia University of Padova
- 2008-2014: course of Molecular Biology II Laurea Magistrale in Biotecnologie Farmaceutiche University of Padova
- 2014 - to date: course of Advanced Molecular Biology, Master degree Pharmaceutical Biotechnologies
- 2016 - to date: course of Protein Engineering, Master degree Pharmaceutical Biotechnologies
- 2017 - 2018: 1 CFU in the course of Molecular Biology, Master degree in Medicine
- 2018 – to date: 2 CFU in the course of Molecular Biology, Master degree in Medicine

Research interest

DS research activity was first devoted to the study of diphtheria toxin mechanism of action in mammalian cells and then of the biochemical and molecular properties of the eukaryotic cytochrome c oxidase. She contributed to determine the nuclear DNA origin of the defect responsible for the Leigh syndrome, a mitochondrial myopathy characterized by cytochrome c oxidase deficiency.

Next, she studied the structure and function of the Photosystem II light harvesting proteins in higher plants.

Since 1999, DS studied the role of the Dystrophin Associated Protein (DAPs) and in particular the role of the sarcoglycan complex in skeletal muscle physiopathology. DS determined and characterized the ecto-ATPase activity of α-sarcoglycan and investigated the function of the extracellular ATP signaling in normal and dystrophic muscles.

In a project funded by the Italian and European Space Agencies, DS studied the signal transduction pathway of muscle atrophy induced by microgravity.

Current research interest regards the trafficking of the sarcoglycan (SG) complex, composed by α, β, γ, δ-SG, and the effects of SG disease-causing point mutations on this process. DS established the involvement of the Endoplasmic Reticulum quality control and the ubiquitin-proteasome system in the pathogenesis of

sarcoglycanopathy. The main research interest of DS is now the evaluation of the efficacy of small molecules, acting either as inhibitors of specific members of the ER associated protein degradation system, or as protein folding correctors, in recovering the sarcoglycan complex. These compounds have the potential to become novel drugs for treating sarcoglycanopathy.
DS has published 56 papers in peer-reviewed international scientific journals. DS has great experience in molecular and cellular biology, biochemistry and muscle physiopathology.

Top five peer-reviewed publications

- Carotti M, Scano M, Fancello I, Richard I, Risato G, Bensalah M, Soardi M and **Sandonà D.** Combined Use of CFTR Correctors in LGMD2D Myotubes Improves Sarcoglycan Complex Recovery (2020) Int. J. Mol. Sci. 21, 1813; doi:10.3390/ijms21051813
- Carotti M, Marsolier J, Soardi M, Bianchini E, Gomiero C, Fecchio C, Henriques SF, Betto R, Sacchetto R, Richard I, **Sandonà D**. Repairing folding-defective α-sarcoglycan mutants by CFTR correctors, a potential therapy for Limb Girdle Muscular Dystrophy 2D. Hum Mol Genet. (2018) 27(6):969-984. doi: 10.1093/hmg/ddy013
- Bianchini E, Fanin M, Mamchaoui K, Betto R, **Sandonà D**. (2014) Unveiling the degradative route of the V247M α-sarcoglycan mutant responsible for LGMD-2D. Hum Mol Genet. 23(14):3746-58 doi: 10.1093/hmg/ddu088
- **Sandonà D**, Desaphy JF, Camerino GM, Bianchini E, Ciciliot S, Danieli-Betto D, Dobrowolny G, Furlan S, Germinario E, Goto K, Gutsmann M, Kawano F, Nakai N, Ohira T, Ohno Y, Picard A, Salanova M, Schiffl G, Blottner D, Musarò A, Ohira Y, Betto R, Conte D, Schiaffino S. (2012) Adaptation of mouse skeletal muscle to long-term microgravity in the MDS mission. PLoS One 7(3):e3323 doi: 10.1371/journal.pone.0033232.
- **Sandonà D**, Danieli-Betto D, Germinario E, Biral D, Martinello T, Lioy A, Tarricone E, Gastaldello S, Betto R. (2005) The T-tubule membrane ATP-operated P2X4 receptor influences contractility of skeletal muscle. FASEB J. 19(9):1184-1186

Grants

- Italian Telethon research grant GGP20097 (239.910 €) 3D modelling of rare muscular diseases, a powerful platform for basic studies and drug validation 2021-2024
- Telethon seed spring program GSP20004_PAsMCT8005 (49.980€) Repurposing CFTR correctors in Allan Herndon Dudley syndrome 2021-2022
- AFM (Association contre les Myopathies) research grant # 23000 (88.700) CFTR correctors to treat sarcoglycanopathy, a repurposing story 2020-2022
- MDA (Muscular dystrophy association) research grant # 577888 (233.162 €) 2019-2020
  "Novel zebrafish models of sarcoglycanopathy. Swimming toward a cure". Role: PI
- Italian Telethon research grant # GGP15140 (314.649 €) 2015-2019
  "Small molecules to rescue folding-defective sarcoglycans: in vivo assessment of novel therapeutic strategies". Role: PI
- AFM (Association contre les Myopathies) research grant, #18620 (71.500 €) 2015-2018
  "Small molecule-based therapy for sarcoglycanopathies. Assessment of efficacy and tolerability in novel animal models". Role: PI
- University of Padova CPDA149821/14 PRAT (35.000 €) 2015-2016
  "New perspectives in sarcoglycanopathy therapy. Assessment of efficacy and tolerability of small molecules in novel animal models" Role: PI
- GEP 12058 Telethon explorative project (34.400 €) 2012-2013
  "Pharmacological rescue of misfolded proteins: innovative approaches for the cure of three muscular diseases". Role: PI

Patents

Sandonà D, Sacchetto R, Bianchini E. Volpe P. Betto R and Mascarello F. A CFTR Corrector For The Teatment Of Genetic Disorders Affecting Striated Muscle.

- Italian patent n.0001414647 (2015)
- US 9987256B2 (2018)
- European Patent No 2925317B (2019)

Richard I and Sandonà D. Combination Treatment Of Sarcoglycanopathies

- PCT/EP2019/050585 (pending)

Pubblications

- Boscaro C, Carotti M, Albiero M, Trenti A, Fadini GP, Trevisi L, **Sandonà D**, Cignarella A, Bolego C. Non-genomic mechanisms in the estrogen regulation of glycolytic protein levels in endothelial cells (2020) FASEB J. 2020 Aug 5. doi: 10.1096/fj.202001130R.
- Carotti M, Scano M, Fancello I, Richard I, Risato G, Bensalah M, Soardi M and **Sandonà D.** Combined Use of CFTR Correctors in LGMD2D Myotubes Improves Sarcoglycan Complex Recovery (2020) Int. J. Mol. Sci. 21, 1813; doi:10.3390/ijms21051813
- Carotti M, Marsolier J, Soardi M, Bianchini E, Gomiero C, Fecchio C, Henriques SF, Betto R, Sacchetto R, Richard I, **Sandonà D**. Repairing folding-defective α-sarcoglycan mutants by CFTR correctors, a potential therapy for Limb Girdle Muscular Dystrophy 2D. Hum Mol Genet. (2018) 27(6):969-984. doi: 10.1093/hmg/ddy013
- Henriques SF, Patissier C, Bourg N, Fecchio C, **Sandonà D**, Marsolier S, Richard I. Different outcome of sarcoglycan missense mutation between human and mouse. PLosOne (2018) 13(1):e0191274 doi: 10.1371/journal.pone.0191274
- Carotti M, Fecchio C, **Sandonà D.** Emerging therapeutic strategies for sarcoglycanopathy Exp Opin in Orphan drugs (2017) 5(5): 381-396 doi: 10.1080/21678707.2017.1307731
- Marsolier J, Laforet P, Pegoraro E, Vissing J, Richard I and Sarcoglycanopathies Working Group. 1st International Workshop on Clinical trial readiness for sarcoglycanopathies 15-16 November 2016, Evry, France. Neuromuscul Disord. (2017) 27(7): 683-692 doi: 10.1016/j.nmd.2017.02.011
- Bianchini E, Testoni S, Gentile A, Calì T, Ottolini D, Villa A, Brini M, Betto R, Mascarello F, Nissen P, **Sandonà D***, Sacchetto R*. (2014) Inhibition of Ubiquitin Proteasome System Rescues the Defective Sarco(endo)plasmic Reticulum Ca2+-ATPase (SERCA1) Protein Causing Chianina Cattle Pseudomyotonia. J Biol Chem. 2014 Oct 6. pii: jbc.M114.576157 doi: 10.1074/jbc
- Bianchini E, Fanin M, Mamchaoui K, Betto R, **Sandonà D**. (2014) Unveiling the degradative route of the V247M α-sarcoglycan mutant responsible for LGMD-2D. Hum Mol Genet. 23(14):3746-58 doi: 10.1093/hmg/ddu088
- Camerino GM, Pierno S, Liantonio A, De Bellis M, Cannone M, Sblendorio V, Conte E, Mele A, Tricarico D, Tavella S, Ruggiu A, Cancedda R, Ohira Y, Danieli-Betto D, Ciciliot S, Germinario E, **Sandonà D**, Betto R, Camerino DC, Desaphy JF. (2013) Effects of pleiotrophin overexpression on mouse skeletal muscles in normal loading and in actual and simulated microgravity. PLoS One 8(8):e72028. doi: 10.1371/journal.pone.0072028
- **Sandonà D**, Desaphy JF, Camerino GM, Bianchini E, Ciciliot S, Danieli-Betto D, Dobrowolny G, Furlan S, Germinario E, Goto K, Gutsmann M, Kawano F, Nakai N, Ohira T, Ohno Y, Picard A, Salanova M, Schiffl G, Blottner D, Musarò A, Ohira Y, Betto R, Conte D, Schiaffino S. (2012) Adaptation of mouse skeletal muscle to long-term microgravity in the MDS mission. PLoS One 7(3):e3323 doi: 10.1371/journal.pone.0033232.
- Martinello T, Baldoin MC, Morbiato L, Paganin M, Tarricone E, Schiavo G, Bianchini E, **Sandonà D**, Betto R. (2011) Extracellular ATP signaling during differentiation of C2C12 skeletal muscle cells: role in proliferation. Mol Cell Biochem. 351(1-2):183-96 doi: 10.1007/s11010-011-0726-4

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- **Sandonà D**, Betto R. (2009) Sarcoglycanopathies: molecular pathogenesis and therapeutic prospects. Expert. Rev. Mol. Med.: 11:e28 doi: 10.1017/S1462399409001203
- Gastaldello S, D'Angelo S, Franzoso S, Fanin M, Angelini C, Betto R, **Sandonà D**. (2008) Inhibition of proteasome activity promotes the correct localization of disease-causing α-sarcoglycan mutants in a heterologous cell system constitutively expressing β-, γ-, and δ-sarcoglycan. Am. J. Path 173(1):170-81 doi: 10.2353/ajpath.2008.071146.
- Zanin M, Germinario E, Dalla Libera L, **Sandonà D,** Sabbadini RA, Betto R, Danieli-Betto D. (2008) Trophic action of sphingosine 1-phosphate in denervated rat soleus muscle. Am J Physiol Cell Physiol. 294(1):C36-46.
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- Danieli-Betto D, Espositi A, Germinario E, **Sandonà D**, Martinello T, Jakubiec-Puka A, Biral D, Betto R. (2005) Deficiency of {alpha}-sarcoglycan differently affects fast- and slow-twitch skeletal muscles. Am. J. Physiol-Regul. Integr. and Comp Physiol. 289(5): R1328-R1337
- **Sandonà D**, Danieli-Betto D, Germinario E, Biral D, Martinello T, Lioy A, Tarricone E, Gastaldello S, Betto R. (2005) The T-tubule membrane ATP-operated P2X4 receptor influences contractility of skeletal muscle. FASEB J. 19(9):1184-1186.
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Functional co-localization of the ecto-ATPase ?-sarcoglycan and P2X receptors. (1999) The ectonucleotidases. Ed. L. Vanduffel Shaker Publishing BV, Maastricht, The Netherlands. 295-304

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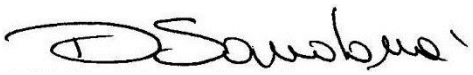

Padova, 01.03.2021