CURRICULUM VITAE

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Date of Birth: November, 1964 (Koege, Denmark)
Citizen: Danish

PROFESSIONAL EXPERIENCE

2014 to date: Associate Professor in Molecular Biology (Bio11), University of Insubria, Department of Biotechnology and Life Sciences, Italy.
2002-2014: Assistant Professor, University of Insubria, Department of Theoretical and Applied Sciences, Italy.
2020 to date: Scientific board member of Centre of Neuroscience, University of Insubria.
2021 to date: Associate Editor in Cellular Neurophysiology, Frontiers in Cellular Neuroscience.
2023: National Scientific Qualification as Full Professor in 05/E2 – Molecular Biology

EDUCATION

1999-2002: Phd in Molecular and Cellular Biology, Open University, London, UK.
1997-1999: Pre-doctoral studies the laboratory of Dr. Vincenzo Zappavigna, San Raffaele Scientific Institute, Milan, Italy.
1996: University Degree in Biochemistry at the University of Copenhagen, Denmark.

GRANTS

1994-1995: Fellowship from the Danish Cancer Society.
1999-2002: Ph.D fellowship from the Danish Research Academy.
2004-2006: Rett Syndrome Research Foundation Grant. “Characterization of novel proteins influencing MeCP2 activity and analysis of their possible involvement in Rett Syndrome”.
2006-2008: Rett Syndrome Research Foundation Grant. "Functional characterization of CDKL5, a novel gene involved in the onset of Rett syndrome".
2006-2008: Progetto ISS “Programma Italia-USA Malattie Rare”. Molecular and functional characterization of the newly identified interaction between the Rett syndrome-associated factor MeCP2 and the pro-apoptotic factor HIPK2.
2008-2010: Prin 2007: “Molecular and Functional Characterization of CDKL5, a novel X-linked kinase, mainly involved in female mental retardation.”
2009-2012: Marie Curie Actions – Network for initial training: “DisChrom – Chromatin diseases: from basic mechanisms to therapy.”
2010-2012: Lejeune Foundation: “Disclosing the brain functions of CDKL5, an X-linked gene involved in severe mental retardation and infantile spasms.”
2013-2015: International Rett Syndrome Foundation. “Investigation of the importance of a hitherto uncharacterized MeCP2 phospho-isoform for neuronal morphogenesis and chromatin related functions”.
2015-2018: Telethon Foundation: “Therapeutic Strategies for CDKL5 disorder”.
2016-2017: University of Pennsylvania Orphan Disease Center, Loulou Foundation: “Predictive Drug Repurposing for CDKL5 Syndrome”.
2017-2018: University of Pennsylvania Orphan Disease Center, Loulou Foundation: “Therapeutic potential of pregnenolone and its synthetic non-metabolized derivative for CDKL5 disorder”.
2018-2020: Airett: “Characterization of the therapeutic potential of neurosteroids for CDKL5-disorder”.
2019-2020: University of Pennsylvania Orphan Disease Center, Loulou Foundation: “Plasma microtubular proteins as potential biomarkers of CDKL5 Deficiency Disorder (CDD).” (partner)
2020-2021: University of Pennsylvania Orphan Disease Center, Loulou Foundation: “Investigation of the microtubule modulator, pregnenolone-methyl-ether (PME), and the synaptic plasticity stimulator, ketamine, as a potential intervention in a mouse model of Cdkl5 deficiency disorder.” (partner)
2021-2024: Telethon Foundation: “GABAA-receptor defects in CDKL5 deficiency disorder: molecular mechanisms and targeting by synthetic neuroactive steroids”.

MEETING ORGANIZATION

- First European Working Group on Rett Syndrome; Busto Arsizio, Italy; April 19-20, 2007
- 6° Word Rett Syndrome Congress; Paris, France; October 10-13 2008
- Second European Working Group on Rett Syndrome; Stresa, Italy; September 17-19, 2009
- FOCUS on CDKL5; Turin, Italy, November 18th-19th, 2016

PUBLICATIONS

1. Berthelsen J*, **Kilstrup-Nielsen C***, Blasi F, Mavilio F, and Zappavigna V. (1999) The subcellular localization of PBX1 and EXD proteins depends on nuclear import and export signals and is modulated by association with PREP1 and HTH. Genes & Dev. 13, 946-953. (*Equal first author) doi: 10.1101/gad.13.8.946.
2. Fognani C, **Kilstrup-Nielsen C**, Berthelsen J, Ferretti E, Zappavigna V, and Blasi F. (2002) Characterization of PREP2, a paralog of PREP1, which defines a novel sub-family of the MEINOX TALE homeodomain transcription factors. Nucl Acids Res 30, 2043-2051. doi: 10.1093/nar/30.9.2043.
3. **Kilstrup-Nielsen C,** Alessio M, Zappavigna V. (2003) PBX1 nuclear export is regulated independently of PBX-MEINOX interaction by PKA phosphorylation of the PBC-B domain. EMBO J. 22, 89-99. doi: 10.1093/emboj/cdg010.
4. Segalla S, Rinaldi L, **Kilstrup-Nielsen C**, Badaracco G, Minucci S, Pelicci PG, Landsberger N. (2003) Retinoic acid receptor alpha fusion to PML affects its transcriptional and chromatin-remodeling properties. Mol Cell Biol. 23, 8795-808. doi: 10.1128/MCB.23.23.8795-8808.2003.
5. Carro S, Bergo A, Mengoni M, Bachi A, Badaracco G, **Kilstrup-Nielsen C**, Landsberger N. (2004) A novel protein, Xenopus p20, influences the stability of MeCP2 through direct interaction. J Biol Chem 279, 25623-25631. doi: 10.1074/jbc.M402571200.
6. Mari F, Azimonti S, Bertani I, Bolognese F, Colombo E, Caselli R, Scala E, Longo I, Grosso S, Pescucci C, Ariani F, Hayek G, Balestri P, Bergo A, Badaracco G, Sapella M, Brocolli V, Renieri A, **Kilstrup-Nielsen C**, Landsberger N. (2005) CDKL5 belongs to the same molecular pathway of MeCP2 and it is responsible for the early-onset seizure variant of Rett syndrome. Hum Mol Genetics 14, 1935-1946. doi: 10.1093/hmg/ddi198.
7. Mari F, **Kilstrup-Nielsen C**, Cambi F, Speciale C, Mencarelli MA, Renieri A. (2005) Genetics and mechanisms of disease in Rett Syndrome. Drug Discovery Today. 2, 419-425.
8. Bertani I, Rusconi, Bolognese F, Forlani G, Conca B, De Monte L, Landsberger N, **Kilstrup-Nielsen C**. (2006). Functional consequences of mutations in CDKL5, an X-linked gene involved in infantile spasms and mental retardation. J.Biol.Chem. 281, 32048-32056. doi: 10.1074/jbc.M606325200.
9. Marchi M, Guarda A, Bergo A, Landsberger N, **Kilstrup-Nielsen C**, Ratto GM, Costa M. (2007) Spatio-temporal dynamics and localization of MeCP2 and pathological mutants in living cells. Epigenetics. 2, 187-197. doi: 10.4161/epi.2.3.5057.
10. Rusconi L, Salvatoni L, Giudici L, Bertani I, **Kilstrup-Nielsen C**, Broccoli V, Landsberger N. (2008) CDKL5 expression is modulated during neuronal development and its subcellular distribution is tightly regulated by the C-terminal tail. J Biol Chem. 283, 30101-30111. doi: 10.1074/jbc.M804613200.
11. Bracaglia G, Conca B, Bergo A, Rusconi L, Zhou Z, Greenberg ME, Landsberger N, Soddu S, **Kilstrup-Nielsen C**. (2009) Methyl-CpG-binding protein 2 is phosphorylated by homeodomain-interacting protein kinase 2 and contributes to apoptosis. EMBO Rep. 10, 1327-1333. doi: 10.1038/embor.2009.217.
12. Ricciardi S, **Kilstrup-Nielsen C**, Bienvenu T, Jacquette A, Landsberger N, Broccoli V. (2009) CDKL5 influences RNA splicing activity by its association to the nuclear speckle molecular machinery. Hum Mol Genet. 18, 4590-4602. doi: 10.1093/hmg/ddp426.
13. Forlani G, Giarda E, Ala U, Di Cunto F, Salani M, Tupler R, ***Kilstrup-Nielsen C**, *Landsberger N (2010) The MeCP2/YY1 interaction regulates ANT1 expression at 4q35: novel hints for Rett syndrome pathogenesis. Hum Mol Genet. 19, 3114-3123. (*Equal last author) doi: 10.1093/hmg/ddq214.
14. Williamson SL, Giudici L, **Kilstrup-Nielsen C**, Gold W, Pelka GJ, Tam PP, Grimm A, Prodi D, Landsberger N, Christodoulou J. (2012) A novel transcript of cyclin-dependent kinase-like 5 (CDKL5) has an alternative C-terminus and is the predominant transcript in brain. Hum Genet. 131:187-200. doi: 10.1007/s00439-011-1058-x.
15. Rusconi L, ***Kilstrup-Nielsen C**, *Landsberger N. (2011) Extrasynaptic N-methyl-D-aspartate (NMDA) receptor stimulation induces cytoplasmic translocation of the CDKL5 kinase and its proteasomal degradation. J Biol Chem. 286:36550-36558. (*Equal last author) doi: 10.1074/jbc.M111.235630.
16. **Kilstrup-Nielsen C**, Rusconi L, La Montanara P, Ciceri D, Bergo A, Bedogni F, Landsberger N. (2012) What we know and would like to know about CDKL5 and its involvement in epileptic encephalopathy. Neural Plast. 2012;2012:728267. doi: 10.1155/2012/728267
17. Ricciardi S, Ungaro F, Hambrock M, Rademacher N, Stefanelli G, Brambilla D, Sessa A, Magagnotti C, Bachi A, Giarda E, Verpelli C, **Kilstrup-Nielsen C**, Sala C, Kalscheuer VM, Broccoli V. (2012) CDKL5 ensures excitatory synapse

stability by reinforcing NGL-1-PSD95 interaction in the postsynaptic compartment and is impaired in patient iPSC-derived neurons. Nat Cell Biol. 14:911-923. doi: 10.1038/ncb2566.

18. Bedogni F, Rossi RL, Galli F, Cobolli Gigli C, Gandaglia A, **Kilstrup-Nielsen C**, Landsberger N. (2014) Rett syndrome and the urge of novel approaches to study MeCP2 functions and mechanisms of action. Neurosci Biobehav Rev. S0149-7634 (14). doi: 10.1016/j.neubiorev.2014.01.011.
19. Bellini E, Pavesi G, Barbiero I, Bergo A, Chandola C, Nawaz MS, Rusconi L, Stefanelli G, Strollo M, Valente MM, ***Kilstrup-Nielsen C**, *Landsberger N. (2014) MeCP2 post-translational modifications: a mechanism to functionally organize a disorganized protein? Frontiers Cell Neurosci. 8:236. (*Equal last author) doi: 10.3389/fncel.2014.00236.
20. Bergo A, Strollo M, Gai M, Barbiero I, Stefanelli G, Sertic S, Cobolli Gigli C, Di Cunto F, ***Kilstrup-Nielsen C**, *Landsberger N. (2015) MeCP2 localizes at the centrosome and is required for proper mitotic spindle organization. J Biol Chem. 290: 3223-3237. (*Equal last author) doi: 10.1074/jbc.M114.608125.
21. La Montanara P, Rusconi L, Locarno A, Forti L, Barbiero I, Chandola C, ***Kilstrup-Nielsen C**, *Landsberger N. (2015) Synaptic synthesis, dephosphorylation and degradation: a novel paradigm for a developmentally regulated NMDA-dependent control of CDKL5. J Biol Chem, 290: 4512-4527. (*Equal last author) doi: 10.1074/jbc.M114.589762.
22. Conti V, Gandaglia A, Galli F, Tirone M, Bellini E, Campana L, **Kilstrup-Nielsen C**, Rovere-Querini P, Brunelli S, Landsberger N. (2015) MeCP2 Affects Skeletal Muscle Growth and Morphology through Non Cell-Autonomous Mechanisms. PLoS One, 10, e0130183. doi: 10.1371/journal.pone.0130183.
23. Bedogni F, Cobolli Gigli C, Pozzi D, Rossi RL, Scaramuzza L, Rossetti G, Pagani M, Matteoli M, **Kilstrup-Nielsen C**, Landsberger N. (2015) Defects during Mecp2 null embryonic cortex development precede the onset of overt neurological symptoms. Cerebral Cortex bhv078. doi: 10.1093/cercor/bhv078.
24. Nawaz MS, Giarda E, Bedogni F, La Montanara P, Ricciardi S, Ciceri D, Landsberger N, Rusconi L, **Kilstrup-Nielsen C**. (2016) CDKL5 and shootin1 interact and concur in regulating neuronal polarization and migration. Plos One 11(2):eO148634. doi: 10.1371/journal.pone.0148634.
25. Cobolli Gigli C, Scaramuzza L, Gandaglia A, Bellini E, **Kilstrup-Nielsen C**, Landsberger N, Bedgoni F. (2016) MeCP2 related studies benefit from the use of CD1 as genetic background. PlosOne, 11, e0153473. doi: 10.1371/journal.pone.0153473.
26. Hector RD, Dando O, Landsberger N, **Kilstrup-Nielsen C**, Kind PC, Bailey MES; Cobb SR. (2016) Characterisation of CDKL5 Transcript Isoforms in Human and Mouse. Plos One, 11, e0157758. doi: 10.1371/journal.pone.0157758.
27. Stefanelli G, Gandaglia A, Costa M, Cheema M, Dimarino D, Barbiero I, **Kilstrup-Nielsen C**, Ausió J, Landsberger N. (2016) Brain phosphorylation of MeCP2 at serine 164 is developmentally regulated and alters its chromatin association. Sci Rep. 6:28295. doi: 10.1038/srep28295.
28. Barbiero I, Valente D, Chandola C, Magi F, Bergo A, Monteonofrio L, Tramarin M, Fazzari M, Soddu S, Landsberger N, Rinaldo C, **Kilstrup-Nielsen C**. (2017) CDKL5 localizes at the centrosome and midbody and is required for faithful cell division. Sci Rep. 7, 6228. doi: 10.1038/s41598-017-05875-z.
29. Barbiero I, Peroni D, Tramarin M, Chandola C, Rusconi L, Landsberger N, **Kilstrup-Nielsen C**. (2017) The neurosteroid pregnenolone reverts microtubule derangement induced by the loss of a functional CDKL5-IQGAP1 complex. Hum Mol Genet 26, 3520-3530. doi: 10.1093/hmg/ddx237.
30. Tramarin M, Rusconi L, Pizzamiglio L, Barbiero I, Peroni D, Scaramuzza L, Guilliams T, Cavalla D, Antonucci F, **Kilstrup-Nielsen C.** (2018) The antidepressant tianeptine reverts synaptic AMPA receptor defects caused by deficiency of CDKL5. Hum Mol Genet. 27, 2052-2063. doi: 10.1093/hmg/ddy108.
31. Vigli D, Rusconi L, Valenti D, La Montanara P, Cosentino L, Lacivita E, Leopoldo M, Amendola E, Gross C, Landsberger N, Laviola G, **Kilstrup-Nielsen C**, Vacca RA, De Filippis B. (2019) Rescue of prepulse inhibition deficit and brain mitochondrial dysfunction by pharmacological stimulation of the central serotonin receptor 7 in a mouse model of CDKL5 disorder. Neuropharmacology. 144, 104-114. doi: 10.1016/j.neuropharm.2018.10.018.
32. Gandaglia A, Brivio E, Carli S, Palmieri M, Bedogni F, Stefanelli G, Bergo A, Leva B, Cattaneo C, Pizzamiglio L, Cicerone M, Bianchi V, **Kilstrup-Nielsen C**, D'Annessa I, Di Marino D, D'Adamo P, Antonucci F, Frasca A, Landsberger N. (2018) A novel Mecp2^{Y120D} knock-in model displays similar behavioral traits but distinct molecular features compared to the Mecp2-null mouse implying precision medicine for the treatments of Rett syndrome. Mol Neurobiol. 56, 4838-4854. doi: 10.1007/s12035-018-1412-2.
33. Zamberletti E, Gabaglio M, Piscitelli F, Brodie JS, Wolley-Roberts M, Barbiero I, Tramarin M, Binelli G, Landsberger N, **Kilstrup-Nielsen C**, Rubino T, Di Marzo V, Parolaro D. (2019) Cannabidivarin completely rescues cognitive deficits and delays neurological and motor defects in male Mecp2 mutant mice. J. Psychopharmacol. 33, 894-907. doi: 10.1177/0269881119844184.
34. Barbiero I, De Rosa R, **Kilstrup-Nielsen C**. (2019) Microtubules: a key to understand and correct neuronal defects in CDKL5 deficiency disorder? Int J Mol Sci, 20, 4075. doi: 10.3390/ijms20174075.

35. Barbiero I, Peroni D, Siniscalchi P, Rusconi L, Tramarin T, De Rosa R, Motta P, Bianchi M, **Kilstrup-Nielsen C**. (2020) Pregnenolone and pregnenolone-methyl-ether rescue neuronal defects caused by dysfunctional CLIP170 in a neuronal model of CDKL5 Deficiency Disorder. Neuropharmacology, 164, 107897. doi: 10.1016/j.neuropharm.2019.107897.
36. Trovò L, Fuchs C, De Rosa R, Barbiero I, Tramarin M, Ciani E, Rusconi L, **Kilstrup-Nielsen C**. (2020) The green tea polyphenol epigallocatechin-3-gallate (EGCG) restores CDKL5-dependent synaptic defects in vitro and in vivo. Neurobiol Dis, 138:104791. doi: 10.1016/j.nbd.2020.104791.
37. Frasca A, Spiombi E, Palmieri M, Valente M, Bergo A, Leva A, **Kilstrup-Nielsen C**, Bianchi F, Di Cunto F, Landsberger N. (2020) MECP2 mutations affect ciliogenesis: a novel perspective for Rett syndrome and related disorders. EMBO Molecular Medicine. 12, e10270. doi: 10.15252/emmm.201910270.
38. Barbiero I, Bianchi M, **Kilstrup-Nielsen C**. (2022) Therapeutic potential of pregnenolone and pregnenolone methyl ether on depressive and CDKL5 deficiency disorders: Focus on microtubule targeting. J Neuroendocrin, 34, e13033. doi: 10.1111/jne.13033.
39. Barbiero I, Zamberletti E, Tramarin M, Gabaglio M, Peroni D, De Rosa R, Baldin S, Bianchi M, Rubino M, **Kilstrup-Nielsen C**. (2022) Pregnenolone-methyl-ether enhances CLIP170 and microtubule functions improving spine maturation and hippocampal deficits related to CDKL5 deficiency. Hum Mol Genetics. doi: 10.1093/hmg/ddac067.
40. De Rosa R, Valastro S, Cambria C, Barbiero I, Puricelli C, Tramarin M, Randi S, Bianchi M, Antonucci F, **Kilstrup-Nielsen C**. (2023) Loss of CDKL5 causes synaptic GABAergic defects that can be restored with the neuroactive steroid Pregnenolone-methyl-ether. Int J Mol Sci. 24, 68. Doi: 10.3390/inms24010068.