



**Charlotte
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UNIVERSITY OF INSUBRIA



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Contact data

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Biography

December 1996: University Degree in Biochemistry at the University of Copenhagen, Denmark.

Experimental thesis: "Identification of a TATA-box in the hIGF1 gene".

December 2003: Ph.D., Molecular and Cellular Biology. Experimental thesis: "Molecular mechanisms underlying the regulation of PBX1 sub-cellular localization".

April 2004 to present: Assistant Professor in Molecular Biology, Department of Structural and Functional Biology, at the University of Insubria, Varese, Italy.

Qualifications and awards

Personal 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".

Research interests

As Assistant Professor at the University of Insubria, I have mainly been occupied in studying the molecular mechanisms involved in Rett Syndrome, a form of X-linked mental retardation. Mutations in the methyl-CpG binding protein, MeCP2, have been found in the major part of patients affected with this syndrome. MeCP2 represses the expression of genes through the binding to methylated DNA and recruitment of chromatin modifying complexes. Several data suggest that more genes are involved in the Rett Syndrome, either directly or as modifiers of MeCP2 activity. Different approaches have been applied to find new factors interacting with MeCP2 and the main focus of my research is aimed at studying the functional role of these.

Teaching experience and appointments

2003 to present:

Laboratory of Experimental Biology (1,25 Credits): a 1st level degree laboratory course in Biological Sciences.

Integrated Laboratory I – unit of Molecular Biology (1 credit): a 1st level degree laboratory course in Biology of Health.

Laboratory of Molecular Biology (3 Credits): a 2nd level degree laboratory course in Biology of Health.

Integrated Laboratory II – unit of Molecular Biology (1 credit): a 2nd degree level laboratory course in Biology Applied to Biomedical Science.

Representative publications

1) Kilstrup-Nielsen C, Berthelsen J, 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. (UK)

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. (UK)

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. (UK)

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. (UK)

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. (UK)

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, Broccoli 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. (UK)

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 (In press). (UK)

Clinical interests

Even if the projects in the laboratory are mainly of basic research, they are focused on investigating different aspects of MeCP2 functions and Rett Syndrome and may therefore prove useful to the development of new therapeutic and diagnostic strategies in the future.