## **ORAL PRESENTATION**



**Open Access** 

## *LMNA* p.R482W mutation related to FPLD2 alters SREBP1-A type lamin interactions in human fibroblasts and adipose stem cells

Brigitte Buendia

*From* 1st French-Italian meeting on laminopathies and other nuclear envelope-related diseases Marseille, France. 15-16 January 2015

SREBP1 (Sterol regulatory element binding protein 1), transcription factor that regulates hundreds of genes involved in lipid metabolism and adipocyte differentiation, is a direct partner of A-type lamins. We show that i) in vitro, the tail regions of prelamin A, lamin A and lamin C bind a polypeptide of SREBP1 and ii) within cells, interactions between wild-type A-type lamins and SREBP1 occur mainly at the nuclear periphery but also within the nucleoplasm. While A-type lamin R482W mutation is responsible for Dunnigan type familial partial lipodystrophy (FPLD2), we show that both overexpression of LMNA p.R482W in primary human preadipocytes and endogenous expression of A-type lamins p.R482W in FPLD2 patient fibroblasts, reduce A-type lamins-SREBP1 in situ interactions and upregulates a large number of SREBP1 target genes [1]. As this LMNA mutant was previously shown to inhibit adipogenic differentiation, we propose that deregulation of SREBP1 by mutated A-type lamins constitutes one underlying mechanism of the physiopathology of FPLD2.

Published: 11 November 2015

## Reference

 Vadrot N, Duband-Goulet I, Cabet E, Attanda W, Barateau A, Vicart P, et al: The p.R482W substitution in A-type lamins deregulates SREBP1 activity in Dunnigan-type familial partial lipodystrophy. Human molecular genetics 2015, 24(7):2096-109.

doi:10.1186/1750-1172-10-S2-O13 Cite this article as: Buendia: *LMNA* p.R482W mutation related to FPLD2 alters SREBP1-A type lamin interactions in human fibroblasts and adipose stem cells. *Orphanet Journal of Rare Diseases* 2015 10(Suppl 2): O13.

Correspondence: brigitte.buendia@univ-paris-diderot.fr

Unité de Biologie Fonctionnelle et Adaptative (BFA), Université Paris Diderot-Paris 7, CNRS, UMR 8251, Paris, France

## Submit your next manuscript to BioMed Central and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar
- Research which is freely available for redistribution

) BioMed Central

Submit your manuscript at www.biomedcentral.com/submit



© 2015 Buendia This is an Open Access article distributed under the terms of the Creative Commons Attribution License (http:// creativecommons.org/licenses/by/4.0), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited. The Creative Commons Public Domain Dedication waiver (http://creativecommons.org/publicdomain/ zero/1.0) applies to the data made available in this article, unless otherwise stated.