



Simposio Internacional: Enfermedades neuromusculares: es el tiempo para el tratamiento

International Symposium: Neuromuscular diseases: It's time for treatment

Madrid, 15 y 16 de noviembre de 2012

Madrid, November 15-16, 2012

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Since 2006 YOUNG GROUP LEADER (INSERM AVENIR) - IGBMC, STRASBOURG, FRANCE :

« Understanding the molecular causes of myotonic dystrophies. »

INSTITUT DE GÉNÉTIQUE ET DE BIOLOGIE MOLÉCULAIRE ET CELLULAIRE,

2003 - 2006 POST-DOCTORAL POSITION - IGBMC, STRASBOURG, FRANCE :

« Regulation of the transcriptionnal bypass of DNA oxidative lesions. »

Supervisor : Prof. Jean-Marc Egly.

1996 - 2003 PHD - UNIVERSITY OF PARIS VII, FRANCE / BCM, HOUSTON, USA :

« Alternative splicing alteration in myotonic dystrophic patients.»

BAYLOR COLLEGE OF MEDICINE, Dept. of Pathology, 77030 Houston, Texas, USA.

Supervisor : Prof. Thomas Cooper.

CENTRE DE GÉNÉTIQUE MOLÉCULAIRE, CNRS UPR 2167, 91190 Gif-sur-Yvette, France.

Supervisor : Dr. Claude Thermes.

MAIN PUBLICATIONS :

- Fugier C, Klein A, Hammer C, Vassilopoulos S, Ivarsson Y, Toussaint A, Tosch V, Vignaud A, Ferry A, Messaddeq N, Kokunai Y, Tsuburaya R, de la Grange P, Dembele D, Francois V, Precigout G, Boulade-Ladame C, Hummel MC, Lopez de Munain A, Sergeant N, Laquerrière A, Thibault C, Deryckere F, Auboeuf D, Garcia L, Zimmermann P, Udd B, Schoser B, Takahashi M, Nishino I, Bassez G, Laporte J, Furling D, Charlet-B N.

Mis-regulation of the alternative splicing of BIN1 is associated with T-tubule alterations and muscle weakness in Myotonic Dystrophy.

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- Rau F, Freyermuth F, FugierC, VilleminJP, JostB, DembeleD, GourdonG, NicoleA, DubocD, WahbiK, DayJW, FujimuraH, TakahashiMP, AuboeufD, DreumontN, Furling D, Charlet-B. N.

Mis-regulation of miR-1 processing is associated with heart defects in Myotonic Dystrophy.

Nature Structural and Molecular Biology. 2011. In press doi:10.1038/nsmb.2067.

- Sellier C., Rau F., Liu Y., Tassone F., Hukema RK., Gattoni R., Schneider A., Richard S., Willemsen R., Elliott DJ., Hagerman PJ., Charlet-B. N.

Sam68 sequestration and partial loss of function are associated with splicing alterations in FXTAS patients.

EMBO. J. 2010. 29(7):1248-61.