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SELECTED PUBLICATIONS

Zurita E, Chagoyen M, Cantero M, Alonso R, González-Neira A, López-Jiménez A, López-Moreno JA, Landel CP, Benítez J, Pazos F, Montoliu L. Genetic polymorphisms among C57BL/6 mouse inbred strains. Transgenic Res. 2011 Jun;20(3):481-9

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Román AC, González-Rico FJ, Moltó E, Hernando H, Neto A, Vicente-Garcia C, Ballestar E, Gómez-Skarmeta JL, Vavrova-Anderson J, White RJ, Montoliu L, Fernández-Salguero PM. Dioxin receptor and SLUG transcription factors regulate the insulator activity of B1 SINE retrotransposons via an RNA polymerase switch. Genome Res. 2011 Mar;21(3):422-32

Martin D, Pantoja C, Fernández Miñán A, Valdes-Quezada C, Moltó E, Matesanz F, Bogdanovi O, de la Calle-Mustienes E, Domínguez O, Taher L, Furlan-Magaril M, Alcina A, Cañón S, Fedetz M, Blasco MA, Pereira PS, Ovcharenko I, Recillas-Targa F, Montoliu L, Manzanares M, Guigó R, Serrano M, Casares F, Gómez-Skarmeta JL. Genome-wide CTCF distribution in vertebrates defines equivalent sites that aid the identification of disease-associated genes. Nat Struct Mol Biol. 2011 Jun;18(6):708-14

Tiana M, Villar D, Pérez-Guijarro E, Gómez-Maldonado L, Moltó E, Fernández-Miñán A, Gómez-Skarmeta JL, Montoliu L, del Peso L. A role for insulator elements in the regulation of gene expression response to hypoxia. Nucleic Acids Res. 2012 Mar;40(5):1916-27

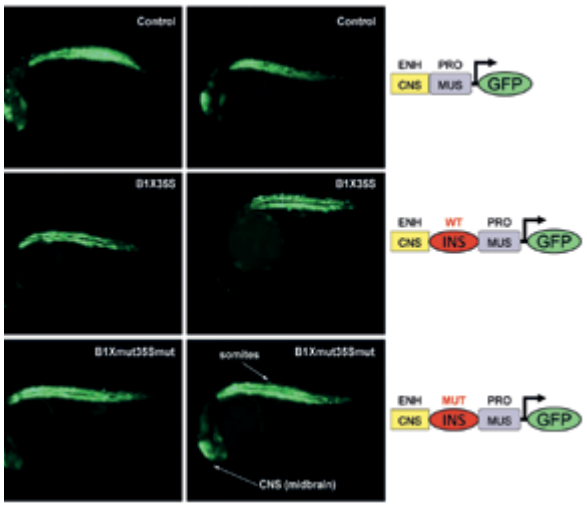
PATENT

P201231296 New animal model for acromatopsia.

Animal models by genetic manipulation

In our laboratory, we want to understand how mammalian expression domains work and how they are organised within genomes. We focus our interest on the identification and characterisation of genomic boundaries or insulators. Insulators can be used effectively in biotechnological applications as spacers, as boundaries, in any gene expression construct for gene transfer experiments. They prevent inappropriate expression patterns of transgenes or gene therapy constructs, by insulating them from neighbouring sequences at the insertion site in the host genome. We are searching for new insulator sequences in vertebrate genomes. We initiate our experiments through *in silico* predictions; insulator candidates are subsequently validated functionally *in vitro* using cells and the enhancer blocking assay. Finally, we carry out *in vivo* studies in transgenic animals, with zebrafish (in collaboration with J.L. Gómez-Skarmeta, Centro Andaluz de Biología de Desarrollo, Seville) and mice bearing appropriate informative constructs. In the last two years, in collaboration with a number of national and international research groups, we described and functionally validated several new types of boundary elements. Our laboratory also generates and analyses new mouse models to study alterations in vision associated with albinism, a rare disease studied in the scope of the CIBERER-ISCIII centre (www.ciberer.es). We also collaborate with Spanish and French associations in support of people with albinism, ALBA (www.albinismo.es) and GENESPOIR (www.genespoir.org). With the group of A. Carracedo (Univ. Santiago de Compostela), we are developing a universal genetic diagnosis for all known albinism-associated genetic mutations (>600).

Our expertise in mouse embryo and sperm cryopreservation enabled participation in the EU FP7 Projects in mouse functional genomics, including INFRAFRONTIER and EMMA, the European Mouse Mutant Archive, whose Spanish node at the CNB is coordinated by Lluís Montoliu. Finally, through collaborations, we have generated a number of additional transgenic mouse models to study human diseases, including Alzheimer's. For this work, we exploited our yeast artificial chromosome (YAC) transgene technology, which has been instrumental in scientific contracts with biotechnological companies.



1 *Functional validation and mechanism of the SINEB1 element X35S working as a boundary in transgenic zebrafish expressing GFP. See Roman et al. (2011) for additional information.*