



ENCONTROS  
SCIENTIA

## What can embryonic development tell us about disease mechanisms? The case of skeletal muscle.

**Sólveig Thorsteinsdóttir**

Group Leader, Development and Evolutionary Morphogenesis Group

Skeletal muscle development starts early in the development of vertebrate embryos and progressively builds the skeletal muscles that make us able to move and breathe. Throughout most of our lives, skeletal muscle is a tissue with remarkable regenerative potential because it contains muscle stem cells, derived from embryonic muscle progenitor cells, which are tucked into a specific “niche” near the existing muscle fibres. When muscles are damaged, some of these cells turn on the muscle developmental programme and repair the damaged muscle fibres.

Congenital muscular dystrophies are diseases where the connection between the muscle fibres and the extracellular matrix molecule laminin 211 (which normally surrounds muscle fibres) is faulty, leading to muscle weakness evident already at birth. Among the most severe ones is LAMA2-congenital muscular dystrophy (LAMA2-CMD) where laminin 211 is absent. Muscle damage and wasting starts early in life and patients rarely survive past their teens or early twenties. There is no treatment or cure.

Since infants with LAMA2-CMD are already affected at birth, the defect underlying the disease must have arisen during development *in utero*. Here we used a mouse model for LAMA2-CMD to study when, during embryonic development, this disease starts to manifest itself and exactly what mechanisms of muscle development and/or maintenance are the first to go wrong.

We find perturbations in two periods of skeletal muscle development in laminin 211-deficient mouse embryos, the most dramatic one being during the later stages of *in utero* development. We see an impaired growth of the muscles concomitantly with a dramatic loss in the number of muscle progenitor cells, but no increase in cell death. Analysis of markers of myogenesis indicate that the absence of laminin 211 leads to precocious differentiation of muscle progenitor cells. Thus laminin 211-deficient mice are not only born with smaller muscles than their normal littermates, but their muscles also have fewer muscle progenitor cells available for muscle growth and repair.

These results are the first to describe defects in laminin 211-deficient mouse muscles before birth. Understanding the molecular mechanisms underlying the loss of progenitor cells in these muscles during development may help develop early intervention therapies which may one day increase the lifespan, improve clinical outcomes and quality of life for LAMA2-CMD patients.

**5ª feira, 19 de Novembro de 2015**

**FCUL (Edif. C6) – 12.00h-13.00h – Sala 6.2.51**

# Centres of moss diversity in southern Africa

## Nonkululo Phephu

PhD student, University of Witwatersrand, Johannesburg, South Africa

I am going to present one chapter of my PhD study, in which the geographical distribution dataset of most recent records was used to identify centres of moss diversity in southern Africa (South Africa, Namibia, Botswana, Lesotho and Swaziland). Five main, and five secondary centres of moss diversity are described, based on the number of moss species per  $\frac{1}{2}$  degree grid square in southern Africa. Although collecting bias is responsible for concentrations of species in many areas, moss diversity is strongly influenced by habitat heterogeneity and mean annual rainfall.

I am conducting a PhD study on the bryophyte flora of southern Africa, at the University of Witwatersrand, Johannesburg. The objectives of my project are to: identify centres of bryophyte (liverworts, mosses and hornworts) diversity and investigate colonization routes of taxa occurring in the north and south hemisphere, and to evaluate to which extent, patterns resulting from long-distance dispersal or from ancient vicariance respectively, are present in the southern African and Portuguese flora (including Macaronesia).

Sampling will take place on both regions but the use of the herbarium collections from several institutions will also support the acquisition of suitable material for the project development.

I work for the South African National Biodiversity Institute (SANBI) as a bryophyte taxonomist at the National Herbarium, Pretoria.

**5<sup>a</sup> feira, 26 de Novembro de 2015**  
**FCUL (Edif. C6) – 12.00h-13.00h – Sala 6.2.51**



# Reprodução *ex-situ* e genética populacional aplicadas à conservação de peixes de água doce nativos ameaçados

**Carla Sousa-Santos**

Investigadora pós-doc MARE-ISPA

Mais de 60% das espécies de peixes de água doce nativas de Portugal são consideradas vulneráveis, em perigo ou criticamente em perigo de extinção e o tamanho das suas populações tem vindo a sofrer um acentuado declínio nos últimos anos. As ameaças à sobrevivência destas espécies são variadas e suas as populações são geralmente sujeitas ao efeito cumulativo de várias destas ameaças. Para agravar a situação, as espécies mais ameaçadas têm áreas de distribuição geográfica muito restritas e, frequentemente, níveis de diversidade genética reduzidos. A preservação dos peixes nativos de Portugal deverá implicar uma abordagem integrada entre medidas de conservação *in-situ* e *ex-situ*, tendo igualmente em conta as especificidades genéticas das populações a salvaguardar.

**5ª feira, 22 de Outubro de 2015**

**FCUL (Edif. C6) – 12.00h-13.00h – Sala 6.2.51**

