Novel tools in cell and molecular biology: -induced pluripotent stem cells in the field of lysosomal storage disorders

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The lysosome is the centre of a group of rare inherited diseases commonly referred to as Lysosomal Storage Disorders (LSDs). LSDs have been long studied; presently, the main objective is to characterize their pathophysiology contributing to their understanding and treatment.
Our main commitment is to help providing the insight required for the development of more specific therapies.
Existing therapies for LSDs constitute a large financial burden, having a significant impact on health systems and family resources. Therefore, in order to progress with new therapies, one needs to first develop effective and economically viable models for testing specific therapeutic approaches.
With the advent of induced pluripotent stem cells (iPSCs) it became possible to establish cellular models for several diseases. In our lab we recently started generating iPSCs from a few lysosomal disorders.
This new tool allows easier access to disease specific cells, with the advantage of preserving the original genotype of the donor cells. The development of precise cellular models, in a non-fully dedicated lab, is a long process. The procedure requires several checkpoints involving specific techniques. The process will be briefly described and the applications of iPSCs will be discussed. With specific disease models, we hope to contribute to the increase of choices in terms of availability of material for developing new therapeutic interventions.

Team collaborators: Ana Joana Duarte, Diogo Ribeiro, Renato Santos, José Bragança
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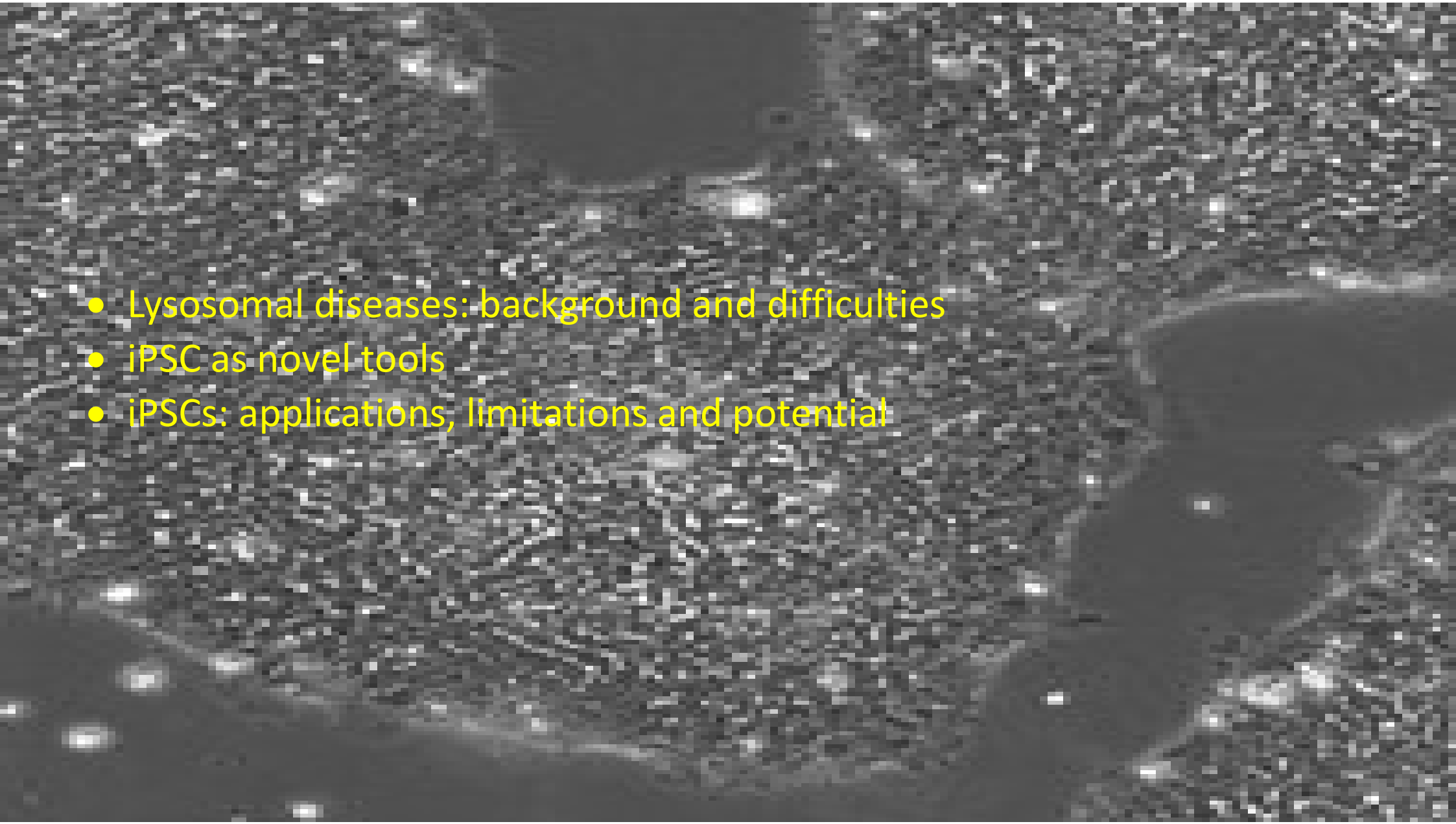
• Lysosomal diseases: background and difficulties
• iPSC as novel tools
• iPSCs: applications, limitations and potential

LSDs are often multysystemic

Lysosomal storage disorders (LSDs) frequently have **multi organ involvement**. The heart, the kidney, the brain, and other organs may be affected.

Future goals in LSDs

- The main objective is to characterize their pathophysiology contributing to their understanding and treatment.
- Our main commitment is to help providing the insight required for the development of more specific therapies.

Present collaborators

Olga Amaral
A. Joana Duarte
Diogo Ribeiro
Renato Santos
José Bragança
(UniAlgarve)

and

Meg Quint and other students
Liliana Matos
J. Inês Santos
Maria F Coutinho
Sandra Alves

Ongoing collaborations

Universidade do Algarve

Universidade do Porto

CECA, Centro de Estudos em Ciência Animal

Instituto Gaslini, Génova

Universidade do Porto_and others

Thank you!

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Convite para o evento comemorativo do dia das doenças raras

Local: INSA-Porto, Rua Alexandre Herculano 321

Dia: 28 de fevereiro 2020

Transmissão simultânea em videoconferência com INSA-Lisboa, informações e programa em https://www.rarediseaseday.org/event/portugal/89

Inscrições, por razoes logísticas, em http://www.insa.min-saude.pt/events/dia-mundial-das-doencas-raras/

Obrigada!

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