

A first step to open the neuronal box of neural progenitor cells

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Introduction

- In the Portuguese population, lysosomal storage diseases (**LSDs**) have a prevalence of 1/1000 (1);
- Gaucher disease (**GD**) is a common LSD usually associated with defective activity of β -glucosylceramidase (encoded by *GBA1*, OMIM#606463);
- Cellular models using human induced pluripotent stem cells (**hiPSCs**) reprogrammed from patient fibroblasts are excellent disease models, presenting the advantage of preserving the patient's genetic background and being able to differentiate them in almost all cell types (2);
- In the case of **GD** it is very important, because we can differentiate **hiPSCs** into cells of the nervous system and study the neuronal involvement in the disease.

Aim

- This work focuses on the differentiation and gene expression characterization of neurons from hiPSCs reprogrammed from type 3 GD (**GD3**) fibroblasts.

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(2) Anderson, R.H., Francis, K.R. (2018) Modeling rare diseases with induced pluripotent stem cell technology. *Mol Cell Probes* Jan5. pii:S0890-8