Neuroinflammation and pain in a mouse model for Fabry disease

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Lysosome are small organelles in the cell and famous for digesting down different kinds of biomolecules, thereby acting as the waste disposal system in the cell. This whole process requires a large machinery with more than 60 enzymes (biocatalysers) to digest all possible types of molecules. If there is a dysfunction in any of these enzymes, the respective molecule cannot be degraded and therefore accumulate.

This is exactly what happens in Fabry Disease. The α-galactosidase A enzyme does not work properly, causing accumulation of a molecule known as globotriaosylceramide (Gb3) within the lysosomes. Dysfunction of this enzyme and the increased metabolite Gb3 affects both the peripheral and the central nervous system, skin, gastrointestinal system and cardiovascular system. Since the information of how to build the enzyme is located on the X-chromosome Fabry disease is hereditary and men are more likely to develop it than women.

There are still many gaps in our knowledge about the processes leading to clinical Fabry disease. Currently, we know that several organs including the brain show Gb3 deposits. However, we do not know how Gb3 is produced, what cell types are key for Gb3 production or if all the cells in the body are able to accumulate Gb3. My main aim in this research project is to explore the contribution and interactions of the nervous system and the immune system in Fabry disease, searching for the possible source of Gb3 accumulation and analyzing the effect of this deposition.

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