

Congenital Disorders of Glycosylation: Why, How & Where sugars matter!

Our bodies are made up of more than a hundred billion (100,000,000,000,000) cells. In turn, cells are formed by several different types of building blocks (molecules), being proteins, sugars and fats (lipids) some of the most important and commonly found. Also, our bodies are made of different cell types, for example heart cells are different from blood cells.

How is that possible? Well, different cells are constituted by distinct building blocks. One of the mechanism that leads proteins and lipids to have different functions are attached-sugars. Indeed, **sugars** (or glycans) are added to proteins and fats in a process called **glycosylation**, thus altering their function and allowing them to do their work within the cell.

How often are proteins and fats modified with sugars? Actually, it is estimated that around **50%** of all our bodies' proteins are modified with sugars.

So, what happens when the mechanism(s) responsible for adding sugars to proteins and/or fats don't work properly? When the mechanisms fail to perform their role, proteins and/or fats can either miss some or all the sugars necessary for them to do their proper job in their cell/tissue/organ. Consequently, the cell/tissue/organ starts to malfunction and disease appears.

These are the underlying causes that explain how **Congenital Disorders of Glycosylation (CDG)**, a family of rare, inherited metabolic disorders with 133 different forms, occur.

CDG are usually **complex and multi-systemic disorders**, meaning that patients frequently have more than one organ affected. **Why is that so?** Various organs are affected in CDG patients, because proteins/fats modified with sugars are present and necessary in almost every organ.

To help understand how defects in the mechanisms that attach sugars to proteins/fats affect each organ, the international research network **CDG & Allies – Professionals and Patient Association International Network (CDG & Allies-PPAIN)** has been compiling medical and scientific information in literature reviews, but has also been developing a **patient-centred research approach**, addressing **clinical, psychological** and social concerns.

In this series of articles, we review the affectation of the:

- 1) **Immune System** (the defence system we have in our bodies that fights bacteria, virus (microorganisms);
- 2) **Heart**
- 3) **Liver** (through a revision of literature and data collection using a patient/caregiver clinical questionnaire)
- 4) **Eyes**

Also, a think tank (an idea factory) research project involving several stakeholders of the CDG community to establish a

- 5) **[People-centred approach to care and research in congenital disorders of glycosylation](#)**

You can access the articles' abstracts [HERE](#).

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