

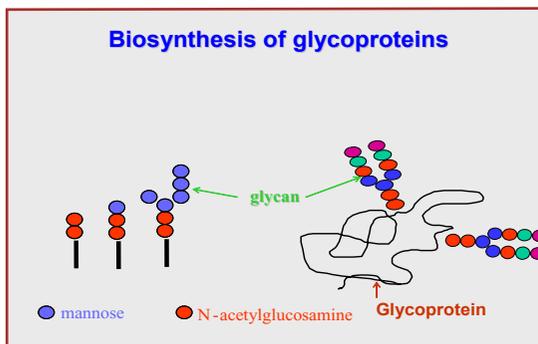
## CONGENITAL DEFECTS OF GLYCOSYLATION (CDG)

### 1. What are congenital defects of glycosylation?

They are genetic errors **in the construction of the sugar part of glycoproteins.**

### 2. What are glycoproteins?

Proteins are composed of long amino acid chains with characteristic spatial shapes that determine their function. A great number of proteins, called **glycoproteins**, are **linked to sugar chains**, which increase their stability, determine its final shape, facilitate their interaction with other proteins and also the differentiation and development of cells. These sugar chains are called **glycans**. Many of them have similar composition and are synthesised in the same way: different sugars are sequentially added to acquire a branched form which is transferred to the protein only when it is correctly completed, that is, when all the sugars are correctly linked.



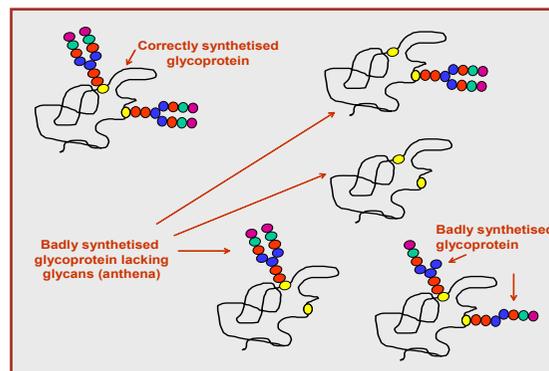
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### 3. What does a metabolic error mean?

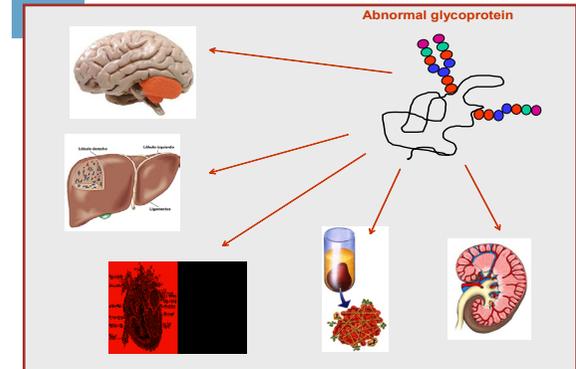
When there is an error in glycoprotein metabolism, one of the reactions leading to the linkage of sugars to proteins does not take place correctly. This affects the final composition of multiple glycoproteins.

### 4. What happens in inborn errors of glycosylation?

Inborn defects of glycosylation are errors in glycoprotein synthesis. They may be caused by the deficiency of any of the proteins (enzymes or transporters) capable of transporting and supplying different sugars in a certain order to the glycan chain that has to be linked to multiple proteins of our body. Since the resulting glycoproteins have different functions and localizations within the cells, every defect in the glycan synthesis will affect multiple proteins simultaneously and cause



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a multisystemic disease, which may involve different organs and systems of the human body.

### 5. Why do we say that these errors are congenital?

We say that these errors are congenital because patients are born with them since they are hereditary. Each of the metabolic reactions involved in the synthesis of the compounds that constitute our body is genetically determined (codified). We all inherit from our parents the right or wrong information that will determine how every metabolic reaction takes place. If we inherit an erroneous or partially altered information, the involved metabolic step will function in a wrong way and this may cause an **inborn error of metabolism, that is, a metabolic disease.**

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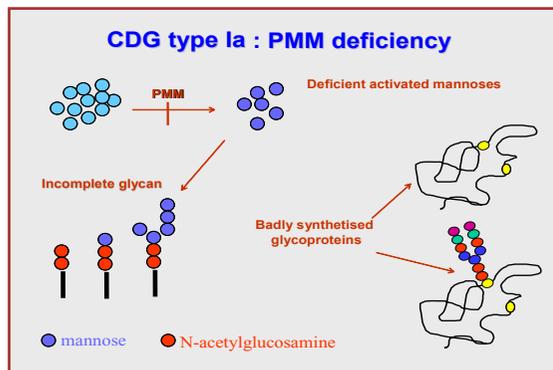
### 6. What happens when a baby is born with a congenital defect of glycosylation?

The baby can already be born with some problems, since the altered glycoproteins can be multiple and diverse and so the life processes may also be altered. However, the clinical presentation and evolution of the baby can be very different depending on the origin of the defect and on its severity.

### 7. How can we diagnose these diseases?

The diagnosis is based on the observation of a number of signs and symptoms of the disease (abnormal distribution of body fat, strabismus, etc...together with failure to thrive, and other multiorganic symptoms, especially neurological).

There are some biochemical markers (the study of abnormal serum transferrin profiles) which are useful to diagnose the diseases after the clinical suspicion. These markers



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also allow the differentiation between the different types of the disease. However, the most frequent defect is the CDG type Ia. It is caused by **deficient activity of the enzyme phosphomannomutase (PMM)**, which is needed to supply a sugar, **mannose**, to the glycoproteins in the synthetic process. The confirmation of the glycosylation defect allows the genetic counselling to the families and the prenatal diagnosis if required.

### 8. Is there a treatment for CDG?

At present there is only treatment for some CDG types which are not very common (CDG Ib, IIc). However, a lot of research is in progress all over the world in order to achieve effective treatment for the most common types of the disease. In any case, supportive treatments are available to ameliorate the quality of life of the patients.

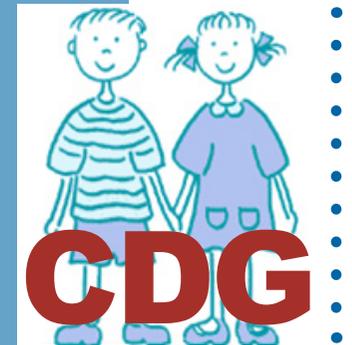


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INBORN ERRORS OF METABOLISM FOLLOW-UP UNIT.  
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