Congenital Disorders of Glycosylation (CDG)

CDG is a growing group of diseases among the 8,000 known rare diseases.

NORMAL GLYCOPROTEIN
The correct sugar transfer to proteins or lipids is essential for their biological function.

INCOMPLETE SUGAR ANTENNAS
This disruption leads to:
- A great variety of more or less severe symptoms;
- Involvement of nearly every organ.

PREVALENCE
Since many cases go unrecognized or misdiagnosed, it is difficult to determine their true frequency.

PMM2-CDG (CDG-ia) is the most common.
The prevalence may be as high as 1 in 20,000.
Childhood mortality: 15-25% in the first two years of life due to severe infections or organ failure.

DIAGNOSIS

CAUSES
Most forms of CDG are inherited as autosomal recessive diseases. This means that two changed copies of a gene are inherited—one from each of the parents. People, with one copy of the gene alteration do not have the condition, and therefore are an unaffected carrier.

TREATMENT
Some forms can be treated specifically, but most forms can not be treated specifically yet.

IMPACT ON PATIENTS AND FAMILY MEMBERS
CDG children and adults are very often hospitalized.
Patients need an interdisciplinary team of professionals.
Many patients need support for almost all daily activities.

CDG are clinical chameleons. Specific symptoms and severity can vary even among individuals with the same subtype and even among members of the same family.

BUT... THERE IS HOPE!
International collaboration between affected families, clinicians and researchers foster and enhance research in this field.

Patient Advocacy Groups are addressing CDG community needs. Many fruitful resources for families and professionals have been elaborated by the Portuguese Association for CDG (APCDG) in collaboration with family members and professionals. The resources are available at apcdg.com

www.apcdg.com