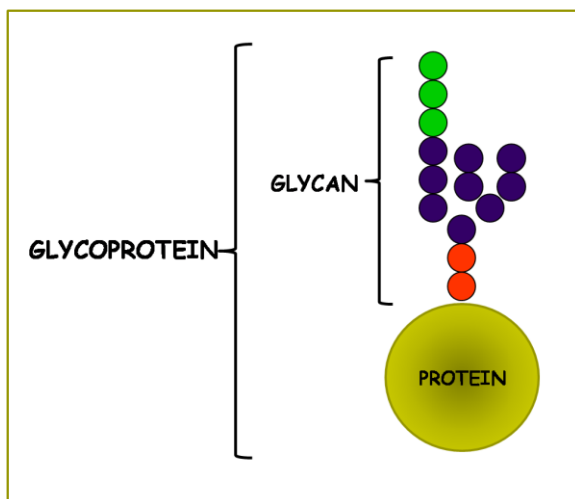


WHAT ARE THE CONGENITAL DISORDERS OF GLYCOSYLATION?

Congenital disorders of glycosylation are a rapidly growing family of inherited disorders affecting the assembly or processing of glycans on glycoconjugates.

WHAT IS GLYCOSYLATION?

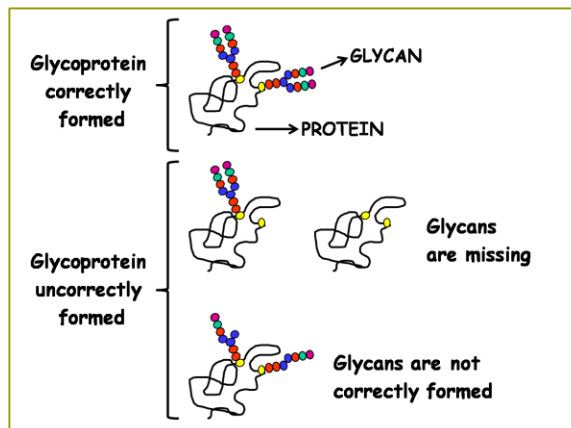
Glycosylation is the enzymatic process that links saccharides to produce glycans, attached to proteins, lipids, or other organic molecules. More than 100 enzymatic steps are involved in the glycosylation pathway.



Adapted from: Errores Congénitos do Metabolismo. Guia divulgativa. Hospital Sant Joan de Déu Barcelona.

WHAT HAPPENS IN THE CASE OF CDG DISORDER?

In patients with CDG, one of these many glycosylation enzymes in the pathway is deficient. All these diseases have in common that a poorly formed glycoprotein with sugar chains missing or incorrect sugar chains attached, can no longer function in the proper way.



Adapted from: Errores Congénitos do Metabolismo. Guia divulgativa. Hospital Sant Joan de Déu Barcelona.

HOW MANY DIFFERENT CDG DISORDERS HAVE BEEN REPORTED?

Nowadays, the CDG field is expanding rapidly. Forty six disorders are currently known: 16 in protein **N-glycosylation**, 11 in protein **O-glycosylation**, 3 in lipid glycosylation and 16 in multiple glycosylation pathways.

HOW IS IT POSSIBLE TO DETECT A CDG CASE?

The CDG can lead to a variety of symptoms affecting multiple systems. In fact, there are different clinical symptoms, findings of diagnostic procedures, and laboratory investigations that might be associated with CDG. The **symptoms** present in patients with inherited glycosylation disorders are summarized:

- Psychomotor retardation
- Stroke-like episodes
- Failure to thrive
- Axial hypotonia

- Strabismus
- Cerebellar hypoplasia
- Dysmorphism (fat pads, inverted nipples)
- Liver disease
- Coagulopathy
- Protein-losing enteropathy

In **adolescence** or **adulthood**, the **symptoms** may include the following clinical features:

- Neurologic evidence of cerebellar dysfunction (ataxia, dysarthria, dysmetria)
- absent puberty in females, small testes in males
- retinitis pigmentosa
- progressive scoliosis
- joint contractures
- peripheral neuropathy with or without muscle wasting

WHICH DIAGNOSTIC PROCEDURES ALLOW THE IDENTIFICATION OF THIS DISEASE?

The common diagnostic test for CDG is **isoelectric focusing (IEF) of serum transferrin**. Although the IEF of transferrin is still the best screening test for CDG, not all CDG types can be detected in this way. There are other diagnostic tests such as: the **thin-layer chromatography of urine** the **functional antithrombin III (AT III) test**.

WHO SHOULD BE SCREENED FOR CDG?

Currently, it is impossible to devise a general guideline around who should be screened for CDG and who should not. Nevertheless it is advisable to screen any child with psychomotor retardation, unexplained multisystem disease,

autoimmune disorders and chronic inflammatory disease, cardiomyopathy, protein-losing enteropathy, cyclic vomiting, anemia with reduced osmotic resistance of the erythrocytes, persistent leukocytosis or low AT III levels for CDG.

IS THERE A TREATMENT FOR CDG SYNDROME?

Only CDG-Ib and CDG II-c are treatable.

SOME CDG EXPERTS AROUND THE WORLD ARE:

Dr Jaak Jaeken (*Faculty of Medicine at the University of Leuven, Belgium*)

Dr Gert Matthijs (*Laboratory for Molecular Diagnostics at the Center for Human Genetics in Leuven, Belgium*)

Dr Hudson Freeze (*Burnham Institute, USA*)

Dr Donna Krasnewich (*Medical Genetics Branch National Institutes of Health, USA*)

Dr Belén Pérez Dueñas (*Hospital Sant Joan de Déu. Esplugues de Llobregat. Barcelona*).

Dr Rafael Artuch (*Hospital Sant Joan de Déu. Esplugues de Llobregat. Barcelona*).

Dr Paz Briones (*Institut de Bioquímica Clínica. Corporació Sanitària Clínic i CSIC. Barcelona. España*)

Dr Célia Pérez-Cerdá (*Centro de diagnóstico de enfermedades moleculares, Universidad Autónoma de Madrid*)

Dr Laura Vilarinho (*Institute of Medical Genetics Jacinto de Magalhães, Portugal*)

Dr Dulce Quelhas (*Institute of Medical Genetics Jacinto de Magalhães, Portugal*)

HOW CAN THE PORTUGUESE (APCDG-DMR) AND SPANISH (AESCDG) ASSOCIATION FOR CDG SYNDROME HELP YOU?

The **APCDG-DMR** and **AESCDG** association are non-profit organizations founded by parents seeking information and support for a group of disorders known as Congenital Disorders of Glycosylation. We want to help people affected with the CDG syndrome. We aim to do it by:

- Identifying people with CDG syndrome and promoting the exchange of information between families and physicians,
- Offering practical help, friendship and support to people living with CDG syndrome, including individuals, families and other carers and supporters,
- Providing high quality up to date information about best practice in treatment and care,
- Promoting greater understanding and awareness of CDG syndrome amongst the general public and amongst medical professionals.

HOW CAN YOU CONTACT AND/OR HELP THE APCDG-DMR OR AESCDG?

The **APCDG-DMR** Association

Attn: Vanessa Ferreira, President

Email: sindromecdg@gmail.com

Webpage: <http://sindromecdg.orgfree.com/>

Facebook:

<http://www.facebook.com/pages/SINDROME-CDG/135220796488836?ref=search>

The **AESCDG** association

Attn: Cristina Lanz, President

Email: aescdg@ono.com

Webpage: <http://webs.ono.com/aescdg/>

Facebook: AESCDG

You can help us becoming a member or making a donation. For more information contact us!

Thanks for helping us!



Congenital Disorder of Glycosylation

(CDG, carbohydrate-deficient glycoprotein syndromes)

“because a large number of enzymes, transporters, and other proteins are involved in glycosylation, it is expected that the great majority of CDG are yet to be identified. –

Jaak Jaeken, discovered the first CDG cases in the 80's”

Author: Vanessa Ferreira (President for the Portuguese CDG Syndrome and other metabolic rare diseases Association)