

# Immunology CDG Questionnaire (ImmunoCDGQ) for CAREGIVERS & PATIENTS

## Informed Consent – EXTENDED VERSION



This is a patient-centric research tool developed to promote understanding, guide research, improve care & management of immunologic involvement among Congenital Disorders Glycosylation (CDG) Patients

This study is part of a research project entitled “*Deciphering immunological aspects of Congenital Disorders of Glycosylation (CDG) using a multidisciplinary approach*”. This is the PhD thesis of the researcher Rita Francisco (more about her [HERE](#)).

Before deciding to participate or not participate, it's important that you understand **why this study is being done, who is conducting it, what are its the aims, how you can benefit from it and potential associated risks.**

By agreeing to participate, you are agreeing with the terms and conditions explained in this document.

## Instructions and Informed Consent for Participants

### 1) About the questionnaire

Glycans (“sugar trees”) play several roles in the immune system. **Congenital disorders of glycosylation (CDG)** are a family of **rare metabolic diseases**, caused by alterations in the glycosylation machinery. CDG are usually multi-systemic, **being immune system involvement** reported and described in a subset of CDG types.

However, literature reports are little detailed and scarce.

Additionally, the underlying molecular mechanisms and the extent of the associated clinical manifestations are highly unknown.

To **gather information** and **build knowledge**, **identify the priorities of CDG families and patients**, as well as to **guide future research projects**, we have developed a **questionnaire online tool** to be administered to CDG patients and caregivers.

This questionnaire focuses on the **physical, functional aspects** as well as on the **impact on quality of life of immune system dysfunction among CDG patients** by assessing main signs, symptoms, and management of immunological alterations.

The questionnaire aims to gather information and build knowledge on the following topics:

- 1) **General CDG immune-related problems, in order to have an overview of the existing immunological problems;**
- 2) **Infections** to understand frequency, type, severity of infections occurring in these patients;
- 3) **Autoimmune diseases** to elucidate the types, frequency and clinical relevance of these conditions among CDG patients;
- 4) **Allergies** to understand the prevalence, types and importance of allergies in this patient population;
- 5) The impact of immune problems on the patients' **Quality of Life**;
- 6) **Vaccination history** to better comprehend the efficacy, side-effects and types of vaccines administered to these patients;
- 7) **CDG symptom checklist** to understand the overall phenotype presentations of CDG patients and compare differences among patients with and without immune system involvement;
- 8) **Awareness and information about immunological affectation in CDG** to generally assess caregivers/patients' knowledge, needs, and management strategies regarding immune system involvement.

Since CDG comprises a family of very rare disorders with **patients geographically dispersed**, to **maximize participation**, and **gather robust** data this questionnaire will be administered online through the platform **Survey Monkey** (<https://www.surveymonkey.com/>).

This questionnaire overcomes substantial obstacles in traditional clinical research by:

- Addressing rare diseases;
- Prioritizing the views of patients and family members;
- Identifying the impact of immunologic dysregulation;
- Improving the standards of care for CDG patients;
- Guiding research projects based on the needs of families and patients.

Also, the questionnaire will be made available in the following **LANGUAGES**:



## 2) What was the questionnaire based on?

The construction of the questionnaire was based on the revision of literature performed by our team and published in 2016 – See [HERE](#)

Also, some questions were inspired by existing immunology questionnaires, namely questionnaires used upon hospital admission or during a doctor's appointment. Additionally, existing validated tools assessing

immunological manifestations, such as the **Comprehensive Early Childhood Allergy Questionnaire (CECAQ)**, **Acute Respiratory Tract Infection Questionnaire (ARTIQ)**, the **Rhinoconjunctivitis Quality of Life Questionnaire (RQLQ)** the **immune system assessment questionnaire (ISAQ)**, were consulted and served as templates either in terms of the structure and/or content of some of the questions present in this questionnaire.

### 3) What are the goals of this questionnaire?

#### General aims:

- To increase knowledge and understanding of the immune system (dys)function in Congenital Disorders of Glycosylation (CDG) patients;
- To develop a web-base, international, patient-centered questionnaire as a reporting tool to assess immune dysfunction in patients with CDG;
- To engage patients from a wide geographical area. The primary rare diseases of interest are CDG, although other rare and common diseases can be included;
- To be a pillar for the future development and use of a specific app for the follow-up of patients, aligning with a major objective of the European Reference Networks ([ERNS](#)). Importantly, such app would allow the collection of data necessary for the natural history of the different types of CDG;
- To improve care and management of immunologic involvement among CDG patients and facilitate early diagnosis and prompt treatment;
- To boost awareness and dissemination of information among families and healthcare key opinion leaders (KOLs);
- To provide a lobby tool for better access to existing or future symptomatic/basic treatments.

#### 3.2. Specific aims:

##### **Medical/Research Objectives**

- To determine the frequency and impact of immunologic problems (infections, allergy, atopy and/or autoimmune diseases) among CDG patients and families, through a questionnaire, and to compare these findings with clinical and published evidence;
- To develop a questionnaire with a high level of caregivers and patients' engagement that improves understanding of immunologic aspects of CDG;
- To propose this study as a model to be used worldwide, in additional CDG-related studies and/or in other diseases;
- To create/ improve guidelines for diagnosis, prevention, management and treatment of immunologic problems among the CDG population.

##### **Lobby/policies/community building objectives**

- To engage patients/families throughout the entire research project, including in the development of the questionnaire;
- To publish the study and its results at international level, to contribute to the implementation of cross-border healthcare directives, and better outcomes for CDG patients;

- This project can be used as a model worldwide, in additional CDG-related studies and/or in other diseases, because the major conclusions and recommendations will be applicable internationally;
- To empower CDG families and patients, whilst promoting and strengthening the sense of community and reinforcing the relationship between patients and professionals.

#### 4) Who is the target population?

**ALL CDG Patients – With or without known immune system affection**

Whether or not you (the CDG patient) have experienced immune system-related signs/symptoms, this questionnaire gives you the opportunity to report your experience. **Questions are adapted automatically according to your responses at key questions.** Generally, no in-depth knowledge is required to complete this questionnaire, questions are about your experience with immunologic symptoms as a CDG patient or caregiver. **At specific questions it is indicated that you should consult the CDG patient last blood and genetic test results to help you answer.**

#### 5) How much time will it take me to complete this survey?

The survey should take about **30 to 40min** to complete (15-20min if the patient reported doesn't have immune problems).

NOTE THAT you don't have to fill in the entire questionnaire all at once. You can stop at any point, how many times you need and finish it when it's convenient to you as long as it is done within the deadline of the project. To ensure that you can return to the question you were at when you left the survey, **MAKE SURE** you use the **SAME DEVICE** (e.g computer or phone) and **INTERNET BROWSER** (e.g. Google Chrome).

#### 6) Can I participate?

You can participate if you are a family member/caregiver of someone suffering from CDG or if you are a CDG patient yourself (in both cases you need to be 18 years old or older).

**Participation is voluntary**, no one can force you to participate against your will. You will not be harmed or hurt in anyway (medically, socially) if you decide not to participate. Only if you consent to participate will you join this study.

#### 7) What do I have to do?

If you are a **CDG patient or family member over 18 years-old**, all you need to do is **answer truthfully** and in accordance with your experience and medical diagnosis to the questions made in this questionnaire.

## Procedure and Protocol

The questionnaire will be made available online through the Survey Monkey (<https://www.surveymonkey.com/>) platform. This questionnaire will also be available in several languages (English, Portuguese, Spanish, French, Italian), so that each participant can choose to answer it in their mother tongue or in the language he/she feels more at ease with.

**The questionnaire is divided in 10 major sections:**

- 1) Stage 1 – Instructions and informed consent for participants
- 2) Stage 2 - Patient Information
- 3) Stage 3 - CDG Immunology checklist
- 4) Stage 4 – CDG immune problems
- 5) Stage 5 - Infection history
- 6) Stage 6 - Autoimmune history
- 7) Stage 7 – Allergy history
- 8) Stage 8 – Quality of Life
- 9) Stage 9 – Vaccination history
- 10) Stage 10 - CDG Symptom checklist

### **A) Risks associated with your participation**

**None**, as all data given by you is treated confidentially and collectively. No interventive measures will be done.

### **B) Benefits associated with your participation**

*The extent, severity, frequency and clinical manifestations of immune system-related problems in CDG patients are widely unknown. By sharing your information with us, we will be able to:*

- 1) Identify the main immunological problems;
- 2) Determine the impact of immunological problems on the patients' quality of life;
- 3) Define future research priorities and design research tasks accordingly;
- 4) Develop more accurate and tailored care guidelines;
- 5) Collect robust data in a reliable and fast manner;
- 6) Consolidate and validate patient-centered research methodologies;
- 7) Improve the care of CDG Patient.

### **C) Will my information be published?**

The survey is **anonymous, and the data will be treated collectively for publications purposes.**

**The survey is anonymous. No personal information will be published.**

**Your participation is protected by the applicable laws of data protection.** Under the scope of this study data will be gathered in an anonymous manner, as the name of the participant will not be collected. Multiple entries

from the same individual will be avoided by selecting the SurveyMonkey option “the questionnaire cannot be answered several times from the same device”.

**Personal data collection abides the Law of Personal (nº67/98 from 26<sup>th</sup> October).**

In case your data is published, it will be treated in the same way, i.e., without it being possible to identify you.

## D) Who is developing this project?

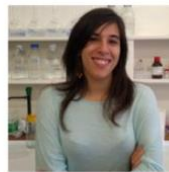
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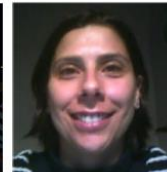
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## Family advisory board of reviewers:



**Barbara Vulso**



**Meral Omurtag**



**Merell Liddle**



**Rana Atwi**



**Tatiana Rijoff**

## E) Remuneration of the research team

The team responsible for the development of this questionnaire is not receiving any payment for this research work. Rita Francisco is a PhD student, who has been awarded a research scholarship to perform her research project, in which this questionnaire is the first task.

## F) Freedom to withdraw from the study

The participation in the study is entirely voluntary. At any stage of the questionnaire you can stop filling it out. You don't need to justify your decision and that won't affect your treatment in anyway.

## G) Study results communication and dissemination

The collected data in this study will be treated collectively, and anonymously. It's planned to be:

- Presented at targeted conferences/congresses;
- Published in a peer-reviewed journal;
- Made available on the CDG & Allies-PPAIN website <http://www.researchcdg.com>;

## Patient Information

**Before you start the questionnaire, please....**

**Make sure you have the LAST/RELEVANT BLOOD TEST and GENETIC TEST RESULTS with you**

**Download the mini & thematic GLOSSARIES of scientific terms explained in Lay-language**  
[HERE](#).

## All the questions asked in this questionnaire are related to the CDG patient.

- If you are a **caregiver**, the following questionnaire asks several questions about the CDG patient that is your relative or for whom you are responsible. Your experience as a caregiver is unique and it can be useful to other families and health professionals working with CDG.
- If you are a CDG **patient**, your experience as a CDG patient is unique and it can be useful to other patients, families and health professionals working with CDG.
  - Please answer all questions based on what you believe is the most appropriate answer and on your experience as a caregiver (direct observation of the patient behaviour and reactions) or as patient (your direct experience).
  - For some questions you may want to rely on the information you have received from the treating physician. You may want to ask your doctor about some of the results or questions in this survey, or for helping you to answer some of the questions.

## Contacts

### If I have any doubts to whom do I talk?

If you have any questions while taking part in this questionnaire, you can contact [rab.francisco@campus.fct.unl.pt](mailto:rab.francisco@campus.fct.unl.pt) or phone number (+351) 925718128

## References

- Monticelli M, Ferro T, Jaeken J et al (2016) Immunological aspects of congenital disorders of glycosylation (CDG): a review. JIMD. (6):765-780.
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