

CDG JOURNEY MAP SURVEY FROM PEOPLE LIVING WITH CDG AND THEIR CAREGIVERS VIEWS

(FAMILIES VERSION)

INTRODUCTION

From the perspective of “community-centered” healthcare, mapping experiences from people living with a certain condition throughout their journey is critical to improve the healthcare pathway.

OBJECTIVE OF THE SURVEY

The present study corresponds to part 2 of an international study aiming to capture the full picture of the people living with a Congenital Disorder of Glycosylation (CDG) from different perspectives: the ones living with CDG, their caregivers and the healthcare professionals involved. This survey has the specific goals:

- To describe the experiences during the quest for CDG diagnosis, and identify CDG-related information needs throughout the CDG journey (since diagnosis)
- To map the level of awareness about the development and dissemination of Clinical Guidelines (CGs) for CDG,
- To collect experiences on participation in CDG clinical research
- To identify current support measures and gaps given by Worldwide CDG patient groups,
- To assess digital solutions tailored for the CDG community.

Why this study?

Our research team has identified a major gap in current CDG knowledge. The true CDG journey is still unknown. We have all heard about family's experiences, firsthand narratives, and medical reports that emerge related to the quest for diagnosis and the struggle for appropriate care. But to our knowledge no study has systematically collected the CDG journey. Mapping CDG experience along the entirety of their disease pathway, from initial signs and symptoms through diagnostics to first definitive treatment, enables a greater understanding of the efficiency of care delivery and identifies opportunities for improvement or, in the case of CDG management, any points that require further clarity or additional interventions.

DISSEMINATION STRATEGY OF THE RESULTS

- Be published in different formats to allow their use for advocacy and policy making at international levels (we plan to publish an article, fact sheets and infographics).
- Better define the need for resources, and allow for better tailored initiatives for people who live with CDG, healthcare professionals or stakeholders' individual needs, helping the person who lives with a certain disease experience and leading to improved individuals outcomes.

DISCLAIMER

If you need any help or additional information about the questionnaire or clarifications on the content, please do not hesitate to contact us at <https://worldcdg.org/contact>, where the authors of this study Paula Videira or Vanessa dos Reis will be available to schedule a SKYPE, WhatsApp CALL, or Zoom meeting.

Can the survey be saved and finished later?

YES. Participants can fill in part of it and then return to it to finish it, using the same device (e.g. mobile phone, laptop) and internet browser. To return to the previous question, please hit the PREV BUTTON, instead of the back button from the internet browser.

Clicking on the "agree" button below indicates that:

- You voluntarily agree to participate;
- You are at least 18 years of age and a person living with CDG or a CDG relatives/caregiver;
- The estimated time to complete the whole survey is 38 minutes;

You understand that the responses to the survey are anonymous and that no information will be collected or processed that allows your identification or that of your relatives.

THEME - PARTICIPANT RELATIONSHIP WITH CDG

1. Please indicate your choice:

☐ Agree (Q2)

☐ Disagree (End of the Survey)

2. Questionnaire completed by (please choose in which capacity you want to answer this questionnaire. If more than one choice qualifies, please check the most appropriate).

- ☐ A person living with CDG
- ☐ A CDG relative (e.g. mother, father, sibling, grandparent)
- ☐ A family caregiver for someone who lives with CDG

3. Do you have multiple children/relatives living with CDG?

☐ Yes (Q16)

☐ No (Q4)

THEME - THE JOURNEY FROM FIRST SIGNS TO A FINAL DIAGNOSIS

4. Which CDG type affects you/your child/children (e.g. PMM2-CDG or CDG Ia, PIGA-CDG, PGM1-CDG, MPI-CDG or CDG Ib and so forth) ?

5. If known, indicate the gene mutation, from the person living with CDG . Note: If you do not know, please write "I don't know".

6. At which age did the CDG symptoms or signs first manifested?

- | | | |
|--|---|---|
| ☐ In the womb (Before Birth) | ☐ 10 - 12 months | ☐ 18 - 21 years old |
| ☐ < 3 months | ☐ 1- 3 years old | ☐ > 21 years old |
| ☐ 3 - 6 months | ☐ 4 - 9 years old | ☐ I don' know |
| ☐ 7 - 9 months | ☐ 10 - 17 years old | |

7. What were the main CDG presenting signs and symptoms (refer to the manifestations that **first led** you or your clinician to **suspect something** was wrong)? **(Please select all that apply)**

- | | | |
|--|---|---|
| ☐ Low muscle tone or floppiness | ☐ Bone manifestations | ☐ Strabismus |
| ☐ Poor growth | ☐ Abnormal brain imaging | ☐ Seizures |
| ☐ Failure to thrive | ☐ Abnormal lab tests | ☐ Stroke-like episodes |
| ☐ Developmental disabilities | ☐ Poor night vision and loss of peripheral vision | ☐ Heart problems |
| ☐ Liver disease and/or with elevated liver enzymes | ☐ Recurrent infections | ☐ Ataxia |
| ☐ Abnormal bleeding or blood clotting | ☐ Feeding problems | ☐ Slurred speech (dysarthria) |
| | | ☐ Other |

8. Concerning the doctors consulted, please tell us

How many doctors (approximately)...

(please select the most appropriate option)

RATING SCALE

1 - 2 3 - 5 6 - 10 11 - 20 > 20 I don't know

were consulted when you SUSPECTED the FIRST SIGNS AND SYMPTOMS of CDG?

☐ ☐ ☐ ☐ ☐ ☐

Did you consult between the FIRST MANIFESTATIONS and the FINAL CDG diagnosis?

☐ ☐ ☐ ☐ ☐ ☐

9. How much time (approximately) did it take to get a FINAL CDG diagnosis? (Please select the most appropriate option)

- | | | |
|-------------------------------------|-----------------------------------|-------------------------------------|
| ☐ < 3 months | ☐ 1 - 3 years | ☐ 10 - 20 years |
| ☐ 3 - 6 months | ☐ 4 - 5 years | ☐ > 20 years |
| ☐ 7 - 12 months | ☐ 6 - 9 years | ☐ I don't know |

10. Was the person with CDG given any other diagnoses, before the FINAL diagnosis was made?

- ☐ Yes (Q11)
- ☐ No (Q12)
- ☐ I don't know (Q12)

11. Which diagnoses were given before the final diagnosis?

12. Do you believe that the CDG FINAL diagnosis could have been made earlier?

- ☐ Yes
- ☐ No
- ☐ I don't know

13. Concerning the medical specialties involved, please tell us the **FIRST to raise the possibility of a CDG diagnosis**

- | | | |
|--|--|--|
| ☐ Geneticist | ☐ Obstetrician | ☐ Gastroenterologist |
| ☐ Neurologist | ☐ General practitioner | ☐ Ophthalmologist |
| ☐ Paediatrician | ☐ Cardiologist | ☐ I don't know |
| ☐ Metabolic specialist | ☐ Endocrinologist | ☐ Other (please specify) |

14. Concerning the medical specialties involved, please tell us the **specialist who gave the FINAL diagnosis**

- | | | |
|--|--|--|
| ☐ Geneticist | ☐ Obstetrician | ☐ Gastroenterologist |
| ☐ Neurologist | ☐ General practitioner | ☐ Ophthalmologist |
| ☐ Paediatrician | ☐ Cardiologist | ☐ I don't know |
| ☐ Metabolic specialist | ☐ Endocrinologist | ☐ Other (please specify) |

15. Did you seek a second opinion to confirm the FINAL CDG diagnosis?

- ☐ Yes (Q31)
- ☐ No (Q31)
- ☐ I don't know (Q31)

MULTIPLE CDG RELATIVES

16. Please specify your kinship with the patients (e.g mother, father, cousin, sibling) and how many of your relatives live with CDG (e.g 2, 3, etc).

17. Which CDG type affects you/your child/children (e.g. PMM2-CDG or CDG Ia, PIGA-CDG, PGM1-CDG, MPI-CDG or CDG Ib and so forth) ?

18. If known, indicate the gene mutation, from the person living with CDG . Note: If you do not know, please write "I don't know".

DISCLAIMER: Please answer the following question only regarding your first relative diagnosed with CDG

19. At which age did the CDG symptoms or signs first manifested?

- | | | |
|--|---|---|
| ☐ In the womb (Before Birth) | ☐ 10 - 12 months | ☐ 18 - 21 years old |
| ☐ < 3 months | ☐ 1- 3 years old | ☐ > 21 years old |
| ☐ 3 - 6 months | ☐ 4 - 9 years old | ☐ I don' know |
| ☐ 7 - 9 months | ☐ 10 - 17 years old | |

20. What were the main CDG presenting signs and symptoms (refer to the manifestations that **first led** you or your clinician to **suspect something** was wrong)? (**Please select all that apply**)

- | | | |
|--|---|---|
| ☐ Low muscle tone or floppiness | ☐ Bone manifestations | ☐ Strabismus |
| ☐ Poor growth | ☐ Abnormal brain imaging | ☐ Seizures |
| ☐ Failure to thrive | ☐ Abnormal lab tests | ☐ Stroke-like episodes |
| ☐ Developmental disabilities | ☐ Poor night vision and loss of peripheral vision | ☐ Heart problems |
| ☐ Liver disease and/or with elevated liver enzymes | ☐ Recurrent infections | ☐ Ataxia |
| ☐ Abnormal bleeding or blood clotting | ☐ Feeding problems | ☐ Slurred speech (dysarthria) |
| | | ☐ Other |

21. Concerning the doctors consulted, please tell us

How many doctors (approximately)...

(please select the most appropriate option)

RATING SCALE

1 - 2 3 - 5 6 - 10 11 - 20 > 20 I don't know

were consulted when you SUSPECTED the FIRST SIGNS AND SYMPTOMS of CDG?

☐ ☐ ☐ ☐ ☐ ☐

Did you consult between the FIRST MANIFESTATIONS and the FINAL CDG diagnosis?

☐ ☐ ☐ ☐ ☐ ☐

22. How much time (approximately) did it take to get a FINAL CDG diagnosis? (Please select the most appropriate option)

- | | | |
|-------------------------------------|---------------------------------------|---|
| ☐ < 3 months | ☐ 1 - 3 years old | ☐ 10 - 20 years old |
| ☐ 3 - 6 months | ☐ 4 - 5 years old | ☐ > 20 years old |
| ☐ 7 - 12 months | ☐ 6 - 9 years old | ☐ I don't know |

23. Was the person with CDG given any other diagnoses, before the FINAL diagnosis was made?

- ☐ Yes (Q24)
- ☐ No (Q25)
- ☐ I don't know (Q25)

24. Which diagnoses were given before the final diagnosis?

25. Do you believe that the CDG FINAL diagnosis could have been made earlier?

- ☐ Yes
- ☐ No
- ☐ I don't know

26. Concerning the medical specialties involved, please tell us the FIRST to raise the possibility of a CDG diagnosis

- | | | |
|--|--|--|
| ☐ Geneticist | ☐ Obstetrician | ☐ Gastroenterologist |
| ☐ Neurologist | ☐ General practitioner | ☐ Ophthalmologist |
| ☐ Paediatrician | ☐ Cardiologist | ☐ I don't know |
| ☐ Metabolic specialist | ☐ Endocrinologist | ☐ Other (please specify) |

27. Concerning the medical specialties involved, please tell us the **specialist who gave the FINAL diagnosis**

- | | | |
|--|--|--|
| ☐ Geneticist | ☐ Obstetrician | ☐ Gastroenterologist |
| ☐ Neurologist | ☐ General practitioner | ☐ Ophthalmologist |
| ☐ Paediatrician | ☐ Cardiologist | ☐ I don't know |
| ☐ Metabolic specialist | ☐ Endocrinologist | ☐ Other (please specify) |

28. Did you seek a second opinion to confirm the FINAL CDG diagnosis?

- ☐ Yes
- ☐ No
- ☐ I don't know

29. Were the diagnosis journeys of the 1st relative diagnosed with CDG and the other(s) very different? Please select all the aspects that differed:

- ☐ Age at which the diagnosis happened
- ☐ Presenting signs and symptoms
- ☐ Time of the diagnosis (how long it took to get a final diagnosis)
- ☐ The way you coped with the diagnosis (e.g. information needs, emotional impact)
- ☐ There were no major differences between the diagnosis
- ☐ Other (Please specify)

30. Could you elaborate and further explain what happened? (Free text)

THEME - ACCESS TO INFORMATION ABOUT CDG AT THE TIME OF DIAGNOSIS AND AT THE PRESENT

31. Which format of information about CDG was provided to you when you first got the CDG diagnosis? Please select all that apply.

- ☐ Printed material (e.g. brochures, leaflet)
- ☐ Referred to website
- ☐ Social media
- ☐ Journal Article
- ☐ Verbal
- ☐ Other

32. How do you and did you access information related to CDG, both at the time of diagnosis and present time?

TD P

- | | | |
|-----------------------|----------------------------------|----------------------------------|
| ☐ | ☒ | Physician or Nurse |
| ☐ | ☒ | Genetic Counsellor or Geneticist |
| ☐ | ☒ | Family Members |
| ☐ | ☒ | Patient Organizations |

TD P

- | | | |
|-----------------------|----------------------------------|-------------------------------------|
| ☐ | ☒ | Other individuals/families with CDG |
| ☐ | ☒ | Internet |
| ☐ | ☒ | Pharmaceutical Industry |
| ☐ | ☒ | Social Media |

TD P

- | | | |
|-----------------------|----------------------------------|--------------------------|
| ☐ | ☒ | I don't know |
| ☐ | ☒ | No information was given |
| ☐ | ☒ | Other |

Abbreviations:

- ☐ TD - Time of Diagnosis
☒ P - Present

THEME - SUPPORT FOR FAMILIES THROUGHOUT PATIENT GROUPS

33. Have you found an organisation or support group specific to CDG within your country?

- ☐ Yes
- ☐ No
- ☐ I don't know

34. How often do you use a certain patient organization as a source of information about CDG?

- ☐ Never
- ☐ Monthly
- ☐ Once or Twice per month
- ☐ Weekly
- ☐ Daily
- ☐ Other

35. How important is the role of patient organisations in providing information about CDG to affected people and families?

- ☐ Not very important ☐ Not important ☐ Important ☐ Very Important ☐ Essential

THEME - CDG-FOCUSED DIGITAL SOLUTIONS THAT MEET THE NEEDS OF THE PEOPLE WHO LIVE WITH CDG

36. Are you on Social media platforms (Facebook, Twitter, and others etc)?

- ☐ Yes (Q37)
- ☐ No (Q39)

37. Are you already familiar with any active social media groups focused on CDG that you consider as good examples?

- ☐ Yes
- ☐ No
- ☐ I don't know

38. What do you think social media can/could do for the CDG community? **(Please select all that apply)**

- | | |
|--|--|
| ☐ Raise CDG awareness | ☐ Get advice of fellow people who live with the same condition |
| ☐ Connect different stakeholders worldwide | ☐ Learn about the latest research news |
| ☐ Learn about clinical trials | ☐ Fundraise for CDG |
| ☐ Optimise Clinical trials (lowering its costs, time, etc) | ☐ To secure a rare disease patient perspective into broader online conversations |
| ☐ Help CDG families at the time of diagnosis | ☐ Other (please specify) |
| ☐ Creation of CDG worldwide organizations | |
| ☐ Broader CDG information | |
| ☐ Promote resources of interest for CDG community | |

THEME - SOCIODEMOGRAPHICS INFORMATION

39. What is your age?

- ☐ 18 - 24 years old
- ☐ 25 - 34 years old
- ☐ 35 - 44 years old
- ☐ 45 - 54 years old
- ☐ 55 - 65 years old
- ☐ Above

40. Are you :

- ☐ Male
- ☐ Female
- ☐ Other (Please Specify)

41. In which country do you live?

- ☐ Afghanistan
- ☐
- ☐ Zimbabwe

42. What is your highest qualification?

- ☐ Less than high school diploma
- ☐ High school diploma or equivalent degree
- ☐ Bachelor's degree
- ☐ Master's degree
- ☐ PhD
- ☐ Other (Please specify)

Thank you for participating in this survey !

Click on **DONE** - at the end of this page - to **FINISH**.

We would be very grateful if you could share a link to <https://worldcdg.org/research-cdg-journey-mapping/survey-2-cdg-experiences-over-time-families-and-professionals-views> on your Facebook and Twitter pages to allow your followers to join and take part in the surveys.

In accordance with the Data Protection laws, you can access, modify, or suppress your information at any time. If you want to exercise this right and obtain information about your data, please contact <https://worldcdg.org/contact>

ACKNOWLEDGMENTS

CDG & Allies – Professionals and Patient Associations International Network (CDG & Allies – PPAIN) deeply acknowledge the participation of the stakeholders that acted as advisors in this survey.

Thank you for taking time to participate in our questionnaire.

We truly value the information you have provided.

The information gained from this survey will be valuable in developing better health services and supports for your child, you and your family.

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SOURCES

1. Zurynski, Y., Deverell, M., Dalkeith, T. et al. (2017). Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays. Orphanet J Rare Dis 12, 68. <https://doi.org/10.1186/s13023-017-0622-4>

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5. Davies, W. (2016) Insights into rare diseases from social media surveys. *Orphanet J Rare Dis* 11, 151. Zurynski, Y., Deverell, M., Dalkeith, T. et al. (2017). Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays. *Orphanet J Rare Dis* 12, 68. <https://doi.org/10.1186/s13023-017-0622-4>