The GENROC Study: Improving the treatment of people with **GEN**etic **R**are disease: an **O**bservational **C**ohort study.

Working with families to increase understanding of growth and development in rare genetic conditions in order to improve clinical care.

Information leaflet for Parent/Carers

University of BRISTOL

We would like to invite you and your child to take part in a research study. It aims to improve the amount of information available about some genetic conditions in order to improve medical care, such as patterns about growth. Before you decide whether you would like to take part, it is important for you to understand why the study is being done and what it will involve. Please read this leaflet carefully. Ask us if there is anything you don't understand or if you want more information. Take the time to decide whether or not you want to take part. Thank you

Why are we doing this study?

- We don't have enough information available about how many genetic conditions affect children
- For example some children may be very small but doctors don't know whether this is related to their genetic condition - this means they don't know whether this needs treatment or not.
- Because of this many families will join social media or online groups and learn more about the conditions this way.

We want to improve everyone's understanding of these conditions by working better with families to gather information about the conditions and by using this information to help doctors to look after genetic conditions better.

This may in turn make it more possible to develop specific treatments in the future.

What are the benefits of taking part in this study?

By taking part you and your child:

- Will help doctors and scientists understand more about your child's rare genetic condition which will help us know the best way to care for your child.
- **We hope that this study will allow us to generate growth curves and other clinically helpful information that may benefit your child.** This sort of information is key to scientists being able to develop new treatments in the future.
- Will help us understand the best way of working with families, syndrome social media groups and getting the most of web based tools
- Will also be helping other families with these conditions now and in the future
- May also be invited to be included in other research studies that you wouldn't have known about otherwise.
- You may also learn a bit about how research studies work.

GENROC

What will I be asked to do if I take part?

First step:

- If you want to take part in the study, you will be asked to fill in an online form to show us that you agree to take part in the study.
- If you would like to hear more about the study, a member of the research team will arrange a time to talk to you over the telephone. They will explain the study to you and your child, and answer any questions you may have. They might ask you about how you are you feeling about the study to make sure you are happy to take part.

Second step:

- Once you and your child have agreed to take part and you have completed the online forms, you will be asked to complete some questions online about your child. This shouldn't take too long. We will contact your child's geneticist and ask them for medical information about your child as well.
- You may also decide to take part in a group in which you answer some questions from the research team. This will not be specifically about your child but more about how you get information about your child's condition and how this could be improved. We will ask for separate consent from you for this and will contact you separately about this.
- Later in the study, you will be asked to upload some further anonymised information about your child to an online portal (special secure website). You will be able to choose whether you want to upload photos of your child to the portal as well.
- We will use your child's NHS number to link to national datasets such as health and education (or in a combined data linkage consortium). More information about what this means is available via the QR code on the next page.

Why have I been asked to take part? You have been asked to take part because your child has a rare genetic condition that we would like to include in our study. **You do not have to take part** in this study. If you want to speak to your genetics doctor or the research team, they will explain more about the study, but this will not commit you to taking part.

If I agree to take part, can I change my mind? Yes. You can leave the study at any point. We will keep any data that we have collected up until the point that you leave the study. Leaving the study at any time will not affect the standard of medical care you will receive

Are there any risks from taking part?

For some people their genetic variant may be unique to them. As part of the study we will be uploading each participant's variant and some other information such as height, weight, gender and clinical features to the DECIPHER website which will allow us to generate growth and development curves (see further information section for a link to more about how we will be working with DECIPHER).In order to protect your child's anonymity no personal identifiers (eg name, date of birth, address) will be included or associated with the information on DECIPHER or in any publication.

If you choose to upload photos of your child to the portal we will use these for analysis. If the analysis shows something important we may then include the de-identified photos in a publication of our research findings. If we wish to publish a photo that would be identifiable (such as a face) we we will contact you to discuss further specific consent to do so.

How will we use information about your child?

How will we use your information?

We will need to use information from you and from your child's medical records and their doctor/geneticist for this research project.

This information will include you and your child's

- Initials
- NHS / CHI number
- Name
- contact details

People will use this information to do the research or to check your records to make sure that the research is being done properly. People who do not need to know who you are will not be able to see your name or contact details. Your data will have a code number instead.

We will keep all information about you safe and secure. You will put all your information on to the REDCAP system. This is a University of Bristol managed system.

Once we have finished the study, we will keep some of the data so we can check the results. We will write our reports in a way that no-one can work out that you took part in the study.

What are your choices about how your information is used?

- You can stop being part of the study at any time, without giving a reason, but we will keep information about you that we already have.
- If you choose to stop taking part in the study, we would like to continue collecting information about your health from central NHS records/ your geneticist. If you do not want this to happen, tell us and we will stop.
- We need to manage your records in specific ways for the research to be reliable. This means that we won't be able to let you see or change the data we hold about you.
- If you agree to take part in this study, you will have the option to take part in future research using your data saved from this study. [REDCap]

Where can you find out more about how your information is used?

You can find out more about how we use your information

- at www.hra.nhs.uk/information-about-patients/
- by asking one of the research team
- by sending an email to data- protection@bristol.ac.uk or
- by ringing us on 0117 3941824.

www.hra.nhs.uk/patientdataandresearch

GDPR and research | University Secretary's Office | University of Bristol

DECIPHER: Some of your child's non identifiable information will be put into the DECIPHER world-wide website at https://deciphergenomics.org. This will be information that is not identifiable, but it will contain your child's genetic variant which may be unique to your child/family. The website will be seen and used by doctors (especially clinical geneticists) and scientists (especially genomic scientists and molecular biologists). The DECIPHER website may be accessed by doctors and scientists and others working in countries outside the UK e.g. USA, Canada and Australia. This will be to help us, other researchers and geneticists, to understand your child's genetic condition better.

Data linkage: With your permission, we will also link the information you provide us with to your child's NHS medical record held by NHS Digital and your child's National Pupil Database records held by the Department for Education (DfE). If we are able to do so we will also link to other nationally held databases in a trusted research environment. This will help us understand your child's physical health needs and educational level without asking you to complete additional questionnaires. We will request access to your child's records for a time period spanning from their year of birth until two years after our study has ended. In order to link to your child's NHS Digital and DfE records we will send your child's personal information (such as name, date of birth, gender, postcode and NHS number or school name) to these agencies in order to access your child's record. The process will follow these agencies' secure encryption transfer protocols in order to maintain confidentiality.

Please scan this QR code for more details about DECIPHER, what data linkage means and how we will use the information.

You can also find the information on this weblink
https://bristolexppsych.eu.qualtrics.com/jfe/form/SV_1LlfuBheVLQILGe

What will happen when the study stops? Your participation in the study will not change the NHS care your child receives. Once you have completed the questionnaires you will continue to receive NHS care. After the study stops and you want to know the study results, let us know and we can send them to you.

What will happen if my child feels unwell during the study? If during the study you or your child start to feel unwell (e.g., if you feel anxious or depressed, or if you have a fever), you should contact your local care providers (e.g., your GP or paediatrician). The research team cannot provide treatment for medical problems you may have. If you do contact the research team about other concerns (e.g., feeling anxious or depressed), the research team will do their best to help. If they feel it's appropriate, they may pass the information on to your local care providers and try to inform you of other services which may help you. The research team may not be able to reply to your queries straight away. This is why you should always contact your GP if you have any concerns.

Does everybody involved in the study have the right police checks? Yes. Those who are working with people within the study have had police checks to make sure they are safe to work with children and adults.

What will happen to the results of the study? This study will give us information on how genetic conditions affect children and how we should care for them. It will help us use digital tools to improve care for rare conditions. We will share the results of the study to help people in the future get the best possible treatment for their condition.

Who is organising and financially supporting the study? This research is organised by Dr Karen Low who is a clinical geneticist in Bristol and a doctoral research fellow at the University of Bristol. The study is funded by the NIHR and is sponsored by the University of Bristol.

What should I do if I have a problem with the study? If you have any problems with this study, please speak to Dr Karen Low, or any member of the clinical team that you know. Dr Low's contact information can be found below. You can also talk to the NHS in the usual way via the Patient Advice and Liaison Services (PALS) in your hospital.

Ethical approval Ethical approval means that we have received a favourable opinion from the NHS Research Ethics Committee. An independent group of people reviewed our study and agreed it was ethical.

Who can I contact for more information?

https://redcap.link/GENROC

Email the study team on genroc-study@bristol.ac.uk

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*"**Unique is delighted to support the GENROC study** which is harnessing the expertise of parents and carers of children with rare genetic conditions to help guide improvements in clinical care."*

Sarah Wynn PhD, Chief Executive Officer, Unique

Understanding Chromosome & Gene Disorders
www.rarechromo.org