ERGO No: 45724
IRAS No: 217804, 265580

The PURA Global Network. Understanding PURA Syndrome. PURA Syndrome Longitudinal Natural History Study.

Study Information Sheet

We would like to invite you to consider giving your permission for your ward/relative/child/person to take part in this research study. Before you decide, it is important for you to understand why the research is being done and what it will involve. This information leaflet is for the person who is able to provide consent or an opinion on behalf of the study participant, when the study participant is a child or an adult who lacks the ability to decide whether to be involved in the study (adults lacking/without capacity).

Please take time to read the following information carefully, and discuss it with others if you wish. Please contact the study administrative team at PURA@soton.ac.uk if there is anything that is not clear or if you would like more information. We would ask that when making this decision you put aside your own views, feelings and wishes about the research and to consider and take into account, what the past and present feelings and wishes of the study participant would have been, had they been able to consent on their own behalf.

What is the purpose of the study?

The aim of this study is to get a better understanding of PURA syndrome and how it affects people over time. We have written a number of medical questionnaires that will collect medical information about the study participant from yourself and their doctor. We intend to collect this information at regular annual intervals. Information collected in questionnaires will be used to learn more about PURA syndrome internationally.
This represents the first ever long-term study of PURA syndrome.

Why is my ward/relative/child eligible for this study?

They are eligible for this research study if they have had genetic testing which confirms PURA syndrome, a 5q31.3 deletion including the PURA gene, or a 5q31.3 duplication including the PURA gene.
We can only process this data if we have a genetic report or letter from a clinician confirming the result. Study participants that live in a Country subject to UK sanctions, a Country on the USA

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State Sponsors of Terrorism list, or are 16 years and over and live in Northern Ireland, are not eligible for inclusion. If you are unsure of whether they are eligible for inclusion, you can email the study administrative team for advice (PURA@soton.ac.uk).

What if my ward/relative/child has died?

If your ward/relative/child had PURA Syndrome or PURA gene duplication or deletion and has died, you can still access the study. When you contact the study administrative team to register for the study, please notify them that your ward/relative/child has died. If your ward/relative/child passes away during the study, please contact the study administrative team at PURA@soton.ac.uk or by accessing the study withdrawal document on the study system.

Who can make the decision for my ward/relative/child to be involved in the study?

From our understanding of PURA Syndrome, children and adults with PURA Syndrome would be unable to make the decision of whether to be involved in this research alone. We ask that you make this decision on their behalf, taking into account their wishes.

If the study participant is a child (under 16 years of age), we ask that one of the parents or legal guardians/custodians of the study participant provide consent. If the parents are divorced or separated, we ask that the main custodian provide consent.

If the study participant is aged 16 years or over, and lives In England or Wales, we ask that a "personal consultee" provides an opinion based on what they know of the study participants interests, wishes and feelings. A personal consultee is someone who cares for, and is interested in the welfare of the study participant other than in a professional capacity. If you are unsure about taking the role of the consultee, you may want to contact the study administrative team or seek independent advice.

If the study participant is aged 16 or over and lives in any Country other than England or Wales, consent will be attained from their legal representative and if not appointed their nearest relative (as defined by the Mental Health (Scotland) Act 1984 relationship hierarchy).

If you initially provide consent for your child whilst under 16 years of age and they have their 16th birthday during the duration of the study, the appropriate consent/personal consultee opinion (as detailed above) will be required to continue study involvement at 16 years of age. If you have any questions regarding this, please contact the study administrative team.

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Does my ward/relative/child have to take part?

No. Participation is entirely optional. We are asking you to make a decision on their behalf.

How do I withdraw my ward/relative/child from the study?

You can withdraw them from the study at any time without providing a reason or affecting the standard of care they will receive. Data that has been submitted up to that point will be included in the research study, however we will not request any further information. You can withdraw them from the study by emailing the study administrative team at PURA@soton.ac.uk or by accessing the study withdrawal document on the study system. On withdrawal, no further information will be able to be added to the study by yourself or their clinician and the account will be closed. We would ask that you also directly update the clinician about withdrawal.

What will happen to my ward/relative/child during the study?

If you decide to take part, you will need to email the study administrative team at PURA@soton.ac.uk. When emailing the study administrative team, you will need to include the name and date of birth, Country of residence of the study participant, and your contact details. The email that you send this information from, is the email the study administrative team will use to contact you. The study administrative team will confirm that the study participant is eligible for the study and then create a study account for them. You will be emailed details on how to access the study website and set a password. Prior to accessing the questionnaires, you will need to sign a consent or personal consultee declaration form (depending on the age and location of the study participant). Over time we will add more questionnaires. We will email you when more questionnaires become available and annually to update the existing questionnaires.

How does my ward/relative/child's clinician get involved?

It may be that the clinician of your ward/relative/child tells you about the study and asks you to provide consent or an opinion on behalf of the study participant. This will allow the clinician to collect information from your ward/relative/childs medical records for this research study. In this case, you can also get involved and also enter information about your ward/relative/child, as explained above.

However in most cases, your clinician will enter the study because you have recommended them. When you access the study we will ask that you provide us with your clinician's work contact details so we can contact them to see if they would be interested in being involved in the study.

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We request that you ask permission from your clinician prior to doing this. For us to be able to link your ward/relative/child's study account with their clinicians account, you will need to give your clinician the study ID so that they can send this to the study administrative team. This ID can be found when you log into your ward/relative/child's account. You will be able to see what your clinician enters and they will be able to see what you enter.

What information will be collected?

We will ask you to provide us with some basic demographic details and your contact details. We will also ask that you upload evidence of the genetic result, this may be the actual test result or a clinicians letter confirming the genetic result. The questionnaires will cover the birth, development and medical history (including puberty). In time, we may ask you to upload photographs or medical reports. We ask that the information you provide is as complete and accurate as possible. We are collecting information that is considered 'sensitive data' or 'special category data'. This includes information such as ethnicity, genetic data and health data. This type of data requires researchers to take additional care in its collection, storage and use.

What is meant by personal information?

We are collecting personally identifiable information. This is any information capable of identifying the study participant. This includes information that directly identifies them, such as their name. It also includes information that can indirectly identify them, for example this is information that when combined with other data, could identify them. For example, a study collecting information on epilepsy may allow them to be directly identified when combined with other information such as age or Country of birth. All research where someone can be identified will be conducted in accordance with data protection laws. We will provide researchers with information about the genetic mutations and medical information collected from the online questionnaires, however we will not provide them with your child's name, address or contact details. The University of Southampton will keep directly identifiable information about your ward/child/relative until the study has ended.

What are the possible benefits of taking part?

We hope to gain a better understanding of PURA syndrome and its course. It is hoped that an improved understanding of PURA syndrome will help in establishing effective management guidelines for clinicians and families.

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What are the possible disadvantages and risks of taking part?

The study does not come at any physical risk to your ward/relative/child. It is important to understand that there may be no direct benefit from this research to your ward/relative/child or their family.

Because PURA syndrome is so rare, there is a risk that a small amount of information may allow you and people who know your ward/relative well, to identify them from the research papers, information on the Foundation website or PURA Syndrome Foundation conference lectures. Every effort will be made to keep this information as private as possible.

How will the information be kept confidential and safe?

All participant details will remain confidential and protected, in compliance with General Data Protection Regulation 2016/679, Data Protection Act 2018 and the University of Southampton data management policy in each case as amended and applicable from time to time. The University of Southampton is the data controller, and is responsible for safely collecting the information, looking after it and using it properly. All personal information is handled according to the University's policies and legal and regulatory requirements. Data protection laws require that personal data is; used lawfully, fairly and in a transparent way, collected only for valid purposes that have clearly been explained to you, proportionate and relevant to the purposes you have been told about, accurate and up to date, kept only as long as necessary and kept securely.

The questionnaires are held on a secure website and the data stored on a secure server, currently in the Netherlands. When an account is created, the study participant is given a 'study ID'. All of the information you enter into the questionnaires is entered under this ID and stored separately to your child's names, genetic reports and contact details. This is called 'pseudonymisation'. We would ask that you do not enter identifying information (such as their name) into the questionnaire answers.

Who will have access to the study information?

The study administrative team will have access to all the information that you enter. This allows them to identify study participants and contact you. The study administrative team may contact you about how the data is being used, provide information about other research opportunities, to notify of additional questionnaires and provide annual questionnaire update reminders. In some occasions, the study administrative team may enter clinical records onto the system on behalf of your clinician or look at the medical records to check the accuracy of the research study.

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Responsible members of the University of Southampton, the data system staff (currently FormsVision) or individuals from regulatory authorities may be given access to monitor and uphold data management regulations.

The University have to ensure that it is in the public interest when collecting and using personally identifiable information. This means that when you agree for the study participant to take part in this research study, we will use the data in the ways needed to conduct and analyse the research study. The data collected will be shared with researchers globally to learn more about PURA syndrome. All researchers will have to apply to the PURA Syndrome Study Advisory Steering Committee for access to the data. Researchers from inside and outside the European Economic Area (EEA) may apply for access to the data. Researchers outside the EEA may be governed by different data laws. Before providing researchers with data, we will ask that they agree to certain data handling, storage and use standards. Researchers wishing to learn from this data will only have access to exported pseudonymised data. They will not receive your contact details or ward/relative/childs name. Outcomes of this research may be shared in reports, publications and conferences.

You can also choose whether we are able to provide your ward/relative/childs name, date of birth and your contact details to the PURA Syndrome Foundation. This information will be stored securely and may be used to update you about Foundation news and events. The Foundation is a global non-profit organization based outside of the European Economic Area, in the United States of America. They provide families with support, educational resources and access to information about the latest medical research on PURA syndrome.

As the data controller, the University of Southampton, may also have to disclose information if required so by law in order to comply with legal obligation, to protect University rights, interests or property, to act in urgent circumstances to protect the personal safety of University staff, students and public, or protect the University against any legal liability.

What happens when the research study finishes?

The length of the study will be regularly reviewed and the end date decided by the PURA Syndrome Foundation. There is currently no definite end date for this study, although you are free to withdraw at any time without giving any reason. The length that data is kept after the study is closed and the use of this data will be in accordance with General Data Protection Regulation 2016/679, Data Protection Act 2018 and the University of Southampton data management policy.

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Who is running the study?

The study administrative team are based at the University of Southampton and University Hospital Southampton NHS Trust. The University of Southampton, United Kingdom, is sponsoring the study. Professor Diana Baralle is the Chief Investigator of the study and data custodian.

Who is funding the study?

The PURA Syndrome Foundation - https://www.purasyndrome.org/

What is the legal basis for this study?

The legal basis for the collecting and processing of this information is to perform a task in the public interest. This means that we have to demonstrate that our research serves the interests of society as a whole. We do this by following the UK Policy Framework for Health and Social Care Research. In this study, the aim would be that your ward/relative/child, their family and their clinician may benefit from the information collected. The University of Southampton endeavours to maintain the highest of standards in the research they conduct. Researchers affiliated to the University of Southampton must comply with the University codes of conduct, policies and procedures to ensure the research complies with University and national regulations and legislation.

What are my rights?

The GDPR and DPA gives you and your ward/relative rights regarding the data entered. This includes, but is not limited to, how you access the information, correction of inaccurate information and withdrawal/erasure of information. These rights may be limited in order for the research to be reliable and accurate.

If you withdraw we may not be able to remove information already obtained. If information has been completely de-identified, it will not be possible to implement specific rights about information.

If you have any questions about how your information is being used or data protection rights requests, please contact the study administrative team at the University of Southampton at PURA@soton.ac.uk.

If you have any concerns regarding the response from the study administrative team or wish to exercise further rights or make a complaint, you can also access the University of Southampton data protection webpage request form at www.southampton.ac.uk/about/governance/subject-

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access request form.page or contact the University's Data protection officer at data.protection@soton.ac.uk.

If you are unhappy with the way your personal information is handled or with the response received from the University of Southampton, you have the right to lodge a complaint with the information commissioners office at Wycliffe House, Water Lane, Wilmslow, SK9 5AF https://ico.org.uk/ The University registration number with this office is Z6801020.

Contacts for further information:

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